Chr	Start	End	Ref	Alt	Gene.refGene	GeneDetail.refGene
Homozygous variants in regions mapped by homozygosity mapping						
1	165173160	165173160	A	G	LMX1A*	.
5	112179055	112179055	T	G	APC**	.
Genome-wide homozygous variants						
5	1411381	1411381	G	A	SLC6A3**	.
6	43336941	43336941	G	A	ZNF318**	.
6	116442293	116442293	C	T	COL10A1**	.
X-linked variants						
X	49126604	49126604	G	A	PPP1R3F***	.
X	111698023	111698023	C	A	RTL4**	.
Possible compound heterozygous variants (Phase unknown)						
1	223177972	223177972	G	A	DISP1**	.
1	223164937	223164937	A	G	DISP1**	.
8	144812619	144812619	C	T	FAM83H**	.
8	144808567	144808567	G	A	FAM83H**	.

*Segregates with hearing loss in the family

**Does not segregate with hearing loss in the family

***Untested for segregation

AAChange.refGene	Func.ensGene	Gene.ensGene.1	GeneDetail.ensGene	ExonicFunc.ensGene	AAChange.ensGene
LMX1A:NM_00117406	exonic	ENSG000001	.	nonsynonym	ENSG00000162761:EI
APC:NM_001127511:e	exonic	ENSG000001	.	nonsynonym	ENSG00000134982:EI
SLC6A3:NM_001044:ex	exonic	ENSG000001	.	nonsynonym	ENSG00000142319:EI
ZNF318:NM_014345:e	exonic	ENSG000001	.	nonsynonym	ENSG00000171467:EI
COL10A1:NM_000493:	exonic	ENSG000001	.	nonsynonym	ENSG00000123500:EI
PPP1R3F:NM_033215:e	exonic	ENSG000000	.	nonsynonym	ENSG00000049769:EI
RTL4:NM_001004308:e	exonic	ENSG000001	.	nonsynonym	ENSG00000187823:EI
DISP1:NM_001350630:	exonic	ENSG000001	.	nonsynonym	ENSG00000154309:EI
DISP1:NM_032890:exo	exonic	ENSG000001	.	nonsynonym	ENSG00000154309:EI
FAM83H:NM_198488:e	exonic	ENSG000001	.	nonsynonym	ENSG00000180921:EI
FAM83H:NM_198488:e	exonic	ENSG000001	.	nonsynonym	ENSG00000180921:EI

avsnp147	SIFT_score	SIFT_pred	Polyphen2_HDIV_score	Polyphen2_HDIV_pred	Polyphen2_HVAR_score	Polyphen2_HVAR_pred	LRT_score	LRT_pred	MutationTaster_score
rs763320093	0	D	1	D	0.996	D	0	D	1
rs757073062	0.339	T	0.947	P	0.707	P	0	D	0.996
rs756571192	0.006	D	0.064	B	0.098	B	0	D	1
.	0	D	1	D	0.968	D	.	.	0.986
.	0	D	1	D	1	D	0	D	1
rs782119228	0.354	T	0.365	B	0.133	B	0.003	U	1
.	0.051	T	0.206	B	0.164	B	0.2	N	1
rs150111973	0.055	T	0.977	D	0.581	P	0	N	1
rs139954139	0.026	D	0.979	D	0.771	P	0	D	1
rs375875154	0.003	D	0.84	P	0.429	B	0.078	N	0.945
rs781867409	0.001	D	0.816	P	0.048	B	0.007	U	1

MutationTaster_pred	MutationAssessor_score	MutationAssessor_pred	FATHMM_score	FATHMM_pred	PROVEAN_score	PROVEAN_pred	CADD_phred	DANN_score	fathmm.MKL_coding_score	fathmm.MKL_coding_pred
D	1.83	L	-2.57	D	-4.19	D	26.8	0.998	0.991	D
D	0.895	L	-2.41	D	0.27	N	22.8	0.987	0.807	D
D	1.35	L	-1.1	T	-3.08	D	23.6	0.997	0.825	D
D	0.55	N	3.75	T	-0.99	N	23	0.985	0.674	D
D	3.985	H	-6.28	D	-5.89	D	24	0.993	0.963	D
D	0	N	0.37	T	-0.29	N	16.4	0.972	0.249	N
N	1.04	L	0.54	T	-0.51	N	21.3	0.989	0.025	N
D	1.385	L	2.33	T	-2.33	N	27.6	1	0.99	D
D	1.15	L	-3.03	D	-1.89	N	24.6	0.998	0.991	D
N	1.795	L	2.73	T	-2.19	N	22.3	0.999	0.391	N
D	1.04	L	1.74	T	-4.81	D	23	0.999	0.701	D

