Supplementary Table 2. Population frequency, ClinVar status, and ACMG/AMP criteria for variants identified by whole exome/genome sequencing

Pt. No.	Gene	Reference transcript	Nucleotide change	HGVS protein change	gnomAD allele frequency	ClinVar status	ACMG/AMP criteria applied	Final classification
1	*ARID1B*	NM_020732.3	c.2201dupG	p.(Ser736Ilefs*27)	Not found	Our first reported case	PVS1, PM2, PM6, PP4	P
2	*SLC9A6*	NM_001330652.2	c.675_676ins106	p.(Asp226Serfs*2)	Not found	Our first reported case	PVS1, PM2, PP4	LP
3	*SYNGAP1*	NM_006772.3	c.1715G>A	p.(Trp572*)	Not found	Our first reported case	PVS1, PM2, PP4	LP
4	*SCN2A*	NM_021007.3	c.4978T>G	p.(Leu1660Val)	Not found	Our first reported case	PM2, PM5, PM6, PP3, PP4	LP
5	*NSD1*	NM_022455.5	c.5798A>G	p.(Asn1933Ser)	Not found	Reported	PM2, PP3, PP4, PP5	LP
6	*DCX*	NM_000555.3	c.358C>T	p.(Arg120*)	<0.00001	Reported	PVS1, PM2, PM6, PP4	P
7	*SLC6A8*	NM_005629.4	c.626_627delCT	p.(Pro209Argfs*87)	Not found	Reported	PVS1, PM2, PP4	P
8	*CSNK2A1*	NM_177559.3	c.593A>G	p.(Lys198Arg)	<0.00001	Reported	PS4, PM2, PP4	LP
9	*TAF1*	NM_001286074.2	c.3701G>A	p.(Arg1234Gln)	Not found	Our first reported case	PM2, PP3, PP4	LP
10	*NR2F1*	NM_005654.6	c.983dupT	p.(Thr329Hisfs*68)	Not found	Our first reported case	PVS1, PM2, PP4	LP
11	*NF1*	NM_001042492.3	c.5812+332A>G	p.(?)	<0.00001	Reported	PS2, PM2, PP4	P
12	*FLNA*	NM_001110556.2	c.2752dup	p.(Asp918Glyfs*13)	Not found	Reported	PVS1, PM2, PM6, PP4	P
13	*DYRK1A*	NM_001396.4	c.665-3C>G	p.(?)	Not found	Not reported	PM2, PM6, PP3, PP4	LP
14	*CASK*	NM_003688.3	c.535del	p.(Arg179Valfs*22)	Not found	Our first reported case	PVS1, PM2, PM6, PP4	P
15	*NEB*	NM_001271208.2	c.[5364G>A]; [21623G>T]	p.(Trp1788*); (Ser7208Ile)	<0.00001/ Not found	Reported/Not reported	PVS1, PM2, PP3, PP4/ PM2, PM3, PP1, PP3, PP4	P/LP
16	*MED12*	NM_005120.3	c.3443G>A	p.(Arg1148His)	Not found	Reported	PS2, PM2, PM5, PP3, PP4	P
17	*CACNA1A*	NM_001127222.2	c.3134C>G	p.(Ser1045*)	Not found	Our first reported case	PVS1, PM2, PM6, PP4	P
18	*CREBBP*	NM_004380.3	c.1270C>T	p.(Arg424*)	Not found	Reported	PVS1, PM2, PM6, PP4	P
19	*CDC42*	NM_001791.4	c.67T>C	p.(Tyr23His)	Not found	Not reported	PS2, PM2, PM5, PP3, PP4	P
20	*LAMA1*	NM_005559.4	c.4252_4255dup	p.(Cys1419*)	<0.00001	Not reported	PVS1, PM2, PM3, PP3, PP4	P
21	*SLC6A8*	NM_005629.4	c.1395_1397del	p.(Gly466del)	Not found	Not reported	PVS1, PM2, PP4,	P
22	*PTPN11*	NM_002834.5	c.922A>G	p.(Asn308Asp)	0.00001	Reported	PS1, PM1, PP4	P
23	*FLNA*	NM_001456.4	c.7895G>T	p.(Ser2640Ile)	Not found	Reported	PM2, PP1, PP3, PP4, PP5	LP
24	*SCARB2*	NM_005506.4	c.994+1G>T	p.(?)	<0.00001	Not reported	PVS1, PM2, PP3, PP4	P
25	*SON*	NM_138927.4	c.1193del	p.(Pro398Leufs*2)	Not found	Not reported	PVS1, PM2, PM6, PP4	P

ACMG/AMP, American College of Medical Genetics and Genomics and Association for Molecular Pathology; LP, likely pathogenic; P, pathogenic; PM, pathogenic moderate evidence; PP, pathogenic supporting evidence; PS, pathogenic strong evidence; Pt., patient; PVS, pathogenic very strong evidence.
Allele frequencies were obtained from the Genome Aggregation Database (gnomAD, https://gnomad.broadinstitute.org). Variants absent from gnomAD were annotated as "not found." ClinVar status was assessed using the ClinVar database (https://www.ncbi.nlm.nih.gov/clinvar). Variant classification was performed according to the ACMG/AMP guidelines.