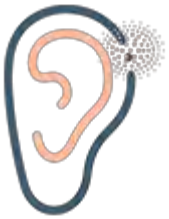
Syndromic and Nonsyndromic Deafness

Precision Panel



Overview

Hearing loss can be defined as conductive or sensorineural. Conductive hearing loss occurs due to dysfunction of the outer or middle ear, which prevents transmission of sound waves from reaching the inner ear. Sensorineural hearing loss, on the other hand, is the result of inner ear or auditory nerve dysfunction preventing neuronal transmission to the brain. In developed countries, approximately 1/1,000 children have severe or profound hearing loss at birth or during childhood. In most cases, hearing loss is a multifactorial disorder caused by genetic and environmental factors. Clinically, it has many different presentations, from mild to profound, including low and high-pitch patterns. Non-syndromic forms are responsible for about 70% of the cases of hereditary etiology and syndromic cases represent 30% of them. Among the patterns of inheritance, autosomal recessive remains the most common form of inheritance, although it can be autosomal dominant, X-linked or mitochondrial.

The Igenomix Syndromic and Nonsyndromic Deafness Precision Panel can be used to make an accurate and directed diagnosis as well as a differential diagnosis of hearing loss ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Syndromic and Nonsyndromic Deafness Precision Panel is indicated for those patients with a clinical suspicion or diagnosis with or without the following manifestations:

- Muffling of speech and other sounds
- Difficulty understanding words
- Trouble hearing consonants
- Needing to turn up the volume of the television or radio
- Associated syndromic features at birth: cardiac findings, renal findings, neurologic abnormalities, skeletal examination findings, craniofacial abnormalities etc
- Withdrawal from conversations
- Avoidance from social settings

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular confirmation for an accurate clinical diagnosis of a symptomatic patient.
- Early initiation of treatment with a multidisciplinary team in the form surgical care of external and middle ear deformities, cochlear implantation and medical care for treatment of middle ear disease, amplification and assistive listening devices.
- Early implementation of speech and language therapy.
- Risk assessment and genetic counselling of asymptomatic family members due to the autosomal dominant mode of inheritance.
- Improvement of delineation of genotype-phenotype correlation.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABCC1	Autosomal Dominant Deafness	AD	98.02	5 of 5
ABHD12	Polyneuropathy, Hearing Loss, Ataxia, Retinitis Pigmentosa, And Cataract	AR	95.77	21 of 21
