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RNASEQ TRIBRO

RNA Sequencing Analysis Report

Transcriptome analysis: expression, differential expression, splicing and fusions

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	[Tissue / cells / extracted RNA]
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	RNASEQ Tribro — transcriptome analysis
Reference	GRCh38 with [GENCODE release] annotation
Study design	[Groups, number of samples, replicates]
Library type	[Poly-A / total RNA (rRNA-depleted) / single-cell]
Questions addressed	[Differential expression / fusions / aberrant splicing / outlier expression]
Pipeline version	[TBO-RNA vX.Y]

RESULT

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

3 Summary of findings

[Main findings in two or three sentences: number of differentially expressed genes, key pathways, fusions or splicing events.]

4 Differentially expressed genes (top)

Gene	log2 fold change	Adjusted p-value	Mean expression	Direction
[GENE]	[ ]	[ ]	[ ]	[Up / Down]

5 Pathway enrichment

Pathway / gene set	Genes	Adjusted p-value	Direction
[Pathway name, database]	[n]	[ ]	[Up / Down]

6 Gene fusions

Fusion	Breakpoints (GRCh38)	Supporting reads	Frame	Known?
[GENE1::GENE2]	[chrN:pos / chrN:pos]	[split / spanning]	[In-frame / out-of-frame]	[COSMIC / novel]

7 Quality metrics

Metric	Result	Acceptance criterion
RNA integrity (RIN / DV200)	[ ]	RIN ≥ 7 or DV200 ≥ 30 % (FFPE)
Reads per sample	[ ] million	≥ 30 million
Uniquely mapped reads	[ ] %	≥ 80 %
rRNA fraction	[ ] %	< 10 %
Sample correlation / outliers	[Pass / samples flagged]	No unexplained outliers

8 Methods

Libraries were prepared with [kit] and sequenced on [platform]. Reads were aligned to GRCh38 with [aligner, version] and quantified with [tool, version]. Differential expression was tested with [method] and p-values adjusted (Benjamini–Hochberg). Fusions were detected with [tool] and splicing analysed with [tool]. Pathway enrichment used [method, database version].

9 Limitations

- Expression reflects the sampled tissue and time point; differences in cell composition can drive apparent expression changes.
- Statistical power depends on the number of replicates; small effects may be missed.
- Fusion and splicing calls should be confirmed by an orthogonal method before clinical use.

10 Recommendations

- Validate key genes by qPCR or an orthogonal assay.
- Integrate with genomic findings (WES/WGS) where available.

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].