Panel de ORL

La pérdida de audición es una condición común en los niños, que afecta a 1 de cada 100 nacidos vivos. En más del 50,0 % de los casos existe una causa genética para este trastorno, de los cuales el 70,0 % presenta hipoacusia no sindrómica. CentoHear incluye genes asociados con la pérdida auditiva sindrómica y no sindrómica. Tanto los genes autosómicos recesivos como los dominantes están incluidos en el panel. Además incluye varios otros síndromes, como Alport, Pendred, Waardenburg, Usher y branchio-oto-renal, entre otros.

SÍNDROMES Y TRASTORNOS COMUNES CUBIERTOS

- Síndrome de Alport
- Síndrome de Coffin-Lowry
- Sordera autosómica recesiva y dominante
- Hipoacusia no sindrómica
- Síndrome de Pendred
- Síndrome de Perrault
- Síndrome de Pfeiffer
- Pérdida auditiva neurosensorial
- Síndrome de Stickler
- Hipoacusia sindrómica
- Síndrome de Usher
- Síndrome de Waardenburg
- Síndrome de wólfram

Nº de genes:	233
Entrega:	25 días
Cobertura:	≥99.5% ≥20x Cobertura media con profundidad ≥150x
Detalles:	Análisis CNV incluido

Resumen de genes y enfermedades asociadas (OMIM) incluidos en este panel:

Genes	OMIM (Genes)	Associated diseases (OMIM)	Inheritance
ABHD12	613599	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract	AR
ACTB	102630	Baraitser-Winter syndrome 1	AD
		Dystonia, juvenile-onset	AD
ACTG1	102560	Baraitser-Winter syndrome 2	AD
		Deafness, autosomal dominant 20/26	AD
ADCY1	103072	Deafness, autosomal recessive 44	AR
ADGRV1	602851	Usher syndrome, type 2C	AR, DD
		Febrile seizures, familial, 4	AD
		Usher syndrome, type 2C, GPR98/PDZD7 digenic	AR, DD
AIFM1	300169	Cowchock syndrome	XLR
		Combined oxidative phosphorylation deficiency 6	XLR
		Deafness, X-linked 5	XLR
		Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy	XLR
ANKH	605145	Chondrocalcinosis 2	AD
		Craniometaphyseal dysplasia	AD
ATP2B2	108733	Deafness, autosomal dominant 82	AD
		Deafness, autosomal recessive 12, modifier of	AR
ATP6V1B1	192132	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss	AR

ATP6V1B2	606939	Zimmermann-Laband syndrome 2	AD
		Deafness, congenital, with onychodystrophy, autosomal dominant	AD
BCS1L	603647	GRACILE syndrome	AR
		Bjornstad syndrome	AR
		Mitochondrial complex III deficiency, nuclear type 1	AR
BDP1	607012	Deafness, autosomal recessive 112	AR
BSND	606412	Bartter syndrome, type 4a	AR
		Sensorineural deafness with mild renal dysfunction	AR
BTD	609019	Biotinidase deficiency	AR
CABP2	607314	Deafness, autosomal recessive 93	AR
CACNA1D	114206	Primary aldosteronism, seizures, and neurologic abnormalities	AD
		Sinoatrial node dysfunction and deafness	AR
CCDC50	611051	?Deafness, autosomal dominant 44	AD
CD151	602243	[Blood group, Raph]	-
		Nephropathy with pretibial epidermolysis bullosa and deafness	-
CD164	603356	?Deafness, autosomal dominant 66	AD
CDC14A	603504	Deafness, autosomal recessive 32, with or without immotile sperm	AR
CDH23	605516	Deafness, autosomal recessive 12	AR
		Usher syndrome, type 1D	AR, DR

		Usher syndrome, type 1D/F digenic	AR, DR
		Pituitary adenoma 5, multiple types	AD
CDKN1C	600856	IMAGE syndrome	AD
		Beckwith-Wiedemann syndrome	AD
CEACAM16	614591	Deafness, autosomal recessive 113	AR
		Deafness, autosomal dominant 4B	AD
CEP78	617110	Cone-rod dystrophy and hearing loss	AR
CHD7	608892	CHARGE syndrome	AD
		Hypogonadotropic hypogonadism 5 with or without anosmia	AD
CHSY1	608183	Temtamy preaxial brachydactyly syndrome	AR
CIB2	605564	Usher syndrome, type IJ	AR
		Deafness, autosomal recessive 48	AR
CISD2	611507	Wolfram syndrome 2	AR
CLDN14	605608	Deafness, autosomal recessive 29	AR
CLIC5	607293	?Deafness, autosomal recessive 103	AR
CLPP	601119	Perrault syndrome 3	AR
CLRN1	606397	Retinitis pigmentosa 61	-
		Usher syndrome, type 3A	AR

