

Supplementary Figures

Detection of aberrant events in RNA sequencing data

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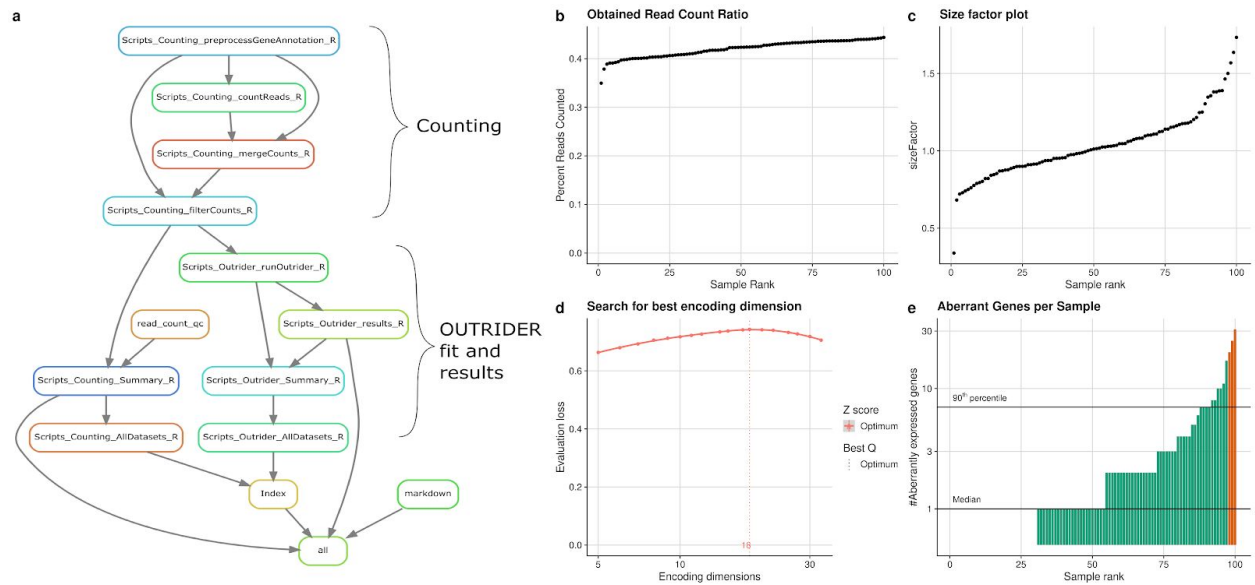


Fig S1 | Counting and OUTRIDER Summary. **a**, Aberrant expression workflow. The two main steps are counting and running the OUTRIDER fit and results. **b**, Percentage of counted reads per sample. **c**, Sorted size factors. Size factors represent the relative sequencing depth of a certain sample. **d**, OUTRIDER evaluation loss for different encoding dimensions q and showing the optimal value (vertical dotted line). **e**, Number of aberrantly expressed genes per sample. Aberrant samples (orange) are samples with too many outlier genes.

