Tribro-Omics gene panel catalogue

200 panels · 1872 unique genes

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CARDIOLOGY 21 GENES

Hypertrophic cardiomyopathy

Sarcomeric and phenocopy genes for unexplained left ventricular hypertrophy

Panel details

Panel code	TBO-GP-001
Clinical area	Cardiology
Number of genes	21
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACTC1***	AD	OMIM:612098, OMIM:612794, OMIM:613424	
2	***ACTN2***	AD	OMIM:612158, OMIM:618654, OMIM:618655	
3	***ALPK3***	AR	OMIM:618052	
4	***CSRP3***	AD	OMIM:607482, OMIM:612124	
5	***FHL1***	XLR, XLD	OMIM:300280, OMIM:300695, OMIM:300696 +2	
6	***FHOD3***	AD	OMIM:619402	
7	***FLNC***	AD	OMIM:609524, OMIM:614065, OMIM:617047	
8	***GLA***	XLR	OMIM:301500	
9	***JPH2***	AD, AR	OMIM:613873, OMIM:619492	
10	***LAMP2***	XLD	OMIM:300257	
11	***MYBPC3***	AD, AR	OMIM:115197, OMIM:615396	
12	***MYH7***	AD, AR	OMIM:160500, OMIM:192600, OMIM:255160 +2	
13	***MYL2***	AD, AR	OMIM:608758, OMIM:619424	
14	***MYL3***	AD, AR	OMIM:608751	
15	***PLN***	AD	OMIM:609909, OMIM:613874	

16	***PRKAG2***	AD	OMIM:194200, OMIM:261740, OMIM:600858
17	***TNNC1***	AD	OMIM:611879, OMIM:613243
18	***TNNI3***	AD, AR	OMIM:115210, OMIM:611880, OMIM:613286 +1
19	***TNNT2***	AD	OMIM:115195, OMIM:601494, OMIM:612422
20	***TPM1***	AD	OMIM:115196, OMIM:611878
21	***TTR***	AD	OMIM:105210, OMIM:115430, OMIM:145680

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY 21 GENES

Dilated cardiomyopathy

Genes for idiopathic and familial dilated cardiomyopathy

Panel details

Panel code	TBO-GP-002
Clinical area	Cardiology
Number of genes	21
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACTC1***	AD	OMIM:612098, OMIM:612794, OMIM:613424	
2	***ACTN2***	AD	OMIM:612158, OMIM:618654, OMIM:618655	
3	***BAG3***	AD	OMIM:612954, OMIM:613881	
4	***DES***	AD, AR	OMIM:181400, OMIM:601419, OMIM:604765	
5	***DSP***	AD, AR	OMIM:605676, OMIM:607450, OMIM:609638 +2	
6	***FLNC***	AD	OMIM:609524, OMIM:614065, OMIM:617047	
7	***JPH2***	AD, AR	OMIM:613873, OMIM:619492	
8	***LDB3***	AD	OMIM:601493, OMIM:609452	
9	***LMNA***	AD, AR	OMIM:115200, OMIM:151660, OMIM:176670 +8	
10	***MYBPC3***	AD, AR	OMIM:115197, OMIM:615396	
11	***MYH7***	AD, AR	OMIM:160500, OMIM:192600, OMIM:255160 +2	

12	***NEXN***	AD	OMIM:613122, OMIM:613876	
13	***PLN***	AD	OMIM:609909, OMIM:613874	
14	***RBM20***	AD	OMIM:613172	
15	***SCN5A***	AD, AR	OMIM:113900, OMIM:272120, OMIM:601144 +5	
16	***TNNC1***	AD	OMIM:611879, OMIM:613243	
17	***TNNI3***	AD, AR	OMIM:115210, OMIM:611880, OMIM:613286 +1	
18	***TNNT2***	AD	OMIM:115195, OMIM:601494, OMIM:612422	
19	***TPM1***	AD	OMIM:115196, OMIM:611878	
20	***TTN***	AD, AR	OMIM:600334, OMIM:603689, OMIM:604145 +3	Large gene; truncating variants in A-band most relevant
21	***VCL***	AD	OMIM:611407, OMIM:613255	

Genes needing specific methods

- ***TTN***: Large gene; truncating variants in A-band most relevant

Notes

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- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY 11 GENES

Arrhythmogenic cardiomyopathy

Desmosomal and non-desmosomal genes for arrhythmogenic right and left ventricular cardiomyopathy

Panel details

Panel code	TBO-GP-003
Clinical area	Cardiology
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CDH2***	AD, AR	OMIM:618920, OMIM:618929, OMIM:619957	
2	***DES***	AD, AR	OMIM:181400, OMIM:601419, OMIM:604765	
3	***DSC2***	AD, AR	OMIM:610476	
4	***DSG2***	AD, AR	OMIM:610193, OMIM:612877	
5	***DSP***	AD, AR	OMIM:605676, OMIM:607450, OMIM:609638 +2	
6	***FLNC***	AD	OMIM:609524, OMIM:614065, OMIM:617047	
7	***JUP***	AD, AR	OMIM:601214, OMIM:611528	
8	***LMNA***	AD, AR	OMIM:115200, OMIM:151660, OMIM:176670 +8	
9	***PKP2***	AD	OMIM:609040	
10	***PLN***	AD	OMIM:609909, OMIM:613874	
11	***TMEM43***	AD	OMIM:604400, OMIM:614302, OMIM:619832	

Notes

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- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY 11 GENES

Left ventricular non-compaction

Genes associated with left ventricular non-compaction, including Barth syndrome

Panel details

Panel code	TBO-GP-004
Clinical area	Cardiology
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACTC1***	AD	OMIM:612098, OMIM:612794, OMIM:613424	
2	***HCN4***	AD	OMIM:163800, OMIM:613123, OMIM:619521	
3	***LDB3***	AD	OMIM:601493, OMIM:609452	
4	***MIB1***	AD	OMIM:615092	
5	***MYBPC3***	AD, AR	OMIM:115197, OMIM:615396	
6	***MYH7***	AD, AR	OMIM:160500, OMIM:192600, OMIM:255160 +2	
7	***NKX2-5***	AD, AR	OMIM:108900, OMIM:187500, OMIM:217095 +3	
8	***PRDM16***	AD	OMIM:615373	
9	***TAFAZZIN***	XLR	OMIM:302060	
10	***TNNT2***	AD	OMIM:115195, OMIM:601494, OMIM:612422	
11	***TPM1***	AD	OMIM:115196, OMIM:611878	

Notes

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- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY · 11 GENES

Restrictive cardiomyopathy

Genes for restrictive cardiomyopathy and infiltrative phenocopies

Panel details

Panel code	TBO-GP-005
Clinical area	Cardiology
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACTC1***	AD	OMIM:612098, OMIM:612794, OMIM:613424	
2	***BAG3***	AD	OMIM:612954, OMIM:613881	
3	***DES***	AD, AR	OMIM:181400, OMIM:601419, OMIM:604765	
4	***FLNC***	AD	OMIM:609524, OMIM:614065, OMIM:617047	
5	***GLA***	XLR	OMIM:301500	
6	***MYH7***	AD, AR	OMIM:160500, OMIM:192600, OMIM:255160 +2	
7	***MYL2***	AD, AR	OMIM:608758, OMIM:619424	
8	***MYL3***	AD, AR	OMIM:608751	
9	***TNNI3***	AD, AR	OMIM:115210, OMIM:611880, OMIM:613286 +1	
10	***TNNT2***	AD	OMIM:115195, OMIM:601494, OMIM:612422	
11	***TTR***	AD	OMIM:105210, OMIM:115430, OMIM:145680	

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY 10 GENES

Long QT syndrome

Genes with established evidence for congenital long QT syndrome

Panel details

Panel code	TBO-GP-006
Clinical area	Cardiology
Number of genes	10
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CACNA1C***	AD	OMIM:601005, OMIM:611875, OMIM:618447 +1	
2	***CALM1***	AD	OMIM:614916, OMIM:616247	
3	***CALM2***	AD	OMIM:616249	
4	***CALM3***	AD	OMIM:618782	
5	***KCNE1***	AD, AR	OMIM:612347, OMIM:613695	
6	***KCNH2***	AD	OMIM:609620, OMIM:613688	
7	***KCNJ2***	AD	OMIM:170390, OMIM:609622, OMIM:613980	
8	***KCNQ1***	AD, AR	OMIM:130650, OMIM:192500, OMIM:220400 +2	
9	***SCN5A***	AD, AR	OMIM:113900, OMIM:272120, OMIM:601144 +5	
10	***TRDN***	AD, AR	OMIM:604772, OMIM:615441	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito

mitochondrial.

- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY · 6 GENES

Brugada syndrome

SCN5A and genes reported in Brugada syndrome

Panel details

Panel code	TBO-GP-007
Clinical area	Cardiology
Number of genes	6
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CACNA1C***	AD	OMIM:601005, OMIM:611875, OMIM:618447 +1	
2	***CACNB2***	AD	OMIM:611876	
3	***GPD1L***	AD	OMIM:611777	
4	***KCND3***	AD	OMIM:607346, OMIM:616399	
5	***SCN1B***	AD, AR	OMIM:604233, OMIM:612838, OMIM:615377 +1	
6	***SCN5A***	AD, AR	OMIM:113900, OMIM:272120, OMIM:601144 +5	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY · 8 GENES

Catecholaminergic polymorphic ventricular tachycardia

Genes for CPVT and related calcium-handling arrhythmias

Panel details

Panel code	TBO-GP-008
Clinical area	Cardiology
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CALM1***	AD	OMIM:614916, OMIM:616247	
2	***CALM2***	AD	OMIM:616249	
3	***CALM3***	AD	OMIM:618782	
4	***CASQ2***	AD, AR	OMIM:604772, OMIM:611938	
5	***KCNJ2***	AD	OMIM:170390, OMIM:609622, OMIM:613980	
6	***RYR2***	AD	OMIM:115000, OMIM:604772	
7	***TECRL***	AR	OMIM:614021	
8	***TRDN***	AD, AR	OMIM:604772, OMIM:615441	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY · 4 GENES

Short QT syndrome

Genes for congenital short QT syndrome

Panel details

Panel code	TBO-GP-009
Clinical area	Cardiology
Number of genes	4
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***KCNH2***	AD	`OMIM:609620, OMIM:613688`	
2	***KCNJ2***	AD	`OMIM:170390, OMIM:609622, OMIM:613980`	
3	***KCNQ1***	AD, AR	`OMIM:130650, OMIM:192500, OMIM:220400 +2`	
4	***SLC4A3***	AD	`OMIM:620231`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY · 13 GENES

Thoracic aortic aneurysm and dissection

Genes for heritable thoracic aortic disease

Panel details

Panel code	TBO-GP-010
Clinical area	Cardiology
Number of genes	13
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACTA2***	AD	OMIM:611788, OMIM:613834, OMIM:614042	
2	***COL3A1***	AD, AR	OMIM:130050, OMIM:618343	
3	***FBN1***	AD	OMIM:102370, OMIM:129600, OMIM:154700 +5	
4	***LOX***	AD	OMIM:617168	
5	***MYH11***	AD, AR	OMIM:132900, OMIM:619350, OMIM:619351	
6	***MYLK***	AD, AR	OMIM:249210, OMIM:613780	
7	***PRKG1***	AD	OMIM:615436	
8	***SMAD2***	AD, AR	OMIM:619656, OMIM:619657	
9	***SMAD3***	AD	OMIM:613795	
10	***TGFB2***	AD	OMIM:614816	
11	***TGFB3***	AD	OMIM:107970, OMIM:615582	
12	***TGFBR1***	AD	OMIM:132800, OMIM:609192	
13	***TGFBR2***	AD	OMIM:133239, OMIM:610168, OMIM:614331	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY | 8 GENES

Marfan and Loeys-Dietz syndromes

Fibrillinopathies and TGF-beta signalling aortopathies

Panel details

Panel code	TBO-GP-011
Clinical area	Cardiology
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***FBN1***	AD	OMIM:102370, OMIM:129600, OMIM:154700 +5	
2	***FBN2***	AD	OMIM:121050, OMIM:616118	
3	***SKI***	AD	OMIM:182212	
4	***SMAD3***	AD	OMIM:613795	
5	***TGFB2***	AD	OMIM:614816	
6	***TGFB3***	AD	OMIM:107970, OMIM:615582	
7	***TGFBR1***	AD	OMIM:132800, OMIM:609192	
8	***TGFBR2***	AD	OMIM:133239, OMIM:610168, OMIM:614331	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY 20 GENES

Ehlers-Danlos syndromes

Genes for classical, vascular, kyphoscoliotic and rarer EDS types

Panel details

Panel code	TBO-GP-012
Clinical area	Cardiology
Number of genes	20
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ADAMTS2*	AR	OMIM:225410	
2	*AEBP1*	AR	OMIM:618000	
3	*B3GALT6*	AR	OMIM:271640, OMIM:609465, OMIM:615349	
4	*B4GALT7*	AR	OMIM:130070	
5	*C1R*	AD	OMIM:130080	
6	*C1S*	AD	OMIM:613783, OMIM:617174	
7	*CHST14*	AR	OMIM:601776	
8	*COL12A1*	AD, AR	OMIM:616470, OMIM:616471	
9	*COL1A1*	AD	OMIM:114000, OMIM:130060, OMIM:166200 +5	
10	*COL1A2*	AD, AR	OMIM:166210, OMIM:166220, OMIM:166710 +4	
11	*COL3A1*	AD, AR	OMIM:130050, OMIM:618343	
12	*COL5A1*	AD	OMIM:130000, OMIM:619329	
13	*COL5A2*	AD	OMIM:130010	
14	*DSE*	AR	OMIM:615539	
15	*FKBP14*	AR	OMIM:614557	

16	***PLOD1***	AR	OMIM:225400
17	***PRDM5***	AR	OMIM:614170
18	***SLC39A13***	AR	OMIM:612350
19	***TNXB***	AD, AR	OMIM:606408, OMIM:615963
20	***ZNF469***	AR	OMIM:229200

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY 5 GENES

Familial hypercholesterolaemia

Genes for monogenic familial hypercholesterolaemia

Panel details

Panel code	TBO-GP-013
Clinical area	Cardiology
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***APOB***	AD, AR	`OMIM:144010, OMIM:615558`	
2	***APOE***	AD, AR	`OMIM:104310, OMIM:269600, OMIM:603075 +3`	
3	***LDLR***	AD, AR	`OMIM:143890`	
4	***LDLRAP1***	AR	`OMIM:603813`	
5	***PCSK9***	AD	`OMIM:603776`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY | 5 GENES

Familial chylomicronaemia

Genes for severe monogenic hypertriglyceridaemia

Panel details

Panel code	TBO-GP-014
Clinical area	Cardiology
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***APOA5***	AD	`OMIM:144650, OMIM:145750`	
2	***APOC2***	AR	`OMIM:207750`	
3	***GPIHBP1***	AR	`OMIM:615947`	
4	***LMF1***	AR	`OMIM:246650`	
5	***LPL***	AD, AR	`OMIM:144250, OMIM:238600`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY · 11 GENES

Pulmonary arterial hypertension

Genes for heritable pulmonary arterial hypertension and veno-occlusive disease

Panel details

Panel code	TBO-GP-015
Clinical area	Cardiology
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACVRL1***	AD	OMIM:600376	
2	***ATP13A3***	AR	OMIM:265400	
3	***BMPR2***	AD	OMIM:178600, OMIM:265450	
4	***CAV1***	AD, AR	OMIM:606721, OMIM:612526, OMIM:615343	
5	***EIF2AK4***	AR	OMIM:234810	
6	***ENG***	AD	OMIM:187300	
7	***GDF2***	AD	OMIM:615506	
8	***KCNK3***	AD	OMIM:615344	
9	***SMAD9***	AD	OMIM:615342	
10	***SOX17***	AD	OMIM:613674	
11	***TBX4***	AD, AR	OMIM:147891, OMIM:601360	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY · 13 GENES

Congenital heart disease

Genes for syndromic and non-syndromic structural heart defects

Panel details

Panel code	TBO-GP-016
Clinical area	Cardiology
Number of genes	13
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*CHD7*	AD	OMIM:214800, OMIM:612370	
2	*ELN*	AD	OMIM:123700, OMIM:185500, OMIM:194050	
3	*GATA4*	AD	OMIM:187500, OMIM:607941, OMIM:614429 +2	
4	*GATA6*	AD, AR	OMIM:187500, OMIM:217095, OMIM:600001 +2	
5	*JAG1*	AD	OMIM:118450, OMIM:187500, OMIM:617992 +1	
6	*MYH6*	AD	OMIM:192600, OMIM:613251, OMIM:613252 +2	
7	*NKX2-5*	AD, AR	OMIM:108900, OMIM:187500, OMIM:217095 +3	
8	*NOTCH1*	AD	OMIM:109730, OMIM:616028	
9	*NR2F2*	AD	OMIM:615779, OMIM:618901	
10	*TBX1*	AD, AR	OMIM:187500, OMIM:188400, OMIM:192430 +1	
11	*TBX20*	AD	OMIM:611363	
12	*TBX5*	AD	OMIM:142900	
13	*ZFPM2*	AD	OMIM:187500, OMIM:610187, OMIM:616067	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY 19 GENES

Noonan syndrome and RASopathies

RAS-MAPK pathway genes for Noonan and related syndromes

Panel details

Panel code	TBO-GP-017
Clinical area	Cardiology
Number of genes	19
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*BRAF*	AD	OMIM:114500, OMIM:115150, OMIM:155600 +4	
2	*CBL*	AD	OMIM:607785, OMIM:613563	
3	*HRAS*	AD	OMIM:109800, OMIM:137550, OMIM:162900 +3	
4	*KRAS*	AD	OMIM:108010, OMIM:109800, OMIM:114480 +9	
5	*LZTR1*	AD, AR	OMIM:605275, OMIM:615670, OMIM:616564	
6	*MAP2K1*	AD	OMIM:155950, OMIM:163950, OMIM:615279	
7	*MAP2K2*	AD	OMIM:615280	
8	*MRAS*	AD	OMIM:618499	
9	*NF1*	AD	OMIM:162200, OMIM:162210, OMIM:193520 +2	
10	*NRAS*	AD	OMIM:114500, OMIM:137550, OMIM:162900 +5	
11	*PPP1CB*	AD	OMIM:617506	
12	*PTPN11*	AD	OMIM:151100, OMIM:156250, OMIM:163950 +1	
13	*RAF1*	AD	OMIM:611553, OMIM:611554, OMIM:615916	
14	*RIT1*	AD	OMIM:615355	
15	*RRAS2*	AD	OMIM:618624	

16	***SHOC2***	AD	OMIM:607721
17	***SOS1***	AD	OMIM:135300, OMIM:610733
18	***SOS2***	AD	OMIM:616559
19	***SPRED1***	AD	OMIM:611431

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY | 5 GENES

Hereditary haemorrhagic telangiectasia

Genes for HHT and related vascular disorders

Panel details

Panel code	TBO-GP-018
Clinical area	Cardiology
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACVRL1***	AD	OMIM:600376	
2	***ENG***	AD	OMIM:187300	
3	***GDF2***	AD	OMIM:615506	
4	***RASA1***	AD	OMIM:605462, OMIM:608354	
5	***SMAD4***	AD	OMIM:139210, OMIM:174900, OMIM:175050 +1	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY | 6 GENES

Hereditary cardiac amyloidosis

Genes for inherited systemic amyloidoses with cardiac involvement

Panel details

Panel code	TBO-GP-019
Clinical area	Cardiology
Number of genes	6
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***APOA1***	AD, AR	`OMIM:618463, OMIM:619836, OMIM:620657`	
2	***B2M***	AD, AR	`OMIM:241600, OMIM:620659`	
3	***FGA***	AD, AR	`OMIM:105200, OMIM:202400`	
4	***GSN***	AD	`OMIM:105120`	
5	***LYZ***	AD	`OMIM:620658`	
6	***TTR***	AD	`OMIM:105210, OMIM:115430, OMIM:145680`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CARDIOLOGY 20 GENES

Sudden cardiac death

Combined cardiomyopathy and channelopathy genes for unexplained sudden cardiac death

Panel details

Panel code	TBO-GP-020
Clinical area	Cardiology
Number of genes	20
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CALM1***	AD	OMIM:614916, OMIM:616247	
2	***CALM2***	AD	OMIM:616249	
3	***CALM3***	AD	OMIM:618782	
4	***CASQ2***	AD, AR	OMIM:604772, OMIM:611938	
5	***DSG2***	AD, AR	OMIM:610193, OMIM:612877	
6	***DSP***	AD, AR	OMIM:605676, OMIM:607450, OMIM:609638 +2	
7	***FLNC***	AD	OMIM:609524, OMIM:614065, OMIM:617047	
8	***KCNH2***	AD	OMIM:609620, OMIM:613688	
9	***KCNQ1***	AD, AR	OMIM:130650, OMIM:192500, OMIM:220400 +2	
10	***LMNA***	AD, AR	OMIM:115200, OMIM:151660, OMIM:176670 +8	
11	***MYBPC3***	AD, AR	OMIM:115197, OMIM:615396	
12	***MYH7***	AD, AR	OMIM:160500, OMIM:192600, OMIM:255160 +2	

13	***PKP2***	AD	OMIM:609040	
14	***PLN***	AD	OMIM:609909, OMIM:613874	
15	***RBM20***	AD	OMIM:613172	
16	***RYR2***	AD	OMIM:115000, OMIM:604772	
17	***SCN5A***	AD, AR	OMIM:113900, OMIM:272120, OMIM:601144 +5	
18	***TNNI3***	AD, AR	OMIM:115210, OMIM:611880, OMIM:613286 +1	
19	***TNNT2***	AD	OMIM:115195, OMIM:601494, OMIM:612422	
20	***TTN***	AD, AR	OMIM:600334, OMIM:603689, OMIM:604145 +3	Large gene; truncating variants in A-band most relevant

Genes needing specific methods

- ***TTN***: Large gene; truncating variants in A-band most relevant

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 13 GENES

Hereditary breast and ovarian cancer

High- and moderate-penetrance breast and ovarian cancer genes

Panel details

Panel code	TBO-GP-021
Clinical area	Cancer predisposition
Number of genes	13
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ATM***	AD, AR	OMIM:114480, OMIM:208900	
2	***BARD1***	AD	OMIM:114480	
3	***BRCA1***	AD, AR	OMIM:114480, OMIM:604370, OMIM:617883	
4	***BRCA2***	AD, AR	OMIM:114480, OMIM:155255, OMIM:176807 +5	
5	***BRIP1***	AD, AR	OMIM:114480, OMIM:609054	
6	***CDH1***	AD	OMIM:114480, OMIM:119580, OMIM:137215 +2	
7	***CHEK2***	AD	OMIM:176807, OMIM:259500, OMIM:609265	
8	***PALB2***	AD, AR	OMIM:610832, OMIM:613348, OMIM:620442	
9	***PTEN***	AD	OMIM:158350, OMIM:176807, OMIM:605309 +2	
10	***RAD51C***	AR	OMIM:613390, OMIM:613399	
11	***RAD51D***	—	1 (Orphanet)	
12	***STK11***	AD	OMIM:155600, OMIM:175200, OMIM:260350 +1	
13	***TP53***	AD	OMIM:114480, OMIM:114500, OMIM:114550 +8	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 5 GENES

Lynch syndrome

Mismatch repair genes and EPCAM

Panel details

Panel code	TBO-GP-022
Clinical area	Cancer predisposition
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***EPCAM***	AD, AR	`OMIM:613217, OMIM:613244`	
2	***MLH1***	AD, AR	`OMIM:158320, OMIM:276300, OMIM:609310`	
3	***MSH2***	AD, AR	`OMIM:120435, OMIM:158320, OMIM:619096`	
4	***MSH6***	AD, AR	`OMIM:608089, OMIM:614350, OMIM:619097`	
5	***PMS2***	AD, AR	`OMIM:614337, OMIM:619101`	Pseudogene PMS2CL

Genes needing specific methods

- ***PMS2***: Pseudogene PMS2CL

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 17 GENES

Hereditary colorectal cancer and polyposis

Genes for Lynch syndrome, adenomatous and hamartomatous polyposis

Panel details

Panel code	TBO-GP-023
Clinical area	Cancer predisposition
Number of genes	17
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***APC***	AD	OMIM:114500, OMIM:114550, OMIM:135290 +3	
2	***AXIN2***	AD	OMIM:114500, OMIM:608615	
3	***BMPR1A***	AD	OMIM:174900, OMIM:610069	
4	***EPCAM***	AD, AR	OMIM:613217, OMIM:613244	
5	***GREM1***	—	1 (Orphanet)	
6	***MLH1***	AD, AR	OMIM:158320, OMIM:276300, OMIM:609310	
7	***MSH2***	AD, AR	OMIM:120435, OMIM:158320, OMIM:619096	
8	***MSH3***	AD, AR	OMIM:608089, OMIM:617100	
9	***MSH6***	AD, AR	OMIM:608089, OMIM:614350, OMIM:619097	
10	***MUTYH***	AR	OMIM:608456, OMIM:613659	
11	***NTHL1***	AR	OMIM:616415	
12	***PMS2***	AD, AR	OMIM:614337, OMIM:619101	Pseudogene PMS2CL
13	***POLD1***	AD	OMIM:612591, OMIM:615381, OMIM:620836	
14	***POLE***	AD, AR	OMIM:615083, OMIM:615139, OMIM:618336	
15	***PTEN***	AD	OMIM:158350, OMIM:176807, OMIM:605309 +2	

16	***SMAD4***	AD	OMIM:139210, OMIM:174900, OMIM:175050 +1
17	***STK11***	AD	OMIM:155600, OMIM:175200, OMIM:260350 +1

Genes needing specific methods

- ***PMS2***: Pseudogene PMS2CL

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 2 GENES

Hereditary diffuse gastric cancer

Genes for hereditary diffuse gastric cancer

Panel details

Panel code	TBO-GP-024
Clinical area	Cancer predisposition
Number of genes	2
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CDH1***	AD	OMIM:114480, OMIM:119580, OMIM:137215 +2	
2	***CTNNA1***	AD	OMIM:608970	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 12 GENES

Hereditary pancreatic cancer

Genes with established pancreatic cancer risk

Panel details

Panel code	TBO-GP-025
Clinical area	Cancer predisposition
Number of genes	12
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ATM***	AD, AR	OMIM:114480, OMIM:208900	
2	***BRCA1***	AD, AR	OMIM:114480, OMIM:604370, OMIM:617883	
3	***BRCA2***	AD, AR	OMIM:114480, OMIM:155255, OMIM:176807 +5	
4	***CDKN2A***	AD	OMIM:155601, OMIM:155755, OMIM:606719	
5	***EPCAM***	AD, AR	OMIM:613217, OMIM:613244	
6	***MLH1***	AD, AR	OMIM:158320, OMIM:276300, OMIM:609310	
7	***MSH2***	AD, AR	OMIM:120435, OMIM:158320, OMIM:619096	
8	***MSH6***	AD, AR	OMIM:608089, OMIM:614350, OMIM:619097	
9	***PALB2***	AD, AR	OMIM:610832, OMIM:613348, OMIM:620442	
10	***PMS2***	AD, AR	OMIM:614337, OMIM:619101	Pseudogene PMS2CL
11	***STK11***	AD	OMIM:155600, OMIM:175200, OMIM:260350 +1	
12	***TP53***	AD	OMIM:114480, OMIM:114500, OMIM:114550 +8	

Genes needing specific methods

- ***PMS2***: Pseudogene PMS2CL

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 12 GENES

Hereditary prostate cancer

Genes for inherited prostate cancer risk

Panel details

Panel code	TBO-GP-026
Clinical area	Cancer predisposition
Number of genes	12
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ATM***	AD, AR	OMIM:114480, OMIM:208900	
2	***BRCA1***	AD, AR	OMIM:114480, OMIM:604370, OMIM:617883	
3	***BRCA2***	AD, AR	OMIM:114480, OMIM:155255, OMIM:176807 +5	
4	***CHEK2***	AD	OMIM:176807, OMIM:259500, OMIM:609265	
5	***EPCAM***	AD, AR	OMIM:613217, OMIM:613244	
6	***HOXB13***	—	–	Cancer risk gene; no Mendelian HPO annotation
7	***MLH1***	AD, AR	OMIM:158320, OMIM:276300, OMIM:609310	
8	***MSH2***	AD, AR	OMIM:120435, OMIM:158320, OMIM:619096	
9	***MSH6***	AD, AR	OMIM:608089, OMIM:614350, OMIM:619097	
10	***PALB2***	AD, AR	OMIM:610832, OMIM:613348, OMIM:620442	

