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| **Table S1: Classification of reported genetic variants** |
| Disease causing variant | * Affected a known disease gene and
* Same variant had previously been described as causing disease, or the variant was similar to known disease-causing variants in that gene and
* Overlap in phenotype between our patient and the published patients with mutations in that gene
* Variants were classified as P, LP or VUS according to ACMG\*
 |
| Incidental finding | * Variant classified as P or LP according to ACMG\* in a disease-causing gene with an associated phenotype which was not (yet) present in the infant or
* Variant classified as VUS according to ACMG\* in an ‘actionable’ disease-causing gene (with an option of treatment and/or surveillance) which was not (yet) present in the infant and
* Associated disease has a childhood onset
 |

P, pathogenic; LP, likely pathogenic; VUS, Variant of Unknown Significance

\*VarSome, an adjusted automated interpretation tool (<https://varsome.com/>)[Kopanos C, et al. VarSome: The human genomic variant search engine. Bioinformatics. 2019;35:1978–1980] was used to classify all variants according to the guidelines of the American College of Medical Genetics (ACMG) [Richards S, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genet Med. 2015 May;17(5):405-24] either or in case of chromsomal deletions (\*) by manual application of the guidelines.