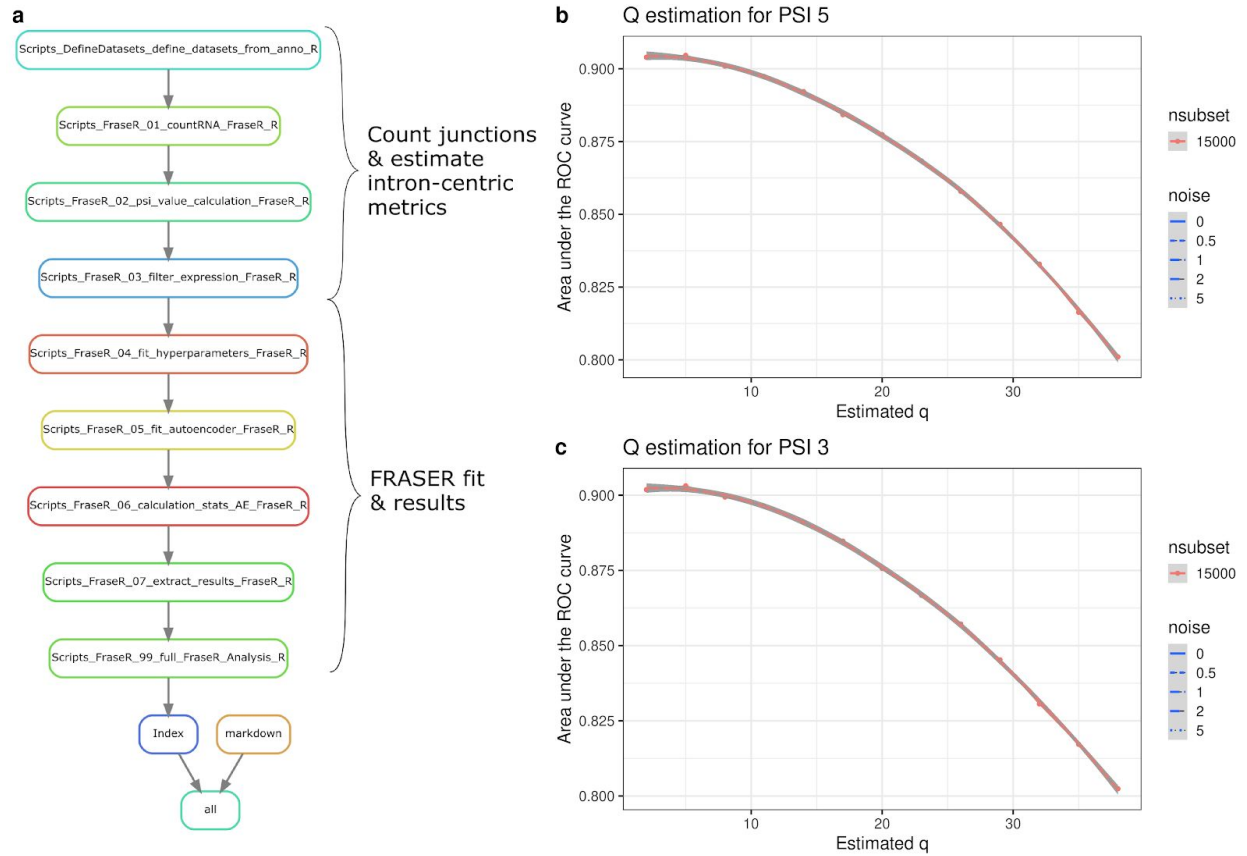


Fig S2 | Aberrant Splicing Module. a, Aberrant splicing workflow. The two main steps are counting the junctions and running the FraseR fit and results. **b-c**, Area under the ROC curve (y-axis) after different encoding dimensions (x-axis) and noise level injections for PSI 3 (b) and PSI 5 (c). The ranking of outliers was bootstrapped to yield 95% confidence bands.

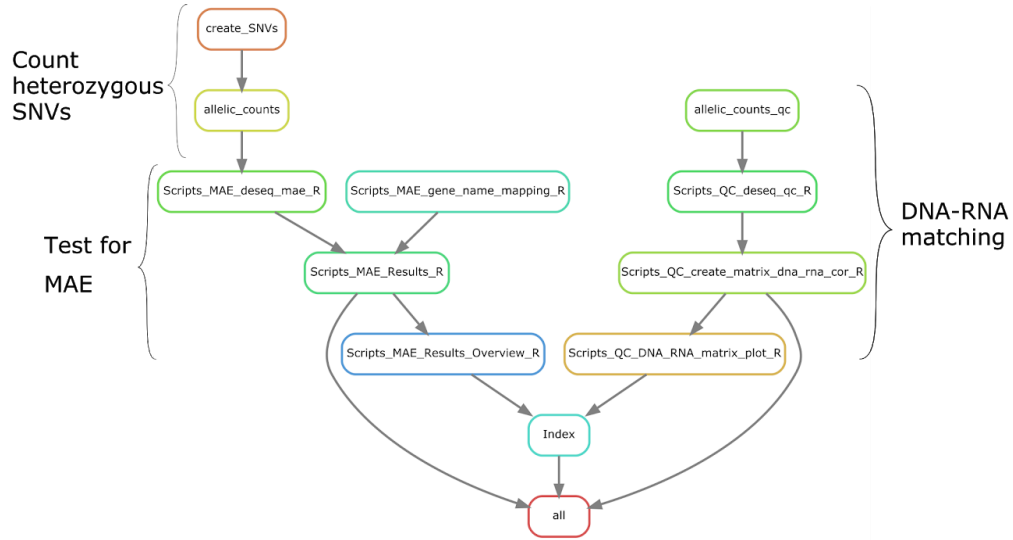


Fig S3 | Mono-allelic expression workflow. It is composed of two parts, the first one tests for heterozygous SNVs that are mono-allelically expressed and the second one matches VCF with BAM files.