ACTB	Baraitser-Winter Syndrome, Becker Nevus Syndrome, Developmental Malformations-Deafness-Dystonia Syndrome	AD	100	40 of 40
ACTG1	Autosomal Dominant Deafness, Baraitser-Winter Cerebrofrontofacial Syndrome	AD	98.59	55 of 55
ADCY1	Autosomal Recessive Deafness	AR	96.91	1 of 1
ADGRV1	Familial Febrile Convulsions, Usher Syndrome	AD,AR	97.53	-
AIFM1	Combined Oxidative Phosphorylation Deficiency, Cowchock Syndrome, X-linked Deafness, Spondyloepimetaphyseal Dysplasia, Severe X-linked Mitochondrial Encephalomyopathy, X-linked Charcot-Marie-Tooth Disease	X,XR,G	100	-
ANKH	Craniometaphyseal Dysplasia, Familial Calcium Pyrophosphate Deposition	AD	100	19 of 19
AP1B1	Ichthyosiform Erythroderma, Corneal Involvement, And Deafness, Mednik Syndrome	AR	100	5 of 5
AP1S1	Mental Retardation, Enteropathy, Deafness, Peripheral Neuropathy, Ichthyosis, And Keratoderma, Mednik Syndrome	AR	99.98	2 of 2
ATP1A3	Cerebellar Ataxia, Areflexia, Pes Cavus, Optic Atrophy, And Sensorineural Hearing Loss	AD	99.94	138 of 138
ATP2B2	Autosomal Recessive Deafness	AR	100	12 of 12
ATP6V0A4	Distal Renal Tubular Acidosis, With Or Without Sensorineural Hearing Loss	AR	100	85 of 85
ATP6V1B1	Renal Tubular Acidosis, Distal, With Progressive Nerve Deafness	AR	100	62 of 62
ATP6V1B2	Autosomal Dominant Deafness-Onychodystrophy Syndrome, Zimmermann-Laband Syndrome	AD	100	5 of 5
BCAP31	Deafness, Dystonia, And Cerebral Hypomyelination, Severe Motor And Intellectual Disabilities-Sensorineural Deafness-Dystonia Syndrome	X,XR,G	100	-
BCS1L	Bjornstad Syndrome, Gracile Syndrome, Leigh Syndrome, Mitochondrial Complex III Deficiency	AR,MI	99.96	40 of 42
BDP1	Autosomal Recessive Deafness	AR	99.3	1 of 1
BSND	Bartter Syndrome, Infantile, With Sensorineural Deafness	AR	99.95	21 of 21
BTD	Biotinidase Deficiency Multiple Carboxylase Deficiency, Late-Onset, Biotinidase Deficiency	AR	100	261 of 262
CABP2	Autosomal Recessive Deafness	AR	99.95	7 of 7
CACNA1D	Primary Aldosteronism, Seizures, And Neurologic Abnormalities, Sinoatrial Node Dysfunction And Deafness	AD,AR	100	18 of 18
CATSPER2	Sensorineural Deafness And Male Infertility	AR	99.87	1 of 1
CCDC50	Autosomal Dominant Deafness	AD	99.98	5 of 5

CD151	Nephropathy With Pretibial Epidermolysis Bullosa And Deafness	AR	100	3 of 3
CD164	Autosomal Dominant Deafness	AD	100	1 of 1
CDC14A	Autosomal Recessive Deafness	AR	99.76	11 of 11
CDH23	Autosomal Recessive Deafness, Usher Syndrome	AD,AR	98	400 of 403
CDKN1C	Beckwith-Wiedemann Syndrome, Image Syndrome	AD	73.58	55 of 76
CEACAM16	Autosomal Dominant Deafness, Autosomal Recessive Deafness	AD,AR	99.81	7 of 7
CEP250	Cone-Rod Dystrophy And Hearing Loss	AR	99.98	7 of 7
CEP78	Cone-Rod Dystrophy And Hearing Loss, Usher Syndrome	AR	99.44	9 of 10
CHD7	Charge Syndrome, Charge Syndrome, Omenn Syndrome	AD	96.25	823 of 896
