Tribro-Omics

Report ID [TBO/RPT/YYYY/NNNN]
Version [v1 · final]
Issued [DD Month YYYY]
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WGS TRIBRO

Whole-Genome Sequencing Report

Germline whole-genome analysis: SNVs, indels, copy-number and structural variants

1 Patient and sample

Patient name	[Surname, First name]
Patient ID / MRN	[ID]
Date of birth	[DD Month YYYY]
Sex	[Female / Male / Unknown]
Sample ID	[TBO-S-YYYY-NNNN]
Sample type	[EDTA blood / extracted DNA / saliva]
Collected	[DD Month YYYY]
Received	[DD Month YYYY]
Referring clinician	[Name, institution]
Clinical indication	[Phenotype summary; HPO terms, e.g. HP:0001250]

2 Test information

Product	WGS Tribro — whole-genome analysis
Genome build	GRCh38
Variant types analysed	SNVs, small indels, CNVs, structural variants, mitochondrial variants [, repeat expansions]
Analysis scope	[Virtual gene panel / phenotype-driven / genome-wide]
Pipeline version	[TBO-WGS vX.Y]
Secondary findings	[Opted in / opted out] (ACMG SF v3.2 gene list)

RESULT

[Positive: pathogenic / likely pathogenic variant identified that explains the phenotype] / [Negative: no reportable variant identified] / [Inconclusive: variant of uncertain significance]

3 Results: SNVs and small indels

Gene	Transcript	Variant (HGVS)	Zygosity	Inheritance	Classification
[GENE]	[NM_ / MANE]	[c.] [p.]	[Het / Hom / Hemi]	[AD / AR / XL / de novo]	[Pathogenic / Likely path. / VUS]

4 Results: copy-number and structural variants

Type	Location (GRCh38)	Size	Genes	Zygosity	Classification
[DEL / DUP / INV / TRA]	[chrN:start-end]	[kb]	[GENES]	[Het / Hom]	[class]

5 Mitochondrial genome

[No reportable mitochondrial variants detected / variant, heteroplasmy level and classification].

6 Interpretation

[Explain how the reported variant(s) relate to the clinical indication, the mode of inheritance and the strength of evidence.]

7 Quality metrics

Metric	Result	Acceptance criterion
Mean depth of coverage	[ ]×	≥ 30×
Genome at ≥ 15×	[ ] %	≥ 90 %
Bases ≥ Q30	[ ] %	≥ 85 %
Contamination estimate	[ ] %	< 2 %
Sex check (genetic vs reported)	[Concordant / discordant]	Concordant

8 Methods

Genomic DNA was sequenced on [platform] with [read length] paired-end reads ([library kit, PCR-free]). Reads were aligned to GRCh38 with [aligner, version]; SNVs and indels were called with [caller, version], CNVs and structural variants with [tools, versions]. Variants were annotated for population frequency

(gnomAD), clinical databases (ClinVar), predicted effect and conservation, filtered according to the analysis scope and classified using ACMG/AMP criteria. Tools, versions and parameters are recorded for every run.

9 Limitations

- Sensitivity is reduced in segmental duplications, highly homologous regions (pseudogenes) and low-complexity sequence.
- Low-level mosaicism (below about 10–20 % allele fraction) may not be detected.
- Balanced rearrangements, repeat expansions and methylation defects are detected only where specifically analysed and stated above.
- Only variants relevant to the analysis scope are reported; other findings are not assessed unless requested.

10 Recommendations

- Genetic counselling for the patient and family.
- Segregation testing in relatives where it would clarify a variant's significance.
- Reanalysis of the genome data in 12–24 months or when the clinical picture changes (see Challenge Card).

11 Sign-off

Role	Name and qualification	Date
Analysed by (bioinformatics)	[Name, qualification]	[DD Month YYYY]
Reviewed by (clinical scientist)	[Name, qualification]	[DD Month YYYY]
Authorised by (laboratory director)	[Name, qualification]	[DD Month YYYY]

12 Important information

This report is intended for the referring clinician and must be interpreted together with the clinical findings, family history and other test results. Variant classification follows the ACMG/AMP 2015 standards (Richards et al., Genet Med 2015) and ClinGen Sequence Variant Interpretation recommendations. Classifications reflect the evidence available on the date of issue and may change. Genetic counselling is recommended when results are discussed with the patient. Report status: [Clinical / Research use only].