11	***PMS2***	AD, AR	OMIM:614337, OMIM:619101	Pseudogene PMS2CL
12	***TP53***	AD	OMIM:114480, OMIM:114500, OMIM:114550 +8	

Genes needing specific methods

- ***HOXB13***: Cancer risk gene; no Mendelian HPO annotation
- ***PMS2***: Pseudogene PMS2CL

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 1 GENES

Li-Fraumeni syndrome

TP53 analysis for Li-Fraumeni syndrome

Panel details

Panel code	TBO-GP-027
Clinical area	Cancer predisposition
Number of genes	1
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***TP53***	AD	`OMIM:114480, OMIM:114500, OMIM:114550 +8`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 8 GENES

Hereditary melanoma

Genes for familial cutaneous and uveal melanoma

Panel details

Panel code	TBO-GP-028
Clinical area	Cancer predisposition
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACD***	AD, AR	OMIM:616553	
2	***BAP1***	AD	OMIM:606661, OMIM:614327, OMIM:619762	
3	***CDK4***	AD	OMIM:609048	
4	***CDKN2A***	AD	OMIM:155601, OMIM:155755, OMIM:606719	
5	***MITF***	AD, AR	OMIM:103500, OMIM:193510, OMIM:614456 +1	
6	***POT1***	AD, AR	OMIM:615848, OMIM:620367, OMIM:620368	
7	***TERF2IP***	—	1 (Orphanet)	
8	***TERT***	AD, AR	OMIM:127550, OMIM:178500, OMIM:601626 +4	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 11 GENES

Hereditary renal cancer

Genes for inherited kidney cancer syndromes

Panel details

Panel code	TBO-GP-029
Clinical area	Cancer predisposition
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***BAP1***	AD	OMIM:606661, OMIM:614327, OMIM:619762	
2	***FH***	AD, AR	OMIM:150800, OMIM:606812	
3	***FLCN***	AD	OMIM:114500, OMIM:135150, OMIM:144700 +2	
4	***MET***	AD, AR	OMIM:114550, OMIM:605074, OMIM:607278 +2	
5	***PTEN***	AD	OMIM:158350, OMIM:176807, OMIM:605309 +2	
6	***SDHB***	AD, AR	OMIM:115310, OMIM:606764, OMIM:606864 +1	
7	***SDHC***	AD	OMIM:605373, OMIM:606764, OMIM:606864	
8	***SDHD***	AD, AR	OMIM:168000, OMIM:606864, OMIM:619167	
9	***TSC1***	AD	OMIM:191100, OMIM:606690, OMIM:607341	
10	***TSC2***	AD	OMIM:606690, OMIM:607341, OMIM:613254	
11	***VHL***	AD, AR	OMIM:144700, OMIM:171300, OMIM:193300 +1	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 12 GENES

Phaeochromocytoma and paraganglioma

Genes for hereditary phaeochromocytoma and paraganglioma

Panel details

Panel code	TBO-GP-030
Clinical area	Cancer predisposition
Number of genes	12
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*EPAS1*	AD	OMIM:611783	
2	*FH*	AD, AR	OMIM:150800, OMIM:606812	
3	*MAX*	AD	OMIM:171300, OMIM:620712	
4	*NF1*	AD	OMIM:162200, OMIM:162210, OMIM:193520 +2	
5	*RET*	AD	OMIM:142623, OMIM:155240, OMIM:162300 +2	
6	*SDHA*	AD, AR	OMIM:252011, OMIM:613642, OMIM:614165 +1	
7	*SDHAF2*	AD	OMIM:601650	
8	*SDHB*	AD, AR	OMIM:115310, OMIM:606764, OMIM:606864 +1	
9	*SDHC*	AD	OMIM:605373, OMIM:606764, OMIM:606864	
10	*SDHD*	AD, AR	OMIM:168000, OMIM:606864, OMIM:619167	
11	*TMEM127*	AD	OMIM:171300	
12	*VHL*	AD, AR	OMIM:144700, OMIM:171300, OMIM:193300 +1	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 4 GENES

Multiple endocrine neoplasia

Genes for MEN1, MEN2, MEN4 and hyperparathyroidism-jaw tumour syndrome

Panel details

Panel code	TBO-GP-031
Clinical area	Cancer predisposition
Number of genes	4
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CDC73***	AD	OMIM:145000, OMIM:145001, OMIM:608266	
2	***CDKN1B***	AD	OMIM:610755	
3	***MEN1***	AD	OMIM:131100	
4	***RET***	AD	OMIM:142623, OMIM:155240, OMIM:162300 +2	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 5 GENES

Hereditary thyroid cancer

Genes for medullary and non-medullary thyroid cancer syndromes

Panel details

Panel code	TBO-GP-032
Clinical area	Cancer predisposition
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***APC***	AD	OMIM:114500, OMIM:114550, OMIM:135290 +3	
2	***DICER1***	AD	OMIM:138800, OMIM:180295, OMIM:601200 +1	
3	***PRKAR1A***	AD	OMIM:101800, OMIM:160980, OMIM:255960 +1	
4	***PTEN***	AD	OMIM:158350, OMIM:176807, OMIM:605309 +2	
5	***RET***	AD	OMIM:142623, OMIM:155240, OMIM:162300 +2	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 1 GENES

Retinoblastoma

RB1 analysis for heritable retinoblastoma

Panel details

Panel code	TBO-GP-033
Clinical area	Cancer predisposition
Number of genes	1
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***RB1***	AD	OMIM:109800, OMIM:180200, OMIM:182280 +1	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 5 GENES

Neurofibromatosis and schwannomatosis

Genes for NF1, NF2-related schwannomatosis and Legius syndrome

Panel details

Panel code	TBO-GP-034
Clinical area	Cancer predisposition
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***LZTR1***	AD, AR	OMIM:605275, OMIM:615670, OMIM:616564	
2	***NF1***	AD	OMIM:162200, OMIM:162210, OMIM:193520 +2	
3	***NF2***	AD	OMIM:101000, OMIM:607174	
4	***SMARCB1***	AD	OMIM:162091, OMIM:609322, OMIM:614608	
5	***SPRED1***	AD	OMIM:611431	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 2 GENES

Tuberous sclerosis complex

TSC1 and TSC2

Panel details

Panel code	TBO-GP-035
Clinical area	Cancer predisposition
Number of genes	2
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***TSC1***	AD	`OMIM:191100, OMIM:606690, OMIM:607341`	
2	***TSC2***	AD	`OMIM:606690, OMIM:607341, OMIM:613254`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 1 GENES

Von Hippel-Lindau disease

VHL analysis

Panel details

Panel code	TBO-GP-036
Clinical area	Cancer predisposition
Number of genes	1
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***VHL***	AD, AR	`OMIM:144700, OMIM:171300, OMIM:193300 +1`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 1 GENES

PTEN hamartoma tumour syndrome

PTEN analysis for Cowden and related syndromes

Panel details

Panel code	TBO-GP-037
Clinical area	Cancer predisposition
Number of genes	1
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***PTEN***	AD	`OMIM:158350, OMIM:176807, OMIM:605309 +2`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 3 GENES

Hamartomatous polyposis

Genes for Peutz-Jeghers and juvenile polyposis

Panel details

Panel code	TBO-GP-038
Clinical area	Cancer predisposition
Number of genes	3
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***BMPR1A***	AD	`OMIM:174900, OMIM:610069`	
2	***SMAD4***	AD	`OMIM:139210, OMIM:174900, OMIM:175050 +1`	
3	***STK11***	AD	`OMIM:155600, OMIM:175200, OMIM:260350 +1`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 8 GENES

Hereditary endometrial cancer

Genes for inherited endometrial cancer risk

Panel details

Panel code	TBO-GP-039
Clinical area	Cancer predisposition
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***EPCAM***	AD, AR	OMIM:613217, OMIM:613244	
2	***MLH1***	AD, AR	OMIM:158320, OMIM:276300, OMIM:609310	
3	***MSH2***	AD, AR	OMIM:120435, OMIM:158320, OMIM:619096	
4	***MSH6***	AD, AR	OMIM:608089, OMIM:614350, OMIM:619097	
5	***PMS2***	AD, AR	OMIM:614337, OMIM:619101	Pseudogene PMS2CL
6	***POLD1***	AD	OMIM:612591, OMIM:615381, OMIM:620836	
7	***POLE***	AD, AR	OMIM:615083, OMIM:615139, OMIM:618336	
8	***PTEN***	AD	OMIM:158350, OMIM:176807, OMIM:605309 +2	

Genes needing specific methods

- ***PMS2***: Pseudogene PMS2CL

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 9 GENES

Myeloid malignancy predisposition

Germline genes for familial leukaemia and myelodysplasia

Panel details

Panel code	TBO-GP-040
Clinical area	Cancer predisposition
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ANKRD26***	AD	OMIM:188000	
2	***CEBPA***	AD	OMIM:601626	
3	***DDX41***	AD	OMIM:616871	
4	***ETV6***	AD	OMIM:601626, OMIM:616216	
5	***GATA2***	AD	OMIM:601626, OMIM:614038, OMIM:614172 +1	
6	***RUNX1***	AD	OMIM:601399, OMIM:601626	
7	***SAMD9***	AD, AR	OMIM:610455, OMIM:617053, OMIM:619041	
8	***SAMD9L***	AD, AR	OMIM:159550, OMIM:252270, OMIM:619806	
9	***TP53***	AD	OMIM:114480, OMIM:114500, OMIM:114550 +8	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 20 GENES

Fanconi anaemia

Genes for Fanconi anaemia complementation groups

Panel details

Panel code	TBO-GP-041
Clinical area	Cancer predisposition
Number of genes	20
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***BRCA2***	AD, AR	OMIM:114480, OMIM:155255, OMIM:176807 +5	
2	***BRIP1***	AD, AR	OMIM:114480, OMIM:609054	
3	***ERCC4***	AR	OMIM:278760, OMIM:610965, OMIM:615272	
4	***FANCA***	AR	OMIM:227650	
5	***FANCB***	XLR	OMIM:300514, OMIM:314390	
6	***FANCC***	AR	OMIM:227645	
7	***FANCD2***	AR	OMIM:227646	
8	***FANCE***	AR	OMIM:600901	
9	***FANCF***	AR	OMIM:603467	
10	***FANCG***	—	OMIM:614082	
11	***FANCI***	AR	OMIM:609053	
12	***FANCL***	AR	OMIM:614083	
13	***MAD2L2***	AR	OMIM:617243	
14	***PALB2***	AD, AR	OMIM:610832, OMIM:613348, OMIM:620442	
15	***RAD51***	AD	OMIM:114480, OMIM:157600, OMIM:614508 +1	

16	***RAD51C***	AR	OMIM:613390, OMIM:613399
17	***RFWD3***	AR	OMIM:617784
18	***SLX4***	AR	OMIM:613951
19	***UBE2T***	AR	OMIM:616435
20	***XRCC2***	AR	OMIM:617247, OMIM:619145, OMIM:619146

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 11 GENES

Hereditary sarcoma

Genes for inherited bone and soft-tissue sarcoma risk

Panel details

Panel code	TBO-GP-042
Clinical area	Cancer predisposition
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***DICER1***	AD	OMIM:138800, OMIM:180295, OMIM:601200 +1	
2	***EXT1***	AD	OMIM:133700, OMIM:215300	
3	***EXT2***	AD, AR	OMIM:133701, OMIM:616682	
4	***NF1***	AD	OMIM:162200, OMIM:162210, OMIM:193520 +2	
5	***RB1***	AD	OMIM:109800, OMIM:180200, OMIM:182280 +1	
6	***SDHA***	AD, AR	OMIM:252011, OMIM:613642, OMIM:614165 +1	
7	***SDHB***	AD, AR	OMIM:115310, OMIM:606764, OMIM:606864 +1	
8	***SDHC***	AD	OMIM:605373, OMIM:606764, OMIM:606864	
9	***SDHD***	AD, AR	OMIM:168000, OMIM:606864, OMIM:619167	
10	***TP53***	AD	OMIM:114480, OMIM:114500, OMIM:114550 +8	
11	***WRN***	AR	OMIM:277700	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 1 GENES

DICER1 syndrome

DICER1 analysis

Panel details

Panel code	TBO-GP-043
Clinical area	Cancer predisposition
Number of genes	1
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***DICER1***	AD	`OMIM:138800, OMIM:180295, OMIM:601200 +1`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 2 GENES

Rhabdoid tumour predisposition

SMARCB1 and SMARCA4

Panel details

Panel code	TBO-GP-044
Clinical area	Cancer predisposition
Number of genes	2
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***SMARCA4***	AD	`OMIM:613325, OMIM:614609, OMIM:620792`	
2	***SMARCB1***	AD	`OMIM:162091, OMIM:609322, OMIM:614608`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 16 GENES

Childhood cancer predisposition

Germline genes for paediatric solid tumours and leukaemia

Panel details

Panel code	TBO-GP-045
Clinical area	Cancer predisposition
Number of genes	16
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ALK*	—	2 (Orphanet)	
2	*APC*	AD	OMIM:114500, OMIM:114550, OMIM:135290 +3	
3	*DICER1*	AD	OMIM:138800, OMIM:180295, OMIM:601200 +1	
4	*ELP1*	AD, AR	OMIM:155255, OMIM:223900	
5	*MEN1*	AD	OMIM:131100	
6	*NF1*	AD	OMIM:162200, OMIM:162210, OMIM:193520 +2	
7	*PHOX2B*	AD	OMIM:209880, OMIM:613013	
8	*PTCH1*	AD	OMIM:109400, OMIM:605462, OMIM:610828	
9	*RB1*	AD	OMIM:109800, OMIM:180200, OMIM:182280 +1	
10	*RET*	AD	OMIM:142623, OMIM:155240, OMIM:162300 +2	
11	*SMARCA4*	AD	OMIM:613325, OMIM:614609, OMIM:620792	
12	*SMARCB1*	AD	OMIM:162091, OMIM:609322, OMIM:614608	
13	*SUFU*	AD, AR	OMIM:155255, OMIM:607174, OMIM:617757 +1	
14	*TP53*	AD	OMIM:114480, OMIM:114500, OMIM:114550 +8	
15	*VHL*	AD, AR	OMIM:144700, OMIM:171300, OMIM:193300 +1	

16	***WT1***	AD	OMIM:106210, OMIM:136680, OMIM:156240 +5

Notes

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- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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CANCER PREDISPOSITION | 3 GENES

Gorlin syndrome

Genes for naevoid basal cell carcinoma syndrome

Panel details

Panel code	TBO-GP-046
Clinical area	Cancer predisposition
Number of genes	3
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***PTCH1***	AD	`OMIM:109400, OMIM:605462, OMIM:610828`	
2	***PTCH2***	AD, AR	`OMIM:155255, OMIM:605462`	
3	***SUFU***	AD, AR	`OMIM:155255, OMIM:607174, OMIM:617757 +1`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 35 GENES

Developmental and epileptic encephalopathy

Genes for early-onset epileptic encephalopathies

Panel details

Panel code	TBO-GP-047
Clinical area	Neurology
Number of genes	35
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ALDH7A1***	AR	OMIM:266100	
2	***ARX***	XLR	OMIM:300004, OMIM:300215, OMIM:300419 +2	
3	***CACNA1A***	AD	OMIM:108500, OMIM:141500, OMIM:183086 +1	
4	***CDKL5***	XLD	OMIM:300672	
5	***CHD2***	AD	OMIM:615369	
6	***DEPDC5***	AD, AR	OMIM:604364, OMIM:620504	
7	***DNM1***	AD, AR	OMIM:616346, OMIM:620352	
8	***FOXG1***	AD	OMIM:613454	
9	***GABRA1***	AD	OMIM:615744	
10	***GABRB3***	AD	OMIM:612269, OMIM:617113	
11	***GNAO1***	AD	OMIM:615473, OMIM:617493	
12	***GRIN2A***	AD	OMIM:245570	
13	***GRIN2B***	AD	OMIM:613970, OMIM:616139	
14	***HCN1***	AD	OMIM:615871, OMIM:618482	
15	***KCNA2***	AD	OMIM:616366	

16	*KCNQ2*	AD	OMIM:121200, OMIM:613720
17	*KCNQ3*	AD	OMIM:121201
18	*KCNT1*	AD	OMIM:614959, OMIM:615005
19	*MECP2*	XLR, XLD	OMIM:300055, OMIM:300260, OMIM:300496 +2
20	*PCDH19*	XL	OMIM:300088
21	*PNPO*	AR	OMIM:610090
22	*PRRT2*	AD	OMIM:128200, OMIM:602066, OMIM:605751
23	*SCN1A*	AD	OMIM:604403, OMIM:607208, OMIM:609634 +1
24	*SCN1B*	AD, AR	OMIM:604233, OMIM:612838, OMIM:615377 +1
25	*SCN2A*	AD	OMIM:607745, OMIM:613721, OMIM:618924
26	*SCN8A*	AD	OMIM:614306, OMIM:614558, OMIM:617080 +1
27	*SLC2A1*	AD, AR	OMIM:601042, OMIM:606777, OMIM:608885 +2
28	*SLC6A1*	AD	OMIM:616421
29	*SPTAN1*	AD	OMIM:613477, OMIM:620528, OMIM:620538 +1
30	*STX1B*	AD	OMIM:616172
31	*STXBP1*	AD	OMIM:612164
32	*SYNGAP1*	AD	OMIM:612621
33	*TSC1*	AD	OMIM:191100, OMIM:606690, OMIM:607341
34	*TSC2*	AD	OMIM:606690, OMIM:607341, OMIM:613254
35	*WWOX*	AR	OMIM:133239, OMIM:614322, OMIM:616211

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 5 GENES

Dravet syndrome and SCN1A spectrum

Genes for Dravet syndrome and its genetic mimics

Panel details

Panel code	TBO-GP-048
Clinical area	Neurology
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***GABRA1***	AD	OMIM:615744	
2	***PCDH19***	XL	OMIM:300088	
3	***SCN1A***	AD	OMIM:604403, OMIM:607208, OMIM:609634 +1	
4	***SCN1B***	AD, AR	OMIM:604233, OMIM:612838, OMIM:615377 +1	
5	***STXBP1***	AD	OMIM:612164	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY · 9 GENES

Familial focal epilepsy

GATOR1 and other genes for familial focal epilepsies

Panel details

Panel code	TBO-GP-049
Clinical area	Neurology
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CHRNA2***	AD	OMIM:610353	
2	***CHRNA4***	AD	OMIM:600513	
3	***CHRNB2***	AD	OMIM:605375	
4	***DEPDC5***	AD, AR	OMIM:604364, OMIM:620504	
5	***GRIN2A***	AD	OMIM:245570	
6	***KCNT1***	AD	OMIM:614959, OMIM:615005	
7	***LGI1***	AD	OMIM:600512	
8	***NPRL2***	AD	OMIM:617116	
9	***NPRL3***	AD	OMIM:617118	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 20 GENES

Hereditary spastic paraplegia

Genes for pure and complex hereditary spastic paraplegias

Panel details

Panel code	TBO-GP-050
Clinical area	Neurology
Number of genes	20
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AP4B1***	AR	OMIM:614066	
2	***AP4E1***	AD, AR	OMIM:184450, OMIM:613744	
3	***AP4M1***	AR	OMIM:612936	
4	***AP4S1***	AR	OMIM:614067	
5	***ATL1***	AD	OMIM:182600, OMIM:613708	
6	***B4GALNT1***	AR	OMIM:609195	
7	***CAPN1***	AR	OMIM:616907	
8	***CYP7B1***	AR	OMIM:270800, OMIM:613812	
9	***DDHD2***	AR	OMIM:615033	
10	***FA2H***	AR	OMIM:612319	
11	***KIF1A***	AD, AR	OMIM:201300, OMIM:610357, OMIM:614213 +1	
12	***KIF5A***	AD	OMIM:604187, OMIM:617235, OMIM:617921	
13	***L1CAM***	XLR	OMIM:303350, OMIM:304100, OMIM:307000	
14	***NIPA1***	AD	OMIM:600363	
15	***PLP1***	XLR	OMIM:312080, OMIM:312920	

16	***REEP1***	AD, AR	OMIM:610250, OMIM:614751, OMIM:620011
17	***SPAST***	AD	OMIM:182601
18	***SPG11***	AR	OMIM:602099, OMIM:604360, OMIM:616668
19	***SPG7***	AD, AR	OMIM:607259
20	***ZFYVE26***	AR	OMIM:270700

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 22 GENES

Hereditary ataxia

Sequence-variant genes for inherited ataxias (repeat-expansion ataxias need separate testing)

Panel details

Panel code	TBO-GP-051
Clinical area	Neurology
Number of genes	22
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*AFG3L2*	AD, AR	OMIM:610246, OMIM:614487, OMIM:618977	
2	*ANO10*	AR	OMIM:613728	
3	*APTX*	AR	OMIM:208920	
4	*ATM*	AD, AR	OMIM:114480, OMIM:208900	
5	*CACNA1A*	AD	OMIM:108500, OMIM:141500, OMIM:183086 +1	
6	*ELOVL4*	AD, AR	OMIM:133190, OMIM:600110, OMIM:614457	
7	*GRID2*	AR	OMIM:616204	
8	*ITPR1*	AD, AR	OMIM:117360, OMIM:206700, OMIM:606658	
9	*KCNC3*	AD	OMIM:605259	
10	*KCND3*	AD	OMIM:607346, OMIM:616399	
11	*MRE11*	AR	OMIM:604391	
12	*PNKP*	AR	OMIM:605589, OMIM:613402, OMIM:616267	
13	*POLG*	AD, AR	OMIM:157640, OMIM:203700, OMIM:258450 +3	
14	*PRKCG*	AD	OMIM:605361	
15	*SACS*	AR	OMIM:270550	

16	***SETX***	AD, AR	OMIM:602433, OMIM:606002
17	***SIL1***	AR	OMIM:248800
18	***SPTBN2***	AD, AR	OMIM:600224, OMIM:615386
19	***SYNE1***	AD, AR	OMIM:610743, OMIM:612998, OMIM:618484
20	***TDP1***	AR	OMIM:607250
21	***TTBK2***	AD	OMIM:604432
22	***TTPA***	AR	OMIM:277460

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 27 GENES

Charcot-Marie-Tooth and hereditary neuropathy

Genes for demyelinating, axonal and intermediate CMT

Panel details

Panel code	TBO-GP-052
Clinical area	Neurology
Number of genes	27
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AARS1***	AD, AR	OMIM:613287, OMIM:616339, OMIM:619661 +1	
2	***BSCL2***	AD, AR	OMIM:269700, OMIM:270685, OMIM:615924 +1	
3	***DNM2***	AD, AR	OMIM:160150, OMIM:606482, OMIM:615368	
4	***EGR2***	AD, AR	OMIM:145900, OMIM:605253, OMIM:607678	
5	***FIG4***	AD, AR	OMIM:216340, OMIM:611228, OMIM:612577 +1	
6	***GARS1***	AD	OMIM:600794, OMIM:601472, OMIM:619042	
7	***GDAP1***	AD, AR	OMIM:214400, OMIM:607706, OMIM:607831 +1	
8	***GJB1***	XLD	OMIM:302800	
9	***HSPB1***	AD	OMIM:606595, OMIM:608634	
10	***HSPB8***	AD	OMIM:158590, OMIM:608673	
11	***IGHMBP2***	AR	OMIM:604320, OMIM:616155	
12	***INF2***	AD	OMIM:613237, OMIM:614455	
13	***LITAF***	AD	OMIM:601098	
14	***LRSAM1***	AD, AR	OMIM:614436	
15	***MFN2***	AD, AR	OMIM:151800, OMIM:601152, OMIM:609260 +1	

16	***MORC2***	AD	OMIM:616688, OMIM:619090
17	***MPZ***	AD, AR	OMIM:118200, OMIM:145900, OMIM:180800 +4
18	***MTMR2***	AR	OMIM:601382
19	***NDRG1***	AR	OMIM:601455
20	***NEFL***	AD, AR	OMIM:607684, OMIM:607734, OMIM:617882
21	***PMP22***	AD, AR	OMIM:118220, OMIM:118300, OMIM:139393 +3
22	***PRX***	AD, AR	OMIM:145900, OMIM:614895
23	***SBF2***	AR	OMIM:604563
24	***SH3TC2***	AD, AR	OMIM:601596, OMIM:613353
25	***SORD***	AR	OMIM:618912
26	***TRPV4***	AD	OMIM:113500, OMIM:156530, OMIM:168400 +7
27	***YARS1***	AD, AR	OMIM:608323, OMIM:619418

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 10 GENES

Spinal muscular atrophy and motor neuronopathy

SMN1/SMN2 dosage plus genes for non-5q spinal muscular atrophies

Panel details

Panel code	TBO-GP-053
Clinical area	Neurology
Number of genes	10
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ASAH1***	AR	OMIM:159950, OMIM:228000	
2	***BICD2***	AD	OMIM:615290, OMIM:618291	
3	***DYNC1H1***	AD	OMIM:158600, OMIM:614228, OMIM:614563	
4	***IGHMBP2***	AR	OMIM:604320, OMIM:616155	
5	***PLEKHG5***	AR	OMIM:611067, OMIM:615376	
6	***SMN1***	AR	OMIM:253300, OMIM:253400, OMIM:253550 +1	Copy-number (dosage) analysis; SMN2 paralogue
7	***SMN2***	AR	OMIM:253400	Copy number reported as modifier
8	***TRPV4***	AD	OMIM:113500, OMIM:156530, OMIM:168400 +7	
9	***UBA1***	XLR	OMIM:301054, OMIM:301830	
10	***VAPB***	AD	OMIM:182980, OMIM:608627	

Genes needing specific methods

- ***SMN1***: Copy-number (dosage) analysis; SMN2 paralogue

- ***SMN2***: Copy number reported as modifier

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 20 GENES

Amyotrophic lateral sclerosis and frontotemporal dementia

Genes for familial ALS and FTD (C9orf72 repeat needs separate testing)

Panel details

Panel code	TBO-GP-054
Clinical area	Neurology
Number of genes	20
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ANXA11***	AD	OMIM:617839, OMIM:619733	
2	***C9orf72***	AD	OMIM:105550	G4C2 repeat expansion analysis required
3	***CCNF***	AD	OMIM:619141	
4	***CHCHD10***	AD	OMIM:615048, OMIM:615911, OMIM:616209	
5	***FUS***	AD	OMIM:608030, OMIM:614782	
6	***GRN***	AD, AR	OMIM:607485, OMIM:614706	
7	***HNRNPA1***	AD	OMIM:610099, OMIM:615424, OMIM:615426	
8	***KIF5A***	AD	OMIM:604187, OMIM:617235, OMIM:617921	
9	***MAPT***	AD, AR	OMIM:168600, OMIM:172700, OMIM:260540 +2	
10	***MATR3***	AD	OMIM:606070	

11	***NEK1***	AD, AR	OMIM:252100, OMIM:263520, OMIM:617892
12	***OPTN***	AD, AR	OMIM:137760, OMIM:613435
13	***PFN1***	AD	OMIM:614808
14	***SOD1***	AD, AR	OMIM:105400, OMIM:618598
15	***SQSTM1***	AD, AR	OMIM:167250, OMIM:616437, OMIM:617145 +1
16	***TARDBP***	AD	OMIM:612069
17	***TBK1***	AD, AR	OMIM:616439, OMIM:617900, OMIM:620880
18	***TUBA4A***	AD	OMIM:616208
19	***UBQLN2***	XLD	OMIM:300857
20	***VCP***	AD	OMIM:167320, OMIM:613954, OMIM:616687