CHSY1	Temtamy Preaxial Brachydactyly Syndrome	AR	96.64	13 of 16
CIB2	Autosomal Recessive Deafness, Usher Syndrome	AR	99.95	16 of 17
CISD2	Wolfram Syndrome	AR	92.92	5 of 5
CLDN14	Autosomal Recessive Deafness	AR	100	17 of 17
CLIC5	Autosomal Recessive Deafness	AR	99.91	2 of 2
CLPP	Perrault Syndrome	AR	99.91	11 of 11
CLRN1	Retinitis Pigmentosa, Usher Syndrome	AD,AR,X,XR,G	99.99	40 of 41
COCH	Autosomal Dominant Deafness, Autosomal Recessive Deafness	AD,AR	99.92	31 of 31
COL11A1	Autosomal Dominant Deafness, Marshall Syndrome, Stickler Syndrome	AD,AR	100	104 of 106
COL11A2	Autosomal Dominant Deafness, Nonsyndromic Sensorineural Deafness, Stickler Syndrome,	AD,AR	99.98	58 of 58
COL2A1	Epiphyseal Dysplasia, Multiple, With Myopia And Conductive Deafness, Legg-Calve-Perthes Disease Stickler Syndrome	AD,MU	100	583 of 583
COL4A3	Alport Syndrome	AD,AR	100	277 of 280
COL4A4	Alport Syndrome	AD,AR	99.95	247 of 251
COL4A5	Alport Syndrome	X,XD,G	99.88	-
COL4A6	X-linked Deafness	X,XR,G	98.72	-
COL9A1	Stickler Syndrome, Multiple Epiphyseal Dysplasia Due To Collagen Anomaly	AD,AR	99.98	8 of 8
COL9A2	Stickler Syndrome, Multiple Epiphyseal Dysplasia Due To Collagen Anomaly	AD,AR	100	16 of 16
COL9A3	Stickler Syndrome, Multiple Epiphyseal Dysplasia Due To Collagen Anomaly	AD	99.98	20 of 20
CRYM	Autosomal Dominant Deafness	AD	96	4 of 4
DACT1	Townes-Brocks Syndrome, Craniorachischisis	AD	98.12	8 of 9
DCAF17	Woodhouse-Sakati Syndrome	AR	98.77	21 of 21
DCDC2	Autosomal Recessive Deafness	AR	99.83	8 of 8
DIABLO	Autosomal Dominant Deafness	AD	100	2 of 2
DIAPH1	Autosomal Dominant Nonsyndromic Sensorineural Deafness	AD,AR	99.94	15 of 15
DIAPH3	Auditory Neuropathy	AD	99.96	7 of 9
DLX5	Split-Hand/Foot Malformation With Sensorineural Hearing Loss	AD,AR	99.98	8 of 8
DMXL2	Autosomal Dominant Deafness	AD,AR	99.83	19 of 23
DNAJC3	Ataxia, Combined Cerebellar And Peripheral, With Hearing Loss And Diabetes Mellitus	AR	99.75	1 of 4
DNMT1	Cerebellar Ataxia, Deafness, And Narcolepsy, Hereditary Sensory Neuropathy	AD	97.87	30 of 30
DSPP	Autosomal Dominant Deafness	AD	99.4	56 of 56
EDN3	Waardenburg Syndrome, Ondine Syndrome	AD,AR	100	20 of 22
EDNRB	Abcd Syndrome, Waardenburg-Shah Syndrome, Waardenburg Syndrome	AD,AR	99.55	70 of 72
EEF1AKNMT	Autosomal Recessive Deafness	AD	99.48	-
ELMOD3	Autosomal Recessive Deafness	AR	99.97	2 of 2
EPS8	Autosomal Recessive Deafnes	AR	99.97	3 of 3
EPS8L2	Autosomal Recessive Deafness	AR	99.27	3 of 3
ESPN	Autosomal Recessive Deafness With Or Without Vestibular Involvement, Usher Syndrome	AR	98.22	22 of 22
ESRP1	Autosomal Recessive Deafness	AR	99.95	2 of 2
ESRRB	Autosomal Recessive Deafness	AR	100	22 of 22
EXOSC2	Short Stature, Hearing Loss, Retinitis Pigmentosa, And Distinctive Facies;	AR	100	3 of 3
EYA1	Branchiootic Syndrome, Branchiootorenal Syndrome, Otofaciocervical Syndrome	AD	100	197 of 199
