

**Supplementary Figure 1. Characterisation of translocation (A, B) and deletion (C, D, E) breakpoints.**

**(A,D)** Integrative Genomics Viewer (IGV) plots. The red vertical line in the top panel indicates which portion of the chromosome is displayed, and the blue line below shows the gene covered by the visible part of the chromosome. The third panel is the coverage over the displayed region, on scale 0-20 reads per base. The bottom panel shows all the aligned reads; gray colour indicates that the read sequence matched the reference, while the mismatched bases are shown in yellow, red, green and blue. Reads with 'rainbow' colour blocks suggest that one end of the read does not align at all, and is only partially mapped. Different colours for the entire reads are used to indicate unexpected mapping pattern, such as read pairs mapping to different chromosomes (**A**), or pairs with unexpectedly large or small insert sizes (**D**) resulting from deletions or insertions.

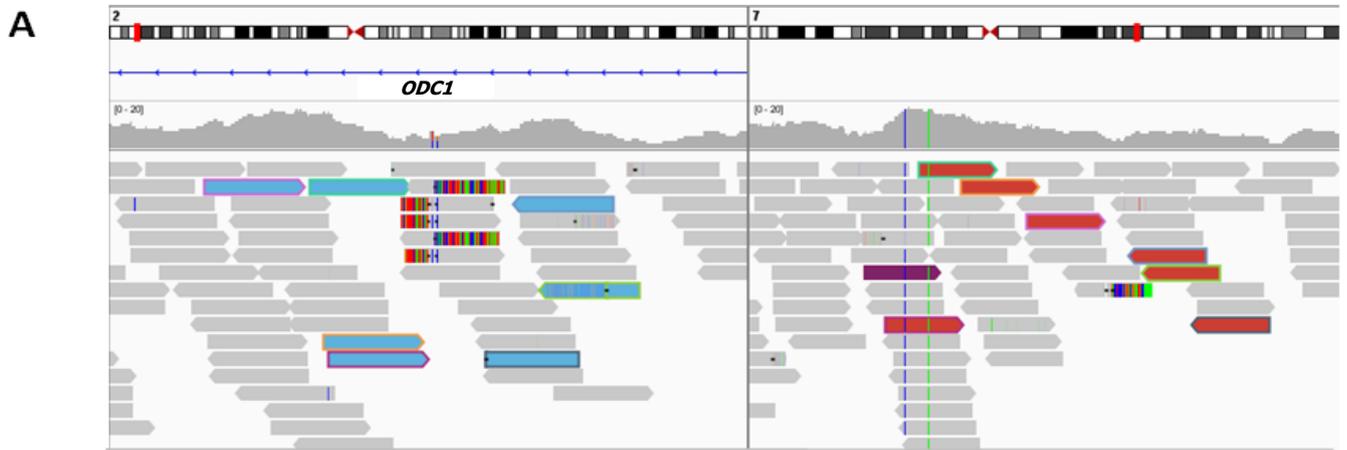
**(A)** IGV plot of family 1 translocation showing 7 read pairs with one of the pair mapping to chromosome 2 (left panel, blue reads), and the other to chromosome 7 (right panel, red reads). Additionally there are 6 partially mapped reads across the breakpoint. Purple read in the right panel is a read with a missing mate pair.

**(B)** Translocation breakpoint junction sequence analysis showing chromosome 7, der(2,7), and chromosome 2 sequences, respectively.

**(C)** Normalised coverage of exome sequence data over the chromosome 7 deletion, showing the exons with reduced coverage in the two affected individuals from family 2 (red and blue lines). Gray lines represent seven unrelated samples sequenced in the same way and used to normalise coverage.

**(D)** IGV plot of the reads around the deletion breakpoints within the *DYNC111* and *SLC25A13* genes in the proband, indicated by 4 read pairs with 106kb insert size (red). The two partially mapped reads resolve the breakpoints at the nucleotide level.

