

Sunshine Genetics Tier 2 Gene – Condition List

Clinical Area	Condition Name	Gene	Inheritance
Hearing Loss	Deafness, autosomal dominant 20/26	ACTG1	AD
Hearing Loss	Baraitser-Winter syndrome 2	ACTG1	AD
Hearing Loss	Combined oxidative phosphorylation deficiency 6	AIFM1	XLR
Hearing Loss	Cowchock syndrome	AIFM1	XLR
Hearing Loss	Deafness, X-linked 5	AIFM1	XLR
Hearing Loss	Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy	AIFM1	XLR
Hearing Loss	Alstrom syndrome	ALMS1	AR
Hearing Loss	KBG syndrome	ANKRD11	AD
Hearing Loss	ATP2B2-related disorders	ATP2B2	AD
Hearing Loss	Bjornstad syndrome or related conditions	BCS1L	AR
Hearing Loss	Deafness, autosomal recessive 93	CABP2	AR
Hearing Loss	CDC14A-related hearing loss	CDC14A	AR
Hearing Loss	Deafness, autosomal dominant 4B	CEACAM16	AD

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Hearing Loss	Deafness, autosomal recessive 113	CEACAM16	AR
Hearing Loss	Cone-rod dystrophy and hearing loss 2	CEP250	AR
Hearing Loss	CEP78-related cone-rod dystrophy and hearing loss	CEP78	AR
Hearing Loss	CLDN14-related hearing loss	CLDN14	AR
Hearing Loss	CLPP-related Perrault syndrome	CLPP	AR
Hearing Loss	COCH-related hearing loss	COCH	AR
Hearing Loss	COL11A1-related disorders	COL11A1	AD
Hearing Loss	Fibrochondrogenesis 1	COL11A1	AR
Hearing Loss	COL11A2-related otospondylomegaepiphyseal dysplasia or deafness	COL11A2	AD
Hearing Loss	COL11A2-related otospondylomegaepiphyseal dysplasia or deafness	COL11A2	AR
Hearing Loss	Fibrochondrogenesis 2	COL11A2	AD
Hearing Loss	X-linked Alport syndrome	COL4A5	XL
Hearing Loss	COL9A1-related disorders	COL9A1	AR

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Hearing Loss	Stickler syndrome, type VI	COL9A3	AR
Hearing Loss	DSPP-related disorders	DSPP	AD
Hearing Loss	Waardenburg syndrome, Type 4B	EDN3	AR
Hearing Loss	Waardenburg syndrome, Type 4A	EDNRB	AR
Hearing Loss	EPS8-related hearing loss	EPS8	AR
Hearing Loss	EPS8L2-related hearing loss	EPS8L2	AR
Hearing Loss	Deafness, autosomal recessive 36	ESPN	AR
Hearing Loss	ESRRB-related hearing loss	ESRRB	AR
Hearing Loss	EYA1-related disorders	EYA1	AD
Hearing Loss	FGF3-related hearing loss with inner ear agenesis, microtia, and microdontia	FGF3	AR
Hearing Loss	GIPC3-related hearing loss	GIPC3	AR
Hearing Loss	GPSM2-related Chudley-McCullough syndrome	GPSM2	AR
Hearing Loss	Deafness, autosomal dominant 28	GRHL2	AD

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Hearing Loss	Ectodermal dysplasia/short stature syndrome	GRHL2	AR
Hearing Loss	GRXCR1-related hearing loss	GRXCR1	AR
Hearing Loss	GRXCR2-related hearing loss	GRXCR2	AR
Hearing Loss	Autosomal dominant deafness-5	GSDME	AD
Hearing Loss	HGF-related hearing loss	HGF	AR
Hearing Loss	D-bifunctional protein deficiency	HSD17B4	AR
Hearing Loss	ILDR1-related hearing loss	ILDR1	AR
Hearing Loss	Deafness, autosomal dominant 69, unilateral or asymmetric	KITLG	AD
Hearing Loss	Waardenburg syndrome, type 2F	KITLG	AR
Hearing Loss	LARS2-related disorders	LARS2	AR
Hearing Loss	LHFPL5-related hearing loss	LHFPL5	AR
Hearing Loss	Deafness, autosomal dominant 7	LMX1A	AD
Hearing Loss	Autosomal recessive deafness 77	LOXHD1	AR