EYA4	Autosomal Dominant Deafness Nonsyndromic, Sensorineural Deafness With Dilated Cardiomyopathy	AD	100	32 of 32

FDXR	Auditory Neuropathy And Optic Atrophy, Optic Atrophy-Ataxia-Peripheral Neuropathy-Global Developmental Delay Syndrome	AR	99.93	23 of 23
FGF3	Congenital Deafness, With Inner Ear Agenesis, Microtia, And Microdontia, Deafness With Labyrinthine Aplasia, Microtia, And Microdontia, Otodental Syndrome	AR	99.81	18 of 18
FGF9	Multiple Synostoses Syndrome	AD	100	2 of 2
FGFR1	Hartsfield Syndrome, Jackson-Weiss Syndrome, Pfeiffer Syndrome, Septo-Optic Dysplasia Spectrum	AD	100	279 of 280
FGFR2	Antley-Bixler Syndrome Without Genital Anomalies Or Disordered Steroidogenesis, Apert Syndrome, Crouzon Syndrome, Jackson-Weiss Syndrome, Lacrimoauriculodentodigital Syndrome, Saethre-Chotzen Syndrome	AD	98	140 of 143
FGFR3	Camptodactyly, Tall Stature, And Hearing Loss Syndrome, Muenke Syndrome	AD,AR	99.89	77 of 78
FKBP14	Ehlers-Danlos Syndrome With Progressive Kyphoscoliosis, Myopathy,and Hearing Loss	AR	99.98	7 of 8
FOXI1	Enlarged Vestibular Aqueduct, Pendred Syndrome	AR	100	11 of 11
GAB1	Autosomal Recessive Deafness	AR	99.84	4 of 5
GATA2	Deafness-Lymphedema-Leukemia Syndrome	AD	100	137 of 142
GATA3	Hypoparathyroidism, Sensorineural Deafness, And Renal Disease	AD	100	81 of 81
GDF5	Multiple Synostoses Syndrome	AD,AR	99.48	48 of 51
GFER	Mitochondrial Progressive Myopathy With Congenital Cataract, Hearing Loss, And Developmental Delay	AR	99.89	6 of 6
GIPC3	Autosomal Recessive Deafness	AR	92.04	21 of 22
GJA1	Oculodentodigital Dysplasia, Craniometaphyseal Dysplasia	AD,AR,MU,O	100	119 of 119
GJB2	Autosomal Dominant Nonsyndromic Sensorineural Deafness, Autosomal Recessive Deafness, Congenital Deafness With Keratopachydermia And Constrictions Offingers And Toes, Palmoplantar Keratoderma-Deafness Syndrome	AD,AR,X,XR,MU,D,G	99.89	413 of 419
GJB3	Autosomal Dominant Deafness, Autosomal Recessive Deafness	AD,AR,MU,D	100	39 of 39
GJB6	Autosomal Dominant Deafness, Autosomal Recessive Deafness, Kid Syndrome	AD,AR,X,XR,MU,D,G	99.89	28 of 28
GPRASP2	X-linked Deafness	X,XR,G	92.99	-
GPSM2	Chudley-Mccullough Syndrome	AR	100	13 of 13
GRAP	Autosomal Recessive Deafness	AR	65.67	1 of 1
GRHL2	Corneal Dystrophy, Posterior Polymorphous, Autosomal Dominant Nonsyndromic Sensorineural Deafnes	AD,AR	100	8 of 11
GRXCR1	Autosomal Recessive Deafness	AR	100	10 of 10
GRXCR2	Autosomal Recessive Deafness,	AR	99.94	2 of 2
GSDME	Autosomal Dominant Nonsyndromic Sensorineural	AD	100	-
HARS1	Charcot-Marie-Tooth Disease, Usher Syndrome	AD,AR	100	-
HARS2	Perrault Syndrome	AR	100	3 of 3
HGF	Congenital Neurosensory Autosomal Recessive Deafness	AR	100	18 of 20
HOMER2	Autosomal Dominant Deafness	AD	99.98	2 of 2