COCH	603196	Deafness, autosomal dominant 9	AD
		Deafness, autosomal recessive 110	AR
COL11A1	120280	Marshall syndrome	AD
		Fibrochondrogenesis 1	AR
		Lumbar disc herniation, susceptibility to	-
		Deafness, autosomal dominant 37	AD
		Stickler syndrome, type II	AD
COL11A2	120290	Deafness, autosomal recessive 53	AR
		Otospondylomegaepiphyseal dysplasia, autosomal recessive	AR
		Fibrochondrogenesis 2	AD, AR
		Otospondylomegaepiphyseal dysplasia, autosomal dominant	AD
		Deafness, autosomal dominant 13	AD
COL2A1	120140	Legg-Calve-Perthes disease	AD
		Stickler syndrome, type I	AD
		Osteoarthritis with mild chondrodysplasia	AD
		Platyspondylic skeletal dysplasia, Torrance type	AD
		Spondyloepiphyseal dysplasia, Stanescu type	AD
		Kniest dysplasia	AD
		Czech dysplasia	AD
		Stickler sydrome, type I, nonsyndromic ocular	AD

		?Vitreoretinopathy with phalangeal epiphyseal dysplasia	AD
		Epiphyseal dysplasia, multiple, with myopia and deafness	AD
		Avascular necrosis of the femoral head	AD
		Spondyloperipheral dysplasia	AD
		Achondrogenesis, type II or hypochondrogenesis	AD
		SMED Strudwick type	AD
		SED congenita	AD
COL4A3	120070	Alport syndrome 3, autosomal dominant	AD
		Hematuria, benign familial	AD
		Alport syndrome 2, autosomal recessive	AR
COL4A4	120131	Alport syndrome 2, autosomal recessive	AR
		Hematuria, familial benign	AD
COL4A5	303630	Alport syndrome 1, X-linked	XLD
COL4A6	303631	Deafness, X-linked 6	XLR
COL9A1	120210	Epiphyseal dysplasia, multiple, 6	AD
		Stickler syndrome, type IV	-
COL9A2	120260	?Stickler syndrome, type V	AR
		Epiphyseal dysplasia, multiple, 2	AD
COL9A3	120270	Epiphyseal dysplasia, multiple, 3, with or without myopathy	AD

		Intervertebral disc disease, susceptibility to	-
		Stickler syndrome, type VI	-
CRYM	123740	Deafness, autosomal dominant 40	AD
DCAF17	612515	Woodhouse-Sakati syndrome	AR
DCDC2	605755	Nephronophthisis 19	AR
		Deafness, autosomal recessive 66	AR
		Sclerosing cholangitis, neonatal	AR
DIABLO	605219	Deafness, autosomal dominant 64	AD
DIAPH1	602121	Seizures, cortical blindness, microcephaly syndrome	AR
		Deafness, autosomal dominant 1, with or without thrombocytopenia	AD
DIAPH3	614567	Auditory neuropathy, autosomal dominant, 1	AD
DLX5	600028	?Split-hand/foot malformation 1 with sensorineural hearing loss	AR
		Split-hand/foot malformation 1	AD
DMXL2	612186	Polyendocrine-polyneuropathy syndrome	AR
		Developmental and epileptic encephalopathy 81	AR
		Deafness, autosomal dominant 71	AD
DNMT1	126375	Neuropathy, hereditary sensory, type IE	AD
		Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant	AD
DSPP	125485	Dentinogenesis imperfecta, Shields type III	AD
		Dentin dysplasia, type II	AD

		Deafness, autosomal dominant 39, with dentinogenesis	AD
		Dentinogenesis imperfecta, Shields type II	AD
EDN3	131242	Waardenburg syndrome, type 4B	AD, AR
		Hirschsprung disease, susceptibility to, 4	AD
		Central hypoventilation syndrome, congenital	AD
EDNRB	131244	Waardenburg syndrome, type 4A	AD, AR
		Hirschsprung disease, susceptibility to, 2	AD
		ABCD syndrome	AR
ELMOD3	615427	Deafness, autosomal dominant 81	AD
		?Deafness, autosomal recessive 88	AR
EPS8	600206	Deafness, autosomal recessive 102	AR
EPS8L2	614988	Deafness autosomal recessive 106	AR
ESPN	606351	Usher syndrome, type 1M	AR
		Deafness, neurosensory, without vestibular involvement, autosomal dominant	AR
		Deafness, autosomal recessive 36	AR
ESRP1	612959	Deafness, autosomal recessive 109	AR
ESRRB	602167	Deafness, autosomal recessive 35	AR
EYA1	601653	Anterior segment anomalies with or without cataract	AD