Genes needing specific methods

- ***C9orf72***: G4C2 repeat expansion analysis required

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 13 GENES

Parkinson disease

Genes for monogenic Parkinson disease and parkinsonism

Panel details

Panel code	TBO-GP-055
Clinical area	Neurology
Number of genes	13
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ATP13A2***	AR	OMIM:606693, OMIM:617225	
2	***DNAJC6***	AR	OMIM:615528	
3	***FBXO7***	AR	OMIM:260300	
4	***GBA1***	AD, AR	OMIM:127750, OMIM:168600, OMIM:230800 +4	Pseudogene GBAP1
5	***LRRK2***	AD	OMIM:607060	
6	***PARK7***	AR	OMIM:606324	
7	***PINK1***	AR	OMIM:605909	
8	***PLA2G6***	AR	OMIM:256600, OMIM:610217, OMIM:612953	
9	***PRKN***	AD, AR	OMIM:167000, OMIM:211980, OMIM:600116	
10	***SNCA***	AD	OMIM:127750, OMIM:168601, OMIM:605543	
11	***SYNJ1***	AR	OMIM:615530, OMIM:617389	
12	***VPS13C***	AR	OMIM:616840	
13	***VPS35***	AD	OMIM:614203	

Genes needing specific methods

- ***GBA1***: Pseudogene GBAP1

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 15 GENES

Dystonia

Genes for isolated and combined dystonias

Panel details

Panel code	TBO-GP-056
Clinical area	Neurology
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ADCY5***	AD, AR	OMIM:606703, OMIM:619647, OMIM:619651	
2	***ANO3***	AD	OMIM:615034	
3	***ATP1A3***	AD	OMIM:128235, OMIM:601338, OMIM:614820 +1	
4	***GCH1***	AD, AR	OMIM:128230, OMIM:233910	
5	***GNAL***	AD	OMIM:615073	
6	***KCTD17***	AD	OMIM:616398	
7	***KMT2B***	AD	OMIM:617284, OMIM:619934	
8	***PNKD***	AD	OMIM:118800	
9	***PRKRA***	AR	OMIM:612067	
10	***SGCE***	AD	OMIM:159900	
11	***SPR***	AD, AR	OMIM:612716	
12	***TH***	AR	OMIM:605407	
13	***THAP1***	AD	OMIM:602629	
14	***TOR1A***	AD, AR	OMIM:128100, OMIM:618947	
15	***VPS16***	AD	OMIM:619291	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 11 GENES

Early-onset dementia

Genes for early-onset Alzheimer disease and frontotemporal dementia

Panel details

Panel code	TBO-GP-057
Clinical area	Neurology
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***APP***	AD	OMIM:104300, OMIM:605714	
2	***C9orf72***	AD	OMIM:105550	G4C2 repeat expansion analysis required
3	***CHMP2B***	AD	OMIM:600795	
4	***GRN***	AD, AR	OMIM:607485, OMIM:614706	
5	***MAPT***	AD, AR	OMIM:168600, OMIM:172700, OMIM:260540 +2	
6	***PSEN1***	AD	OMIM:172700, OMIM:600274, OMIM:607822 +2	
7	***PSEN2***	AD	OMIM:606889, OMIM:613697	
8	***TBK1***	AD, AR	OMIM:616439, OMIM:617900, OMIM:620880	
9	***TREM2***	AR	OMIM:618193	
10	***TYROBP***	AR	OMIM:221770	
11	***VCP***	AD	OMIM:167320, OMIM:613954, OMIM:616687	

Genes needing specific methods

- ***C9orf72***: G4C2 repeat expansion analysis required

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 26 GENES

Leukodystrophy

Genes for inherited white-matter disorders

Panel details

Panel code	TBO-GP-058
Clinical area	Neurology
Number of genes	26
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*AARS2*	AR	OMIM:614096, OMIM:615889	
2	*ABCD1*	XLR	OMIM:300100	
3	*ARSA*	AR	OMIM:250100	
4	*ASPA*	AR	OMIM:271900	
5	*CLCN2*	AD, AR	OMIM:605635, OMIM:607628, OMIM:615651	
6	*CSF1R*	AD, AR	OMIM:221820, OMIM:618476	
7	*DARS2*	AR	OMIM:611105	
8	*EIF2B1*	AR	OMIM:603896	
9	*EIF2B2*	AR	OMIM:620312	
10	*EIF2B3*	AR	OMIM:620313	
11	*EIF2B4*	AR	OMIM:620314	
12	*EIF2B5*	AR	OMIM:620315	
13	*GALC*	AR	OMIM:245200	
14	*GFAP*	AD	OMIM:203450	
15	*HEPACAM*	AD, AR	OMIM:604004, OMIM:613925, OMIM:613926	

16	***HTRA1***	AD, AR	`OMIM:600142, OMIM:610149, OMIM:616779`
17	***LMNB1***	AD	`OMIM:169500, OMIM:619179`
18	***MLC1***	AR	`OMIM:604004`
19	***NOTCH3***	AD	`OMIM:125310, OMIM:130720, OMIM:615293`
20	***PLP1***	XLR	`OMIM:312080, OMIM:312920`
21	***POLR1C***	AR	`OMIM:248390, OMIM:616494`
22	***POLR3A***	AR	`OMIM:264090, OMIM:607694`
23	***POLR3B***	AD, AR	`OMIM:607694, OMIM:614381, OMIM:619742`
24	***PSAP***	AD, AR	`OMIM:249900, OMIM:610539, OMIM:611721 +2`
25	***SUMF1***	AR	`OMIM:272200`
26	***TUBB4A***	AD	`OMIM:128101, OMIM:612438`

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 25 GENES

Mitochondrial disease (nuclear genes)

Nuclear-encoded genes for mitochondrial disorders (mtDNA analysed separately)

Panel details

Panel code	TBO-GP-059
Clinical area	Neurology
Number of genes	25
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACAD9***	AR	OMIM:611126	
2	***COQ2***	AD, AR	OMIM:146500, OMIM:607426	
3	***COQ8A***	AR	OMIM:612016	
4	***COX10***	AR	OMIM:619046	
5	***DGUOK***	AR	OMIM:251880, OMIM:617068, OMIM:617070	
6	***ETHE1***	AR	OMIM:602473	
7	***FBXL4***	AR	OMIM:615471	
8	***MPV17***	AR	OMIM:256810, OMIM:618400	
9	***MTFMT***	AR	OMIM:614947, OMIM:618248	
10	***NDUFS1***	AR	OMIM:618226	
11	***NDUFS4***	AR, Mito	OMIM:252010	
12	***NDUFV1***	AR	OMIM:618225	
13	***OPA1***	AD, AR	OMIM:125250, OMIM:165500, OMIM:210000 +1	
14	***PDHA1***	XLD	OMIM:312170	
15	***POLG***	AD, AR	OMIM:157640, OMIM:203700, OMIM:258450 +3	

16	***POLG2***	AD, AR	OMIM:610131, OMIM:618528, OMIM:619425
17	***RRM2B***	AD, AR	OMIM:268315, OMIM:612075, OMIM:613077
18	***SCO2***	AD, AR	OMIM:604377, OMIM:608908
19	***SLC25A4***	AD, AR	OMIM:609283, OMIM:615418, OMIM:617184
20	***SUCLA2***	AR	OMIM:612073
21	***SUCLG1***	AR	OMIM:245400
22	***SURF1***	AR, Mito	OMIM:220110, OMIM:616684
23	***TK2***	AR	OMIM:609560, OMIM:617069
24	***TWNK***	AD, AR	OMIM:271245, OMIM:607459, OMIM:609286 +1
25	***TYMP***	AR	OMIM:603041

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 29 GENES

Muscular dystrophy

Genes for Duchenne/Becker, limb-girdle, Emery-Dreifuss and congenital muscular dystrophies

Panel details

Panel code	TBO-GP-060
Clinical area	Neurology
Number of genes	29
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ANO5***	AD, AR	OMIM:166260, OMIM:611307, OMIM:613319	
2	***CAPN3***	AD, AR	OMIM:253600, OMIM:618129	
3	***CAV3***	AD	OMIM:123320, OMIM:192600, OMIM:606072 +2	
4	***COL6A1***	AD, AR	OMIM:158810, OMIM:254090	
5	***COL6A2***	AD, AR	OMIM:255600, OMIM:620725, OMIM:620727	
6	***COL6A3***	AD, AR	OMIM:616411, OMIM:620726, OMIM:620728	
7	***DES***	AD, AR	OMIM:181400, OMIM:601419, OMIM:604765	
8	***DMD***	XLR	OMIM:300376, OMIM:302045, OMIM:310200	Exon deletions/duplications by dosage analysis
9	***DNAJB6***	AD	OMIM:603511	
10	***DYSF***	AR	OMIM:253601, OMIM:254130, OMIM:606768	
11	***EMD***	XLR	OMIM:310300	

12	***FHL1***	XLR, XLD	OMIM:300280, OMIM:300695, OMIM:300696 +2	
13	***FKRP***	AR	OMIM:236670, OMIM:606612, OMIM:607155 +1	
14	***FKTN***	AR	OMIM:236670, OMIM:253800, OMIM:611588 +2	
15	***GAA***	AR	OMIM:232300	
16	***LAMA2***	AR	OMIM:607855, OMIM:618138	
17	***LARGE1***	AR	OMIM:236670, OMIM:608840, OMIM:613154	
18	***LMNA***	AD, AR	OMIM:115200, OMIM:151660, OMIM:176670 +8	
19	***MYOT***	AD	OMIM:609200	
20	***POMGNT1***	AR	OMIM:253280, OMIM:613151, OMIM:613157 +1	
21	***POMT1***	AR	OMIM:236670, OMIM:609308, OMIM:613155	
22	***POMT2***	AR	OMIM:236670, OMIM:613150, OMIM:613156 +1	
23	***SGCA***	AR	OMIM:608099	
24	***SGCB***	AR	OMIM:604286	
25	***SGCD***	AD, AR	OMIM:601287, OMIM:606685	
26	***SGCG***	AR	OMIM:253700	
27	***SMCHD1***	AD	OMIM:158901, OMIM:603457	FSHD1 (D4Z4) tested separately
28	***TCAP***	AD, AR	OMIM:601954, OMIM:607487	
29	***TRIM32***	AR	OMIM:254110, OMIM:615988	

Genes needing specific methods

- ***DMD***: Exon deletions/duplications by dosage analysis
- ***SMCHD1***: FSHD1 (D4Z4) tested separately

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

This product uses the Human Phenotype Ontology (Köhler et al., hpo.jax.org). © Tribro-Omics · TBO-GP-060 · 11 November 2026

NEUROLOGY · 17 GENES

Congenital myopathy

Genes for nemaline, core, centronuclear and other congenital myopathies

Panel details

Panel code	TBO-GP-061
Clinical area	Neurology
Number of genes	17
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACTA1***	AD, AR	OMIM:161800, OMIM:616852, OMIM:620265 +1	
2	***BIN1***	AR	OMIM:255200	
3	***CFL2***	AR	OMIM:610687	
4	***DNM2***	AD, AR	OMIM:160150, OMIM:606482, OMIM:615368	
5	***KLHL40***	AR	OMIM:615348	
6	***KLHL41***	AR	OMIM:615731	
7	***LMOD3***	AR	OMIM:616165	
8	***MTM1***	XLR	OMIM:310400	
9	***MYH7***	AD, AR	OMIM:160500, OMIM:192600, OMIM:255160 +2	
10	***NEB***	AR	OMIM:256030, OMIM:619334	Triplicated region; reduced sensitivity
11	***RYR1***	AD, AR	OMIM:117000, OMIM:145600, OMIM:255320 +1	
12	***SELENON***	AR	OMIM:602771	

13	***STAC3***	AR	OMIM:255995	
14	***TNNT1***	AD, AR	OMIM:605355, OMIM:620386, OMIM:620389	
15	***TPM2***	AD	OMIM:108120, OMIM:609285	
16	***TPM3***	AD, AR	OMIM:255310, OMIM:609284	
17	***TTN***	AD, AR	OMIM:600334, OMIM:603689, OMIM:604145 +3	Large gene; truncating variants in A-band most relevant

Genes needing specific methods

- ***NEB***: Triplicated region; reduced sensitivity
- ***TTN***: Large gene; truncating variants in A-band most relevant

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

This product uses the Human Phenotype Ontology (Köhler et al., hpo.jax.org). © Tribro-Omics · TBO-GP-061 · 11 November 2026

NEUROLOGY | 20 GENES

Congenital myasthenic syndromes

Genes for congenital myasthenic syndromes

Panel details

Panel code	TBO-GP-062
Clinical area	Neurology
Number of genes	20
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AGRN***	AR	OMIM:615120	
2	***ALG14***	AR	OMIM:616227, OMIM:619031, OMIM:619036	
3	***ALG2***	AR	OMIM:607906, OMIM:616228	
4	***CHAT***	AR	OMIM:254210	
5	***CHRNA1***	AD, AR	OMIM:253290, OMIM:601462, OMIM:608930	
6	***CHRNB1***	AD, AR	OMIM:616313, OMIM:616314	
7	***CHRND***	AD, AR	OMIM:253290, OMIM:616321, OMIM:616322 +1	
8	***CHRNE***	AD, AR	OMIM:605809, OMIM:608931, OMIM:616324	
9	***COL13A1***	AR	OMIM:616720	
10	***COLQ***	AR	OMIM:603034	
11	***DOK7***	AR	OMIM:254300, OMIM:618389	
12	***DPAGT1***	AR	OMIM:608093, OMIM:614750	
13	***GFPT1***	AR	OMIM:608931, OMIM:610542	
14	***GMPPB***	AR	OMIM:615350, OMIM:615351, OMIM:615352	
15	***LRP4***	AD, AR	OMIM:212780, OMIM:614305, OMIM:616304	

16	***MUSK***	AR	OMIM:208150, OMIM:616325
17	***PREPL***	AR	OMIM:616224
18	***RAPSN***	AR	OMIM:616326, OMIM:618388
19	***SCN4A***	AD, AR	OMIM:168300, OMIM:170400, OMIM:170500 +5
20	***SLC5A7***	AD, AR	OMIM:158580, OMIM:617143

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 4 GENES

Periodic paralysis and non-dystrophic myotonia

Skeletal muscle ion channel genes

Panel details

Panel code	TBO-GP-063
Clinical area	Neurology
Number of genes	4
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CACNA1S***	AD, AR	OMIM:170400, OMIM:188580, OMIM:601887 +1	
2	***CLCN1***	AD, AR	OMIM:160800, OMIM:255700	
3	***KCNJ2***	AD	OMIM:170390, OMIM:609622, OMIM:613980	
4	***SCN4A***	AD, AR	OMIM:168300, OMIM:170400, OMIM:170500 +5	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 9 GENES

Rett and Rett-like disorders

Genes for Rett syndrome and clinically overlapping disorders

Panel details

Panel code	TBO-GP-064
Clinical area	Neurology
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CDKL5***	XLD	OMIM:300672	
2	***FOXG1***	AD	OMIM:613454	
3	***GRIN2B***	AD	OMIM:613970, OMIM:616139	
4	***MECP2***	XLR, XLD	OMIM:300055, OMIM:300260, OMIM:300496 +2	
5	***MEF2C***	AD	OMIM:613443	
6	***SLC9A6***	XLR	OMIM:300243	
7	***STXBP1***	AD	OMIM:612164	
8	***SYNGAP1***	AD	OMIM:612621	
9	***TCF4***	AD	OMIM:610954, OMIM:613267	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 9 GENES

Angelman-like disorders

Sequence-variant genes for Angelman and its mimics (methylation testing separate)

Panel details

Panel code	TBO-GP-065
Clinical area	Neurology
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CDKL5***	XLD	OMIM:300672	
2	***EHMT1***	AD	OMIM:610253	
3	***FOXG1***	AD	OMIM:613454	
4	***MBD5***	AD	OMIM:156200	
5	***MECP2***	XLR, XLD	OMIM:300055, OMIM:300260, OMIM:300496 +2	
6	***SLC9A6***	XLR	OMIM:300243	
7	***TCF4***	AD	OMIM:610954, OMIM:613267	
8	***UBE3A***	AD	OMIM:105830	
9	***ZEB2***	AD	OMIM:235730	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 57 GENES

Intellectual disability (core genes)

High-yield genes for intellectual disability and developmental delay

Panel details

Panel code	TBO-GP-066
Clinical area	Neurology
Number of genes	57
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ADNP***	AD	OMIM:615873	
2	***ANKRD11***	AD	OMIM:148050	
3	***ARID1B***	AD	OMIM:135900	
4	***ASXL3***	AD	OMIM:615485	
5	***AUTS2***	AD	OMIM:615834	
6	***CASK***	XLD	OMIM:300422, OMIM:300749, OMIM:300908	
7	***CDKL5***	XLD	OMIM:300672	
8	***CHAMP1***	AD	OMIM:616579	
9	***CHD2***	AD	OMIM:615369	
10	***CHD7***	AD	OMIM:214800, OMIM:612370	
11	***CHD8***	AD	OMIM:615032	
12	***CREBBP***	AD	OMIM:180849, OMIM:618332	
13	***CTCF***	AD	OMIM:615502	
14	***CTNNB1***	AD, AR	OMIM:114500, OMIM:114550, OMIM:132600 +4	
15	***DDX3X***	XLR, XLD	OMIM:300958	

16	*DLG4*	AD	OMIM:618793
17	*DNMT3A*	AD	OMIM:601626, OMIM:615879, OMIM:618724
18	*DYRK1A*	AD	OMIM:614104
19	*EHMT1*	AD	OMIM:610253
20	*EP300*	AD	OMIM:114500, OMIM:180849, OMIM:613684 +1
21	*FOXP1*	AD	OMIM:613670
22	*GRIN2B*	AD	OMIM:613970, OMIM:616139
23	*HNRNPU*	AD	OMIM:617391
24	*KAT6A*	AD	OMIM:616268
25	*KAT6B*	AD	OMIM:603736, OMIM:606170
26	*KDM5C*	XLR	OMIM:300534
27	*KMT2A*	AD	OMIM:605130
28	*KMT2C*	AD	OMIM:617768
29	*KMT2D*	AD	OMIM:147920, OMIM:620186
30	*MECP2*	XLR, XLD	OMIM:300055, OMIM:300260, OMIM:300496 +2
31	*MED13L*	AD	OMIM:616789
32	*MYT1L*	AD	OMIM:616521
33	*NAA10*	XLR, XLD	OMIM:300855, OMIM:309800
34	*NIPBL*	AD	OMIM:122470
35	*NSD1*	AD	OMIM:117550
36	*PACS1*	AD	OMIM:615009
37	*POGZ*	AD	OMIM:616364
38	*PPP2R5D*	AD	OMIM:616355
39	*PTEN*	AD	OMIM:158350, OMIM:176807, OMIM:605309 +2
40	*PURA*	AD	OMIM:616158
41	*SATB2*	AD	OMIM:612313
42	*SCN2A*	AD	OMIM:607745, OMIM:613721, OMIM:618924
43	*SETD5*	AD	OMIM:615761
44	*SHANK3*	AD	OMIM:606232, OMIM:613950
45	*SMARCA2*	AD	OMIM:601358, OMIM:619293
46	*SMARCA4*	AD	OMIM:613325, OMIM:614609, OMIM:620792
47	*SMC1A*	XLR, XLD	OMIM:300590, OMIM:301044
48	*SON*	AD	OMIM:617140

49	***STXBP1***	AD	OMIM:612164
50	***SYNGAP1***	AD	OMIM:612621
51	***TBR1***	AD	OMIM:606053
52	***TCF4***	AD	OMIM:610954, OMIM:613267
53	***TRIO***	AD	OMIM:617061, OMIM:618825
54	***USP9X***	XLR, XLD	OMIM:300919, OMIM:300968
55	***WAC***	AD	OMIM:616708
56	***ZBTB18***	AD	OMIM:612337
57	***ZEB2***	AD	OMIM:235730

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 28 GENES

Autism spectrum disorder

Genes with strong evidence in syndromic and non-syndromic autism

Panel details

Panel code	TBO-GP-067
Clinical area	Neurology
Number of genes	28
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ADNP***	AD	OMIM:615873	
2	***ANKRD11***	AD	OMIM:148050	
3	***ARID1B***	AD	OMIM:135900	
4	***ASH1L***	AD	OMIM:617796	
5	***CHD2***	AD	OMIM:615369	
6	***CHD8***	AD	OMIM:615032	
7	***CNTNAP2***	AR	OMIM:610042	
8	***CTNNB1***	AD, AR	OMIM:114500, OMIM:114550, OMIM:132600 +4	
9	***CUL3***	AD	OMIM:614496, OMIM:619239	
10	***DEAF1***	AD, AR	OMIM:615828, OMIM:617171	
11	***DYRK1A***	AD	OMIM:614104	
12	***FOXP1***	AD	OMIM:613670	
13	***GRIN2B***	AD	OMIM:613970, OMIM:616139	
14	***KDM6B***	AD	OMIM:618505	
15	***KMT2C***	AD	OMIM:617768	

16	***KMT5B***	AD	OMIM:617788
17	***MECP2***	XLR, XLD	OMIM:300055, OMIM:300260, OMIM:300496 +2
18	***MED13L***	AD	OMIM:616789
19	***NRXN1***	AR	OMIM:614325
20	***POGZ***	AD	OMIM:616364
21	***PTEN***	AD	OMIM:158350, OMIM:176807, OMIM:605309 +2
22	***SCN2A***	AD	OMIM:607745, OMIM:613721, OMIM:618924
23	***SETD5***	AD	OMIM:615761
24	***SHANK3***	AD	OMIM:606232, OMIM:613950
25	***SYNGAP1***	AD	OMIM:612621
26	***TBR1***	AD	OMIM:606053
27	***TSC1***	AD	OMIM:191100, OMIM:606690, OMIM:607341
28	***TSC2***	AD	OMIM:606690, OMIM:607341, OMIM:613254

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 15 GENES

Primary microcephaly

Genes for autosomal recessive primary microcephaly and related disorders

Panel details

Panel code	TBO-GP-068
Clinical area	Neurology
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ASPM*	AR	OMIM:608716	
2	*CASK*	XLD	OMIM:300422, OMIM:300749, OMIM:300908	
3	*CDK5RAP2*	AR	OMIM:604804	
4	*CDK6*	AR	OMIM:616080	
5	*CEP135*	AR	OMIM:614673	
6	*CEP152*	AR	OMIM:613823, OMIM:614852	
7	*CPAP*	AR	OMIM:608393, OMIM:613676	
8	*KNL1*	AR	OMIM:604321	
9	*MCPH1*	AR	OMIM:251200	
10	*NDE1*	AR	OMIM:605013, OMIM:614019	
11	*PCNT*	AR	OMIM:210720	
12	*PHC1*	AR	OMIM:615414	
13	*STIL*	AR	OMIM:612703	
14	*WDR62*	AR	OMIM:604317	
15	*ZNF335*	AR	OMIM:615095	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 10 GENES

Overgrowth and macrocephaly

Genes for Sotos, Weaver, Tatton-Brown-Rahman and related overgrowth syndromes

Panel details

Panel code	TBO-GP-069
Clinical area	Neurology
Number of genes	10
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***BRWD3***	XLR	OMIM:300659	
2	***CHD8***	AD	OMIM:615032	
3	***DNMT3A***	AD	OMIM:601626, OMIM:615879, OMIM:618724	
4	***EED***	AD	OMIM:617561	
5	***EZH2***	AD	OMIM:277590	
6	***GPC3***	AD, XLR	OMIM:194070, OMIM:312870	
7	***GPC4***	AD, XLR	OMIM:194070, OMIM:301026, OMIM:312870	
8	***NFIX***	AD	OMIM:602535, OMIM:614753	
9	***NSD1***	AD	OMIM:117550	
10	***PTEN***	AD	OMIM:158350, OMIM:176807, OMIM:605309 +2	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito

mitochondrial.

- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 21 GENES

Malformations of cortical development

Genes for lissencephaly, polymicrogyria, heterotopia and dysgyria

Panel details

Panel code	TBO-GP-070
Clinical area	Neurology
Number of genes	21
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ADGRG1*	AR	OMIM:606854, OMIM:615752	
2	*AKT3*	AD	OMIM:615937	
3	*ARFGEF2*	AR	OMIM:608097	
4	*ARX*	XLR	OMIM:300004, OMIM:300215, OMIM:300419 +2	
5	*COL4A1*	AD	OMIM:175780, OMIM:180000, OMIM:611773 +1	
6	*COL4A2*	AD	OMIM:614483	
7	*DCX*	XL	OMIM:300067	
8	*DYNC1H1*	AD	OMIM:158600, OMIM:614228, OMIM:614563	
9	*FLNA*	XLR, XLD	OMIM:300048, OMIM:300049, OMIM:300244 +6	
10	*KIF5C*	AD	OMIM:615282	
11	*MTOR*	AD	OMIM:607341, OMIM:616638	
12	*NDE1*	AR	OMIM:605013, OMIM:614019	
13	*PAFAH1B1*	AD	OMIM:607432	
14	*PIK3R2*	AD	OMIM:603387	
15	*RELN*	AD, AR	OMIM:257320, OMIM:600512, OMIM:616436	

16	***TUBA1A***	AD	OMIM:611603
17	***TUBB2B***	AD	OMIM:610031
18	***TUBB3***	AD	OMIM:600638, OMIM:614039
19	***TUBG1***	AD	OMIM:615412
20	***VLDLR***	AR	OMIM:224050
21	***WDR62***	AR	OMIM:604317

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 5 GENES

Congenital hydrocephalus

Genes for inherited hydrocephalus

Panel details

Panel code	TBO-GP-071
Clinical area	Neurology
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CCDC88C***	AD, AR	`OMIM:236600, OMIM:616053`	
2	***L1CAM***	XLR	`OMIM:303350, OMIM:304100, OMIM:307000`	
3	***MPDZ***	AR	`OMIM:615219`	
4	***SMARCC1***	AD	`OMIM:620241`	
5	***TRIM71***	AD	`OMIM:618667`	

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 24 GENES

Joubert syndrome and related ciliopathies

Genes for Joubert syndrome and Meckel-Gruber spectrum

Panel details

Panel code	TBO-GP-072
Clinical area	Neurology
Number of genes	24
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AHI1***	AR	OMIM:608629	
2	***ARL13B***	AR	OMIM:612291	
3	***B9D1***	AR	OMIM:614209, OMIM:617120	
4	***B9D2***	AR	OMIM:614175	
5	***CC2D2A***	AR	OMIM:612284, OMIM:612285, OMIM:619111 +1	
6	***CEP290***	AR	OMIM:610188, OMIM:610189, OMIM:611134 +2	
7	***CEP41***	AR	OMIM:614464	
8	***CPLANE1***	AR	OMIM:277170, OMIM:614615	
9	***CSPP1***	AR	OMIM:615636	
10	***INPP5E***	AR	OMIM:213300, OMIM:610156	
11	***KIAA0586***	AR	OMIM:616490, OMIM:616546	
12	***KIF7***	AR	OMIM:200990, OMIM:607131, OMIM:614120	
13	***MKS1***	AR	OMIM:249000, OMIM:615990, OMIM:617121	
14	***NPHP1***	AR	OMIM:256100, OMIM:266900, OMIM:609583	
15	***OFD1***	XLR, XLD	OMIM:300209, OMIM:300424, OMIM:300804 +1	

16	***PDE6D***	AR	OMIM:615665
17	***RPGRIP1L***	AR	OMIM:611560, OMIM:611561, OMIM:619113
18	***TCTN1***	AR	OMIM:614173
19	***TCTN2***	AR	OMIM:613885, OMIM:616654
20	***TCTN3***	AR	OMIM:258860, OMIM:614815
21	***TMEM138***	AR	OMIM:614465
22	***TMEM216***	AR	OMIM:603194, OMIM:608091
23	***TMEM231***	AR	OMIM:614970, OMIM:615397
24	***TMEM67***	AR	OMIM:216360, OMIM:602152, OMIM:607361 +3

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY 13 GENES

Neuronal ceroid lipofuscinosis

Genes for the neuronal ceroid lipofuscinoses (CLN disorders)

Panel details

Panel code	TBO-GP-073
Clinical area	Neurology
Number of genes	13
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ATP13A2***	AR	OMIM:606693, OMIM:617225	
2	***CLN3***	AR	OMIM:204200	
3	***CLN5***	AR	OMIM:256731	
4	***CLN6***	AR	OMIM:204300, OMIM:601780	
5	***CLN8***	AR	OMIM:600143, OMIM:610003	
6	***CTSD***	AR	OMIM:610127	
7	***CTSF***	AR	OMIM:615362	
8	***DNAJC5***	AD	OMIM:162350	
9	***GRN***	AD, AR	OMIM:607485, OMIM:614706	
10	***KCTD7***	AR	OMIM:611726	
11	***MFSD8***	AR	OMIM:610951, OMIM:616170	
12	***PPT1***	AR	OMIM:256730	
13	***TPP1***	AR	OMIM:204500, OMIM:609270	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 15 GENES