HOXB1	Facial Paresis, Hereditary Congenital	AR	98.81	6 of 6
HSD17B4	Perrault Syndrome	AR	99.52	85 of 85
IARS2	Cataracts, Growth Hormone Deficiency, Sensory Neuropathy, Sensorineural Hearing Loss, And Skeletal Dysplasia	AR	99.95	11 of 11
IGF1	Growth Delay Due To Insulin-like Growth Factor Type 1 Deficiency	AR	100	7 of 8
ILDR1	Autosomal Recessive Neurosensory Deafness	AR	100	31 of 31
KARS1	Charcot-Marie-Tooth Disease, Autosomal Recessive Deafness	AR	100	34 of 34
KCNE1	Jervell And Lange-Nielsen Syndrome, Jervell And Lange-Nielsen Syndrome, Romano-Ward Syndrome	AD,AR	100	53 of 53
KCNJ10	Enlarged Vestibular Aqueduct, Pendred Syndrome, Seizures, Sensorineural Deafness, Ataxia, Mental Retardation, East Syndrome	AR	93.53	27 of 32
KCNQ1	Beckwith-Wiedemann Syndrome, Jervell And Lange-Nielsen Syndrome, Romano-Ward Syndrome	AD,AR	93.23	600 of 624
KCNQ4	Autosomal Dominant Deafness	AD	93.09	45 of 46
KIT	Mast Cell Disease, Systemic Mastocytosis With Associated Hematologic Neoplasm	AD	100	112 of 112
KITLG	Autosomal Dominant Deafness , Waardenburg Syndrome	AD	99.93	10 of 10
LARS2	Hydrops, Lactic Acidosis, And Sideroblastic Anemia, Perrault Syndrome	AR	99.99	20 of 20
LHFPL5	Autosomal Recessive Deafness	AR	100	17 of 17

LHX3	Sensorineural Deafness, With Pituitary Dwarfism, Hypothyroidism	AR	99.97	18 of 19
LMX1A	Autosomal Dominant Nonsyndromic Sensorineural Deafness	AD	100	4 of 4
LOXHD1	Autosomal Recessive Deafness	AR	99.98	97 of 97
LRP2	Donnai-Barrow Syndrome	AR	99.99	58 of 58
LRTOMT	Autosomal Recessive Deafness	AR	94.7	20 of 21
MAF	Ayme-Gripp Syndrome, Cataract-Microcornea Syndrome	AD	75.14	23 of 23
MAFB	Duane Retraction Syndrome With Or Without Deafness	AD	98.63	24 of 24
MAN2B1	Alpha-Mannosidosis, Infantile Form	AR	100	149 of 149
MANBA	Beta-Mannosidosis	AR	99.98	20 of 20
MAP3K7	Cardiospondylocarpofacial Syndrome, Frontometaphyseal Dysplasia	AD	99.96	13 of 13
MARVELD2	Autosomal Recessive Deafness	AR	100	18 of 19
MCM2	Autosomal Dominant Deafness	AD	100	1 of 1
MET	Autosomal Recessive Deafness	AD,AR	99.8	41 of 41
MGP	Keutel Syndrome	AR	99.93	7 of 7
MIR96	Autosomal Dominant Deafness	AD	-	-
MITF	Coloboma, Osteopetrosis, Microphthalmia, Macrocephaly, Albinism, And Deafness, Tietz Syndrome, Waardenburg Syndrome	AD,AR	100	72 of 72
MPZL2	Autosomal Recessive Deafness	AR	100	4 of 4
MSRB3	Autosomal Recessive Deafness	AR	100	4 of 4
MYH14	Autosomal Dominant Nonsyndromic Sensorineural Deafness, Peripheral Neuropathy, Myopathy, Hoarseness, And Hearing Loss	AD	99.97	52 of 52
MYH9	Autosomal Dominant Nonsyndromic Sensorineural Deafness	AD	100	144 of 145
MYO15A	Deafness, Neurosensory, Autosomal Recessive Neurosensory Deafness	AR	99.12	306 of 307
MYO3A	Autosomal Recessive Deafness	AR	99.67	21 of 21
MYO6	Autosomal Dominant Deafness, Autosomal Recessive Deafness	AD,AR	100	74 of 75