		Branchiootorenal syndrome 1, with or without cataracts	AD
		?Otofaciocervical syndrome	AD
		Branchiootic syndrome 1	AD
EYA4	603550	Deafness, autosomal dominant 10	AD
		Cardiomyopathy, dilated, 1J	AD
FDXR	103270	Auditory neuropathy and optic atrophy	AR
FGF3	164950	Deafness, congenital with inner ear agenesis, microtia, and microdontia	AR
FGFR1	136350	Osteoglophonic dysplasia	AD
		Trigonocephaly 1	AD
		Pfeiffer syndrome	AD
		Encephalocraniocutaneous lipomatosis, somatic mosaic	-
		Hypogonadotropic hypogonadism 2 with or without anosmia	AD
		Jackson-Weiss syndrome	AD
		Hartsfield syndrome	AD
FGFR2	176943	Crouzon syndrome	AD
		Saethre-Chotzen syndrome	AD
		Craniofacial-skeletal-dermatologic dysplasia	AD
		Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis	AD
		Gastric cancer, somatic	-

		LADD syndrome	AD
		Beare-Stevenson cutis gyrata syndrome	AD
		Scaphocephaly, maxillary retrusion, and mental retardation	-
		Apert syndrome	AD
		Bent bone dysplasia syndrome	AD
		Pfeiffer syndrome	AD
		Jackson-Weiss syndrome	AD
FGFR3	134934	Achondroplasia	AD
		CATSHL syndrome	AD, AR
		Thanatophoric dysplasia, type I	AD
		Bladder cancer, somatic	-
		Hypochondroplasia	AD
		SADDAN	AD
		Colorectal cancer, somatic	-
		Crouzon syndrome with acanthosis nigricans	AD
		Cervical cancer, somatic	-
		Nevus, epidermal, somatic	-
		Thanatophoric dysplasia, type II	AD
		Spermatocytic seminoma, somatic	-
		Muenke syndrome	AD

		LADD syndrome	AD
FOXI1	601093	Enlarged vestibular aqueduct	AR
GAB1	604439	Deafness, autosomal recessive 26	AR
GATA3	131320	Hypoparathyroidism, sensorineural deafness, and renal dysplasia	AD
GIPC3	608792	Deafness, autosomal recessive 15	AR
GJA1	121014	Oculodentodigital dysplasia, autosomal recessive	AR
		Atrioventricular septal defect 3	AD
		Syndactyly, type III	AD
		Craniometaphyseal dysplasia, autosomal recessive	AR
		Palmoplantar keratoderma with congenital alopecia	AD
		Oculodentodigital dysplasia	AD
		Hypoplastic left heart syndrome 1	AR
		Erythrokeratodermia variabilis et progressiva 3	AD
GJB2	121011	Keratoderma, palmoplantar, with deafness	AD
		Keratitis-ichthyosis-deafness syndrome	AD
		Deafness, autosomal dominant 3A	AD
		Hystrix-like ichthyosis with deafness	AD
		Bart-Pumphrey syndrome	AD
		Vohwinkel syndrome	AD
		Deafness, autosomal recessive 1A	AR, DD

GJB3	603324	Deafness, autosomal dominant 2B	AD
		Erythrokeratodermia variabilis et progressiva 1	AD, AR
		Deafness, digenic, GJB2/GJB3	AR, DD
GJB6	604418	Deafness, autosomal recessive 1B	AR
		Deafness, digenic GJB2/GJB6	AR, DD
		Ectodermal dysplasia 2, Clouston type	AD
		Deafness, autosomal dominant 3B	AD
GPRASP2	300969	Deafness, X-linked 7	XLR
GPSM2	609245	Chudley-McCullough syndrome	AR
GRHL2	608576	Deafness, autosomal dominant 28	AD
		Ectodermal dysplasia/short stature syndrome	AR
		Corneal dystrophy, posterior polymorphous, 4	AD
GRXCR1	613283	Deafness, autosomal recessive 25	AR
GRXCR2	615762	Deafness, autosomal recessive 101	AR
GSDME	608798	Deafness, autosomal dominant 5	AD
HARS1	142810	Usher syndrome type 3B	AR
		Charcot-Marie-Tooth disease, axonal, type 2W	AD
HARS2	600783	Perrault syndrome 2	AR
HGF	142409	Deafness, autosomal recessive 39	AR
HOMER2	604799	Deafness, autosomal dominant 68	AD
HOXB1	142968	Facial paresis, hereditary congenital, 3	AR

