

Description of plots in Family Summary

1. Summary Table:

This table lists all the chromosomes with abnormal (non-reference) transcription states that are identified in all cells from each family ("family_id"). Cell A and cell B denote MN cell and MN sister (for generation 1) and MN daughters (for generation 2). Cousin cells (for generation 2 only) are denoted as c1 and c2 (when available). Each row shows the normalized transcription level of the parental homolog ("MN_hap") of the chromosome with non-reference transcription in each cell. Note: Not all chromosomes with inferred abnormal transcription states arise from micronucleation. For details on the calculation of transcription levels, the identification of non-reference transcription states, and the inference of micronuclear chromosomes, see the Methods.

2. Genome Wide Allele Specific Expression:

This plot shows the normalized allelic expression levels of each chromosome in each cell of the family. The allelic expression is represented as the chromosomal average of the gene-level transcription ratio (observed transcription in each cell to the mean transcription level in control cells). The transcription levels from both parental homologs (A and B) are stacked to show both allelic transcription and the combined transcription yield of each chromosome.

The remaining plots are grouped for each chromosome with non-reference transcription as listed in the Summary Table. Plots for each chromosome include the following:

3a. Total (left) and allelic expression (right) in 10Mb bins across each chromosome:

The (normalized) expression is calculated similar to the chromosomal average but in 10Mb intervals. Blue and red dots represent the expression of each parental haplotype according to the color scheme shown in 2.

3b. (Left) Gene-level transcriptional variation:

This plot shows the ratio of the expression of each gene (combined expression from both homologs) compared to the mean expression level in control cells (TPM ratio). Each data point corresponds to a single gene: The x-axis value represents its mean TPM in control cells; the y-axis value shows the TPM ratio in a cell.

3c. (Right) Cumulative transcription of genes from low to high transcription

This plot shows the cumulative sum of TPM of genes from low to high transcription (based on the mean expression as in 3b) in a single cell in comparison to the cumulative TPM calculated from control cells. For normal transcription, the mean TPM ratio should be close to 1 (as shown in 3b), which corresponds to a slope close to 1 in the cumulative TPM plot (3c). The cumulative sum averages out transcriptional and technical noise (esp. for genes with low transcription levels) and reveals the average change in the total transcription output corresponding to a change in the number of actively transcribing chromosomes. The three lines represent the cumulative TPM values predicted for normal disomy (thick dash line), monosomy (thin dash line below the thick dash line), and trisomy (thin dash line above the thick dash line)