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Hearing Loss	LRTOMT-related hearing loss	LRTOMT	AR
Hearing Loss	MARVELD2-related hearing loss	MARVELD2	AR
Hearing Loss	MIR96-related hearing loss	MIR96	AD
Hearing Loss	Waardenburg syndrome, type 2A / Tietz albinism-deafness syndrome / COMMAD syndrome	MITF	AD
Hearing Loss	MPZL2-related hearing loss	MPZL2	AR
Hearing Loss	MSRB3-related hearing loss	MSRB3	AR
Hearing Loss	MYH14-related hearing loss	MYH14	AD
Hearing Loss	MYH9-related disorders	MYH9	AD
Hearing Loss	MYO15A-related nonsyndromic hearing loss and deafness	MYO15A	AR
Hearing Loss	MYO3A-related hearing loss	MYO3A	AD
Hearing Loss	MYO3A-related hearing loss	MYO3A	AR
Hearing Loss	MYO6-related deafness	MYO6	AD
Hearing Loss	MYO6-related deafness	MYO6	AR

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Hearing Loss	Deafness, autosomal dominant 67	OSBPL2	AD
Hearing Loss	OTOA-related nonsyndromic hearing loss	OTOA	AR
Hearing Loss	Deafness, autosomal recessive 18B	OTOG	AR
Hearing Loss	Deafness, autosomal recessive 84B	OTOGL	AR
Hearing Loss	PAX3-Related Waardenburg Syndrome	PAX3	AD
Hearing Loss	PAX3-Related Waardenburg Syndrome	PAX3	AR
Hearing Loss	PDZD7-related disorders	PDZD7	AR
Hearing Loss	PJVK-related nonsyndromic hearing loss	PJVK	AR
Hearing Loss	POU3F4-related nonsyndromic hearing loss	POU3F4	XL
Hearing Loss	Phosphoribosylpyrophosphate synthetase I superactivity	PRPS1	XL
Hearing Loss	PRPS1 deficiency disorder	PRPS1	XL
Hearing Loss	PTPRQ-related hearing loss	PTPRQ	AR
Hearing Loss	Autosomal recessive Deafness, 24	RDX	AR

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Hearing Loss	RIPOR2-related hearing loss	RIPOR2	AR
Hearing Loss	S1PR2-related hearing loss	S1PR2	AR
Hearing Loss	SERPINB6-related hearing loss	SERPINB6	AR
Hearing Loss	Branchiootic syndrome 3	SIX1	AD
Hearing Loss	Deafness, autosomal dominant 23	SIX1	AD
Hearing Loss	SLC17A8-related nonsyndromic hearing loss	SLC17A8	AD
Hearing Loss	SLITRK6-related hearing loss and myopia	SLITRK6	AR
Hearing Loss	SMPX-related hearing loss	SMPX	XL
Hearing Loss	SOX10-related disorders	SOX10	AD
Hearing Loss	Deafness-16	STRC	AR
Hearing Loss	SYNE4-related nonsyndromic hearing loss	SYNE4	AR
Hearing Loss	TBC1D24-related disorders	TBC1D24	AR
Hearing Loss	Treacher Collins syndrome 1	TCOF1	AD

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Hearing Loss	TECTA-related deafness	TECTA	AD
Hearing Loss	TECTA-related deafness	TECTA	AR
Hearing Loss	TIMM8A-related Mohr-Tranebjaerg syndrome	TIMM8A	XLR
Hearing Loss	TMC1-related nonsyndromic hearing loss	TMC1	AD
Hearing Loss	TMC1-related nonsyndromic hearing loss	TMC1	AR
Hearing Loss	TMIE-related hearing loss	TMIE	AR
Hearing Loss	TMPRSS3-related nonsyndromic hearing loss	TMPRSS3	AR
Hearing Loss	TPRN-related hearing loss	TPRN	AR
Hearing Loss	Deafness, autosomal recessive 28	TRIOBP	AR
Hearing Loss	Leber congenital amaurosis with early-onset deafness	TUBB4B	AD
Hearing Loss	TWINK-related disorders	TWINK	AR
Hearing Loss	WFS1-related disorders	WFS1	AD
Hearing Loss	WFS1-related disorders	WFS1	AR

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Epilepsy	GABA-transaminase deficiency	ABAT	AR
Epilepsy	Succinic Semialdehyde Dehydrogenase Deficiency	ALDH5A1	AR
Epilepsy	Developmental and epileptic encephalopathy 8	ARHGEF9	XL
Epilepsy	Asparagine synthetase deficiency	ASNS	AR
Epilepsy	Canavan disease	ASPA	AR
Epilepsy	BRAT1-related disorders	BRAT1	AR
Epilepsy	Developmental and epileptic encephalopathy 42	CACNA1A	AD
Epilepsy	CACNA1A-related disorders	CACNA1A	AD
Epilepsy	Episodic ataxia, type 2	CACNA1A	AD
Epilepsy	Familial hemiplegic migraine 1	CACNA1A	AD
Epilepsy	CACNA1E-associated disorders	CACNA1E	AD
Epilepsy	Early infantile encephalopathy type 2	CDKL5	XLD
Epilepsy	DNM1-related developmental and epileptic encephalopathy	DNM1	AD