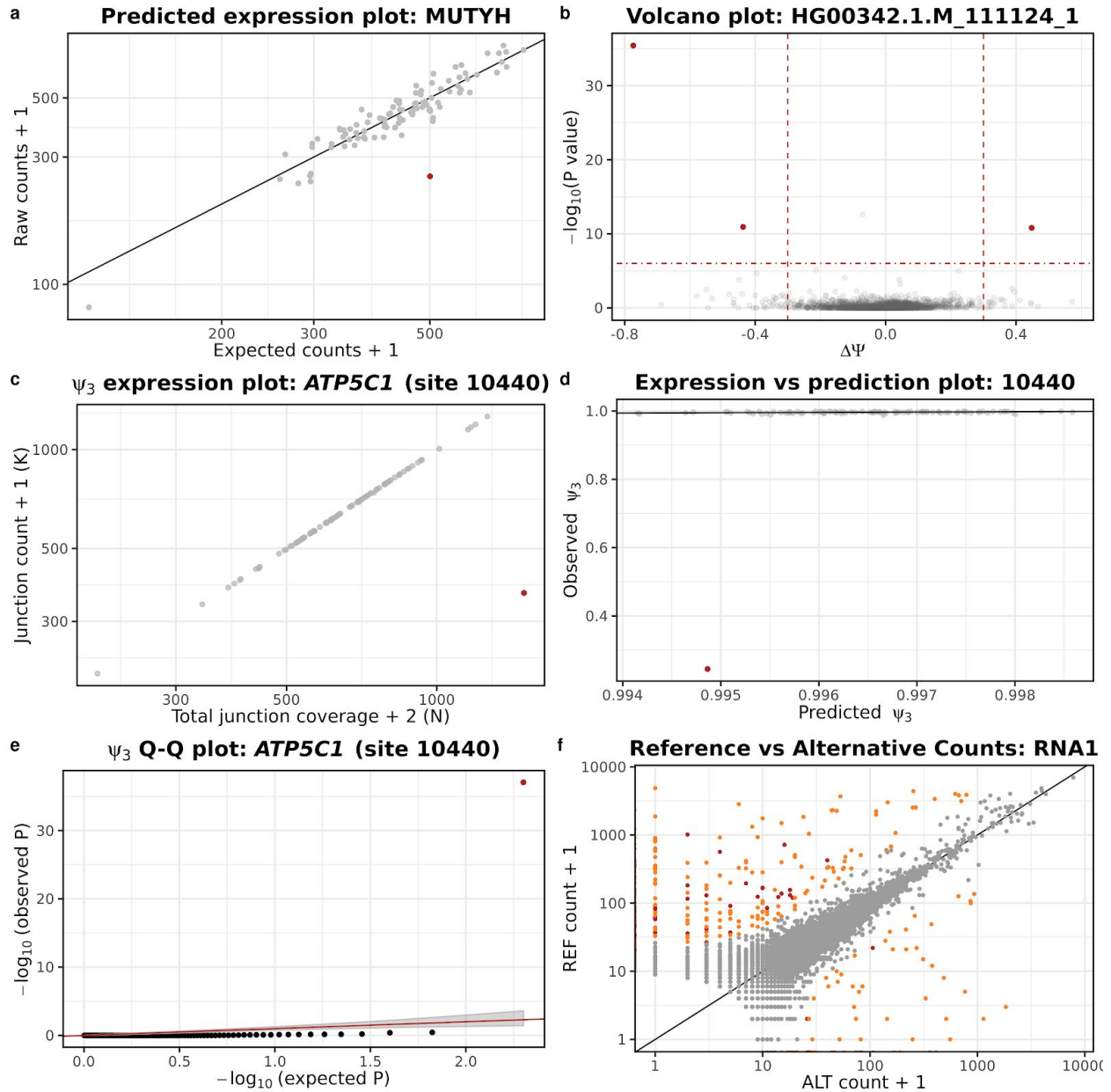


Fig. S4 | a, Raw (y-axis) versus expected (x-axis) counts of gene *MUTYH* showing one outlier (red). **b**, Negative log-transformed nominal P values (y-axis) versus $\Delta\psi_3$ values (x-axis) derived from all the splice sites (aggregated by gene) of sample HG00342.1.M_111124_1. Outliers are marked in red. **c**, Junction read counts (K , y-axis) plotted against the total split read coverage at the acceptor site (N , x-axis), of one junction in gene *ATP5C1*, which is the most severe outlier of panel (b). **d**, Observed (y-axis) versus expected (FRASER-predicted, x-axis) ψ_3 values of the same junction. **e**, Quantile-quantile plot of observed P values ($-\log_{10}$ scale, y-axis) against expected P values ($-\log_{10}$ scale, x-axis), with a 95% confidence band (gray) of the same junction. **f**, Counts assigned to the alternative (y-axis) versus reference allele (x-axis), highlighted by significance (orange) and significance and rarity (red), of sample HG00096.