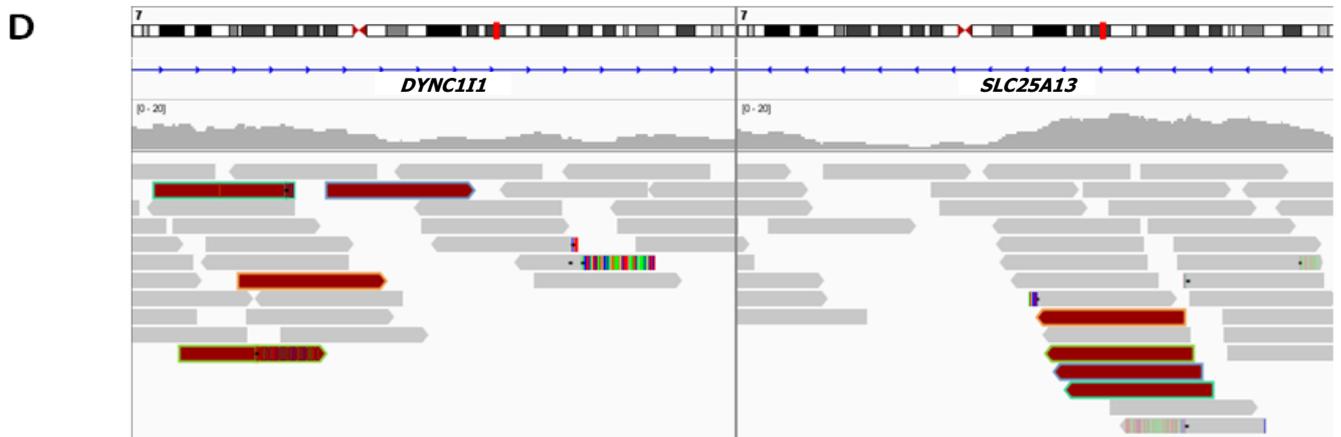
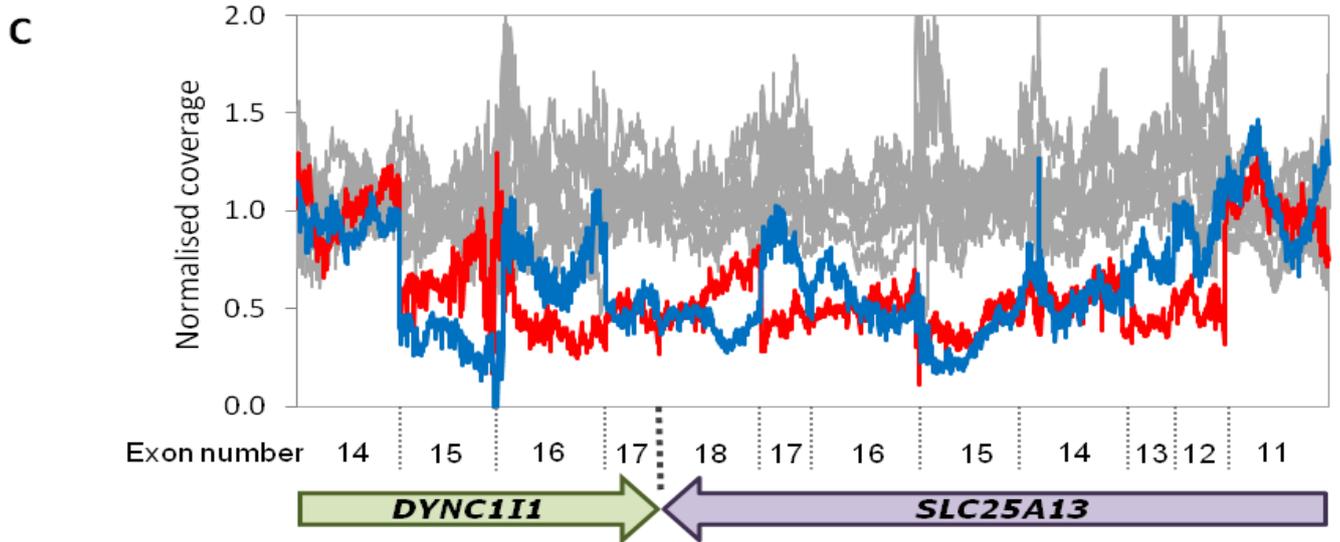
**(E)** Deletion breakpoint junction sequence analysis showing proximal, recombined and distal sequences, respectively.



**B**

chr7:96,229,309

chr7 (-)	TCTAAATTAATCCAGTAGCAACAGACTCGCGTGTTTC
Proband 1 der(2,7)	TCTAAATTAATCCAGTAGCAACCTCTGCCTCACTGCAACCTCTGCCTCCCGGGTTC
chr2 (+)	GCAGCGGTGCAATCTTGGCTCACTGCAACCTCTGCCTCCCGGGTTC
	chr2:10,585,833



**E**

chr7:95,704,809

chr7 (+)	TTACCTCCTGGAAGAGCAGTAATCCTCTTTAAAATATTTGCACGCATTCC
Proband 2 sequence	TTACCTCCTGGAAGAGCAGTAATCCTCTTCTTTAAAATAACCTTCATTCTAATG
	CTTGTGGTGGAGCACTACAAGATTCTTTAAAATAACCTTCATTCTAATG
chr7 (+)	chr7:95,810,746