

Supplementary material

Supplementary Table 1. List of genes in targeted capture experiments.

Supplementary Table 2. Parent-child segregation of SNPs in Cases 2-4 to confirm parentage

This was investigated where one parent is homozygous for the non-reference allele and the other is homozygous for the reference allele by finding the total number of sites where the child is heterozygous. The child should be heterozygous in almost 100% cases. This was performed for the mother-proband (maternal concordance) and father- proband (paternal concordance) and averaged (Column A average heterozygous concordance). This shows that the child is heterozygous at >99% of sites as would be expected for a child and their biological parents. Additionally, where the parents are both homozygous non-reference then, again as expected, the child is homozygous non-reference at >99% of sites (Column B: homozygousnonreference concordance).