Pontocerebellar hypoplasia

Genes for pontocerebellar hypoplasia types 1–13

Panel details

Panel code	TBO-GP-074
Clinical area	Neurology
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AMPD2***	AR	OMIM:615686, OMIM:615809	
2	***CASK***	XLD	OMIM:300422, OMIM:300749, OMIM:300908	
3	***CHMP1A***	AR	OMIM:614961	
4	***CLP1***	AR	OMIM:615803	
5	***EXOSC3***	AR	OMIM:614678	
6	***EXOSC8***	AR	OMIM:616081	
7	***PCLO***	AR	OMIM:608027	
8	***RARS2***	AR	OMIM:611523	
9	***SEPSECS***	AR	OMIM:613811	
10	***TSEN15***	AR	OMIM:617026	
11	***TSEN2***	AR	OMIM:612389	
12	***TSEN34***	AR	OMIM:612390	
13	***TSEN54***	AR	OMIM:225753, OMIM:277470, OMIM:610204	
14	***VPS53***	AR	OMIM:615851	
15	***VRK1***	AR	OMIM:607596, OMIM:620542	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 14 GENES

Hereditary sensory and autonomic neuropathy

Genes for HSAN and congenital insensitivity to pain

Panel details

Panel code	TBO-GP-075
Clinical area	Neurology
Number of genes	14
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ATL1***	AD	OMIM:182600, OMIM:613708	
2	***DNMT1***	AD	OMIM:604121, OMIM:614116	
3	***ELP1***	AD, AR	OMIM:155255, OMIM:223900	
4	***NGF***	AR	OMIM:608654	
5	***NTRK1***	AR	OMIM:256800	
6	***PRDM12***	AR	OMIM:616488	
7	***RAB7A***	AD	OMIM:600882	
8	***RETREG1***	AR	OMIM:201300, OMIM:613115	
9	***SCN10A***	AD	OMIM:615551	
10	***SCN11A***	AD	OMIM:615548, OMIM:615552	
11	***SCN9A***	AD, AR	OMIM:133020, OMIM:167400, OMIM:201300 +1	
12	***SPTLC1***	AD	OMIM:162400, OMIM:620285	
13	***SPTLC2***	AD	OMIM:613640	
14	***WNK1***	AD, AR	OMIM:201300, OMIM:614492	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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NEUROLOGY | 6 GENES

Episodic ataxia and hemiplegic migraine

Ion channel and transporter genes for paroxysmal neurological disorders

Panel details

Panel code	TBO-GP-076
Clinical area	Neurology
Number of genes	6
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ATP1A2***	AD, AR	OMIM:104290, OMIM:602481, OMIM:619602 +1	
2	***CACNA1A***	AD	OMIM:108500, OMIM:141500, OMIM:183086 +1	
3	***KCNA1***	AD	OMIM:160120	
4	***PRRT2***	AD	OMIM:128200, OMIM:602066, OMIM:605751	
5	***SCN1A***	AD	OMIM:604403, OMIM:607208, OMIM:609634 +1	
6	***SLC1A3***	AD	OMIM:612656	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC · 8 GENES

Urea cycle disorders

Genes for urea cycle defects and hyperammonaemia

Panel details

Panel code	TBO-GP-077
Clinical area	Metabolic
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ARG1***	AR	OMIM:207800	
2	***ASL***	AR	OMIM:207900	
3	***ASS1***	AR	OMIM:215700	
4	***CPS1***	AR	OMIM:237300	
5	***NAGS***	AR	OMIM:237310	
6	***OTC***	XLR	OMIM:311250	
7	***SLC25A13***	AR	OMIM:603471, OMIM:605814	
8	***SLC25A15***	AR	OMIM:238970	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC 13 GENES

Organic acidaemias

Genes for methylmalonic, propionic, isovaleric and related acidaemias

Panel details

Panel code	TBO-GP-078
Clinical area	Metabolic
Number of genes	13
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACAT1***	AR	OMIM:203750	
2	***GCDH***	AR	OMIM:231670	
3	***HMGCL***	AR	OMIM:246450	
4	***IVD***	AR	OMIM:243500	
5	***MCCC1***	AR	OMIM:210200	
6	***MCCC2***	AR	OMIM:210210	
7	***MMAA***	AR	OMIM:251100	
8	***MMAB***	AR	OMIM:251110	
9	***MMACHC***	AR	OMIM:277400	
10	***MMADHC***	AR	OMIM:277410	
11	***MMUT***	AR	OMIM:251000	
12	***PCCA***	AR	OMIM:606054	
13	***PCCB***	AR	OMIM:606054	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC | 14 GENES

Fatty acid oxidation disorders

Genes for mitochondrial fatty acid oxidation and carnitine disorders

Panel details

Panel code	TBO-GP-079
Clinical area	Metabolic
Number of genes	14
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACAD9***	AR	OMIM:611126	
2	***ACADM***	AR	OMIM:201450	
3	***ACADS***	AR	OMIM:201470	
4	***ACADVL***	AR	OMIM:201475	
5	***CPT1A***	AR	OMIM:255120	
6	***CPT2***	AD, AR	OMIM:255110, OMIM:600649, OMIM:608836 +1	
7	***ETFA***	AR	OMIM:231680	
8	***ETFB***	AR	OMIM:231680	
9	***ETFDH***	AR	OMIM:231680	
10	***HADH***	AR	OMIM:231530, OMIM:609975	
11	***HADHA***	AR	OMIM:609015, OMIM:609016	
12	***HADHB***	AR	OMIM:620300	
13	***SLC22A5***	AR	OMIM:212140	
14	***SLC25A20***	AR	OMIM:212138	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC · 18 GENES

Aminoacidopathies

Genes for phenylketonuria, tyrosinaemia, maple syrup urine disease and homocystinuria

Panel details

Panel code	TBO-GP-080
Clinical area	Metabolic
Number of genes	18
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AMT***	AR	OMIM:620398	
2	***BCKDHA***	AR	OMIM:248600	
3	***BCKDHB***	AR	OMIM:620698	
4	***CBS***	AR	OMIM:236200	
5	***DBT***	AR	OMIM:620699	
6	***DLD***	AR	OMIM:246900	
7	***DNAJC12***	AR	OMIM:617384	
8	***FAH***	AR	OMIM:276700	
9	***GCH1***	AD, AR	OMIM:128230, OMIM:233910	
10	***GLDC***	AR	OMIM:605899	
11	***HGD***	AR	OMIM:203500	
12	***HPD***	AD, AR	OMIM:140350, OMIM:276710	
13	***MTHFR***	AD, AR	OMIM:181500, OMIM:188050, OMIM:236250 +1	
14	***PAH***	AR	OMIM:261600	
15	***PCBD1***	AR	OMIM:264070	

16	***PTS***	AR	OMIM:261640
17	***QDPR***	AR	OMIM:261630
18	***TAT***	AR	OMIM:276600

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC 18 GENES

Glycogen storage disease

Genes for hepatic and muscle glycogen storage disorders

Panel details

Panel code	TBO-GP-081
Clinical area	Metabolic
Number of genes	18
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AGL***	AR	OMIM:232400	
2	***ENO3***	AR	OMIM:612932	
3	***G6PC1***	AR	OMIM:232200	
4	***GAA***	AR	OMIM:232300	
5	***GBE1***	AR	OMIM:232500, OMIM:263570	
6	***GYG1***	AR	OMIM:613507, OMIM:616199	
7	***GYS1***	AR	OMIM:611556	
8	***GYS2***	AR	OMIM:240600	
9	***LDHA***	AR	OMIM:612933	
10	***PFKM***	AR	OMIM:232800	
11	***PGAM2***	AR	OMIM:261670	
12	***PGM1***	AR	OMIM:614921	
13	***PHKA2***	XLR	OMIM:306000	
14	***PHKB***	AR	OMIM:261750	
15	***PHKG2***	AR	OMIM:613027	

16	***PYGL***	AR	OMIM:232700
17	***PYGM***	AR	OMIM:232600
18	***SLC37A4***	AD, AR	OMIM:232220, OMIM:232240, OMIM:619525

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC · 34 GENES

Lysosomal storage disorders

Genes for sphingolipidoses, mucopolysaccharidoses, oligosaccharidoses and related disorders

Panel details

Panel code	TBO-GP-082
Clinical area	Metabolic
Number of genes	34
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AGA***	AR	OMIM:208400	
2	***ARSA***	AR	OMIM:250100	
3	***ARSB***	AR	OMIM:253200	
4	***CTSA***	AR	OMIM:256540	
5	***CTSK***	AR	OMIM:265800	
6	***FUCA1***	AR	OMIM:230000	
7	***GAA***	AR	OMIM:232300	
8	***GALC***	AR	OMIM:245200	
9	***GALNS***	AR	OMIM:253000	
10	***GBA1***	AD, AR	OMIM:127750, OMIM:168600, OMIM:230800 +4	Pseudogene GBAP1
11	***GLA***	XLR	OMIM:301500	
12	***GLB1***	AR	OMIM:230500, OMIM:230600, OMIM:230650 +1	
13	***GNPTAB***	AR	OMIM:252500, OMIM:252600	
14	***GNPTG***	AR	OMIM:252605	
15	***GNS***	AR	OMIM:252940	

16	***GUSB***	AR	OMIM:253220
17	***HEXA***	AR	OMIM:272800
18	***HEXB***	AR	OMIM:268800
19	***HGSNAT***	AR	OMIM:252930, OMIM:616544
20	***HYAL1***	AR	OMIM:601492
21	***IDS***	XLR	OMIM:309900
22	***IDUA***	AR	OMIM:607014, OMIM:607015, OMIM:607016
23	***LIPA***	AR	OMIM:278000, OMIM:620151
24	***MAN2B1***	AR	OMIM:248500
25	***MANBA***	AR	OMIM:248510
26	***NAGA***	AR	OMIM:609241, OMIM:609242
27	***NAGLU***	AD, AR	OMIM:252920, OMIM:616491
28	***NEU1***	AR	OMIM:256550
29	***NPC1***	AR	OMIM:257220
30	***NPC2***	AR	OMIM:607625
31	***PSAP***	AD, AR	OMIM:249900, OMIM:610539, OMIM:611721 +2
32	***SGSH***	AR	OMIM:252900
33	***SMPD1***	AR	OMIM:257200, OMIM:607616
34	***SUMF1***	AR	OMIM:272200

Genes needing specific methods

- ***GBA1***: Pseudogene GBAP1

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC 11 GENES

Mucopolysaccharidoses

Genes for MPS types I–IX

Panel details

Panel code	TBO-GP-083
Clinical area	Metabolic
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ARSB***	AR	OMIM:253200	
2	***GALNS***	AR	OMIM:253000	
3	***GLB1***	AR	OMIM:230500, OMIM:230600, OMIM:230650 +1	
4	***GNS***	AR	OMIM:252940	
5	***GUSB***	AR	OMIM:253220	
6	***HGSNAT***	AR	OMIM:252930, OMIM:616544	
7	***HYAL1***	AR	OMIM:601492	
8	***IDS***	XLR	OMIM:309900	
9	***IDUA***	AR	OMIM:607014, OMIM:607015, OMIM:607016	
10	***NAGLU***	AD, AR	OMIM:252920, OMIM:616491	
11	***SGSH***	AR	OMIM:252900	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC · 20 GENES

Peroxisomal disorders

Genes for Zellweger spectrum and single peroxisomal enzyme defects

Panel details

Panel code	TBO-GP-084
Clinical area	Metabolic
Number of genes	20
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCD1***	XLR	OMIM:300100	
2	***ACOX1***	AD, AR	OMIM:264470, OMIM:618960	
3	***AGPS***	AR	OMIM:600121	
4	***AMACR***	AR	OMIM:214950, OMIM:614307	
5	***GNPAT***	AR	OMIM:222765	
6	***HSD17B4***	AR	OMIM:233400, OMIM:261515	
7	***PEX1***	AR	OMIM:214100, OMIM:234580, OMIM:601539	
8	***PEX10***	AR	OMIM:614870, OMIM:614871	
9	***PEX12***	AR	OMIM:266510, OMIM:614859	
10	***PEX13***	AR	OMIM:614883, OMIM:614885	
11	***PEX14***	AR	OMIM:614887	
12	***PEX16***	AR	OMIM:614876, OMIM:614877	
13	***PEX19***	AR	OMIM:614886	
14	***PEX2***	AR	OMIM:614866, OMIM:614867	
15	***PEX26***	AR	OMIM:614872, OMIM:614873	

16	***PEX3***	AR	OMIM:614882, OMIM:617370
17	***PEX5***	AR	OMIM:202370, OMIM:214110, OMIM:616716
18	***PEX6***	AD, AR	OMIM:614862, OMIM:614863, OMIM:616617
19	***PEX7***	AR	OMIM:215100, OMIM:266500, OMIM:614879
20	***PHYH***	AR	OMIM:266500

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

This product uses the Human Phenotype Ontology (Köhler et al., hpo.jax.org). © Tribro-Omics · TBO-GP-084 · 11 November 2026

METABOLIC 29 GENES

Congenital disorders of glycosylation

Genes for N-linked and multiple-pathway glycosylation defects

Panel details

Panel code	TBO-GP-085
Clinical area	Metabolic
Number of genes	29
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ALG1***	AR	OMIM:608540	
2	***ALG11***	AR	OMIM:613661	
3	***ALG12***	AR	OMIM:607143	
4	***ALG13***	XL	OMIM:300884	
5	***ALG2***	AR	OMIM:607906, OMIM:616228	
6	***ALG3***	AR	OMIM:601110	
7	***ALG6***	AR	OMIM:603147	
8	***ALG8***	AD, AR	OMIM:608104, OMIM:617874	
9	***ALG9***	AR	OMIM:263210, OMIM:608776	
10	***ATP6V0A2***	AR	OMIM:219200, OMIM:278250	
11	***COG1***	AR	OMIM:611209	
12	***COG4***	AD, AR	OMIM:613489, OMIM:618150	
13	***COG6***	AR	OMIM:614576, OMIM:615328	
14	***COG7***	AR	OMIM:608779	
15	***COG8***	AR	OMIM:611182	

16	***DOLK***	AR	OMIM:610768
17	***DPAGT1***	AR	OMIM:608093, OMIM:614750
18	***DPM1***	AR	OMIM:608799
19	***MOGS***	AR	OMIM:606056
20	***MPI***	AR	OMIM:602579
21	***NGLY1***	AR	OMIM:615273
22	***PGM1***	AR	OMIM:614921
23	***PMM2***	AR	OMIM:212065
24	***SLC35A2***	XLD	OMIM:300896
25	***SLC35C1***	AR	OMIM:266265
26	***SRD5A3***	AR	OMIM:612379, OMIM:612713
27	***SSR4***	XLR	OMIM:300934
28	***STT3A***	AD, AR	OMIM:615596, OMIM:619714
29	***STT3B***	AR	OMIM:615597

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC · 8 GENES

Copper and iron overload disorders

Genes for Wilson disease, Menkes disease and haemochromatosis

Panel details

Panel code	TBO-GP-086
Clinical area	Metabolic
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ATP7A***	XLR	OMIM:300489, OMIM:304150, OMIM:309400	
2	***ATP7B***	AR	OMIM:277900	
3	***CP***	AR	OMIM:604290	
4	***HAMP***	AR	OMIM:613313	
5	***HFE***	AR	OMIM:235200	
6	***HJV***	AR	OMIM:602390	
7	***SLC40A1***	AD	OMIM:606069	
8	***TFR2***	AR	OMIM:604250	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC 3 GENES

Cerebral creatine deficiency

Genes for creatine synthesis and transport defects

Panel details

Panel code	TBO-GP-087
Clinical area	Metabolic
Number of genes	3
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***GAMT***	AR	OMIM:612736	
2	***GATM***	AD, AR	OMIM:134600, OMIM:612718	
3	***SLC6A8***	XLR	OMIM:300352	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC · 15 GENES

Neurotransmitter disorders

Genes for monoamine, GABA and vitamin B6-dependent neurotransmitter disorders

Panel details

Panel code	TBO-GP-088
Clinical area	Metabolic
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABAT***	AR	OMIM:613163	
2	***ALDH5A1***	AR	OMIM:271980	
3	***ALDH7A1***	AR	OMIM:266100	
4	***DBH***	AR	OMIM:223360	
5	***DDC***	AR	OMIM:608643	
6	***DNAJC12***	AR	OMIM:617384	
7	***GCH1***	AD, AR	OMIM:128230, OMIM:233910	
8	***MAOA***	XLR	OMIM:300615	
9	***PNPO***	AR	OMIM:610090	
10	***PTS***	AR	OMIM:261640	
11	***QDPR***	AR	OMIM:261630	
12	***SLC18A2***	AR	OMIM:618049	
13	***SLC6A3***	AR	OMIM:613135	
14	***SPR***	AD, AR	OMIM:612716	
15	***TH***	AR	OMIM:605407	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC | 5 GENES

Disorders of vitamin D metabolism

Genes for vitamin D-dependent rickets

Panel details

Panel code	TBO-GP-089
Clinical area	Metabolic
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CYP24A1***	AR	OMIM:143880	
2	***CYP27B1***	AR	OMIM:264700	
3	***CYP2R1***	AR	OMIM:600081	
4	***CYP3A4***	AD	OMIM:619073	
5	***VDR***	AR	OMIM:277440	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC | 9 GENES

Hypoparathyroidism and hypocalcaemia

Genes for inherited hypoparathyroidism and autosomal dominant hypocalcaemia

Panel details

Panel code	TBO-GP-090
Clinical area	Metabolic
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AIRE***	AD, AR	OMIM:240300	
2	***CASR***	AD, AR	OMIM:145980, OMIM:239200, OMIM:601198 +1	
3	***FAM111A***	AD	OMIM:127000, OMIM:602361	
4	***GATA3***	AD	OMIM:146255	
5	***GCM2***	AD, AR	OMIM:617343, OMIM:618883	
6	***GNA11***	AD	OMIM:145981, OMIM:615361	
7	***PTH***	AD, AR	OMIM:146200	
8	***TBCE***	AR	OMIM:241410, OMIM:244460, OMIM:617207	
9	***TBX1***	AD, AR	OMIM:187500, OMIM:188400, OMIM:192430 +1	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC | 10 GENES

Maturity-onset diabetes of the young

Genes for MODY and monogenic diabetes in adolescents and adults

Panel details

Panel code	TBO-GP-091
Clinical area	Metabolic
Number of genes	10
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCC8***	AD, AR	OMIM:125853, OMIM:240800, OMIM:256450 +2	
2	***CEL***	AD	OMIM:609812	
3	***GCK***	AD, AR	OMIM:125851, OMIM:125853, OMIM:602485 +1	
4	***HNF1A***	AD	OMIM:125853, OMIM:142330, OMIM:144700 +3	
5	***HNF1B***	AD	OMIM:125853, OMIM:137920, OMIM:144700	
6	***HNF4A***	AD	OMIM:125850, OMIM:125853, OMIM:616026	
7	***INS***	AD, AR	OMIM:125852, OMIM:613370, OMIM:616214 +1	
8	***KCNJ11***	AD, AR	OMIM:601820, OMIM:610582, OMIM:616329 +1	
9	***NEUROD1***	AD	OMIM:125853, OMIM:606394	
10	***PDX1***	AD, AR	OMIM:125853, OMIM:260370, OMIM:606392	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito

mitochondrial.

- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC · 17 GENES

Neonatal diabetes

Genes for permanent and transient neonatal diabetes

Panel details

Panel code	TBO-GP-092
Clinical area	Metabolic
Number of genes	17
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCC8***	AD, AR	OMIM:125853, OMIM:240800, OMIM:256450 +2	
2	***EIF2AK3***	AR	OMIM:226980	
3	***FOXP3***	XLR	OMIM:304790	
4	***GATA4***	AD	OMIM:187500, OMIM:607941, OMIM:614429 +2	
5	***GATA6***	AD, AR	OMIM:187500, OMIM:217095, OMIM:600001 +2	
6	***GCK***	AD, AR	OMIM:125851, OMIM:125853, OMIM:602485 +1	
7	***GLIS3***	AR	OMIM:610199	
8	***IER3IP1***	AR	OMIM:614231	
9	***INS***	AD, AR	OMIM:125852, OMIM:613370, OMIM:616214 +1	
10	***KCNJ11***	AD, AR	OMIM:601820, OMIM:610582, OMIM:616329 +1	
11	***MNX1***	AD	OMIM:176450	
12	***NEUROG3***	AR	OMIM:610370	
13	***PDX1***	AD, AR	OMIM:125853, OMIM:260370, OMIM:606392	
14	***PTF1A***	AR	OMIM:609069, OMIM:615935	
15	***RFX6***	AR	OMIM:615710	

16	***SLC2A2***	AD, AR	OMIM:125853, OMIM:227810
17	***ZFP57***	AD	OMIM:601410

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC | 10 GENES

Congenital hyperinsulinism

Genes for hyperinsulinaemic hypoglycaemia

Panel details

Panel code	TBO-GP-093
Clinical area	Metabolic
Number of genes	10
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCC8***	AD, AR	OMIM:125853, OMIM:240800, OMIM:256450 +2	
2	***GCK***	AD, AR	OMIM:125851, OMIM:125853, OMIM:602485 +1	
3	***GLUD1***	AD	OMIM:606762	
4	***HADH***	AR	OMIM:231530, OMIM:609975	
5	***HK1***	AD, AR	OMIM:235700, OMIM:605285, OMIM:617460 +1	
6	***HNF1A***	AD	OMIM:125853, OMIM:142330, OMIM:144700 +3	
7	***HNF4A***	AD	OMIM:125850, OMIM:125853, OMIM:616026	
8	***KCNJ11***	AD, AR	OMIM:601820, OMIM:610582, OMIM:616329 +1	
9	***SLC16A1***	AD, AR	OMIM:245340, OMIM:610021, OMIM:616095	
10	***UCP2***	—	1 (Orphanet)	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito

mitochondrial.

- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC · 11 GENES

Lipodystrophy

Genes for congenital and familial partial lipodystrophy

Panel details

Panel code	TBO-GP-094
Clinical area	Metabolic
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AGPAT2***	AR	OMIM:608594	
2	***AKT2***	AD	OMIM:125853, OMIM:240900	
3	***BSCL2***	AD, AR	OMIM:269700, OMIM:270685, OMIM:615924 +1	
4	***CAV1***	AD, AR	OMIM:606721, OMIM:612526, OMIM:615343	
5	***CAVIN1***	AR	OMIM:613327	
6	***CIDEC***	AR	OMIM:615238	
7	***LMNA***	AD, AR	OMIM:115200, OMIM:151660, OMIM:176670 +8	
8	***PLIN1***	AD	OMIM:613877	
9	***POLD1***	AD	OMIM:612591, OMIM:615381, OMIM:620836	
10	***PPARG***	AD, AR	OMIM:125853, OMIM:601665, OMIM:604367	
11	***ZMPSTE24***	AR	OMIM:275210, OMIM:608612	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC | 5 GENES

Galactose and fructose disorders

Genes for galactosaemia and hereditary fructose intolerance

Panel details

Panel code	TBO-GP-095
Clinical area	Metabolic
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ALDOB***	AR	OMIM:229600	
2	***FBP1***	AR	OMIM:229700	
3	***GALE***	AR	OMIM:230350, OMIM:620776	
4	***GALK1***	AR	OMIM:230200	
5	***GALT***	AR	OMIM:230400	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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METABOLIC · 15 GENES

Cobalamin and folate disorders

Genes for inherited disorders of vitamin B12 and folate metabolism

Panel details

Panel code	TBO-GP-096
Clinical area	Metabolic
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCD4***	AR	OMIM:614857	
2	***AMN***	AR	OMIM:618882	
3	***CBLIF***	AR	OMIM:261000	
4	***CD320***	AR	OMIM:613646	
5	***CUBN***	AR	OMIM:261100, OMIM:618884	
6	***DHFR***	AR	OMIM:613839	
7	***FOLR1***	AR	OMIM:613068	
8	***LMBRD1***	AR	OMIM:277380	
9	***MMACHC***	AR	OMIM:277400	
10	***MMADHC***	AR	OMIM:277410	
11	***MTHFR***	AD, AR	OMIM:181500, OMIM:188050, OMIM:236250 +1	
12	***MTR***	AR	OMIM:250940, OMIM:601634	
13	***MTRR***	AR	OMIM:236270, OMIM:601634	
14	***SLC46A1***	AR	OMIM:229050	
15	***TCN2***	AR	OMIM:275350	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL | 9 GENES

Polycystic kidney disease

Genes for autosomal dominant and recessive polycystic kidney disease

Panel details

Panel code	TBO-GP-097
Clinical area	Renal
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ALG8***	AD, AR	OMIM:608104, OMIM:617874	
2	***DNAJB11***	AD	OMIM:618061	
3	***DZIP1L***	AR	OMIM:617610	
4	***GANAB***	AD	OMIM:600666	
5	***HNF1B***	AD	OMIM:125853, OMIM:137920, OMIM:144700	
6	***IFT140***	AR	OMIM:266920, OMIM:617781	
7	***PKD1***	AD	OMIM:173900	Pseudogene region; long-range methods for exons 1–33
8	***PKD2***	AD	OMIM:613095	
9	***PKHD1***	AR	OMIM:263200	

Genes needing specific methods

- ***PKD1***: Pseudogene region; long-range methods for exons 1–33

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL · 5 GENES

Alport syndrome

Type IV collagen genes for Alport syndrome and thin basement membrane nephropathy

Panel details

Panel code	TBO-GP-098
Clinical area	Renal
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***COL4A3***	AD, AR	OMIM:104200, OMIM:620320, OMIM:620536	
2	***COL4A4***	AD, AR	OMIM:141200, OMIM:203780	
3	***COL4A5***	XLD	OMIM:301050	
4	***COL4A6***	XLR	OMIM:300914	
5	***MYH9***	AD	OMIM:155100, OMIM:603622	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL | 24 GENES

Steroid-resistant nephrotic syndrome

Podocyte and glomerular genes for nephrotic syndrome and FSGS

Panel details

Panel code	TBO-GP-099
Clinical area	Renal
Number of genes	24
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ACTN4*	AD	OMIM:603278	
2	*ARHGDIA*	AR	OMIM:615244	
3	*CD2AP*	—	OMIM:607832	
4	*COQ2*	AD, AR	OMIM:146500, OMIM:607426	
5	*COQ6*	AR	OMIM:614650	
6	*COQ8B*	AR	OMIM:615573	
7	*CRB2*	AR	OMIM:219730, OMIM:616220	
8	*INF2*	AD	OMIM:613237, OMIM:614455	
9	*ITGA3*	AR	OMIM:614748	
10	*ITGB4*	AR	OMIM:226730, OMIM:619816	
11	*LAMB2*	AR	OMIM:609049, OMIM:614199	
12	*LMX1B*	AD, AR	OMIM:161200, OMIM:256020	
13	*MYO1E*	AR	OMIM:614131	
14	*NPHS1*	AR	OMIM:256300	
15	*NPHS2*	AR	OMIM:600995	

16	***NUP205***	AR	OMIM:616893
17	***NUP93***	AR	OMIM:616892
18	***PDSS2***	AR	OMIM:614652
19	***PLCE1***	AR	OMIM:610725
20	***PTPRO***	AR	OMIM:614196
21	***SGPL1***	AR	OMIM:617575
22	***SMARCAL1***	AR	OMIM:242900
23	***TRPC6***	AD	OMIM:603965
24	***WT1***	AD	OMIM:106210, OMIM:136680, OMIM:156240 +5

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL 16 GENES

Nephronophthisis

Genes for nephronophthisis and renal ciliopathies

Panel details

Panel code	TBO-GP-100
Clinical area	Renal
Number of genes	16
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ANKS6*	AR	OMIM:615382	
2	*CEP164*	AR	OMIM:614845	
3	*CEP290*	AR	OMIM:610188, OMIM:610189, OMIM:611134 +2	
4	*GLIS2*	AR	OMIM:611498	
5	*INVS*	AR	OMIM:602088	
6	*IQCB1*	AR	OMIM:609254	
7	*NEK8*	AD, AR	OMIM:613824, OMIM:615415, OMIM:620903	
8	*NPHP1*	AR	OMIM:256100, OMIM:266900, OMIM:609583	
9	*NPHP3*	AR	OMIM:208540, OMIM:267010, OMIM:604387	
10	*NPHP4*	AR	OMIM:606966, OMIM:606996	
11	*RPGRIP1L*	AR	OMIM:611560, OMIM:611561, OMIM:619113	
12	*SDCCAG8*	AR	OMIM:613615, OMIM:615993	
13	*TMEM67*	AR	OMIM:216360, OMIM:602152, OMIM:607361 +3	
14	*TTC21B*	AD, AR	OMIM:613819, OMIM:613820	
15	*WDR19*	AR	OMIM:614376, OMIM:614377, OMIM:614378 +2	

