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

Whole-Exome Sequencing Report

Germline exome analysis for rare and inherited disease; singleton or trio

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	WES Tribro — whole-exome analysis
Genome build	GRCh38
Exome capture	[Capture kit and version]
Analysis design	[Singleton / trio (proband, mother, father)]
Analysis scope	[Phenotype-driven (HPO) / virtual panel: name, version]
Pipeline version	[TBO-WES vX.Y]

RESULT

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

3 Results

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 Segregation (trio analysis)

Variant	Proband	Mother	Father	Inference
[GENE c.]	[Het]	[Not detected]	[Not detected]	[De novo]

5 Interpretation

[State whether the finding explains the phenotype, the ACMG evidence codes applied (e.g. PS2, PM2_Supporting, PP3) and any residual uncertainty.]

6 Quality metrics

Metric	Result	Acceptance criterion
Mean target depth	[ ]×	≥ 100×
Targets at ≥ 20×	[ ] %	≥ 95 %
Duplicate reads	[ ] %	< 20 %
Contamination estimate	[ ] %	< 2 %
Sex and relatedness check	[Concordant / discordant]	Concordant

7 Methods

Exonic regions were enriched with [capture kit] and sequenced on [platform]. Reads were aligned to GRCh38 with [aligner, version]; variants were called with [caller, version] and exome CNVs with [tool, version]. Variants were annotated (gnomAD, ClinVar, in silico predictors), prioritised against the HPO terms and inheritance model, and classified using ACMG/AMP criteria.

8 Limitations

- Exome sequencing does not reliably detect deep intronic and regulatory variants, repeat expansions, balanced rearrangements or most structural variants.
- Exome CNV calling is limited to multi-exon events; single-exon CNVs may be missed.
- Regions with low coverage or high homology are listed on request; variants there may be missed.
- Mosaic variants below about 15–20 % allele fraction may not be detected.

9 Recommendations

- Genetic counselling.
- Confirmation of the reported variant(s) by an orthogonal method where required by local policy.
- Consider genome sequencing or reanalysis (Challenge Card) if the result is negative and suspicion remains.

10 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]

11 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].