MYO7A	Autosomal Dominant Nonsyndromic Sensorineural Deafness, Autosomal Recessive Neurosensory Deafness, Usher Syndrome	AD,AR	100	579 of 580
NARS2	Combined Oxidative Phosphorylation Deficiency, Autosomal Recessive Deafness	AR	99.63	13 of 13
NDP	Norrie Disease, Coats Disease	X,XR,G	100	NA of NA
NLRP3	Cinca Syndrome, Autosomal Dominant Deafness, Muckle-Wells Syndrome, Cinca Syndrome	AD	100	152 of 152
NOG	Multiple Synostoses Syndrome, Proximal Symphalangism, Tarsal-Carpal Coalition Syndrome	AD	99.89	61 of 62
OPA1	Behr Syndrome, Mitochondrial DNA Depletion Syndrome, Optic Atrophy With Or Without Deafness	AD,AR	99.98	397 of 402
OSBPL2	Autosomal Dominant Deafness	AD	99.98	4 of 4
OTOA	Autosomal Recessive Deafness	AR	79.48	34 of 36
OTOF	Autosomal Recessive Deafness	AR	100	200 of 200
OTOG	Autosomal Recessive Deafness	AR	99.95	11 of 11
OTOGL	Autosomal Recessive Deafness	AR	98.52	20 of 24
P2RX2	Autosomal Dominant Deafness	AD	99.14	4 of 4
PAX3	Craniofacial-Deafness-Hand Syndrome, Waardenburg Syndrome	AD,AR	99.98	157 of 157
PBX1	Congenital Anomalies Of Kidney And Urinary Tract Syndrome With Or Without Hearing Loss	AD	98	18 of 18
PCDH15	Autosomal Recessive Deafness, Usher Syndrome	AR	99.36	152 of 158
PDE1C	Autosomal Dominant Deafness	AD	99.98	1 of 1
PDZD7	Autosomal Recessive Deafness, Usher Syndrome	AR	100	28 of 28
PEX1	Sensorineural Hearing Loss, With Enamel Hypoplasia And Nail Defects, Zellweger Syndrome, Infantile Refsum Disease	AR	97.02	126 of 134
PEX26	Infantile Refsum Disease , Neonatal Adrenoleukodystrophy, Zellweger Syndrome	AR	100	29 of 29
PEX6	Heimler Syndrome, Autosomal Recessive Spinocerebellar Ataxia-Blindness-Deafness Syndrome, Zellweger Syndrome	AD,AR	99.94	105 of 108
PJVK	Autosomal Recessive Deafnes	AR	100	-
PLOD3	Bone Fragility With Contractures, Arterial Rupture, And Deafness	AR	100	6 of 6
PLS1	Autosomal Dominant Deafness	AD	99.92	5 of 5
PMP22	Charcot-Marie-Tooth Disease And Deafness	AD,AR	97.82	110 of 110

PNPT1	Combined Oxidative Phosphorylation Deficiency, Autosomal Recessive Deafness	AR	99.93	26 of 26
POLD1	Mandibular Hypoplasia, Deafness, Progeroid Features, And Lipodystrophy Syndrome	AD	100	40 of 41
POLR1A	Choanal Atresia-Hearing Loss-Cardiac Defects-Craniofacial Dysmorphism Syndrome	AD	99.8	6 of 6
POLR1C	Mandibulofacial Dysostosis, Treacher-Collins Syndrome	AR	99.99	35 of 35
POLR1D	Treacher Collins Syndrome	AD,AR	100	23 of 23
POU3F4	X-linked Deafness, Xq21 Microdeletion Syndrome	X,XR,G	99.98	-
POU4F3	Autosomal Dominant Deafness	AD	100	36 of 36
PPIP5K2	Autosomal Recessive Deafness	AR	97.86	4 of 4
PRPS1	Arts Syndrome, Charcot-Marie-Tooth Disease, X-linked Deafness, Lethal Ataxia With Deafness And Optic Atrophy	X,XR,G	100	-
