The detailed data are in supplement table1.

Annotation of the 18 suspected variants

NO	Chr	Start	End	Ref	Alt	Func.refGene	Gene.refGene	ExonicFunc.refGene	AAChange.refGene	OMIM phenotype	MAF gnomAD_exome_ALL	REVEL	SIFT		Polyphen2	
													Score	Prediction	Score	Prediction
1	chr13	20797486	20797486	C	G	exonic	GJB6	nonsynonymous SNV	GJB6:NM_001110221:exon3:c.G134C:p.G45A GJB6:NM_006783:exon3:c.G134C:p.G45A, GJB6:NM_001110220:exon4:c.G134C:p.G45A GJB6:NM_001110219:exon5:c.G134C:p.G45A	Name=Deafness; autosomal dominant 3B; 612643 （3）; Autosomal dominant; Deafness; autosomal recessive 1B; 612645 （3）; Autosomal recessive; Deafness; digenic GJB2/GJB6; 220290 （3）; Autosomal recessive; Ectodermal dysplasia 2; Clouston type; 129500（3）;	0	0.861	0	D	1	D

										Autosomal dominant						
2	chr14	91779741	91779741	G	A	exonic	CCDC88C	nonsynonymous SNV	CCDC88C:NM_001080414:exon15:c.C2419T:p.R807W	Name=Hydrocephalus; nonsyndromic; autosomal recessive; 236600 （ 3 ） ; Autosomal recessive; ?Spinocerebellar ataxia 40; 616053 （ 3 ） ; Autosomal dominant	0.0000168	0	0.01	D	0.776	P
3	chr14	55369143	55369143	C	T	exonic	GCH1	nonsynonymous SNV	GCH1:NM_000161:exon1:c.G239A:p.S80N, GCH1:NM_001024024:exon1:c.G239A:p.S80N GCH1:NM_001024070:exon1:c.G239A:p.S80N GCH1:NM_001024071:exon1:c.G239A:p.S80N	Name=Dystonia; DOPA-responsive; with or without hyperphenylalaninemia; 128230 （ 3 ） ; Autosomal recessive; Autosomal dominant; Hyperphenylalaninemia; BH4-deficient; B; 233910（3）; Autosomal recessive	0.00008603	0	0.29	T	0.002	B

4	chr8	145223221	145223221	G	A	exonic	MROH1	nonsynonymous SNV	MROH1:NM_001099281:exon3:c.G46A:p.A16T, MROH1:NM_001288814:exon3:c.G46A:p.A16T, MROH1:NM_001099280:exon4:c.G46A:p.A16T, MROH1:NM_032450:exon4:c.G46A:p.A16T	Not record	0.0005	0	0.75	T	0.555	P
5	chr8	139793199	139793199	G	C	exonic	COL22A1	nonsynonymous SNV	COL22A1:NM_152888:exon13:c.C1621G:p.P541A	Not record	0.0003	0	0.19	T	0.949	D
6	chr12	109725904	109725	C	T	exonic	FOXN4	nonsynonymous	FOXN4:NM_213596:exon4:c.G314A:p.R105	Not record	0.00001415	0	0.35	T	0.001	B

			904					SNV	Q							
7	chr12	110437560	110437560	C	G	exonic	ANKRD13A	nonsynonymous SNV	ANKRD13A:NM_033121:exon1:c.C67G:p.R23G	Not record	0.0002	0	0.25	T	0.667	P
8	chr11	125775454	125775454	C	T	exonic	DDX25	nonsynonymous SNV	DDX25:NM_013264:exon3:c.C137T:p.S46F	Not record	0.0002	0	0.11	T	0.216	B
9	chr14	74995691	74995691	C	T	exonic	LTBP2	nonsynonymous SNV	LTBP2:NM_000428:exon11:c.G2122A:p.E708K	Name=Glaucoma 3; primary congenital; D; 613086（3）; Microspherophakia and/or megalocornea; with ectopia lentis and with or without secondary glaucoma; 251750（3）; Autosomal recessive; ?Weill-Marchesani syndrome 3; recessive; 614819	0.00008417	0	0.9	T	0.001	B

										（3）; Autosomal recessive						
10	chr14	88411994	88411994	C	T	exonic	GALC	nonsynonymous SNV	GALC:NM_001201401:exon13:c.G1504A:p.E502K, GALC:NM_000153:exon14:c.G1573A:p.E525K, GALC:NM_001201402:exon14:c.G1495A:p.E499K	Name=Krabbe disease; 245200 （3）; Autosomal recessive	0.0002	0	0.87	T	0.021	B
11	chr4	92456571	92456711	T	C	exonic	USP17L17	nonsynonymous SNV	USP17L17:NM_001256857:exon1:c.T67C:p.S23P	Not record	0.2839	0	0.25	T	0	Not available
12	chr22	37693732	37693732	G	A	intronic	CYTH4	Not available	Not available	Not record	0.0002	0	0	Not available	0	Not available

13	chr14	60213041	60213041	C	T	exonic	RTN1	nonsynonymous SNV	RTN1:NM_021136:exon2:c.G400A:p.V134I	Not record	0.00002439	0	0.64	T	0.001	B
14	chr22	32003941	32003941	G	A	exonic	SFI1	nonsynonymous SNV	SFI1:NM_001258325:exon20:c.G2011A:p.E671K, SFI1:NM_001258326:exon20:c.G1930A:p.E644K, SFI1:NM_001258327:exon20:c.G1894A:p.E632K, SFI1:NM_014775:exon21:c.G2083A:p.E695K, SFI1:NM_001007467:exon22:c.G2176A:p.E72	Not record	0.0005	0	0.07	T	0.999	D

									6K							
15	chr12	121880596	121880596	T	C	exonic	KDM2B	nonsynonymous SNV	KDM2B:NM_001005366:exon18:c.A2441G:p.N814S, KDM2B:NM_032590:exon19:c.A2648G:p.N883S	Not record	0.00001193	0	0.44	T	0.002	B
16	chr14	55647464	55647464	T	G	exonic	DLGAP5	nonsynonymous SNV	DLGAP5:NM_001146015:exon6:c.A613C:p.M205L, DLGAP5:NM_014750:exon6:c.A613C:p.M205L	Not record	0.00008121	0	0.68	T	0.001	B
17	chr18	12097679	12097679	G	A	exonic	ANKRD62	nonsynonymous SNV	ANKRD62:NM_001277333:exon5:c.G655A:p.V219I	Not record	0	0	1	T	0	Not available

18	chr12	112130610	112130610	C	T	exonic	ACAD10	stopgain	ACAD10:NM_001136538:exon2:c.C97T:p.R33X,ACAD10:NM_025247:exon2:c.C97T:p.R33X	Not record	0.0005	0	1	T	0	Not available

Note: Func.refGene: The functional region of the reference gene where the variant is located; Gene.refGene: The name of the reference gene where the variant is located; ExonicFunc.refGene: Types of variants in exonic regions; Omim_phenotype: The phenotype corresponding to the gene (not the variant) in the OMIM database; MAF gnomAD_exome_ALL: Minor Allele Frequency of each variant in Genome Aggregation Database; REVEL (Rare Exome Variant Ensemble Learner) : the predicted score range is 0-1; The SIFT (Sorting Intolerant From Tolerant) score range is 0-1, the position with score < 0.05 is predicted as harmful D: deleterious, and the position with score ≥ 0.05 is predicted as harmless T: tolerated;Polyphen2 (Polymorphism Phenotyping v2)_ Hvar prediction results D: potentially harmful, score ≥ 0.909, P: potentially harmful, score≤ 0.447, B: benign.