Sunshine Genetics Tier 2 Gene – Condition List

Sunshine Genetics
FSU Florida Institute For Pediatric Rare Diseases

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	Fibrochondrogenesis 2	COL11A2	AD
Hearing Loss	COL11A2-related otospondylomegaepiphyseal dysplasia or deafness	COL11A2	AR
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	TWNK-related disorders	TWNK	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	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
Epilepsy	Developmental and epileptic encephalopathy 54	HNRNPU	AD
Epilepsy	KCNA2-related neurological disorders	KCNA2	AD

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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
Epilepsy	PNKP-related disorders	PNKP	AR
Epilepsy	PPT1-related neuronal ceroid lipofuscinosis	PPT1	AR

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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
Epilepsy	Developmental and epileptic encephalopathy 117	SNAP25	AD
Epilepsy	Generalized epilepsy with febrile seizures plus, type 9	STX1B	AD

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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
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	Angelman syndrome	UBE3A	AD
Miscellaneous	Prader-Willi syndrome	15q11-13 region	NA