16	***ZNF423***	AD, AR	OMIM:614844

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL 16 GENES

Congenital anomalies of the kidney and urinary tract

Genes for CAKUT and renal hypodysplasia

Panel details

Panel code	TBO-GP-101
Clinical area	Renal
Number of genes	16
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***BMP4***	AD	OMIM:600625, OMIM:607932	
2	***DSTYK***	AD, AR	OMIM:270750, OMIM:610805	
3	***EYA1***	AD	OMIM:113650, OMIM:166780, OMIM:602588	
4	***FRAS1***	AR	OMIM:219000	
5	***FREM2***	AR	OMIM:123570, OMIM:617666	
6	***GATA3***	AD	OMIM:146255	
7	***GREB1L***	AD	OMIM:617805, OMIM:619274	
8	***GRIP1***	AR	OMIM:617667	
9	***HNF1B***	AD	OMIM:125853, OMIM:137920, OMIM:144700	
10	***PAX2***	AD	OMIM:120330, OMIM:616002	
11	***RET***	AD	OMIM:142623, OMIM:155240, OMIM:162300 +2	
12	***ROBO2***	AD	OMIM:610878	
13	***SALL1***	AD	OMIM:107480	
14	***SIX1***	AD	OMIM:113650, OMIM:605192, OMIM:608389	
15	***SIX5***	AD	OMIM:610896	

16	***TBX18***	AD	OMIM:143400

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL · 8 GENES

Atypical haemolytic uraemic syndrome

Complement and coagulation genes for aHUS and C3 glomerulopathy

Panel details

Panel code	TBO-GP-102
Clinical area	Renal
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***C3***	AD, AR	OMIM:612925, OMIM:613779	
2	***CD46***	AD, AR	OMIM:612922	
3	***CFB***	AD, AR	OMIM:612924, OMIM:615561	
4	***CFH***	AD, AR	OMIM:126700, OMIM:235400, OMIM:609814 +1	
5	***CFHR5***	AD	OMIM:614809	
6	***CFI***	AD, AR	OMIM:610984, OMIM:612923, OMIM:615439	
7	***DGKE***	AR	OMIM:615008	
8	***THBD***	AD	OMIM:612926, OMIM:614486	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL · 23 GENES

Salt-wasting and inherited tubulopathies

Genes for Bartter, Gitelman, pseudohypoaldosteronism and magnesium-wasting disorders

Panel details

Panel code	TBO-GP-103
Clinical area	Renal
Number of genes	23
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***BSND***	AR	OMIM:602522	
2	***CASR***	AD, AR	OMIM:145980, OMIM:239200, OMIM:601198 +1	
3	***CLCNKA***	—	OMIM:613090	
4	***CLCNKB***	AR	OMIM:607364, OMIM:613090	
5	***CLDN16***	AR	OMIM:248250	
6	***CLDN19***	AR	OMIM:248190	
7	***CNNM2***	AD, AR	OMIM:613882, OMIM:616418	
8	***CUL3***	AD	OMIM:614496, OMIM:619239	
9	***FXYD2***	AD	OMIM:154020	
10	***HNF1B***	AD	OMIM:125853, OMIM:137920, OMIM:144700	
11	***KCNJ1***	AR	OMIM:241200	
12	***KCNJ10***	AR	OMIM:274600, OMIM:600791, OMIM:612780	
13	***KLHL3***	AD, AR	OMIM:614495	
14	***MAGED2***	XLR	OMIM:300971	
15	***NR3C2***	AD	OMIM:177735, OMIM:605115	

16	***SCNN1A***	AD, AR	OMIM:264350, OMIM:613021, OMIM:618126
17	***SCNN1B***	AD, AR	OMIM:177200, OMIM:211400, OMIM:620125
18	***SCNN1G***	AD, AR	OMIM:613071, OMIM:618114, OMIM:620126
19	***SLC12A1***	AR	OMIM:601678
20	***SLC12A3***	AR	OMIM:263800
21	***TRPM6***	AR	OMIM:602014
22	***WNK1***	AD, AR	OMIM:201300, OMIM:614492
23	***WNK4***	AD	OMIM:614491

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL · 6 GENES

Renal tubular acidosis

Genes for distal and proximal renal tubular acidosis

Panel details

Panel code	TBO-GP-104
Clinical area	Renal
Number of genes	6
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ATP6V0A4***	AR	OMIM:602722	
2	***ATP6V1B1***	AR	OMIM:267300	
3	***CA2***	AR	OMIM:259730	
4	***FOXI1***	AR	OMIM:274600, OMIM:600791	
5	***SLC4A1***	AD, AR	OMIM:166900, OMIM:179800, OMIM:185020 +2	
6	***SLC4A4***	AR	OMIM:604278	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL · 21 GENES

Nephrolithiasis and nephrocalcinosis

Genes for monogenic kidney stone disease and nephrocalcinosis

Panel details

Panel code	TBO-GP-105
Clinical area	Renal
Number of genes	21
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ADCY10***	AD	OMIM:143870	
2	***AGXT***	AR	OMIM:259900	
3	***APRT***	AR	OMIM:614723	
4	***ATP6V0A4***	AR	OMIM:602722	
5	***ATP6V1B1***	AR	OMIM:267300	
6	***CASR***	AD, AR	OMIM:145980, OMIM:239200, OMIM:601198 +1	
7	***CLCN5***	XLR	OMIM:300009, OMIM:300554, OMIM:308990 +1	
8	***CLDN16***	AR	OMIM:248250	
9	***CLDN19***	AR	OMIM:248190	
10	***CYP24A1***	AR	OMIM:143880	
11	***GRHPR***	AR	OMIM:260000	
12	***HOGA1***	AR	OMIM:613616	
13	***OCRL***	XLR	OMIM:300555, OMIM:309000	
14	***SLC22A12***	AR	OMIM:220150	
15	***SLC2A9***	AD, AR	OMIM:612076	

16	***SLC34A1***	AD, AR	OMIM:612286, OMIM:613388, OMIM:616963
17	***SLC34A3***	AR	OMIM:241530
18	***SLC3A1***	AD, AR	OMIM:220100
19	***SLC4A1***	AD, AR	OMIM:166900, OMIM:179800, OMIM:185020 +2
20	***SLC7A9***	AD, AR	OMIM:220100
21	***XDH***	AR	OMIM:278300

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL · 7 GENES

Renal Fanconi syndrome and cystinosis

Genes for generalised proximal tubulopathy

Panel details

Panel code	TBO-GP-106
Clinical area	Renal
Number of genes	7
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CLCN5***	XLR	OMIM:300009, OMIM:300554, OMIM:308990 +1	
2	***CTNS***	AR	OMIM:219750, OMIM:219800, OMIM:219900	
3	***EHHADH***	AD	OMIM:615605	
4	***FAH***	AR	OMIM:276700	
5	***HNF4A***	AD	OMIM:125850, OMIM:125853, OMIM:616026	
6	***OCRL***	XLR	OMIM:300555, OMIM:309000	
7	***SLC34A1***	AD, AR	OMIM:612286, OMIM:613388, OMIM:616963	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL · 6 GENES

Autosomal dominant tubulointerstitial kidney disease

Genes for ADTKD (MUC1 VNTR needs separate testing)

Panel details

Panel code	TBO-GP-107
Clinical area	Renal
Number of genes	6
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***DNAJB11***	AD	OMIM:618061	
2	***HNF1B***	AD	OMIM:125853, OMIM:137920, OMIM:144700	
3	***MUC1***	AD	OMIM:174000	VNTR variants need specific assay
4	***REN***	AD, AR	OMIM:267430, OMIM:613092	
5	***SEC61A1***	AD	OMIM:617056, OMIM:620670, OMIM:620674	
6	***UMOD***	AD	OMIM:162000	

Genes needing specific methods

- ***MUC1***: VNTR variants need specific assay

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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RENAL 4 GENES

Diabetes insipidus

Genes for central and nephrogenic diabetes insipidus

Panel details

Panel code	TBO-GP-108
Clinical area	Renal
Number of genes	4
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AQP2***	AD, AR	OMIM:125800	
2	***AVP***	AD	OMIM:125700	
3	***AVPR2***	XLR	OMIM:300539, OMIM:304800	
4	***WFS1***	AD, AR	OMIM:116400, OMIM:125853, OMIM:222300 +2	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY · 34 GENES

Retinitis pigmentosa

Genes for autosomal dominant, recessive and X-linked retinitis pigmentosa

Panel details

Panel code	TBO-GP-109
Clinical area	Ophthalmology
Number of genes	34
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCA4***	AD, AR	OMIM:153800, OMIM:248200, OMIM:601718 +1	
2	***BEST1***	AD, AR	OMIM:153700, OMIM:193220, OMIM:611809 +1	
3	***CERKL***	AR	OMIM:608380	
4	***CNGA1***	AD, AR, XLR	OMIM:268000, OMIM:613756	
5	***CNGB1***	AR	OMIM:613767	
6	***CRB1***	AD, AR, XL	OMIM:172870, OMIM:600105, OMIM:613835	
7	***CRX***	AD, AR, XLR	OMIM:120970, OMIM:268000, OMIM:613829	
8	***EYS***	AR	OMIM:602772	
9	***FAM161A***	AR	OMIM:606068	
10	***IMPDH1***	AD	OMIM:180105, OMIM:613837	
11	***KLHL7***	AD, AR	OMIM:612943, OMIM:617055	
12	***LRAT***	AD, AR, XLR	OMIM:268000, OMIM:613341	
13	***MAK***	AR	OMIM:614181	
14	***MERTK***	AR	OMIM:613862	
15	***NR2E3***	AD, AR	OMIM:268100, OMIM:611131	

16	***NRL***	AD	OMIM:613750
17	***PCARE***	AR	OMIM:613428
18	***PDE6A***	AR	OMIM:613810
19	***PDE6B***	AD, AR	OMIM:163500, OMIM:613801
20	***PRPF3***	AD	OMIM:601414
21	***PRPF31***	AD	OMIM:600138
22	***PRPF8***	AD	OMIM:600059
23	***PRPH2***	AD, AR	OMIM:136880, OMIM:169150, OMIM:608133 +2
24	***RHO***	AD, AR	OMIM:136880, OMIM:610445, OMIM:613731
25	***RLBP1***	AD, AR	OMIM:136880, OMIM:607475, OMIM:607476
26	***ROM1***	AD, AR, XLR	OMIM:268000, OMIM:608133
27	***RP1***	AD, AR	OMIM:180100
28	***RP2***	XL	OMIM:312600
29	***RPE65***	AD, AR	OMIM:204100, OMIM:613794, OMIM:618697
30	***RPGR***	XLR	OMIM:300029, OMIM:300455, OMIM:300834 +1
31	***SNRNP200***	AD	OMIM:610359
32	***TOPORS***	AD	OMIM:609923
33	***TULP1***	AR	OMIM:600132, OMIM:613843
34	***USH2A***	AR	OMIM:276901, OMIM:613809

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY · 17 GENES

Leber congenital amaurosis

Genes for LCA and early-onset severe retinal dystrophy

Panel details

Panel code	TBO-GP-110
Clinical area	Ophthalmology
Number of genes	17
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AIPL1***	AD, AR, XLR	OMIM:268000, OMIM:604393	
2	***CEP290***	AR	OMIM:610188, OMIM:610189, OMIM:611134 +2	
3	***CRB1***	AD, AR, XL	OMIM:172870, OMIM:600105, OMIM:613835	
4	***CRX***	AD, AR, XLR	OMIM:120970, OMIM:268000, OMIM:613829	
5	***GUCY2D***	AD, AR	OMIM:204000, OMIM:215500, OMIM:601777 +1	
6	***IMPDH1***	AD	OMIM:180105, OMIM:613837	
7	***IQCB1***	AR	OMIM:609254	
8	***KCNJ13***	AD, AR	OMIM:193230, OMIM:614186	
9	***LCA5***	AR	OMIM:604537	
10	***LRAT***	AD, AR, XLR	OMIM:268000, OMIM:613341	
11	***NMNAT1***	AR	OMIM:608553, OMIM:619260	
12	***RD3***	AR	OMIM:610612	
13	***RDH12***	AD, AR	OMIM:612712	
14	***RPE65***	AD, AR	OMIM:204100, OMIM:613794, OMIM:618697	
15	***RPGRIP1***	AR	OMIM:608194, OMIM:613826	

16	***SPATA7***	AR	`OMIM:604232`
17	***TULP1***	AR	`OMIM:600132, OMIM:613843`

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY | 10 GENES

Stargardt disease and macular dystrophy

Genes for inherited macular dystrophies

Panel details

Panel code	TBO-GP-111
Clinical area	Ophthalmology
Number of genes	10
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCA4***	AD, AR	OMIM:153800, OMIM:248200, OMIM:601718 +1	
2	***BEST1***	AD, AR	OMIM:153700, OMIM:193220, OMIM:611809 +1	
3	***CRX***	AD, AR, XLR	OMIM:120970, OMIM:268000, OMIM:613829	
4	***EFEMP1***	AD, AR	OMIM:126600, OMIM:611276, OMIM:620780	
5	***ELOVL4***	AD, AR	OMIM:133190, OMIM:600110, OMIM:614457	
6	***GUCA1A***	AD	OMIM:602093	
7	***PROM1***	AD, AR	OMIM:603786, OMIM:608051, OMIM:612095 +1	
8	***PRPH2***	AD, AR	OMIM:136880, OMIM:169150, OMIM:608133 +2	
9	***RP1L1***	AD, AR	OMIM:613587, OMIM:618826	
10	***TIMP3***	AD	OMIM:136900	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito

mitochondrial.

- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY | 14 GENES

Cone-rod dystrophy and achromatopsia

Genes for cone and cone-rod dysfunction syndromes

Panel details

Panel code	TBO-GP-112
Clinical area	Ophthalmology
Number of genes	14
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCA4***	AD, AR	OMIM:153800, OMIM:248200, OMIM:601718 +1	
2	***ATF6***	AR	OMIM:616517	
3	***CACNA1F***	XLR	OMIM:300071, OMIM:300476, OMIM:300600	
4	***CDHR1***	AR	OMIM:613660	
5	***CNGA3***	AR	OMIM:216900	
6	***CNGB3***	AR	OMIM:262300	
7	***CRX***	AD, AR, XLR	OMIM:120970, OMIM:268000, OMIM:613829	
8	***GNAT2***	AR	OMIM:613856	
9	***GUCY2D***	AD, AR	OMIM:204000, OMIM:215500, OMIM:601777 +1	
10	***KCNV2***	AR	OMIM:610356	
11	***PDE6C***	AR	OMIM:613093	
12	***PDE6H***	AD, AR	OMIM:610024	
13	***PROM1***	AD, AR	OMIM:603786, OMIM:608051, OMIM:612095 +1	
14	***RPGRIP1***	AR	OMIM:608194, OMIM:613826	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY | 9 GENES

Usher syndrome

Genes for Usher syndrome types 1–3

Panel details

Panel code	TBO-GP-113
Clinical area	Ophthalmology
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ADGRV1***	AD, AR	OMIM:604352, OMIM:605472	
2	***CDH23***	AD, AR	OMIM:601067, OMIM:601386, OMIM:617540	
3	***CLRN1***	AD, AR, XLR	OMIM:268000, OMIM:276902, OMIM:614180	
4	***MYO7A***	AD, AR	OMIM:276900, OMIM:600060, OMIM:601317	
5	***PCDH15***	AR	OMIM:601067, OMIM:602083, OMIM:609533	
6	***USH1C***	AR	OMIM:276900, OMIM:276904, OMIM:602092	
7	***USH1G***	AR	OMIM:606943	
8	***USH2A***	AR	OMIM:276901, OMIM:613809	
9	***WHRN***	AR	OMIM:607084, OMIM:611383	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY | 19 GENES

Bardet-Biedl syndrome

Genes for Bardet-Biedl syndrome

Panel details

Panel code	TBO-GP-114
Clinical area	Ophthalmology
Number of genes	19
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ARL6***	AD, AR, XLR	OMIM:209900, OMIM:268000, OMIM:600151 +1	
2	***BBIP1***	AR	OMIM:615995	
3	***BBS1***	AR	OMIM:209900	
4	***BBS10***	AR	OMIM:615987	
5	***BBS12***	AR	OMIM:615989	
6	***BBS2***	AR	OMIM:615981, OMIM:616562	
7	***BBS4***	AR	OMIM:615982	
8	***BBS5***	AR	OMIM:615983	
9	***BBS7***	AR	OMIM:615984	
10	***BBS9***	AR	OMIM:615986	
11	***CEP290***	AR	OMIM:610188, OMIM:610189, OMIM:611134 +2	
12	***IFT172***	AR	OMIM:615630, OMIM:616394, OMIM:619471	
13	***IFT27***	AR	OMIM:615996	
14	***LZTFL1***	AR	OMIM:615994	
15	***MKKS***	AR	OMIM:236700, OMIM:605231	

16	***MKS1***	AR	OMIM:249000, OMIM:615990, OMIM:617121
17	***SDCCAG8***	AR	OMIM:613615, OMIM:615993
18	***TTC8***	AR	OMIM:613464, OMIM:615985
19	***WDPCP***	AR	OMIM:217085, OMIM:615992

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY · 25 GENES

Congenital cataract

Genes for isolated and syndromic congenital cataract

Panel details

Panel code	TBO-GP-115
Clinical area	Ophthalmology
Number of genes	25
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***BFSP1***	AD, AR	OMIM:611391	
2	***BFSP2***	AD	OMIM:611597	
3	***CRYAA***	AD, AR	OMIM:604219	
4	***CRYAB***	AD, AR	OMIM:608810, OMIM:613763, OMIM:613869 +1	
5	***CRYBA1***	AD	OMIM:600881	
6	***CRYBA4***	AD	OMIM:610425	
7	***CRYBB1***	AD, AR	OMIM:611544	
8	***CRYBB2***	AD	OMIM:601547	
9	***CRYBB3***	AD, AR	OMIM:609741	
10	***CRYGC***	AD	OMIM:604307	
11	***CRYGD***	AD	OMIM:115700	
12	***CRYGS***	AD	OMIM:116100	
13	***CYP27A1***	AR	OMIM:213700	
14	***EPHA2***	AD	OMIM:116600	
15	***FOXE3***	AD, AR	OMIM:610256, OMIM:617349	

16	***GALK1***	AR	OMIM:230200
17	***GJA3***	AD	OMIM:601885
18	***GJA8***	AD	OMIM:116200, OMIM:612474
19	***HSF4***	AD	OMIM:116800
20	***LIM2***	AR	OMIM:615277
21	***MAF***	AD	OMIM:601088, OMIM:610202
22	***MIP***	AD	OMIM:615274
23	***NHS***	XLD	OMIM:302200, OMIM:302350
24	***OCRL***	XLR	OMIM:300555, OMIM:309000
25	***PITX3***	AD, AR	OMIM:107250, OMIM:610623

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY · 8 GENES

Glaucoma

Genes for primary congenital and juvenile open-angle glaucoma

Panel details

Panel code	TBO-GP-116
Clinical area	Ophthalmology
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CYP1B1***	AR	OMIM:231300, OMIM:600975, OMIM:617315	
2	***FOXC1***	AD	OMIM:601631, OMIM:602482	
3	***LTBP2***	AR	OMIM:251750, OMIM:600975, OMIM:613086 +1	
4	***MYOC***	AD	OMIM:137750	
5	***OPTN***	AD, AR	OMIM:137760, OMIM:613435	
6	***PITX2***	AD	OMIM:137600, OMIM:180500, OMIM:180550	
7	***TBK1***	AD, AR	OMIM:616439, OMIM:617900, OMIM:620880	
8	***TEK***	AD, AR	OMIM:600195, OMIM:600975, OMIM:617272	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY | 7 GENES

Anterior segment dysgenesis and aniridia

Genes for aniridia, Axenfeld-Rieger and Peters anomaly

Panel details

Panel code	TBO-GP-117
Clinical area	Ophthalmology
Number of genes	7
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***B3GLCT***	AR	OMIM:261540	
2	***CYP1B1***	AR	OMIM:231300, OMIM:600975, OMIM:617315	
3	***FOXC1***	AD	OMIM:601631, OMIM:602482	
4	***FOXE3***	AD, AR	OMIM:610256, OMIM:617349	
5	***PAX6***	AD	OMIM:106210, OMIM:120200, OMIM:120430 +5	
6	***PITX2***	AD	OMIM:137600, OMIM:180500, OMIM:180550	
7	***PITX3***	AD, AR	OMIM:107250, OMIM:610623	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY · 12 GENES

Inherited optic neuropathy

Nuclear genes for optic atrophy (LHON mtDNA variants tested separately)

Panel details

Panel code	TBO-GP-118
Clinical area	Ophthalmology
Number of genes	12
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACO2***	AR	OMIM:614559, OMIM:616289	
2	***AFG3L2***	AD, AR	OMIM:610246, OMIM:614487, OMIM:618977	
3	***DNM1L***	AD, AR	OMIM:610708, OMIM:614388	
4	***MFN2***	AD, AR	OMIM:151800, OMIM:601152, OMIM:609260 +1	
5	***NR2F1***	AD	OMIM:615722	
6	***OPA1***	AD, AR	OMIM:125250, OMIM:165500, OMIM:210000 +1	
7	***OPA3***	AD, AR	OMIM:165300, OMIM:258501	
8	***RTN4IP1***	AR	OMIM:616732	
9	***SPG7***	AD, AR	OMIM:607259	
10	***SSBP1***	AD	OMIM:165510	
11	***TMEM126A***	AR	OMIM:612989	
12	***WFS1***	AD, AR	OMIM:116400, OMIM:125853, OMIM:222300 +2	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY | 18 GENES

Albinism

Genes for oculocutaneous and ocular albinism and syndromic forms

Panel details

Panel code	TBO-GP-119
Clinical area	Ophthalmology
Number of genes	18
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*AP3B1*	AR	OMIM:608233	
2	*AP3D1*	AR	OMIM:617050	
3	*BLOC1S3*	AR	OMIM:614077	
4	*BLOC1S6*	AR	OMIM:614171	
5	*DTNBP1*	AR	OMIM:614076	
6	*GPR143*	XLR	OMIM:300500, OMIM:300814	
7	*HPS1*	AR	OMIM:203300	
8	*HPS3*	AR	OMIM:614072	
9	*HPS4*	AR	OMIM:614073	
10	*HPS5*	AR	OMIM:614074	
11	*HPS6*	AR	OMIM:614075	
12	*LRMDA*	AR	OMIM:615179	
13	*LYST*	AR	OMIM:214500	
14	*OCA2*	AR	OMIM:203200	
15	*SLC24A5*	AR	OMIM:113750	

16	***SLC45A2***	AR	OMIM:606574
17	***TYR***	AR	OMIM:203100, OMIM:606952
18	***TYRP1***	AR	OMIM:203290

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY 17 GENES

Microphthalmia, anophthalmia and coloboma

Genes for developmental eye disorders

Panel details

Panel code	TBO-GP-120
Clinical area	Ophthalmology
Number of genes	17
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ALDH1A3*	AR	OMIM:615113	
2	*BMP4*	AD	OMIM:600625, OMIM:607932	
3	*FOXE3*	AD, AR	OMIM:610256, OMIM:617349	
4	*GDF3*	AD	OMIM:613702, OMIM:613703, OMIM:613704	
5	*GDF6*	AD, AR	OMIM:118100, OMIM:613094, OMIM:613703 +2	
6	*MAB21L2*	AD, AR	OMIM:615877	
7	*MITF*	AD, AR	OMIM:103500, OMIM:193510, OMIM:614456 +1	
8	*OTX2*	AD	OMIM:610125, OMIM:613986	
9	*PAX6*	AD	OMIM:106210, OMIM:120200, OMIM:120430 +5	
10	*RARB*	AD, AR	OMIM:615524	
11	*RAX*	AR	OMIM:611038	
12	*SMOC1*	AR	OMIM:206920	
13	*SOX2*	AD	OMIM:206900	
14	*STRA6*	AR	OMIM:601186	
15	*TENM3*	AR	OMIM:615145	

16	***VSX2***	AR	OMIM:610092, OMIM:610093
17	***YAP1***	AD	OMIM:120433

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY | 14 GENES

Congenital stationary night blindness

Genes for complete and incomplete CSNB

Panel details

Panel code	TBO-GP-121
Clinical area	Ophthalmology
Number of genes	14
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CABP4***	AR	OMIM:610427	
2	***CACNA1F***	XLR	OMIM:300071, OMIM:300476, OMIM:300600	
3	***GNAT1***	AD, AR	OMIM:610444, OMIM:616389	
4	***GPR179***	AR	OMIM:614565	
5	***GRK1***	AR	OMIM:613411	
6	***GRM6***	AR	OMIM:257270	
7	***LRIT3***	AR	OMIM:615058	
8	***NYX***	XLR	OMIM:310500	
9	***PDE6B***	AD, AR	OMIM:163500, OMIM:613801	
10	***RDH5***	AD, AR	OMIM:136880	
11	***RHO***	AD, AR	OMIM:136880, OMIM:610445, OMIM:613731	
12	***SAG***	AD, AR	OMIM:258100, OMIM:613758, OMIM:620228	
13	***SLC24A1***	AR	OMIM:613830	
14	***TRPM1***	AR	OMIM:613216	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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OPHTHALMOLOGY · 11 GENES

Corneal dystrophies

Genes for epithelial, stromal and endothelial corneal dystrophies

Panel details

Panel code	TBO-GP-122
Clinical area	Ophthalmology
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CHST6***	AR	OMIM:217800	
2	***COL8A2***	AD	OMIM:136800, OMIM:609140	
3	***GSN***	AD	OMIM:105120	
4	***KRT12***	AD	OMIM:122100	
5	***KRT3***	AD	OMIM:618767	
6	***PIKFYVE***	AD	OMIM:121850	
7	***SLC4A11***	AD, AR	OMIM:217400, OMIM:217700, OMIM:613268	
8	***TACSTD2***	AR	OMIM:204870	
9	***TGFBI***	AD	OMIM:121820, OMIM:121900, OMIM:122200 +4	
10	***UBIAD1***	AD	OMIM:121800	
11	***ZEB1***	AD	OMIM:609141, OMIM:613270	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HEARING AND AIRWAY | 32 GENES

Non-syndromic hearing loss

Genes for congenital and progressive non-syndromic hearing loss

Panel details

Panel code	TBO-GP-123
Clinical area	Hearing and airway
Number of genes	32
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ACTG1*	AD	OMIM:604717, OMIM:614583	
2	*CDH23*	AD, AR	OMIM:601067, OMIM:601386, OMIM:617540	
3	*CLDN14*	AR	OMIM:614035	
4	*COCH*	AD, AR	OMIM:601369, OMIM:618094	
5	*COL11A2*	AD, AR	OMIM:184840, OMIM:215150, OMIM:601868 +2	
6	*DIAPH1*	AD, AR	OMIM:124900, OMIM:616632	
7	*EYA4*	AD	OMIM:601316, OMIM:605362	
8	*GJB2*	AD, AR, XLR	OMIM:124500, OMIM:148210, OMIM:148350 +5	
9	*GJB6*	AD, AR, XLR	OMIM:129500, OMIM:220290, OMIM:304400 +2	
10	*GRXCR1*	AR	OMIM:613285	
11	*GSDME*	AD	OMIM:600994	
12	*KCNQ4*	AD	OMIM:600101	
13	*LOXHD1*	AR	OMIM:613079	