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Epilepsy	DNM1-related developmental and epileptic encephalopathy	DNM1	AR
Epilepsy	FLNA-related otopalatodigital spectrum disorders	FLNA	XL
Epilepsy	GABRA1-related disorders	GABRA1	AD
Epilepsy	Developmental and epileptic encephalopathy 43	GABRB3	AD
Epilepsy	Dystonia, DOPA-responsive	GCH1	AD
Epilepsy	Tetrahydrobiopterin deficiency	GCH1	AR
Epilepsy	GNAO1-related neurodevelopmental disorder	GNAO1	AD
Epilepsy	GRIN1-related neurodevelopmental disorders	GRIN1	AD
Epilepsy	GRIN1-related neurodevelopmental disorders	GRIN1	AR
Epilepsy	Focal epilepsy and speech disorder (FESD) with or without impaired intellectual development	GRIN2A	AD
Epilepsy	GRIN2B-related neurodevelopmental disorders	GRIN2B	AD
Epilepsy	Developmental and epileptic encephalopathy 46	GRIN2D	AD
Epilepsy	Methylmalonic acidemia and homocystinuria type cblX	HCFC1	XLR

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Epilepsy	Developmental and epileptic encephalopathy 54	HNRNPU	AD
Epilepsy	KCNA2-related neurological disorders	KCNA2	AD
Epilepsy	Developmental and epileptic encephalopathy-26	KCNB1	AD
Epilepsy	KCNH1-related disorder	KCNH1	AD
Epilepsy	KCNT1-related epilepsy	KCNT1	AD
Epilepsy	Hyperglycinemia, lactic acidosis, and seizures	LIAS	AR
Epilepsy	MECP2-related neurodevelopmental disorder	MECP2	XL
Epilepsy	MOCS2-related molybdenum cofactor deficiency B	MOCS2	AR
Epilepsy	Smith-Kingsmore syndrome	MTOR	AD
Epilepsy	PAFAH1B1-Related Lissencephaly / Subcortical Band Heterotopia	PAFAH1B1	AD
Epilepsy	Developmental and epileptic encephalopathy 9	PCDH19	XLD
Epilepsy	PIGA-related disorders	PIGA	XLR
Epilepsy	PLCB1-related genetic developmental and epileptic encephalopathy	PLCB1	AR

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Epilepsy	PNKP-related disorders	PNKP	AR
Epilepsy	PPT1-related neuronal ceroid lipofuscinosis	PPT1	AR
Epilepsy	PSAT1-related phosphoserine aminotransferase deficiency	PSAT1	AR
Epilepsy	PSPH-related phosphoserine phosphatase deficiency	PSPH	AR
Epilepsy	Neurodevelopmental disorder with neonatal respiratory insufficiency, hypotonia, and feeding difficulties	PURA	AD
Epilepsy	RARS2-related pontocerebellar hypoplasia	RARS2	AR
Epilepsy	Developmental and epileptic encephalopathy 64	RHOBTB2	AD
Epilepsy	Developmental and epileptic encephalopathy 64	RHOBTB2	AR
Epilepsy	SCN2A-related complex neurodevelopmental disorder	SCN2A	AD
Epilepsy	SCN3A-related neurological disorders	SCN3A	AD
Epilepsy	SLC25A22-related developmental and epileptic encephalopathy	SLC25A22	AR
Epilepsy	Developmental and epileptic encephalopathy 85, with or without midline brain defects	SMC1A	XL
Epilepsy	Cornelia de Lange syndrome 2	SMC1A	XL

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Epilepsy	Developmental and epileptic encephalopathy 117	SNAP25	AD
Epilepsy	Generalized epilepsy with febrile seizures plus, type 9	STX1B	AD
Epilepsy	Autosomal dominant intellectual developmental disorder 5	SYNGAP1	AD
Epilepsy	Developmental and epileptic encephalopathy 18	SZT2	AR
Epilepsy	TBL1XR1-related neurodevelopmental disorders	TBL1XR1	AD
Epilepsy	Lissencephaly 3	TUBA1A	AD
Epilepsy	Angelman syndrome	UBE3A	AD
Miscellaneous	Wilson disease	ATP7B	AR
Miscellaneous	GM1-gangliosidosis or related conditions	GLB1	AR
Miscellaneous	Tay-Sachs disease	HEXA	AR
Miscellaneous	Sandhoff disease	HEXB	AR
Miscellaneous	LDLR-related familial hypercholesterolemia	LDLR	AR
Miscellaneous	Prader-Willi syndrome	15q11-13 region	NA

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