PTPRQ	Autosomal Dominant Deafness, Autosomal Recessive Deafness	AD,AR	94.47	33 of 34
RDX	Autosomal Recessive Deafness	AR	99.99	14 of 14
REST	Autosomal Dominant Deafness	AD	99.83	15 of 16
RIPOR2	Autosomal Recessive Deafness	AR	96.14	-
RMND1	Combined Oxidative Phosphorylation Deficiency	AR	99.67	15 of 16
ROR1	Autosomal Recessive Deafness	AR	97.2	2 of 2
RPGR	Retinitis Pigmentosa And Sinorespiratory Infections Withor Without Deafness	X,XR,G	94	-
RPS6KA3	Coffin-Lowry Syndrome	X,XD,G	99.95	-
S1PR2	Autosomal Recessive Deafness	AR	100	3 of 3
SALL1	Townes-Brocks Syndrome	AD	100	85 of 86
SALL4	Duane-Radial Ray Syndrome, Ivic Syndrome, Acro-Renal-Ocular Syndrome	AD	100	54 of 54
SEMA3E	Charge Syndrome	AD,AR	99.81	6 of 7
SERAC1	3-a Methylglutaconic Aciduria With Deafness	AR	99.93	53 of 53
SERPINB6	Autosomal Recessive Deafness	AR	100	3 of 3
SIX1	Branchiootic Syndrome, Branchiootorenal Syndrome, Autosomal Dominant Deafness	AD	73	20 of 20
SIX5	Branchiootorenal Syndrome	AD	93.16	11 of 11
SLC12A1	Bartter Syndrome	AR	99	90 of 95
SLC17A8	Deafness, Autosomal Dominant Deafness	AD	100	8 of 8
SLC19A2	Thiamine-Responsive Megaloblastic Anemia Syndrome	AR	99.99	67 of 68
SLC26A4	Enlarged Vestibular Aqueduct, Pendred Syndrome	AR	100	577 of 581
SLC26A5	Autosomal Recessive Deafness	AR	100	9 of 9
SLC29A3	Histiocytosis-Lymphadenopathy Plus Syndrome, Dysosteosclerosis	AR	100	32 of 32
SLC33A1	Congenital Cataracts, Hearing Loss, And Neurodegeneration	AD,AR	99.44	9 of 9
SLC44A4	Autosomal Dominant Deafness	AD	99.69	-
SLC4A11	Corneal Endothelial Dystrophy And Perceptive Deafness Syndrome	AD,AR	99.98	108 of 109
SLC52A2	Brown-Vialetto-Van Laere Syndrome, Autosomal Recessive Spinocerebellar Ataxia-Blindness-Deafness Syndrome	AR	100	31 of 32
SLC52A3	Progressive Bulbar Palsy With Sensorineural Deafness	AR	100	43 of 43
SLITRK6	Cochlear Deafness With Myopia And Intellectual Impairment	AR	99.52	9 of 9
SMAD4	Myhre Syndrome, Generalized Juvenile Polyposis/Juvenile Polyposis Coli	AD	99.56	136 of 136
SMPX	X-linked Deafness	X,XD,G	100	-
SNAI2	Waardenburg Syndrome	AD,AR	99.79	1 of 2
SOX10	Peripheral Demyelinating Neuropathy, Waardenburg Syndrome	AD	99.74	139 of 147
SOX2	Anophthalmia/Microphthalmia-Esophageal Atresia Syndrome, Septo-optic Dysplasia Spectrum	AD	99.91	78 of 78
SPATA5	Microcephaly-Intellectual Disability-Sensorineural Hearing Loss-Epilepsy-Abnormal Muscle Tone Syndrome	AR	99.83	30 of 30
SPNS2	Autosomal Recessive Deafness	AR	93.06	2 of 2
SPTBN4	Congenital Myopathy With Neuropathy And Deafness	AR	99.26	10 of 10
STRC	Autosomal Recessive Sensorineural Deafness And Male Infertility	AR	47.3	35 of 62
SUCLA2	Mitochondrial DNA Depletion Syndrome	AR	100	27 of 27
SUCLG1	Mitochondrial DNA Depletion Syndrome	AR	100	34 of 34
SYNE4	Autosomal Recessive Deafness	AR	100	2 of 2
