SUPPLEMENTARY MATERIAL

DETAILS OF THE IDENTIFIED GENETIC VARIANT IN THE PATIENT

Whole exome sequencing revealed a novel heterozygous missense likely pathogenic variant in *GCH1* gene (NM_001024024.1:c.449G>A;p.Gly150Glu). The variant is a novel variant, previously not found either in 1,000 genome project, ExAC database, gnomAD database or in the internal database in heterozygous/homozygous state. The position is strongly conserved. The variant is located in mutational hotspot, metal ion binding Zinc and it has 44 missense/in-frame variants. In silico assessments predicted deleterious by Sorting Intolerant from Tolerant (SIFT) and probably damaging by Polymorphism Phenotyping v2 (PolyPHEN-2). The Combined Annotation-Dependent Depletion (CADD) Phred score was 28. With these evidences, and according to the American College of Medical Genetics and Genomics (ACMG) criteria, the variant was classified likely pathogenic ($PM_{1,2}$, PP_{22}). Sanger sequencing in mother is pending.