14	***MYO15A***	AR	OMIM:600316	
15	***MYO3A***	AR	OMIM:607101	
16	***MYO6***	AD, AR	OMIM:606346, OMIM:607821	
17	***MYO7A***	AD, AR	OMIM:276900, OMIM:600060, OMIM:601317	
18	***OTOA***	AR	OMIM:607039	Pseudogene region
19	***OTOF***	AR	OMIM:601071	
20	***PCDH15***	AR	OMIM:601067, OMIM:602083, OMIM:609533	
21	***POU3F4***	XLR	OMIM:304400	
22	***POU4F3***	AD	OMIM:602459	
23	***PTPRQ***	AD, AR	OMIM:613391, OMIM:617663	
24	***SLC26A4***	AR	OMIM:274600, OMIM:600791	
25	***STRC***	AR	OMIM:603720, OMIM:611102, OMIM:612997	Pseudogene; dosage analysis
26	***TECTA***	AD, AR	OMIM:601543, OMIM:603629	
27	***TMC1***	AD, AR	OMIM:600974, OMIM:606705	
28	***TMPRSS3***	AR	OMIM:601072	
29	***TPRN***	AR	OMIM:613307	
30	***TRIOBP***	AR	OMIM:609823	
31	***USH1C***	AR	OMIM:276900, OMIM:276904, OMIM:602092	
32	***WFS1***	AD, AR	OMIM:116400, OMIM:125853, OMIM:222300 +2	

Genes needing specific methods

- ***OTOA***: Pseudogene region
- ***STRC***: Pseudogene; dosage analysis

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

This product uses the Human Phenotype Ontology (Köhler et al., hpo.jax.org). © Tribro-Omics · TBO-GP-123 · 11 November 2026

HEARING AND AIRWAY | 19 GENES

Syndromic hearing loss

Genes for Waardenburg, Pendred, branchio-oto-renal, CHARGE, Alport and Jervell-Lange-Nielsen syndromes

Panel details

Panel code	TBO-GP-124
Clinical area	Hearing and airway
Number of genes	19
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CHD7***	AD	OMIM:214800, OMIM:612370	
2	***COL4A3***	AD, AR	OMIM:104200, OMIM:620320, OMIM:620536	
3	***COL4A4***	AD, AR	OMIM:141200, OMIM:203780	
4	***COL4A5***	XLD	OMIM:301050	
5	***EDN3***	AD, AR	OMIM:613265, OMIM:613712	
6	***EDNRB***	AD, AR	OMIM:277580, OMIM:600155, OMIM:600501	
7	***EYA1***	AD	OMIM:113650, OMIM:166780, OMIM:602588	
8	***FOXI1***	AR	OMIM:274600, OMIM:600791	
9	***KCNE1***	AD, AR	OMIM:612347, OMIM:613695	
10	***KCNJ10***	AR	OMIM:274600, OMIM:600791, OMIM:612780	
11	***KCNQ1***	AD, AR	OMIM:130650, OMIM:192500, OMIM:220400 +2	
12	***MITF***	AD, AR	OMIM:103500, OMIM:193510, OMIM:614456 +1	
13	***PAX3***	AD, AR	OMIM:122880, OMIM:148820, OMIM:193500 +1	
14	***SIX1***	AD	OMIM:113650, OMIM:605192, OMIM:608389	

15	***SIX5***	AD	OMIM:610896
16	***SLC26A4***	AR	OMIM:274600, OMIM:600791
17	***SNAI2***	—	2 (Orphanet)
18	***SOX10***	AD	OMIM:609136, OMIM:611584, OMIM:613266
19	***WFS1***	AD, AR	OMIM:116400, OMIM:125853, OMIM:222300 +2

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HEARING AND AIRWAY | 29 GENES

Primary ciliary dyskinesia

Genes for primary ciliary dyskinesia and related motile ciliopathies

Panel details

Panel code	TBO-GP-125
Clinical area	Hearing and airway
Number of genes	29
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*CCDC39*	AR	OMIM:613807	
2	*CCDC40*	AR	OMIM:613808	
3	*CCNO*	AR	OMIM:615872	
4	*CFAP298*	AR	OMIM:615500	
5	*DNAAF1*	AR	OMIM:613193	
6	*DNAAF11*	AR	OMIM:614935	
7	*DNAAF19*	AR	OMIM:614679	
8	*DNAAF2*	AR	OMIM:612518	
9	*DNAAF3*	AR	OMIM:606763	
10	*DNAAF4*	AD, AR	OMIM:127700, OMIM:615482	
11	*DNAAF5*	AR	OMIM:614874	
12	*DNAAF6*	XLR	OMIM:300991	
13	*DNAH11*	AR	OMIM:611884	
14	*DNAH5*	AR	OMIM:608644	
15	*DNAI1*	AR	OMIM:244400	

16	***DNAI2***	AR	OMIM:612444
17	***DRC1***	AR	OMIM:615294, OMIM:620222
18	***GAS8***	AR	OMIM:616726
19	***HYDIN***	AR	OMIM:608647
20	***MCIDAS***	AR	OMIM:618695
21	***ODAD1***	AR	OMIM:615067
22	***ODAD2***	AR	OMIM:615451
23	***ODAD3***	AR	OMIM:616037
24	***ODAD4***	AR	OMIM:617092
25	***RSPH1***	AR	OMIM:615481
26	***RSPH4A***	AR	OMIM:612649
27	***RSPH9***	AR	OMIM:612650
28	***SPAG1***	AR	OMIM:615505
29	***ZMYND10***	AR	OMIM:615444

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HEARING AND AIRWAY | 4 GENES

Cystic fibrosis and CFTR-related disorders

CFTR with epithelial sodium channel genes for CF-like presentations

Panel details

Panel code	TBO-GP-126
Clinical area	Hearing and airway
Number of genes	4
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CFTR***	AD, AR	OMIM:167800, OMIM:211400, OMIM:219700 +1	
2	***SCNN1A***	AD, AR	OMIM:264350, OMIM:613021, OMIM:618126	
3	***SCNN1B***	AD, AR	OMIM:177200, OMIM:211400, OMIM:620125	
4	***SCNN1G***	AD, AR	OMIM:613071, OMIM:618114, OMIM:620126	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HEARING AND AIRWAY | 7 GENES

Hereditary pancreatitis

Genes for hereditary and idiopathic chronic pancreatitis

Panel details

Panel code	TBO-GP-127
Clinical area	Hearing and airway
Number of genes	7
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CASR***	AD, AR	`OMIM:145980, OMIM:239200, OMIM:601198 +1`	
2	***CEL***	AD	`OMIM:609812`	
3	***CFTR***	AD, AR	`OMIM:167800, OMIM:211400, OMIM:219700 +1`	
4	***CPA1***	—	`1 (Orphanet)`	
5	***CTRC***	AD	`OMIM:167800`	
6	***PRSS1***	AD	`OMIM:167800`	
7	***SPINK1***	AD, AR	`OMIM:167800, OMIM:608189`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY 8 GENES

Haemoglobinopathies and thalassaemia

Globin genes and modifiers (alpha-globin deletions need dosage analysis)

Panel details

Panel code	TBO-GP-128
Clinical area	Haematology
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ATRX***	XLR, XLD	OMIM:300448, OMIM:301040, OMIM:309580	
2	***HBA1***	AD	OMIM:140700, OMIM:604131, OMIM:613978 +2	Deletions need dosage analysis
3	***HBA2***	AD	OMIM:140700, OMIM:604131, OMIM:613978 +1	Deletions need dosage analysis
4	***HBB***	AD, AR	OMIM:140700, OMIM:141749, OMIM:603902 +5	
5	***HBD***	—	1 (Orphanet)	
6	***HBG1***	AD	OMIM:141749	
7	***HBG2***	AD	OMIM:141749, OMIM:613977	
8	***KLF1***	AD	OMIM:613673	

Genes needing specific methods

- ***HBA1***: Deletions need dosage analysis
- ***HBA2***: Deletions need dosage analysis

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY 24 GENES

Inherited bleeding disorders

Genes for coagulation factor deficiencies, von Willebrand disease and platelet function disorders

Panel details

Panel code	TBO-GP-129
Clinical area	Haematology
Number of genes	24
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*F10*	AR	OMIM:227600	
2	*F11*	AD, AR	OMIM:612416	
3	*F13A1*	AD, AR	OMIM:188050, OMIM:613225	
4	*F13B*	AR	OMIM:613235	
5	*F2*	AD, AR	OMIM:188050, OMIM:601367, OMIM:613679 +1	
6	*F5*	AD, AR	OMIM:188055, OMIM:227400, OMIM:600880 +2	
7	*F7*	AR	OMIM:227500	
8	*F8*	XLR, XLD	OMIM:301071, OMIM:306700	Intron 22 and intron 1 inversions need specific assay
9	*F9*	XLR	OMIM:300807, OMIM:306900	
10	*FGA*	AD, AR	OMIM:105200, OMIM:202400	
11	*FGB*	AR	OMIM:202400	
12	*FGG*	AR	OMIM:202400	
13	*GGCX*	AR	OMIM:277450, OMIM:610842	

14	***GP1BA***	AD, AR	OMIM:153670, OMIM:177820, OMIM:231200 +1
15	***GP1BB***	AR	OMIM:231200
16	***GP9***	AR	OMIM:231200
17	***ITGA2B***	AD, AR	OMIM:187800, OMIM:273800
18	***ITGB3***	AD, AR	OMIM:619267, OMIM:619271
19	***LMAN1***	AR	OMIM:227300
20	***MCFD2***	AR	OMIM:613625
21	***SERPINE1***	AD, AR	OMIM:613329
22	***SERPINF2***	AR	OMIM:262850
23	***VKORC1***	AD, AR	OMIM:122700, OMIM:607473
24	***VWF***	AD, AR	OMIM:193400, OMIM:277480, OMIM:613554

Genes needing specific methods

- ***F8***: Intron 22 and intron 1 inversions need specific assay

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY | 5 GENES

Inherited thrombophilia

Genes for natural anticoagulant deficiencies and common thrombophilic variants

Panel details

Panel code	TBO-GP-130
Clinical area	Haematology
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***F2***	AD, AR	`OMIM:188050, OMIM:601367, OMIM:613679 +1`	
2	***F5***	AD, AR	`OMIM:188055, OMIM:227400, OMIM:600880 +2`	
3	***PROC***	AD, AR	`OMIM:176860, OMIM:612304`	
4	***PROS1***	AD, AR	`OMIM:612336, OMIM:614514`	
5	***SERPINC1***	AD, AR	`OMIM:613118`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY 25 GENES

Inherited thrombocytopenia

Genes for inherited platelet number disorders

Panel details

Panel code	TBO-GP-131
Clinical area	Haematology
Number of genes	25
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ACTN1*	AD	OMIM:615193	
2	*ANKRD26*	AD	OMIM:188000	
3	*CYCS*	AD	OMIM:612004	
4	*DIAPH1*	AD, AR	OMIM:124900, OMIM:616632	
5	*ETV6*	AD	OMIM:601626, OMIM:616216	
6	*FLI1*	AD, AR	OMIM:617443	
7	*FLNA*	XLR, XLD	OMIM:300048, OMIM:300049, OMIM:300244 +6	
8	*GFI1B*	AD, AR	OMIM:187900	
9	*GP1BA*	AD, AR	OMIM:153670, OMIM:177820, OMIM:231200 +1	
10	*GP1BB*	AR	OMIM:231200	
11	*GP9*	AR	OMIM:231200	
12	*HOXA11*	AD	OMIM:605432	
13	*ITGA2B*	AD, AR	OMIM:187800, OMIM:273800	
14	*ITGB3*	AD, AR	OMIM:619267, OMIM:619271	
15	*MECOM*	AD	OMIM:616738	

16	***MPL***	AD, AR	OMIM:254450, OMIM:601977, OMIM:604498
17	***MYH9***	AD	OMIM:155100, OMIM:603622
18	***NBEAL2***	AR	OMIM:139090
19	***PRKACG***	AR	OMIM:616176
20	***RBM8A***	AR	OMIM:274000
21	***RUNX1***	AD	OMIM:601399, OMIM:601626
22	***SRC***	AD	OMIM:114500, OMIM:616937
23	***THPO***	AD, AR	OMIM:187950, OMIM:620478, OMIM:620481
24	***TUBB1***	AD	OMIM:613112
25	***WAS***	XLR	OMIM:300299, OMIM:301000, OMIM:313900

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY 13 GENES

Telomere biology disorders

Genes for dyskeratosis congenita and telomeropathies

Panel details

Panel code	TBO-GP-132
Clinical area	Haematology
Number of genes	13
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACD***	AD, AR	OMIM:616553	
2	***CTC1***	AR	OMIM:612199	
3	***DKC1***	XLR, XLD	OMIM:301108, OMIM:305000	
4	***NHP2***	AR	OMIM:224230, OMIM:613987	
5	***NOP10***	AD, AR	OMIM:224230, OMIM:620400, OMIM:620425	
6	***PARN***	AD, AR	OMIM:616353, OMIM:616371	
7	***POT1***	AD, AR	OMIM:615848, OMIM:620367, OMIM:620368	
8	***RTEL1***	AD, AR	OMIM:615190, OMIM:616373	
9	***STN1***	AR	OMIM:617341	
10	***TERC***	AD	OMIM:127550, OMIM:614743	Non-coding RNA gene
11	***TERT***	AD, AR	OMIM:127550, OMIM:178500, OMIM:601626 +4	
12	***TINF2***	AD	OMIM:127550, OMIM:268130, OMIM:613990	
13	***WRAP53***	AR	OMIM:613988	

Genes needing specific methods

- ***TERC***: Non-coding RNA gene

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY 19 GENES

Diamond-Blackfan anaemia

Ribosomal protein and related genes for Diamond-Blackfan anaemia

Panel details

Panel code	TBO-GP-133
Clinical area	Haematology
Number of genes	19
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***GATA1***	XLR	OMIM:159595, OMIM:300367, OMIM:300835 +2	
2	***RPL11***	AD	OMIM:612562	
3	***RPL15***	AD	OMIM:615550	
4	***RPL18***	AD	OMIM:618310	
5	***RPL26***	AD	OMIM:614900	
6	***RPL27***	AD	OMIM:617408	
7	***RPL35***	AD	OMIM:618312	
8	***RPL35A***	AD	OMIM:612528	
9	***RPL5***	AD	OMIM:612561	
10	***RPS10***	AD	OMIM:613308	
11	***RPS15A***	AD	OMIM:618313	
12	***RPS17***	AD	OMIM:612527	
13	***RPS19***	AD	OMIM:105650	
14	***RPS24***	AD	OMIM:610629	
15	***RPS26***	AD	OMIM:613309	

16	***RPS27***	AD	OMIM:617409
17	***RPS29***	AD	OMIM:615909
18	***RPS7***	AD	OMIM:612563
19	***TSR2***	XLR	OMIM:300946

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY 17 GENES

Congenital haemolytic anaemia

Red cell membrane, enzyme and hydration disorder genes

Panel details

Panel code	TBO-GP-134
Clinical area	Haematology
Number of genes	17
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ALDOA***	AR	OMIM:611881	
2	***ANK1***	AD, AR	OMIM:182900	
3	***EPB41***	AD, AR	OMIM:611804	
4	***EPB42***	AR	OMIM:612690	
5	***G6PD***	XLD	OMIM:300908	
6	***GPI***	AR	OMIM:613470	
7	***GSS***	AR	OMIM:231900, OMIM:266130	
8	***HK1***	AD, AR	OMIM:235700, OMIM:605285, OMIM:617460 +1	
9	***KCNN4***	AD	OMIM:616689	
10	***NT5C3A***	AR	OMIM:266120	
11	***PGK1***	XLR	OMIM:300653	
12	***PIEZO1***	AD, AR	OMIM:194380, OMIM:616843	
13	***PKLR***	AD, AR	OMIM:102900, OMIM:266200	
14	***SLC4A1***	AD, AR	OMIM:166900, OMIM:179800, OMIM:185020 +2	
15	***SPTA1***	AD, AR	OMIM:130600, OMIM:266140, OMIM:270970	

16	***SPTB***	AD	`OMIM:616649, OMIM:617948`
17	***TPI1***	AR	`OMIM:615512`

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY | 14 GENES

Congenital dyserythropoietic and sideroblastic anaemia

Genes for CDA and congenital sideroblastic anaemias

Panel details

Panel code	TBO-GP-135
Clinical area	Haematology
Number of genes	14
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCB7***	XLR	OMIM:301310	
2	***ALAS2***	XLR, XLD	OMIM:300751, OMIM:300752	
3	***CDAN1***	AR	OMIM:224120	
4	***CDIN1***	AR	OMIM:615631	
5	***GLRX5***	AR	OMIM:616859, OMIM:616860	
6	***HSPA9***	AD, AR	OMIM:182170, OMIM:616854	
7	***KIF23***	AD	OMIM:105600	
8	***KLF1***	AD	OMIM:613673	
9	***PUS1***	AR	OMIM:600462	
10	***SEC23B***	AD, AR	OMIM:224100, OMIM:616858	
11	***SLC19A2***	AR	OMIM:249270	
12	***SLC25A38***	AR	OMIM:205950	
13	***TRNT1***	AR	OMIM:616084, OMIM:616959	
14	***YARS2***	AR	OMIM:613561	

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY 14 GENES

Severe congenital neutropenia

Genes for congenital neutropenias

Panel details

Panel code	TBO-GP-136
Clinical area	Haematology
Number of genes	14
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CLPB***	AD, AR	OMIM:616271, OMIM:619813, OMIM:619835	
2	***CSF3R***	AD, AR	OMIM:162830, OMIM:617014	
3	***CXCR4***	AD	OMIM:193670	
4	***ELANE***	AD	OMIM:162800, OMIM:202700	
5	***G6PC3***	AR	OMIM:612541	
6	***GFI1***	AD	OMIM:607847, OMIM:613107	
7	***HAX1***	AR	OMIM:610738	
8	***JAGN1***	AR	OMIM:616022	
9	***SBDS***	AR	OMIM:260400, OMIM:609135	
10	***SLC37A4***	AD, AR	OMIM:232220, OMIM:232240, OMIM:619525	
11	***SRP54***	AD, AR	OMIM:260400, OMIM:618752	
12	***TAFAZZIN***	XLR	OMIM:302060	
13	***VPS45***	AR	OMIM:615285	
14	***WAS***	XLR	OMIM:300299, OMIM:301000, OMIM:313900	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY 13 GENES

Haemophagocytic lymphohistiocytosis

Genes for familial and syndromic HLH

Panel details

Panel code	TBO-GP-137
Clinical area	Haematology
Number of genes	13
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*AP3B1*	AR	OMIM:608233	
2	*CD27*	AR	OMIM:615122	
3	*ITK*	AR	OMIM:613011	
4	*LYST*	AR	OMIM:214500	
5	*MAGT1*	XLR	OMIM:300853, OMIM:301031	
6	*NLRC4*	AD	OMIM:616050, OMIM:616115	
7	*PRF1*	AR	OMIM:603553, OMIM:605027, OMIM:609135	
8	*RAB27A*	AR	OMIM:607624	
9	*SH2D1A*	XLR	OMIM:308240	
10	*STX11*	AR	OMIM:603552	
11	*STXBP2*	AR	OMIM:613101	
12	*UNC13D*	AR	OMIM:608898	
13	*XIAP*	XLR	OMIM:300635, OMIM:308240	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY | 9 GENES

Porphyrias

Haem biosynthesis genes for acute and cutaneous porphyrias

Panel details

Panel code	TBO-GP-138
Clinical area	Haematology
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ALAD***	AR	OMIM:612740	
2	***ALAS2***	XLR, XLD	OMIM:300751, OMIM:300752	
3	***CLPX***	AD	OMIM:618015	
4	***CPOX***	AD, AR	OMIM:121300, OMIM:618892	
5	***FECH***	AR	OMIM:177000	
6	***HMBS***	AD, AR	OMIM:176000, OMIM:620704, OMIM:620711	
7	***PPOX***	AD, AR	OMIM:176200, OMIM:620483	
8	***UROD***	AD, AR	OMIM:176100	
9	***UROS***	AR	OMIM:263700	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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HAEMATOLOGY 10 GENES

Methaemoglobinaemia and erythrocytosis

Genes for congenital methaemoglobinaemia and familial erythrocytosis

Panel details

Panel code	TBO-GP-139
Clinical area	Haematology
Number of genes	10
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***BPGM***	AR	OMIM:222800	
2	***CYB5A***	AR	OMIM:250790	
3	***CYB5R3***	AR	OMIM:250800	
4	***EGLN1***	AD	OMIM:609820	
5	***EPAS1***	AD	OMIM:611783	
6	***EPOR***	AD	OMIM:133100	
7	***HBA1***	AD	OMIM:140700, OMIM:604131, OMIM:613978 +2	Deletions need dosage analysis
8	***HBA2***	AD	OMIM:140700, OMIM:604131, OMIM:613978 +1	Deletions need dosage analysis
9	***HBB***	AD, AR	OMIM:140700, OMIM:141749, OMIM:603902 +5	
10	***VHL***	AD, AR	OMIM:144700, OMIM:171300, OMIM:193300 +1	

Genes needing specific methods

- ***HBA1***: Deletions need dosage analysis
- ***HBA2***: Deletions need dosage analysis

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY | 18 GENES

Severe combined immunodeficiency

Genes for SCID and leaky SCID

Panel details

Panel code	TBO-GP-140
Clinical area	Immunology
Number of genes	18
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ADA*	AR	OMIM:102700	
2	*AK2*	AR	OMIM:267500	
3	*CD247*	AR	OMIM:610163	
4	*CD3D*	AR	OMIM:615617	
5	*CD3E*	AR	OMIM:615615	
6	*CORO1A*	AR	OMIM:615401	
7	*DCLRE1C*	AR	OMIM:602450, OMIM:603554	
8	*IL2RG*	XLR	OMIM:300400, OMIM:312863	
9	*IL7R*	AR	OMIM:608971	
10	*JAK3*	AR	OMIM:600802	
11	*LIG4*	AR	OMIM:254500, OMIM:606593	
12	*NHEJ1*	—	1 (Orphanet)	
13	*PRKDC*	AR	OMIM:615966	
14	*PTPRC*	AR	OMIM:619924	
15	*RAG1*	AR	OMIM:233650, OMIM:601457, OMIM:603554	

16	***RAG2***	AR	OMIM:233650, OMIM:601457, OMIM:603554	
17	***RMRP***	AR	OMIM:250250, OMIM:250460, OMIM:607095	Non-coding RNA gene
18	***ZAP70***	AR	OMIM:269840, OMIM:617006	

Genes needing specific methods

- ***RMRP***: Non-coding RNA gene

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY | 19 GENES

Common variable immunodeficiency

Genes for predominantly antibody deficiencies

Panel details

Panel code	TBO-GP-141
Clinical area	Immunology
Number of genes	19
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AICDA***	AR	OMIM:605258	
2	***BTK***	XLR	OMIM:300755, OMIM:307200	
3	***CD19***	AD, AR	OMIM:240500, OMIM:613493	
4	***CD40***	AR	OMIM:606843	
5	***CD40LG***	XLR	OMIM:308230	
6	***CD81***	AR	OMIM:613496	
7	***CR2***	AD, AR	OMIM:240500, OMIM:614699	
8	***CTLA4***	AD	OMIM:140300, OMIM:152700, OMIM:616100	
9	***ICOS***	AD, AR	OMIM:240500, OMIM:607594	
10	***IKZF1***	AD	OMIM:616873	
11	***LRBA***	AR	OMIM:614700	
12	***MS4A1***	AR	OMIM:613495	
13	***NFKB1***	AD	OMIM:616576	
14	***NFKB2***	AD	OMIM:615577	
15	***PIK3CD***	AD, AR	OMIM:613328, OMIM:615513, OMIM:619281	

16	***PIK3R1***	AD, AR	OMIM:269880, OMIM:615214, OMIM:616005
17	***TNFRSF13B***	AD, AR	OMIM:240500, OMIM:609529
18	***TNFRSF13C***	AD, AR	OMIM:240500, OMIM:613494
19	***UNG***	AR	OMIM:608106

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY | 6 GENES

Chronic granulomatous disease

NADPH oxidase complex genes

Panel details

Panel code	TBO-GP-142
Clinical area	Immunology
Number of genes	6
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CYBA***	AR	OMIM:233690	
2	***CYBB***	XLR	OMIM:300645, OMIM:306400	
3	***CYBC1***	AR	OMIM:618935	
4	***NCF1***	AR	OMIM:233700	Pseudogenes NCF1B/NCF1C
5	***NCF2***	AR	OMIM:233710	
6	***NCF4***	AR	OMIM:613960	

Genes needing specific methods

- ***NCF1***: Pseudogenes NCF1B/NCF1C

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY | 13 GENES

Autoinflammatory and periodic fever syndromes

Genes for monogenic autoinflammatory diseases

Panel details

Panel code	TBO-GP-143
Clinical area	Immunology
Number of genes	13
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ADA2*	AR	OMIM:182410, OMIM:615688	
2	*IL1RN*	AR	OMIM:612852, OMIM:613659	
3	*IL36RN*	AR	OMIM:614204	
4	*LPIN2*	AR	OMIM:609628	
5	*MEFV*	AD, AR	OMIM:134610, OMIM:249100, OMIM:608068	
6	*MVK*	AD, AR	OMIM:175900, OMIM:260920, OMIM:610377	
7	*NLRP12*	AD	OMIM:611762	
8	*NLRP3*	AD	OMIM:120100, OMIM:148200, OMIM:191900 +2	
9	*NOD2*	AD	OMIM:186580, OMIM:266600, OMIM:617321	
10	*PSMB8*	AR	OMIM:256040	
11	*PSTPIP1*	AD	OMIM:601979, OMIM:604416	
12	*TNFAIP3*	AD	OMIM:616744	
13	*TNFRSF1A*	AD	OMIM:142680	

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY 18 GENES

Complement deficiencies

Genes for deficiencies of the classical, lectin and alternative pathways

Panel details

Panel code	TBO-GP-144
Clinical area	Immunology
Number of genes	18
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***C1QA***	AR	OMIM:613652	
2	***C1QB***	AR	OMIM:620321	
3	***C1QC***	AR	OMIM:620322	
4	***C1R***	AD	OMIM:130080	
5	***C1S***	AD	OMIM:613783, OMIM:617174	
6	***C2***	AR	OMIM:217000	
7	***C3***	AD, AR	OMIM:612925, OMIM:613779	
8	***C5***	AR	OMIM:609536	
9	***C6***	AR	OMIM:612446	
10	***C7***	AR	OMIM:610102	
11	***C8A***	AR	OMIM:613790	
12	***C8B***	AR	OMIM:613789	
13	***C9***	AD	OMIM:613825, OMIM:615591	
14	***CFD***	AR	OMIM:613912	
15	***CFH***	AD, AR	OMIM:126700, OMIM:235400, OMIM:609814 +1	

16	***CFI***	AD, AR	`OMIM:610984, OMIM:612923, OMIM:615439`
17	***CFP***	XLR	`OMIM:312060`
18	***MASP2***	AR	`OMIM:613791`

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY | 7 GENES

Hereditary angioedema

Genes for hereditary angioedema with normal and low C1 inhibitor

Panel details

Panel code	TBO-GP-145
Clinical area	Immunology
Number of genes	7
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ANGPT1***	AD	OMIM:619361	
2	***F12***	AD, AR	OMIM:234000, OMIM:610618	
3	***HS3ST6***	AD	OMIM:619367	
4	***KNG1***	AD, AR	OMIM:228960, OMIM:619363	
5	***MYOF***	AD	OMIM:619366	
6	***PLG***	AD, AR	OMIM:217090, OMIM:619360	
7	***SERPING1***	AD, AR	OMIM:106100, OMIM:120790	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY · 11 GENES

Immune dysregulation

Genes for IPEX-like, ALPS and autoimmune polyendocrinopathy disorders

Panel details

Panel code	TBO-GP-146
Clinical area	Immunology
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AIRE***	AD, AR	OMIM:240300	
2	***CASP10***	AD	OMIM:603909, OMIM:605027, OMIM:613659	
3	***CTLA4***	AD	OMIM:140300, OMIM:152700, OMIM:616100	
4	***FAS***	AD	OMIM:601859	
5	***FASLG***	AD	OMIM:211980, OMIM:601859	
6	***FOXP3***	XLR	OMIM:304790	
7	***IL2RA***	AR	OMIM:601942, OMIM:606367	
8	***LRBA***	AR	OMIM:614700	
9	***STAT1***	AD, AR	OMIM:613796, OMIM:614162, OMIM:614892	
10	***STAT3***	AD	OMIM:147060, OMIM:615952	
11	***STAT5B***	AD, AR	OMIM:245590, OMIM:618985	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY · 13 GENES