TBC1D24	Autosomal Dominant Deafness, Autosomal Recessive Deafness, Doors Syndrome	AD,AR	100	80 of 80
TBL1Y	Y-linked Deafness	Y,G	44.6	-
TBX1	Digeorge Syndrome, Velocardiofacial Syndrome	AD,AR	88.7	35 of 42

TBX22	Charge-like Syndrome, Cleft Palate, Abruzzo-Erickson Syndrome	X,G	99.94	-
TCOF1	Treacher Collins-Franceschetti Syndrome	AD	100	326 of 327
TECTA	Autosomal Dominant Nonsyndromic Sensorineural Deafness	AD,AR	99.96	149 of 149
TFAP2A	Branchiooculofacial Syndrome	AD	98.61	37 of 37
TIMM8A	Mohr-Tranebjaerg Syndrome	X,XR,G	100	-
TJP2	Progressive Familial Intrahepatic Cholestasis	AR	99.85	43 of 43
TMC1	Autosomal Dominant Deafness, Autosomal Recessive Neurosensory Deafness	AD,AR	100	106 of 107
TMEM132E	Autosomal Recessive Deafness	AR	99.8	1 of 1
TMIE	Autosomal Recessive Deafness	AR	96.56	9 of 10
TMPRSS3	Childhood-Onset Neurosensory Deafness	AR	100	85 of 85
TNC	Autosomal Dominant Deafness	AD	99.98	7 of 7
TPRN	Autosomal Recessive Deafness	AR	75.75	7 of 12
TRAPPC12	Early-Onset Progressive Encephalopathy-Hearing Loss-Pons Hypoplasia-Brain Atrophy Syndrome	AR	99.98	3 of 3
TRIOBP	Autosomal Recessive Deafness	AR	98.48	42 of 42
TRMU	Aminoglycoside-Induced Deafness, Liver Failure, Acute Infantile, Mitochondrial Myopathy With Reversible Cytochrome C Oxidase Deficiency	AR,MI	100	25 of 25
TRNE	Maternally-Inherited Diabetes And Deafness	-	-	-
TRNK	Maternally-Inherited Diabetes And Deafness, Mitochondrial DNA-Related Cardiomyopathy And Hearing Loss	MI	-	-
TRNL1	Kearns-Sayre Syndrome, Maternally-Inherited Diabetes And Deafness, Mitochondrial DNA-Associated Leigh Syndrome	MI	-	-
TRNS1	Deafness, Aminoglycoside-Induced, Mitochondrial Complex Iv Deficiency, Palmoplantar Keratoderma-Deafness Syndrome	AR,MI	-	-
TRRAP	Autosomal Dominant Deafness	AD	99.98	46 of 46
TSPEAR	Autosomal Recessive Deafness	AR	100	9 of 9
TUBB4B	Leber Congenital Amaurosis With Early-Onset Deafness	AD	100	3 of 3
TWNK	Infantile-Onset Spinocerebellar Ataxia, Perrault Syndrome, Progressive External Ophthalmoplegia With Mitochondrial DNA Deletions	AD,AR	-	-
TXNL4A	Burn-Mckeown Syndrome, Choanal Atresia-Hearing Loss-Cardiac Defects-Craniofacial Dysmorphism Syndrome	AR	80.96	4 of 4
TYR	Oculocutaneous Albinism	AR	99.77	437 of 455
USH1C	Autosomal Recessive Neurosensory Deafness, Usher Syndrome	AR	99.97	79 of 79
USH1G	Usher Syndrome	AR	100	35 of 35
USH2A	Retinitis Pigmentosa, Usher Syndrome	AR	100	1286 of 1314
VCAN	Wagner Syndrome	AD	99.91	11 of 21
WBP2	Autosomal Recessive Deafness	AR	80.97	3 of 3
WFS1	Autosomal Dominant Deafness, Wolfram Syndrome	AD,AR	99.97	390 of 395
WHRN	Autosomal Recessive Deafness, Usher Syndrome	AR	99.94	-

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology

Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

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