MetaSVM_score	MetaSVM_pred	MetaLR_score	MetaLR_pred	integrated_fitCons_score	integrated_confidence_value	GERP..._RS	phyloP7way_vertebrate	phyloP20way_mammalian	phastCons7way_vertebrate	phastCons20way_mammalian
0.655	D	0.739	D	0.487	0	5.14	1.062	1.199	0.998	1
0.072	D	0.606	D	0.706	0	4.81	0.991	1.061	1	1
-0.406	T	0.357	T	0.516	0	4.5	0.917	0.893	0.999	0.997
-0.945	T	0.013	T	0.243	2	2.96	0.846	0.726	0.559	0.779
0.966	D	0.993	D	0.554	0	5.12	0.871	0.935	0.999	0.997
-1.084	T	0.065	T	.	.	2.6	0.546	0.929	0.997	1
-1.014	T	0.103	T	.	.	1.19	0.598	0.935	0.482	0.804
-1.189	T	0.06	T	0.651	0	5.95	0.917	0.998	0.983	0.992
0.652	D	0.755	D	0.638	0	5.64	1.062	1.199	1	1
-1.084	T	0.039	T	0.635	0	3.07	0.598	0.935	0.432	0.998
-1.012	T	0.045	T	0.66	0	3.94	0.913	0.947	0.026	0.016

SiPhy_29way_logOdds	dbscSNV_ADA_SCORE	dbscSNV_RF_SCORE
14.785	.	.
12.05	.	.
14.709	.	.
9.486	.	.
19.108	.	.
8.929	.	.
5.811	.	.
20.372	.	.
16.152	.	.
7.962	.	.
8.349	.	.

LIM homeobox transcription factor 1 alpha [Source:HGNC Symbol;Acc:HGNC:6653]
APC, WNT signaling pathway regulator [Source:HGNC Symbol;Acc:HGNC:583]

solute carrier family 6 member 3 [Source:HGNC Symbol;Acc:HGNC:11049]
zinc finger protein 318 [Source:HGNC Symbol;Acc:HGNC:13578]
collagen type X alpha 1 chain [Source:HGNC Symbol;Acc:HGNC:2185]

protein phosphatase 1 regulatory subunit 3F [Source:HGNC Symbol;Acc:HGNC:14944]
retrotransposon Gag like 4 [Source:HGNC Symbol;Acc:HGNC:25214]

dispatched RND transporter family member 1 [Source:HGNC Symbol;Acc:HGNC:19711]
dispatched RND transporter family member 1 [Source:HGNC Symbol;Acc:HGNC:19711]
family with sequence similarity 83 member H [Source:HGNC Symbol;Acc:HGNC:24797]
family with sequence similarity 83 member H [Source:HGNC Symbol;Acc:HGNC:24797]

Clinical Genomic Database (CGD) condition description	Genotype	gnomAD_genome_ALL
	1/1	3.23E-05
Familial adenomatous polyposis; Gardner syndrome; Desmoid disease, here	1/1	.
Parkinsonism-dystonia, infantile	1/1	.
	1/1	.
Metaphyseal chondrodysplasia, Schmid type	1/1	.
	1/1	.
	1/1	.
	0/1	0.0009
	0/1	.
Amelogenesis imperfecta, type 3	0/1	0.0002
Amelogenesis imperfecta, type 3	0/1	.

gnomAD_genome_AFR	gnomAD_genome_AMR	gnomAD_genome_ASJ	gnomAD_genome_EAS	gnomAD_genome_FIN	gnomAD_genome_NFE	gnomAD_genome_OTH	gnomAD_exome_ALL	gnomAD_exome_AFR	gnomAD_exome_AMR
0	0	0	0	0	6.66E-05	0	1.63E-05	0	0
.	.	.	.	.	.	.	4.07E-06	0	0
.	.	.	.	.	.	.	6.56E-06	0	0
.	.	.	.	.	.	.	.	.	.
.	.	.	.	.	.	.	.	.	.
.	.	.	.	.	.	.	2.58E-05	0	0
.	.	.	.	.	.	.	.	.	.
0	0	0	0	0	0.0019	0	0.0003	0	5.96E-05
.	.	.	.	.	.	.	6.51E-05	0	0
0.0001	0	0	0	0	0.0003	0	0.0004	6.59E-05	2.98E-05
.	.	.	.	.	.	.	2.66E-05	0	0

gnomAD_exome_ASJ	gnomAD_exome_EAS	gnomAD_exome_FIN	gnomAD_exome_NFE	gnomAD_exome_OTH	gnomAD_exome_SAS	PopFreqMax	GME_AF	GME_NWA	GME_NEA
0	0	0	2.69E-05	0.0002	0	0	.	.	.
0	0	0	0	0	3.25E-05	0.0001	.	.	.
0	0	0	0	0	4.36E-05	0.0001	.	.	.
.	.	.	.	.	.	.	.	.	.
.	.	.	.	.	.	.	.	.	.
0	0	0	0	0.0011	0	0	0.00072	0	0
.	.	.	.	.	.	.	.	.	.
0.0011	5.80E-05	0	0.0004	0	0.0005	0.0011	0.0005	0	0
0.0011	0	0	4.49E-05	0	0	0.0001	.	.	.
0	5.82E-05	0	3.61E-05	0.0006	0.003	0.003	0.00101	0	0
0	0	0	2.01E-05	0	0.0001	0.0001	.	.	.

GME_AP	GME_Israel	GME_SD	GME_TP	GME_CA
.	.	.	.	.
.	.	.	.	.
.	.	.	.	.
.	.	.	.	.
.	.	.	.	.
0.00407	0	0	0	0
.	.	.	.	.
0.00292	0	0	0	0
.	.	.	.	.
0	0	0	0	0.00379
.	.	.	.	.