Mendelian susceptibility to mycobacterial disease

Genes of the IL-12/IFN-gamma axis

Panel details

Panel code	TBO-GP-147
Clinical area	Immunology
Number of genes	13
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CYBB***	XLR	OMIM:300645, OMIM:306400	
2	***IFNGR1***	AD, AR	OMIM:209950, OMIM:600263, OMIM:615978	
3	***IFNGR2***	AR	OMIM:614889	
4	***IKBKG***	XLR, XLD	OMIM:300291, OMIM:300636, OMIM:301081 +1	Pseudogene IKBKGP1
5	***IL12B***	AR	OMIM:614890	
6	***IL12RB1***	AR	OMIM:614891	
7	***IRF8***	AD, AR	OMIM:226990, OMIM:614893	
8	***ISG15***	AR	OMIM:616126	
9	***JAK1***	AD	OMIM:618999	
10	***RORC***	AR	OMIM:616622	
11	***SPPL2A***	AR	OMIM:619549	
12	***STAT1***	AD, AR	OMIM:613796, OMIM:614162, OMIM:614892	
13	***TYK2***	AR	OMIM:611521	

Genes needing specific methods

- ***IKBKG***: Pseudogene IKBKGP1

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY | 9 GENES

Hyper-IgE syndromes

Genes for autosomal dominant and recessive hyper-IgE syndromes

Panel details

Panel code	TBO-GP-148
Clinical area	Immunology
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CARD11***	AD, AR	OMIM:615206, OMIM:616452, OMIM:617638	
2	***DOCK8***	AR	OMIM:243700	
3	***IL6R***	AR	OMIM:618944	
4	***IL6ST***	AD, AR	OMIM:618523, OMIM:619750, OMIM:619751 +1	
5	***PGM3***	AR	OMIM:615816	
6	***SPINK5***	AR	OMIM:256500	
7	***STAT3***	AD	OMIM:147060, OMIM:615952	
8	***TYK2***	AR	OMIM:611521	
9	***ZNF341***	AR	OMIM:618282	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY · 14 GENES

Inborn errors of antiviral immunity

Genes predisposing to severe viral infection

Panel details

Panel code	TBO-GP-149
Clinical area	Immunology
Number of genes	14
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*GATA2*	AD	OMIM:601626, OMIM:614038, OMIM:614172 +1	
2	*IFNAR1*	AR	OMIM:619935	
3	*IFNAR2*	AR	OMIM:616669	
4	*IRF3*	AD	OMIM:616532	
5	*IRF7*	AR	OMIM:616345	
6	*IRF9*	AR	OMIM:618648	
7	*POLR3A*	AR	OMIM:264090, OMIM:607694	
8	*STAT1*	AD, AR	OMIM:613796, OMIM:614162, OMIM:614892	
9	*STAT2*	AR	OMIM:616636, OMIM:618886	
10	*TBK1*	AD, AR	OMIM:616439, OMIM:617900, OMIM:620880	
11	*TICAM1*	AD, AR	OMIM:614850	
12	*TLR3*	AD, AR	OMIM:613002	
13	*TRAF3*	AD	OMIM:614849	
14	*UNC93B1*	AR	OMIM:610551	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY | 9 GENES

Agammaglobulinaemia

Genes for X-linked and autosomal agammaglobulinaemia

Panel details

Panel code	TBO-GP-150
Clinical area	Immunology
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***BLNK***	AR	OMIM:613502	
2	***BTK***	XLR	OMIM:300755, OMIM:307200	
3	***CD79A***	AR	OMIM:613501	
4	***CD79B***	AR	OMIM:612692	
5	***IGHM***	AR	OMIM:601495	
6	***IGLL1***	AR	OMIM:613500	
7	***LRRC8A***	AD	OMIM:613506	
8	***PIK3R1***	AD, AR	OMIM:269880, OMIM:615214, OMIM:616005	
9	***TCF3***	AD, AR	OMIM:616941, OMIM:619824	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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IMMUNOLOGY | 8 GENES

Combined immunodeficiency with syndromic features

Genes for Wiskott-Aldrich, DOCK8 deficiency and DNA repair immunodeficiencies

Panel details

Panel code	TBO-GP-151
Clinical area	Immunology
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ARPC1B***	AR	OMIM:617718	
2	***ATM***	AD, AR	OMIM:114480, OMIM:208900	
3	***BLM***	AR	OMIM:210900	
4	***DOCK2***	AR	OMIM:616433	
5	***DOCK8***	AR	OMIM:243700	
6	***NBN***	AR	OMIM:251260, OMIM:609135, OMIM:613065	
7	***WAS***	XLR	OMIM:300299, OMIM:301000, OMIM:313900	
8	***WIPF1***	AR	OMIM:614493	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 20 GENES

Osteogenesis imperfecta

Genes for dominant and recessive osteogenesis imperfecta

Panel details

Panel code	TBO-GP-152
Clinical area	Skeletal and connective tissue
Number of genes	20
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*BMP1*	AR	OMIM:614856	
2	*CCDC134*	AR	OMIM:619795	
3	*COL1A1*	AD	OMIM:114000, OMIM:130060, OMIM:166200 +5	
4	*COL1A2*	AD, AR	OMIM:166210, OMIM:166220, OMIM:166710 +4	
5	*CREB3L1*	AR	OMIM:616229	
6	*CRTAP*	AR	OMIM:610682	
7	*FKBP10*	AR	OMIM:259450, OMIM:610968	
8	*IFITM5*	AD	OMIM:610967	
9	*KDELR2*	AR	OMIM:619131	
10	*MBTPS2*	XLR	OMIM:300918, OMIM:301014, OMIM:308205 +1	
11	*MESD*	AR	OMIM:618644	
12	*P3H1*	AR	OMIM:610915	
13	*PLS3*	—	OMIM:306950	
14	*PPIB*	AR	OMIM:259440	
15	*SERPINF1*	AR	OMIM:613982	

16	***SERPINH1***	AR	OMIM:610504, OMIM:613848
17	***SP7***	AR	OMIM:613849
18	***SPARC***	AR	OMIM:616507
19	***TMEM38B***	AR	OMIM:615066
20	***WNT1***	AR	OMIM:615220

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 22 GENES

Skeletal dysplasia

Genes for common skeletal dysplasias

Panel details

Panel code	TBO-GP-153
Clinical area	Skeletal and connective tissue
Number of genes	22
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACAN***	AD, AR	OMIM:165800, OMIM:608361, OMIM:612813	
2	***ARSL***	XLR	OMIM:302950	
3	***COL10A1***	AD	OMIM:156500	
4	***COL11A1***	AD, AR	OMIM:154780, OMIM:228520, OMIM:604841 +1	
5	***COL11A2***	AD, AR	OMIM:184840, OMIM:215150, OMIM:601868 +2	
6	***COL2A1***	AD	OMIM:108300, OMIM:132450, OMIM:150600 +13	
7	***COMP***	AD	OMIM:132400, OMIM:177170, OMIM:619161	
8	***EXT1***	AD	OMIM:133700, OMIM:215300	
9	***EXT2***	AD, AR	OMIM:133701, OMIM:616682	
10	***FGFR1***	AD	OMIM:101600, OMIM:123150, OMIM:147950 +4	
11	***FGFR2***	AD	OMIM:101200, OMIM:101400, OMIM:101600 +8	
12	***FGFR3***	AD, AR	OMIM:100800, OMIM:109800, OMIM:114500 +11	
13	***IHH***	AD, AR	OMIM:112500, OMIM:607778	
14	***MATN3***	AD, AR	OMIM:140600, OMIM:607078, OMIM:608728	
15	***NPR2***	AD, AR	OMIM:602875, OMIM:615923, OMIM:616255	

16	***PTH1R***	AD, AR	OMIM:125350, OMIM:156400, OMIM:215045 +1
17	***ROR2***	AD, AR	OMIM:113000, OMIM:268310
18	***RUNX2***	AD	OMIM:119600, OMIM:156510
19	***SHOX***	AD, AR, XL	OMIM:127300, OMIM:249700, OMIM:300582
20	***SLC26A2***	AR	OMIM:222600, OMIM:226900, OMIM:256050 +1
21	***SOX9***	AD	OMIM:114290, OMIM:278850, OMIM:616425
22	***TRPV4***	AD	OMIM:113500, OMIM:156530, OMIM:168400 +7

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 14 GENES

Craniosynostosis

Genes for syndromic and non-syndromic craniosynostosis

Panel details

Panel code	TBO-GP-154
Clinical area	Skeletal and connective tissue
Number of genes	14
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ALX4***	AD, AR	OMIM:609597, OMIM:613451, OMIM:615529	
2	***EFNB1***	XLD	OMIM:304110	
3	***ERF***	AD	OMIM:600775, OMIM:617180	
4	***FGFR1***	AD	OMIM:101600, OMIM:123150, OMIM:147950 +4	
5	***FGFR2***	AD	OMIM:101200, OMIM:101400, OMIM:101600 +8	
6	***FGFR3***	AD, AR	OMIM:100800, OMIM:109800, OMIM:114500 +11	
7	***IL11RA***	AR	OMIM:614188	
8	***MSX2***	AD	OMIM:168500, OMIM:168550, OMIM:604757	
9	***POR***	AD, AR	OMIM:201750, OMIM:207410, OMIM:613571	
10	***RAB23***	AR	OMIM:201000	
11	***SKI***	AD	OMIM:182212	
12	***SMAD6***	AD	OMIM:179300, OMIM:614823, OMIM:617439	
13	***TCF12***	AD	OMIM:615314, OMIM:619718	
14	***TWIST1***	AD	OMIM:101400, OMIM:123100, OMIM:180750 +1	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 7 GENES

Stickler syndrome and type II collagenopathies

Genes for Stickler syndrome

Panel details

Panel code	TBO-GP-155
Clinical area	Skeletal and connective tissue
Number of genes	7
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***COL11A1***	AD, AR	`OMIM:154780, OMIM:228520, OMIM:604841 +1`	
2	***COL11A2***	AD, AR	`OMIM:184840, OMIM:215150, OMIM:601868 +2`	
3	***COL2A1***	AD	`OMIM:108300, OMIM:132450, OMIM:150600 +13`	
4	***COL9A1***	AD, AR	`OMIM:614134, OMIM:614135`	
5	***COL9A2***	AD, AR	`OMIM:600204, OMIM:614284`	
6	***COL9A3***	AD, AR	`OMIM:600969, OMIM:620022`	
7	***LOXL3***	AR	`OMIM:619781`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 7 GENES

Hypophosphataemic rickets

Genes for X-linked and autosomal hypophosphataemic rickets

Panel details

Panel code	TBO-GP-156
Clinical area	Skeletal and connective tissue
Number of genes	7
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CLCN5***	XLR	OMIM:300009, OMIM:300554, OMIM:308990 +1	
2	***DMP1***	AR	OMIM:241520	
3	***ENPP1***	AD, AR	OMIM:125853, OMIM:208000, OMIM:601665 +2	
4	***FAM20C***	AR	OMIM:259775	
5	***FGF23***	AD, AR	OMIM:193100, OMIM:617993	
6	***PHEX***	XLD	OMIM:307800	
7	***SLC34A3***	AR	OMIM:241530	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 11 GENES

Osteopetrosis and sclerosing bone disorders

Genes for osteopetrosis and high bone mass disorders

Panel details

Panel code	TBO-GP-157
Clinical area	Skeletal and connective tissue
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*CA2*	AR	OMIM:259730	
2	*CLCN7*	AD, AR	OMIM:166600, OMIM:611490, OMIM:618541	
3	*LEMD3*	AD	OMIM:166700	
4	*LRP5*	AD, AR	OMIM:133780, OMIM:144750, OMIM:259770 +3	
5	*OSTM1*	AR	OMIM:259720	
6	*PLEKHM1*	AD, AR	OMIM:611497, OMIM:618107	
7	*SNX10*	AR	OMIM:615085	
8	*SOST*	AD, AR	OMIM:122860, OMIM:269500	
9	*TCIRG1*	AR	OMIM:259700	
10	*TNFRSF11A*	AD, AR	OMIM:174810, OMIM:602080, OMIM:612301	
11	*TNFSF11*	AR	OMIM:259710	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 8 GENES

Limb malformations

Genes for polydactyly, split hand/foot and radial ray anomalies

Panel details

Panel code	TBO-GP-158
Clinical area	Skeletal and connective tissue
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***GLI3***	AD	OMIM:146510, OMIM:174200, OMIM:174700 +1	
2	***HOXD13***	AD	OMIM:113200, OMIM:113300, OMIM:186000 +2	
3	***LMBR1***	AD, AR	OMIM:135750, OMIM:186200, OMIM:190605 +1	
4	***SALL4***	AD	OMIM:147750, OMIM:607323	
5	***SHH***	AD	OMIM:142945, OMIM:147250, OMIM:611638	
6	***TBX5***	AD	OMIM:142900	
7	***TP63***	AD	OMIM:103285, OMIM:106260, OMIM:129400 +4	
8	***WNT10B***	AD, AR	OMIM:225300, OMIM:617073	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 11 GENES

Arthrogryposis

Genes for distal arthrogryposis and fetal akinesia spectrum

Panel details

Panel code	TBO-GP-159
Clinical area	Skeletal and connective tissue
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CHRNG***	AR	OMIM:253290, OMIM:265000	
2	***ECEL1***	AR	OMIM:615065	
3	***FBN2***	AD	OMIM:121050, OMIM:616118	
4	***MYH3***	AD, AR	OMIM:178110, OMIM:193700, OMIM:618436 +1	
5	***MYH8***	AD	OMIM:158300, OMIM:608837	
6	***NALCN***	AD, AR	OMIM:615419, OMIM:616266	
7	***PIEZO2***	AD, AR	OMIM:108145, OMIM:114300, OMIM:248700 +1	
8	***TNNI2***	AD	OMIM:601680	
9	***TNNT3***	AD	OMIM:618435	
10	***TPM2***	AD	OMIM:108120, OMIM:609285	
11	***ZC4H2***	XLR, XLD	OMIM:301041, OMIM:314580	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 10 GENES

Cutis laxa

Genes for inherited cutis laxa

Panel details

Panel code	TBO-GP-160
Clinical area	Skeletal and connective tissue
Number of genes	10
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ALDH18A1***	AD, AR	OMIM:219150, OMIM:601162, OMIM:616586 +1	
2	***ATP6V0A2***	AR	OMIM:219200, OMIM:278250	
3	***ATP6V1A***	AD, AR	OMIM:617403, OMIM:618012	
4	***ATP6V1E1***	AR	OMIM:617402	
5	***ATP7A***	XLR	OMIM:300489, OMIM:304150, OMIM:309400	
6	***EFEMP2***	AR	OMIM:614437	
7	***ELN***	AD	OMIM:123700, OMIM:185500, OMIM:194050	
8	***FBLN5***	AD, AR	OMIM:219100, OMIM:608895, OMIM:614434 +1	
9	***LTBP4***	AR	OMIM:613177	
10	***PYCR1***	AR	OMIM:612940, OMIM:614438	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito

mitochondrial.

- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 15 GENES

Short stature

Genes for monogenic short stature and growth hormone axis disorders

Panel details

Panel code	TBO-GP-161
Clinical area	Skeletal and connective tissue
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACAN***	AD, AR	OMIM:165800, OMIM:608361, OMIM:612813	
2	***FGFR3***	AD, AR	OMIM:100800, OMIM:109800, OMIM:114500 +11	
3	***GH1***	AD, AR	OMIM:173100, OMIM:262400, OMIM:262650 +1	
4	***GHR***	AD, AR	OMIM:143890, OMIM:262500, OMIM:604271	
5	***GHRHR***	AR	OMIM:618157	
6	***HMGA2***	AD	OMIM:618908	
7	***IGF1***	AR	OMIM:608747	
8	***IGF1R***	AD, AR	OMIM:270450	
9	***IGF2***	AD	OMIM:130650, OMIM:180860, OMIM:194070 +1	
10	***IGFALS***	—	OMIM:615961	
11	***NPR2***	AD, AR	OMIM:602875, OMIM:615923, OMIM:616255	
12	***POU1F1***	AD, AR	OMIM:613038	
13	***PROP1***	AR	OMIM:262600	
14	***SHOX***	AD, AR, XL	OMIM:127300, OMIM:249700, OMIM:300582	
15	***STAT5B***	AD, AR	OMIM:245590, OMIM:618985	

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 9 GENES

Multiple epiphyseal and spondyloepiphyseal dysplasia

Genes for epiphyseal dysplasias

Panel details

Panel code	TBO-GP-162
Clinical area	Skeletal and connective tissue
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CANT1***	AR	OMIM:251450, OMIM:617719	
2	***COL2A1***	AD	OMIM:108300, OMIM:132450, OMIM:150600 +13	
3	***COL9A1***	AD, AR	OMIM:614134, OMIM:614135	
4	***COL9A2***	AD, AR	OMIM:600204, OMIM:614284	
5	***COL9A3***	AD, AR	OMIM:600969, OMIM:620022	
6	***COMP***	AD	OMIM:132400, OMIM:177170, OMIM:619161	
7	***MATN3***	AD, AR	OMIM:140600, OMIM:607078, OMIM:608728	
8	***SLC26A2***	AR	OMIM:222600, OMIM:226900, OMIM:256050 +1	
9	***TRAPPC2***	XLR	OMIM:313400	

Notes

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- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 2 GENES

Heterotopic ossification

Genes for fibrodysplasia ossificans progressiva and progressive osseous heteroplasia

Panel details

Panel code	TBO-GP-163
Clinical area	Skeletal and connective tissue
Number of genes	2
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACVR1***	AD	`OMIM:135100`	
2	***GNAS***	AD	`OMIM:103580, OMIM:166350, OMIM:174800 +5`	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 14 GENES

Joint hypermobility and connective tissue disorders

Genes for heritable connective tissue disorders with hypermobility

Panel details

Panel code	TBO-GP-164
Clinical area	Skeletal and connective tissue
Number of genes	14
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*COL1A1*	AD	OMIM:114000, OMIM:130060, OMIM:166200 +5	
2	*COL1A2*	AD, AR	OMIM:166210, OMIM:166220, OMIM:166710 +4	
3	*COL3A1*	AD, AR	OMIM:130050, OMIM:618343	
4	*COL5A1*	AD	OMIM:130000, OMIM:619329	
5	*COL5A2*	AD	OMIM:130010	
6	*FBN1*	AD	OMIM:102370, OMIM:129600, OMIM:154700 +5	
7	*FKBP14*	AR	OMIM:614557	
8	*PLOD1*	AR	OMIM:225400	
9	*SKI*	AD	OMIM:182212	
10	*SMAD3*	AD	OMIM:613795	
11	*TGFBR1*	AD	OMIM:132800, OMIM:609192	
12	*TGFBR2*	AD	OMIM:133239, OMIM:610168, OMIM:614331	
13	*TNXB*	AD, AR	OMIM:606408, OMIM:615963	
14	*ZNF469*	AR	OMIM:229200	

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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SKELETAL AND CONNECTIVE TISSUE | 8 GENES

Ectodermal dysplasia

Genes for hypohidrotic and other ectodermal dysplasias

Panel details

Panel code	TBO-GP-165
Clinical area	Skeletal and connective tissue
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***EDA***	XLR, XLD	OMIM:305100, OMIM:313500	
2	***EDAR***	AD, AR	OMIM:129490, OMIM:224900	
3	***EDARADD***	AD, AR	OMIM:129490, OMIM:224900, OMIM:614940 +1	
4	***GJB6***	AD, AR, XLR	OMIM:129500, OMIM:220290, OMIM:304400 +2	
5	***IKBKG***	XLR, XLD	OMIM:300291, OMIM:300636, OMIM:301081 +1	Pseudogene IKBKGP1
6	***PKP1***	AR	OMIM:604536	
7	***TP63***	AD	OMIM:103285, OMIM:106260, OMIM:129400 +4	
8	***WNT10A***	AD, AR	OMIM:150400, OMIM:224750, OMIM:257980	

Genes needing specific methods

- ***IKBKG***: Pseudogene IKBKGP1

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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DERMATOLOGY | 15 GENES

Epidermolysis bullosa

Genes for simplex, junctional, dystrophic and Kindler epidermolysis bullosa

Panel details

Panel code	TBO-GP-166
Clinical area	Dermatology
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CD151***	AR	OMIM:609057	
2	***COL17A1***	AD, AR	OMIM:122400, OMIM:619787	
3	***COL7A1***	AD, AR	OMIM:131705, OMIM:131750, OMIM:131850 +4	
4	***DST***	AR	OMIM:614653, OMIM:615425	
5	***EXPH5***	AR	OMIM:615028	
6	***FERMT1***	AR	OMIM:173650	
7	***ITGA6***	AR	OMIM:619817	
8	***ITGB4***	AR	OMIM:226730, OMIM:619816	
9	***KLHL24***	AD, AR	OMIM:617294, OMIM:620236	
10	***KRT14***	AD, AR	OMIM:125595, OMIM:131760, OMIM:131800 +3	
11	***KRT5***	AD, AR	OMIM:131760, OMIM:131960, OMIM:179850 +5	
12	***LAMA3***	AR	OMIM:226700, OMIM:245660, OMIM:619783 +1	
13	***LAMB3***	AD, AR	OMIM:104530, OMIM:226650, OMIM:226700	
14	***LAMC2***	AR	OMIM:226700, OMIM:619785, OMIM:619786	
15	***PLEC***	AD, AR	OMIM:131950, OMIM:226670, OMIM:226730 +3	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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DERMATOLOGY | 20 GENES

Ichthyosis

Genes for congenital and autosomal recessive ichthyoses

Panel details

Panel code	TBO-GP-167
Clinical area	Dermatology
Number of genes	20
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ABCA12*	AR	OMIM:242500, OMIM:601277	
2	*ALOX12B*	AR	OMIM:242100	
3	*ALOXE3*	AR	OMIM:242100, OMIM:606545	
4	*CERS3*	AR	OMIM:615023	
5	*CLDN1*	AR	OMIM:607626	
6	*CYP4F22*	AR	OMIM:604777	
7	*FLG*	AD, AR	OMIM:146700	
8	*KRT1*	AD, AR	OMIM:113800, OMIM:146590, OMIM:600962 +3	
9	*KRT10*	AD, AR	OMIM:146600, OMIM:607602, OMIM:609165 +2	
10	*KRT2*	AD	OMIM:146800	
11	*LIPN*	AR	OMIM:613943	
12	*NIPAL4*	AR	OMIM:612281	
13	*PNPLA1*	AR	OMIM:615024	
14	*SDR9C7*	AR	OMIM:617574	
15	*SLC27A4*	AR	OMIM:608649	

16	***SPINK5***	AR	OMIM:256500
17	***ST14***	AR	OMIM:602400
18	***STS***	XLR	OMIM:308100
19	***SULT2B1***	AR	OMIM:617571
20	***TGM1***	AR	OMIM:242300

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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DERMATOLOGY · 15 GENES

Palmoplantar keratoderma

Genes for inherited palmoplantar keratodermas

Panel details

Panel code	TBO-GP-168
Clinical area	Dermatology
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AQP5***	AD	OMIM:600231	
2	***CTSC***	AR	OMIM:170650, OMIM:245000, OMIM:245010	
3	***DSG1***	AD, AR	OMIM:148700, OMIM:615508	
4	***DSP***	AD, AR	OMIM:605676, OMIM:607450, OMIM:609638 +2	
5	***GJB2***	AD, AR, XLR	OMIM:124500, OMIM:148210, OMIM:148350 +5	
6	***JUP***	AD, AR	OMIM:601214, OMIM:611528	
7	***KRT1***	AD, AR	OMIM:113800, OMIM:146590, OMIM:600962 +3	
8	***KRT16***	AD	OMIM:167200, OMIM:613000	
9	***KRT17***	AD	OMIM:167210, OMIM:184500	
10	***KRT6A***	AD	OMIM:615726	
11	***KRT6B***	AD	OMIM:615728	
12	***KRT9***	AD	OMIM:144200	
13	***SERPINB7***	AR	OMIM:615598	
14	***SLURP1***	AR	OMIM:248300	
15	***TRPV3***	AD	OMIM:614594, OMIM:616400	

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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DERMATOLOGY · 9 GENES

Inherited hair disorders

Genes for hypotrichosis and hair shaft disorders

Panel details

Panel code	TBO-GP-169
Clinical area	Dermatology
Number of genes	9
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***APCDD1***	AD	OMIM:605389	
2	***CDSN***	AD, AR	OMIM:146520, OMIM:270300	
3	***DSG4***	AR	OMIM:607903	
4	***HR***	AR	OMIM:203655, OMIM:209500	
5	***KRT81***	AD	OMIM:158000	
6	***KRT83***	AD, AR	OMIM:158000, OMIM:617756	
7	***KRT86***	AD	OMIM:158000	
8	***LIPH***	AR	OMIM:604379	
9	***LPAR6***	AR	OMIM:278150	

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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DERMATOLOGY · 11 GENES

Xeroderma pigmentosum and Cockayne syndrome

Nucleotide excision repair genes

Panel details

Panel code	TBO-GP-170
Clinical area	Dermatology
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***DDB2***	AR	OMIM:278740	
2	***ERCC2***	AR	OMIM:278730, OMIM:601675, OMIM:610756	
3	***ERCC3***	AR	OMIM:610651, OMIM:616390	
4	***ERCC4***	AR	OMIM:278760, OMIM:610965, OMIM:615272	
5	***ERCC5***	AR	OMIM:278780, OMIM:616570	
6	***ERCC6***	AD, AR	OMIM:133540, OMIM:211980, OMIM:214150 +3	
7	***ERCC8***	AR	OMIM:216400, OMIM:614621	
8	***POLH***	AR	OMIM:278750	
9	***UVSSA***	AR	OMIM:614640	
10	***XPA***	AR	OMIM:278700	
11	***XPC***	AR	OMIM:278720	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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DERMATOLOGY | 7 GENES

Piebaldism and pigmentary disorders

Genes for piebaldism and neural crest pigmentation disorders

Panel details

Panel code	TBO-GP-171
Clinical area	Dermatology
Number of genes	7
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***EDN3***	AD, AR	OMIM:613265, OMIM:613712	
2	***EDNRB***	AD, AR	OMIM:277580, OMIM:600155, OMIM:600501	
3	***KIT***	AD	OMIM:154800, OMIM:172800, OMIM:273300 +2	
4	***MITF***	AD, AR	OMIM:103500, OMIM:193510, OMIM:614456 +1	
5	***PAX3***	AD, AR	OMIM:122880, OMIM:148820, OMIM:193500 +1	
6	***SNAI2***	—	2 (Orphanet)	
7	***SOX10***	AD	OMIM:609136, OMIM:611584, OMIM:613266	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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DERMATOLOGY | 5 GENES

Pachyonychia congenita

Keratin genes for pachyonychia congenita

Panel details

Panel code	TBO-GP-172
Clinical area	Dermatology
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***KRT16***	AD	OMIM:167200, OMIM:613000	
2	***KRT17***	AD	OMIM:167210, OMIM:184500	
3	***KRT6A***	AD	OMIM:615726	
4	***KRT6B***	AD	OMIM:615728	
5	***KRT6C***	AD	OMIM:615735	

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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DERMATOLOGY 7 GENES

Vascular malformations

Genes for capillary, venous and cerebral cavernous malformations

Panel details

Panel code	TBO-GP-173
Clinical area	Dermatology
Number of genes	7
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CCM2***	AD	OMIM:603284	
2	***EPHB4***	AD	OMIM:617300, OMIM:618196	
3	***GLMN***	AD	OMIM:138000	
4	***KRIT1***	AD	OMIM:116860	
5	***PDCD10***	AD	OMIM:603285	
6	***RASA1***	AD	OMIM:605462, OMIM:608354	
7	***TEK***	AD, AR	OMIM:600195, OMIM:600975, OMIM:617272	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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ENDOCRINOLOGY AND REPRODUCTION | 15 GENES

