Supplementary

Table S1 List of VUS, PP or P variants

Gene symbol	OMIM code	Inheritance	HG 19 location	Transcript	Nucleotide and amino acid	Zygotic state	Population frequency	ACMG variation classification	Related diseases	Variation source
ADA2	607575	AR	chr22:17669245	NM_017424	c.1065C>A(p.F355L)	Heterozygote	0.003; East Asia	VUS	Autoimmune vasculitis-immunodeficiency-Blood deficiency syndrome/?Sneddon syndrome	Father
ADAR	146920	AR/AD	chr1:154560739	NM_001111	c.2886-5T>C	Heterozygote	<0.001	VUS	Aicardi Goutieres syndrome/ Dyschromatosis symmetrica hereditaria	Father
B4GALT1	137060	AR	chr9:33120603	NM_001497	c.650C>T(p.A217V)	Heterozygote	0.001; East Asia	VUS	Congenital disorder of glycosylation type IId	Mother
DUOX2	606759	AR	chr15:45396563	NM_014080	c.2335G>A(p.V779M)	Heterozygote	0.004; East Asia	VUS	Thyroid hormone production disorder type 6	Mother
FBN1	134797	AD	chr15:48717589	NM_000138	c.7430A>G(p.Q2477R)	Heterozygote	<0.001	VUS	Acromegaly dysplasia/Marfan syndrome	Father
G6PD	305900	XL	chrX:153762317	NM_000402	c.793C>T(p.L265F)	Heterozygote	<0.001	VUS	Glucose-6-phosphate Dehydrogenase	Mother
GYS2	138571	AR	chr12:21699327	NM_021957	c.1500T>A(p.C500*)	Heterozygote	–	PP	Hepatic glycogen storage type 0a	Mother
SLC37A4	602671	AR	chr11:118897728	NM_001164278	c.703G>T(p.V235L)	Heterozygote	0.001; East Asia	VUS	Glycogen storage disease type 1b/ Glycogen storage disease type 1c	Mother
SPATA5	613940	AR	chr4:123857310	NM_145207	c.1333C>T(p.R445*)	Heterozygote	<0.001	PP	Epilepsy-hearing impairment-mental retardation syndrome	Father
SRD5A2	607306	AR	chr2:31754395	NM_000348	c.680G>A(p.R227Q)	Heterozygote	0.006; East Asia	P variants	5α-reductase deficiency type 2	Mother
STEAP3	609671	AD	chr2:31754395	NM_182915	c.1492G>A(p.V498I)	Heterozygote	<0.001	VUS	Hypochromic anemia with iron overload type 2	Father

AR, autosomal recessive; AD, autosomal dominant; XL, X-linked; VUS, variant uncertain significance; PP, possibly pathogenic; P variants, pathogenic variants.

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