

Description of Additional Supplementary Files

File name: Supplementary Data 1

Description: List of previous clinical assays that were performed for each proband, including exome sequencing (ES), whole genome sequencing (WGS) and chromosomal microarray (CMA).

File name: Supplementary Data 2

Description: Summary of leading rare GA4K alleles in phrank rankings and that map to OMIM genes, together with their pathogenic potential.

File name: Supplementary Data 3

Description: Sex and age reporting for study participants.