Congenital hypothyroidism

Genes for thyroid dysgenesis and dyshormonogenesis

Panel details

Panel code	TBO-GP-174
Clinical area	Endocrinology and reproduction
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*DUOX2*	AR	OMIM:607200	
2	*DUOXA2*	AR	OMIM:274900	
3	*FOXE1*	AD, AR	OMIM:241850, OMIM:616534	
4	*GLIS3*	AR	OMIM:610199	
5	*IGSF1*	XLR	OMIM:300888	
6	*IYD*	AR	OMIM:274800	
7	*NKX2-1*	AD	OMIM:118700, OMIM:188550, OMIM:610978	
8	*PAX8*	AD	OMIM:218700	
9	*SLC26A4*	AR	OMIM:274600, OMIM:600791	
10	*SLC5A5*	AR	OMIM:274400	
11	*TG*	AR	OMIM:274700, OMIM:608175	
12	*TPO*	AR	OMIM:274500	
13	*TRHR*	AR	OMIM:618573	
14	*TSHB*	AR	OMIM:275100	
15	*TSHR*	AD, AR	OMIM:275200, OMIM:603373, OMIM:609152	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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TBO-GP-175
Version 1.0 · 11 November 2026
Genome build GRCh38
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ENDOCRINOLOGY AND REPRODUCTION | 14 GENES

Congenital adrenal hyperplasia and adrenal insufficiency

Genes for CAH, familial glucocorticoid deficiency and adrenal hypoplasia

Panel details

Panel code	TBO-GP-175
Clinical area	Endocrinology and reproduction
Number of genes	14
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AAAS***	AR	OMIM:231550	
2	***ABCD1***	XLR	OMIM:300100	
3	***CYP11A1***	—	OMIM:613743	
4	***CYP11B1***	AD, AR	OMIM:103900, OMIM:202010	CYP11B1/CYP11B2 chimera analysis
5	***CYP17A1***	AR	OMIM:202110	
6	***CYP21A2***	AR	OMIM:201910	Pseudogene CYP21A1P; specific method
7	***HSD3B2***	AR	OMIM:201810	
8	***MC2R***	AR	OMIM:202200	
9	***MRAP***	AR	OMIM:607398	
10	***NNT***	AR	OMIM:614736	
11	***NR0B1***	XLR	OMIM:300018, OMIM:300200	
12	***NR5A1***	AD	OMIM:612964, OMIM:612965, OMIM:613957 +1	

13	***POR***	AD, AR	`OMIM:201750, OMIM:207410, OMIM:613571`
14	***STAR***	AR	`OMIM:201710`

Genes needing specific methods

- ***CYP11B1***: CYP11B1/CYP11B2 chimera analysis
- ***CYP21A2***: Pseudogene CYP21A1P; specific method

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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ENDOCRINOLOGY AND REPRODUCTION | 20 GENES

Hypogonadotropic hypogonadism and Kallmann syndrome

Genes for congenital hypogonadotropic hypogonadism

Panel details

Panel code	TBO-GP-176
Clinical area	Endocrinology and reproduction
Number of genes	20
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ANOS1***	XLR	OMIM:308700	
2	***CHD7***	AD	OMIM:214800, OMIM:612370	
3	***DUSP6***	AD, AR	OMIM:146110, OMIM:615269	
4	***FEZF1***	AR	OMIM:146110, OMIM:616030	
5	***FGF8***	AD	OMIM:612702	
6	***FGFR1***	AD	OMIM:101600, OMIM:123150, OMIM:147950 +4	
7	***GNRH1***	AR	OMIM:146110, OMIM:614841	
8	***GNRHR***	AR	OMIM:146110	
9	***HS6ST1***	AD	OMIM:614880	
10	***IL17RD***	AD, AR	OMIM:615267	
11	***KISS1***	AR	OMIM:614842	
12	***KISS1R***	AD, AR	OMIM:176400, OMIM:614837	
13	***NSMF***	AD	OMIM:614838	
14	***PROK2***	AD	OMIM:610628	

15	***PROKR2***	AD	OMIM:244200
16	***SEMA3A***	AD	OMIM:614897
17	***SOX10***	AD	OMIM:609136, OMIM:611584, OMIM:613266
18	***TAC3***	AR	OMIM:614839
19	***TACR3***	AR	OMIM:614840
20	***WDR11***	AD, AR	OMIM:146110, OMIM:614858, OMIM:620237

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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ENDOCRINOLOGY AND REPRODUCTION | 16 GENES

Differences of sex development

Genes for 46,XY and 46,XX differences of sex development

Panel details

Panel code	TBO-GP-177
Clinical area	Endocrinology and reproduction
Number of genes	16
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AR***	XLR	OMIM:300068, OMIM:300633, OMIM:312300 +1	
2	***CYP11A1***	—	OMIM:613743	
3	***CYP17A1***	AR	OMIM:202110	
4	***DHH***	AR	OMIM:233420, OMIM:607080	
5	***HSD17B3***	AR	OMIM:264300	
6	***LHCGR***	AR	OMIM:176410, OMIM:238320	
7	***MAP3K1***	AD	OMIM:613762	
8	***NR0B1***	XLR	OMIM:300018, OMIM:300200	
9	***NR5A1***	AD	OMIM:612964, OMIM:612965, OMIM:613957 +1	
10	***POR***	AD, AR	OMIM:201750, OMIM:207410, OMIM:613571	
11	***SOX9***	AD	OMIM:114290, OMIM:278850, OMIM:616425	
12	***SRD5A2***	AR	OMIM:264600	
13	***SRY***	XLD, YL	OMIM:400044, OMIM:400045	
14	***STAR***	AR	OMIM:201710	
15	***WT1***	AD	OMIM:106210, OMIM:136680, OMIM:156240 +5	

16	***ZFPM2***	AD	OMIM:187500, OMIM:610187, OMIM:616067

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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ENDOCRINOLOGY AND REPRODUCTION | 12 GENES

Combined pituitary hormone deficiency

Genes for congenital hypopituitarism

Panel details

Panel code	TBO-GP-178
Clinical area	Endocrinology and reproduction
Number of genes	12
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*GH1*	AD, AR	OMIM:173100, OMIM:262400, OMIM:262650 +1	
2	*GHRHR*	AR	OMIM:618157	
3	*GLI2*	AD	OMIM:610829, OMIM:615849	
4	*HESX1*	AD, AR	OMIM:182230	
5	*LHX3*	AR	OMIM:221750	
6	*LHX4*	AD	OMIM:262700	
7	*OTX2*	AD	OMIM:610125, OMIM:613986	
8	*POU1F1*	AD, AR	OMIM:613038	
9	*PROP1*	AR	OMIM:262600	
10	*SOX2*	AD	OMIM:206900	
11	*SOX3*	XL	OMIM:300123, OMIM:312000	
12	*TBX19*	AR	OMIM:201400	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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ENDOCRINOLOGY AND REPRODUCTION | 10 GENES

Monogenic obesity

Genes of the leptin-melanocortin pathway

Panel details

Panel code	TBO-GP-179
Clinical area	Endocrinology and reproduction
Number of genes	10
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ADCY3***	AR	OMIM:617885	
2	***BDNF***	—	2 (Orphanet)	
3	***LEP***	AR	OMIM:614962	
4	***LEPR***	AR	OMIM:614963	
5	***MC4R***	AD, AR	OMIM:618406	
6	***NTRK2***	AD	OMIM:613886, OMIM:617830	
7	***PCSK1***	AR	OMIM:600955	
8	***POMC***	AD, AR	OMIM:601665, OMIM:609734	
9	***SH2B1***	—	3 (Orphanet)	
10	***SIM1***	—	3 (Orphanet)	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito

mitochondrial.

- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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ENDOCRINOLOGY AND REPRODUCTION | 14 GENES

Primary ovarian insufficiency

Genes for primary ovarian insufficiency (FMR1 premutation needs repeat testing)

Panel details

Panel code	TBO-GP-180
Clinical area	Endocrinology and reproduction
Number of genes	14
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***BMP15***	XL	OMIM:300510	
2	***FIGLA***	—	OMIM:612310	
3	***FMR1***	XLD	OMIM:300623, OMIM:300624, OMIM:311360	CGG repeat analysis required
4	***FOXL2***	AD	OMIM:110100, OMIM:608996	
5	***GDF9***	AR	OMIM:618014	
6	***HFM1***	AR	OMIM:615724	
7	***MCM8***	AR	OMIM:612885	
8	***MCM9***	AR	OMIM:616185	
9	***MSH5***	AR	OMIM:617442, OMIM:619937	
10	***NOBOX***	AD	OMIM:611548	
11	***NR5A1***	AD	OMIM:612964, OMIM:612965, OMIM:613957 +1	
12	***PSMC3IP***	AR	OMIM:614324	
13	***STAG3***	AR	OMIM:615723, OMIM:619672	

14	***SYCE1***	AR	OMIM:616947, OMIM:616950

Genes needing specific methods

- ***FMR1***: CGG repeat analysis required

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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ENDOCRINOLOGY AND REPRODUCTION | 12 GENES

Male infertility

Genes for monogenic male infertility (Y-chromosome microdeletions tested separately)

Panel details

Panel code	TBO-GP-181
Clinical area	Endocrinology and reproduction
Number of genes	12
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***AR***	XLR	OMIM:300068, OMIM:300633, OMIM:312300 +1	
2	***AURKC***	AR	OMIM:243060	
3	***CFAP43***	AD, AR	OMIM:236690, OMIM:617592	
4	***CFAP44***	AR	OMIM:617593	
5	***CFTR***	AD, AR	OMIM:167800, OMIM:211400, OMIM:219700 +1	
6	***DNAH1***	AR	OMIM:617576, OMIM:617577	
7	***DPY19L2***	AR	OMIM:613958	
8	***NR5A1***	AD	OMIM:612964, OMIM:612965, OMIM:613957 +1	
9	***SPATA16***	AR	OMIM:102530	
10	***SYCP3***	AD	OMIM:270960	
11	***TEX11***	XLR	OMIM:309120	
12	***ZMYND15***	AR	OMIM:615842	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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ENDOCRINOLOGY AND REPRODUCTION | 15 GENES

Monogenic hypertension

Genes for familial hyperaldosteronism, Liddle, Gordon and apparent mineralocorticoid excess

Panel details

Panel code	TBO-GP-182
Clinical area	Endocrinology and reproduction
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*CACNA1D*	AD, AR	OMIM:614896, OMIM:615474	
2	*CACNA1H*	AD	OMIM:617027	
3	*CLCN2*	AD, AR	OMIM:605635, OMIM:607628, OMIM:615651	
4	*CUL3*	AD	OMIM:614496, OMIM:619239	
5	*CYP11B1*	AD, AR	OMIM:103900, OMIM:202010	CYP11B1/CYP11B2 chimera analysis
6	*CYP17A1*	AR	OMIM:202110	
7	*HSD11B2*	AR	OMIM:218030	
8	*KCNJ5*	AD	OMIM:613485, OMIM:613677	
9	*KLHL3*	AD, AR	OMIM:614495	
10	*NR3C2*	AD	OMIM:177735, OMIM:605115	
11	*PDE3A*	AD	OMIM:112410	
12	*SCNN1B*	AD, AR	OMIM:177200, OMIM:211400, OMIM:620125	

13	***SCNN1G***	AD, AR	OMIM:613071, OMIM:618114, OMIM:620126
14	***WNK1***	AD, AR	OMIM:201300, OMIM:614492
15	***WNK4***	AD	OMIM:614491

Genes needing specific methods

- ***CYP11B1***: CYP11B1/CYP11B2 chimera analysis

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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ENDOCRINOLOGY AND REPRODUCTION | 4 GENES

Thyroid hormone resistance and transport

Genes for reduced sensitivity to thyroid hormone

Panel details

Panel code	TBO-GP-183
Clinical area	Endocrinology and reproduction
Number of genes	4
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***SECISBP2***	AR	OMIM:609698	
2	***SLC16A2***	XLR	OMIM:300523	
3	***THRA***	AD	OMIM:614450	
4	***THRB***	AD, AR	OMIM:145650, OMIM:188570, OMIM:274300	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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ENDOCRINOLOGY AND REPRODUCTION | 6 GENES

Primary hyperparathyroidism

Genes for familial and syndromic primary hyperparathyroidism

Panel details

Panel code	TBO-GP-184
Clinical area	Endocrinology and reproduction
Number of genes	6
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CASR***	AD, AR	OMIM:145980, OMIM:239200, OMIM:601198 +1	
2	***CDC73***	AD	OMIM:145000, OMIM:145001, OMIM:608266	
3	***CDKN1B***	AD	OMIM:610755	
4	***GCM2***	AD, AR	OMIM:617343, OMIM:618883	
5	***MEN1***	AD	OMIM:131100	
6	***RET***	AD	OMIM:142623, OMIM:155240, OMIM:162300 +2	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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ENDOCRINOLOGY AND REPRODUCTION | 7 GENES

Syndromic diabetes

Genes for Wolfram and other syndromic forms of diabetes

Panel details

Panel code	TBO-GP-185
Clinical area	Endocrinology and reproduction
Number of genes	7
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CISD2***	AR	OMIM:604928	
2	***EIF2AK3***	AR	OMIM:226980	
3	***HNF1B***	AD	OMIM:125853, OMIM:137920, OMIM:144700	
4	***INSR***	AD, AR	OMIM:246200, OMIM:262190, OMIM:609968	
5	***PPARG***	AD, AR	OMIM:125853, OMIM:601665, OMIM:604367	
6	***SLC19A2***	AR	OMIM:249270	
7	***WFS1***	AD, AR	OMIM:116400, OMIM:125853, OMIM:222300 +2	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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GASTROENTEROLOGY AND HEPATOLOGY | 16 GENES

Inherited cholestasis

Genes for progressive familial intrahepatic cholestasis, Alagille syndrome and bile acid synthesis defects

Panel details

Panel code	TBO-GP-186
Clinical area	Gastroenterology and hepatology
Number of genes	16
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCB11***	AR	OMIM:601847, OMIM:605479	
2	***ABCB4***	AD, AR	OMIM:600803, OMIM:602347, OMIM:614972	
3	***AKR1D1***	AR	OMIM:235555	
4	***ATP8B1***	AD, AR	OMIM:147480, OMIM:211600, OMIM:243300	
5	***CYP7B1***	AR	OMIM:270800, OMIM:613812	
6	***HSD3B7***	AR	OMIM:607765	
7	***JAG1***	AD	OMIM:118450, OMIM:187500, OMIM:617992 +1	
8	***KIF12***	AR	OMIM:619662	
9	***MYO5B***	AR	OMIM:251850, OMIM:619868	
10	***NOTCH2***	AD	OMIM:102500, OMIM:610205	
11	***NR1H4***	AR	OMIM:617049	
12	***SLC10A1***	AR	OMIM:619256	
13	***TJP2***	AR	OMIM:607748, OMIM:615878	
14	***USP53***	AR	OMIM:619658	
15	***VIPAS39***	AR	OMIM:613404	

16	***VPS33B***	AR	OMIM:208085, OMIM:620009, OMIM:620010

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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GASTROENTEROLOGY AND HEPATOLOGY | 5 GENES

Inherited liver disease

Genes for alpha-1 antitrypsin deficiency, Wilson disease, haemochromatosis and citrin deficiency

Panel details

Panel code	TBO-GP-187
Clinical area	Gastroenterology and hepatology
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCB4***	AD, AR	OMIM:600803, OMIM:602347, OMIM:614972	
2	***ATP7B***	AR	OMIM:277900	
3	***HFE***	AR	OMIM:235200	
4	***SERPINA1***	AR	OMIM:613490	
5	***SLC25A13***	AR	OMIM:603471, OMIM:605814	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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GASTROENTEROLOGY AND HEPATOLOGY | 15 GENES

Very-early-onset inflammatory bowel disease

Monogenic causes of IBD-like disease in early childhood

Panel details

Panel code	TBO-GP-188
Clinical area	Gastroenterology and hepatology
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CTLA4***	AD	OMIM:140300, OMIM:152700, OMIM:616100	
2	***CYBB***	XLR	OMIM:300645, OMIM:306400	
3	***DOCK8***	AR	OMIM:243700	
4	***FOXP3***	XLR	OMIM:304790	
5	***IL10***	—	OMIM:180300	
6	***IL10RA***	AR	OMIM:613148	
7	***IL10RB***	AR	OMIM:612567	
8	***LRBA***	AR	OMIM:614700	
9	***NCF1***	AR	OMIM:233700	Pseudogenes NCF1B/NCF1C
10	***NCF2***	AR	OMIM:233710	
11	***SKIC2***	AR	OMIM:614602	
12	***SKIC3***	AR	OMIM:222470	
13	***TTC7A***	AR	OMIM:243150	
14	***WAS***	XLR	OMIM:300299, OMIM:301000, OMIM:313900	
15	***XIAP***	XLR	OMIM:300635, OMIM:308240	

Genes needing specific methods

- ***NCF1***: Pseudogenes NCF1B/NCF1C

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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GASTROENTEROLOGY AND HEPATOLOGY | 8 GENES

Hirschsprung disease

Genes for isolated and syndromic Hirschsprung disease

Panel details

Panel code	TBO-GP-189
Clinical area	Gastroenterology and hepatology
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***EDN3***	AD, AR	OMIM:613265, OMIM:613712	
2	***EDNRB***	AD, AR	OMIM:277580, OMIM:600155, OMIM:600501	
3	***GDNF***	AD	OMIM:613711	
4	***KIFBP***	AR	OMIM:609460	
5	***PHOX2B***	AD	OMIM:209880, OMIM:613013	
6	***RET***	AD	OMIM:142623, OMIM:155240, OMIM:162300 +2	
7	***SOX10***	AD	OMIM:609136, OMIM:611584, OMIM:613266	
8	***ZEB2***	AD	OMIM:235730	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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GASTROENTEROLOGY AND HEPATOLOGY | 15 GENES

Congenital diarrhoea and enteropathy

Genes for congenital diarrhoeal disorders

Panel details

Panel code	TBO-GP-190
Clinical area	Gastroenterology and hepatology
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*DGAT1*	AR	OMIM:615863	
2	*EPCAM*	AD, AR	OMIM:613217, OMIM:613244	
3	*GUCY2C*	AD, AR	OMIM:614616, OMIM:614665	
4	*MYO5B*	AR	OMIM:251850, OMIM:619868	
5	*NEUROG3*	AR	OMIM:610370	
6	*PCSK1*	AR	OMIM:600955	
7	*SI*	AR	OMIM:222900	
8	*SKIC2*	AR	OMIM:614602	
9	*SKIC3*	AR	OMIM:222470	
10	*SLC26A3*	AR	OMIM:214700	
11	*SLC5A1*	AR	OMIM:606824	
12	*SLC9A3*	AR	OMIM:616868	
13	*SPINT2*	AR	OMIM:270420	
14	*STX3*	AR	OMIM:619445, OMIM:619446	
15	*TTC7A*	AR	OMIM:243150	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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GASTROENTEROLOGY AND HEPATOLOGY | 4 GENES

Inherited hyperbilirubinaemia

Genes for Gilbert, Crigler-Najjar, Dubin-Johnson and Rotor syndromes

Panel details

Panel code	TBO-GP-191
Clinical area	Gastroenterology and hepatology
Number of genes	4
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ABCC2***	AR	OMIM:237500	
2	***SLCO1B1***	PGx	OMIM:237450	
3	***SLCO1B3***	—	OMIM:237450	
4	***UGT1A1***	AR	OMIM:143500, OMIM:218800, OMIM:237900 +1	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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GASTROENTEROLOGY AND HEPATOLOGY | 5 GENES

Hypobetalipoproteinaemia

Genes for abetalipoproteinaemia and familial hypobetalipoproteinaemia

Panel details

Panel code	TBO-GP-192
Clinical area	Gastroenterology and hepatology
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ANGPTL3***	AR	OMIM:605019	
2	***APOB***	AD, AR	OMIM:144010, OMIM:615558	
3	***MTTP***	AR	OMIM:200100	
4	***PCSK9***	AD	OMIM:603776	
5	***SAR1B***	AR	OMIM:246700	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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GASTROENTEROLOGY AND HEPATOLOGY | 7 GENES

Cholesterol biosynthesis disorders

Genes for Smith-Lemli-Opitz and related sterol disorders

Panel details

Panel code	TBO-GP-193
Clinical area	Gastroenterology and hepatology
Number of genes	7
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***DHCR24***	AR	OMIM:602398	
2	***DHCR7***	AR	OMIM:270400	
3	***EBP***	XLR, XLD	OMIM:300960, OMIM:302960	
4	***LBR***	AD, AR	OMIM:169400, OMIM:215140, OMIM:613471 +1	
5	***MSMO1***	AR	OMIM:616834	
6	***NSDHL***	XLR, XLD	OMIM:300831, OMIM:308050	
7	***SC5D***	AR	OMIM:607330	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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PULMONOLOGY | 11 GENES

Surfactant dysfunction and childhood interstitial lung disease

Genes for surfactant disorders and developmental lung disease

Panel details

Panel code	TBO-GP-194
Clinical area	Pulmonology
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	*ABCA3*	AR	OMIM:610921	
2	*CSF2RA*	XL	OMIM:300770	
3	*CSF2RB*	AR	OMIM:614370	
4	*FOXF1*	AD	OMIM:265380	
5	*MARS1*	AD, AR	OMIM:615486, OMIM:616280, OMIM:619692 +1	
6	*NKX2-1*	AD	OMIM:118700, OMIM:188550, OMIM:610978	
7	*SFTPA1*	AD	OMIM:619611	
8	*SFTPA2*	AD	OMIM:178500	
9	*SFTPB*	AR	OMIM:265120	
10	*SFTPC*	AD	OMIM:178500, OMIM:610913	
11	*TBX4*	AD, AR	OMIM:147891, OMIM:601360	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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PULMONOLOGY | 8 GENES

Familial pulmonary fibrosis

Telomere and surfactant genes for familial interstitial lung disease

Panel details

Panel code	TBO-GP-195
Clinical area	Pulmonology
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***DKC1***	XLR, XLD	OMIM:301108, OMIM:305000	
2	***NAF1***	AD	OMIM:620365	
3	***PARN***	AD, AR	OMIM:616353, OMIM:616371	
4	***RTEL1***	AD, AR	OMIM:615190, OMIM:616373	
5	***SFTPA2***	AD	OMIM:178500	
6	***SFTPC***	AD	OMIM:178500, OMIM:610913	
7	***TERC***	AD	OMIM:127550, OMIM:614743	Non-coding RNA gene
8	***TERT***	AD, AR	OMIM:127550, OMIM:178500, OMIM:601626 +4	

Genes needing specific methods

- ***TERC***: Non-coding RNA gene

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.

- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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PULMONOLOGY | 5 GENES

Hereditary pneumothorax and cystic lung disease

Genes for Birt-Hogg-Dubé and connective tissue causes of pneumothorax

Panel details

Panel code	TBO-GP-196
Clinical area	Pulmonology
Number of genes	5
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***COL3A1***	AD, AR	OMIM:130050, OMIM:618343	
2	***FBN1***	AD	OMIM:102370, OMIM:129600, OMIM:154700 +5	
3	***FLCN***	AD	OMIM:114500, OMIM:135150, OMIM:144700 +2	
4	***TSC1***	AD	OMIM:191100, OMIM:606690, OMIM:607341	
5	***TSC2***	AD	OMIM:606690, OMIM:607341, OMIM:613254	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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MULTISYSTEM SYNDROMES | 8 GENES

Cornelia de Lange syndrome and cohesinopathies

Genes for Cornelia de Lange syndrome and related cohesin disorders

Panel details

Panel code	TBO-GP-197
Clinical area	Multisystem syndromes
Number of genes	8
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ANKRD11***	AD	OMIM:148050	
2	***BRD4***	AD	OMIM:620568	
3	***ESCO2***	AR	OMIM:216100, OMIM:268300	
4	***HDAC8***	XLD	OMIM:300882	
5	***NIPBL***	AD	OMIM:122470	
6	***RAD21***	AD, AR	OMIM:611376, OMIM:614701	
7	***SMC1A***	XLR, XLD	OMIM:300590, OMIM:301044	
8	***SMC3***	AD	OMIM:610759	

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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MULTISYSTEM SYNDROMES | 11 GENES

Kabuki, Rubinstein-Taybi and Coffin-Siris syndromes

Chromatin regulator genes for overlapping syndromes

Panel details

Panel code	TBO-GP-198
Clinical area	Multisystem syndromes
Number of genes	11
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ARID1A***	AD	OMIM:614607	
2	***ARID1B***	AD	OMIM:135900	
3	***CREBBP***	AD	OMIM:180849, OMIM:618332	
4	***EP300***	AD	OMIM:114500, OMIM:180849, OMIM:613684 +1	
5	***KDM6A***	AD, XLD	OMIM:147920, OMIM:300867	
6	***KMT2A***	AD	OMIM:605130	
7	***KMT2D***	AD	OMIM:147920, OMIM:620186	
8	***SMARCA4***	AD	OMIM:613325, OMIM:614609, OMIM:620792	
9	***SMARCB1***	AD	OMIM:162091, OMIM:609322, OMIM:614608	
10	***SMARCE1***	AD	OMIM:607174, OMIM:616938	
11	***SOX11***	AD	OMIM:615866	

Notes

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- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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MULTISYSTEM SYNDROMES | 15 GENES

Pharmacogenomics core

Genes with CPIC guideline drug–gene pairs (CYP2D6 structural variants and HLA typing need dedicated methods)

Panel details

Panel code	TBO-GP-199
Clinical area	Multisystem syndromes
Number of genes	15
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***CYP2B6***	PGx	Pharmacogene	
2	***CYP2C19***	PGx	Pharmacogene	
3	***CYP2C9***	AD	OMIM:122700	
4	***CYP2D6***	AR	OMIM:608902	Structural variants and hybrids need dedicated method
5	***CYP3A5***	PGx	OMIM:145500	
6	***CYP4F2***	PGx	Pharmacogene	
7	***DPYD***	AR	OMIM:274270	
8	***G6PD***	XLD	OMIM:300908	
9	***HLA-B***	PGx	OMIM:106300	HLA typing method
10	***IFNL3***	PGx	Pharmacogene	
11	***NUDT15***	PGx	Pharmacogene	
12	***SLCO1B1***	PGx	OMIM:237450	
13	***TPMT***	AR	OMIM:610460	

14	***UGT1A1***	AR	OMIM:143500, OMIM:218800, OMIM:237900 +1
15	***VKORC1***	AD, AR	OMIM:122700, OMIM:607473

Genes needing specific methods

- ***CYP2D6***: Structural variants and hybrids need dedicated method
- ***HLA-B***: HLA typing method

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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MULTISYSTEM SYNDROMES | 12 GENES

Fetal akinesia and multiple pterygium

Genes for fetal akinesia deformation sequence

Panel details

Panel code	TBO-GP-200
Clinical area	Multisystem syndromes
Number of genes	12
Available as	PANEL Tribro (targeted) or virtual panel on WES Tribro / WGS Tribro data
Variant types	SNVs and small indels; CNVs where coverage allows (see notes)
Turnaround and price	[On request]

Genes

#	Gene	Inheritance (HPO)	Linked disorders (OMIM)	Technical note
1	***ACTA1***	AD, AR	OMIM:161800, OMIM:616852, OMIM:620265 +1	
2	***CHRNA1***	AD, AR	OMIM:253290, OMIM:601462, OMIM:608930	
3	***CHRND***	AD, AR	OMIM:253290, OMIM:616321, OMIM:616322 +1	
4	***CHRNG***	AR	OMIM:253290, OMIM:265000	
5	***DOK7***	AR	OMIM:254300, OMIM:618389	
6	***ERBB3***	AD, AR	OMIM:133180, OMIM:243180, OMIM:607598	
7	***GLE1***	AR	OMIM:253310, OMIM:611890	
8	***MUSK***	AR	OMIM:208150, OMIM:616325	
9	***NEB***	AR	OMIM:256030, OMIM:619334	Triplicated region; reduced sensitivity
10	***PIEZO2***	AD, AR	OMIM:108145, OMIM:114300, OMIM:248700 +1	
11	***RAPSN***	AR	OMIM:616326, OMIM:618388	

12	***RYR1***	AD, AR	OMIM:117000, OMIM:145600, OMIM:255320 +1

Genes needing specific methods

- ***NEB***: Triplicated region; reduced sensitivity

Notes

- Gene content curated by Tribro-Omics from published gene–disease evidence. Review by a clinical scientist is required before diagnostic use.
- Inheritance and linked disorders are taken from Human Phenotype Ontology gene annotations (release 2025-01-16); AD autosomal dominant, AR autosomal recessive, XL/XLR/XLD X-linked, Mito mitochondrial.
- Gene symbols follow HGNC nomenclature. Coverage gaps are reported per case.

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