HSD17B4	601860	Perrault syndrome 1	AR
		D-bifunctional protein deficiency	AR
ILDR1	609739	Deafness, autosomal recessive 42	AR
KARS1	601421	Charcot-Marie-Tooth disease, recessive intermediate, B	AR
		Deafness, congenital, and adult-onset progressive leukoencephalopathy	AR
		Leukoencephalopathy, progressive, infantile-onset, with or without deafness	AR
		Deafness, autosomal recessive 89	AR
KCNE1	176261	Jervell and Lange-Nielsen syndrome 2	AR
		Long QT syndrome 5	AD
KCNJ10	602208	Enlarged vestibular aqueduct, digenic	AR
		SESAME syndrome	AR
KCNQ1	607542	Long QT syndrome 1, acquired, susceptibility to	AD
		Jervell and Lange-Nielsen syndrome	AR
		Atrial fibrillation, familial, 3	AD
		Short QT syndrome 2	AD
		Long QT syndrome 1	AD
KCNQ4	603537	Deafness, autosomal dominant 2A	AD
KIT	164920	Gastrointestinal stromal tumor, familial	AD, IC
		Germ cell tumors, somatic	-
		Piebaldism	AD
		Leukemia, acute myeloid, somatic	-

		Mastocytosis, systemic, somatic	-
		Mastocytosis, cutaneous	AD
KITLG	184745	[Skin/hair/eye pigmentation 7, blond/brown hair]	-
		Deafness, autosomal dominant 69, unilateral or asymmetric	AD
		Hyperpigmentation with or without hypopigmentation	AD
		Waardenburg syndrome, type 2F	-
LARS2	604544	Hydrops, lactic acidosis, and sideroblastic anemia	AR
		Perrault syndrome 4	AR
LHFPL5	609427	Deafness, autosomal recessive 67	AR
LOXHD1	613072	Deafness, autosomal recessive 77	AR
LRP2	600073	Donnai-Barrow syndrome	AR
LRTOMT	612414	Deafness, autosomal recessive 63	AR
MAN2B1	609458	Mannosidosis, alpha-, types I and II	AR
MANBA	609489	Mannosidosis, beta	AR
MARVELD2	610572	Deafness, autosomal recessive 49	AR
MCM2	116945	Deafness, autosomal dominant 70	AD
MET	164860	Hepatocellular carcinoma, childhood type, somatic	-
		Deafness, autosomal recessive 97	AR
		Osteofibrous dysplasia, susceptibility to	AD
		Renal cell carcinoma, papillary, 1, familial and somatic	-
		Arthrogryposis, distal, type 11	-

MGP	154870	Keutel syndrome	AR
MITF	156845	Melanoma, cutaneous malignant, susceptibility to, 8	-
		Waardenburg syndrome, type 2A	AD
		Waardenburg syndrome/ocular albinism, digenic	-
		Tietz albinism-deafness syndrome	AD
		COMMAD syndrome	AR
MPZL2	604873	Deafness, autosomal recessive 111	AR
MSRB3	613719	Deafness, autosomal recessive 74	AR
MYH14	608568	Peripheral neuropathy, myopathy, hoarseness, and hearing loss	AD
		Deafness, autosomal dominant 4A	AD
MYH9	160775	Deafness, autosomal dominant 17	AD
		Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss	AD
MYO15A	602666	Deafness, autosomal recessive 3	AR
MYO3A	606808	Deafness, autosomal recessive 30	AR
MYO6	600970	Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy	AD
		Deafness, autosomal dominant 22	AD
		Deafness, autosomal recessive 37	AR
MYO7A	276903	Deafness, autosomal dominant 11	AD
		Usher syndrome, type 1B	AR
		Deafness, autosomal recessive 2	AR
NARS2	612803	Deafness, autosomal recessive 94	AR

		Combined oxidative phosphorylation deficiency 24	AR
NDP	300658	Norrie disease	XLR
		Exudative vitreoretinopathy 2, X-linked	XLD, XLR
NLRP3	606416	CINCA syndrome	AD
		Deafness, autosomal dominant 34, with or without inflammation	AD
		Keratoendothelitis fugax hereditaria	AD
		Familial cold inflammatory syndrome 1	AD
		Muckle-Wells syndrome	AD
OPA1	605290	Optic atrophy 1	AD
		Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type)	AR
		Glaucoma, normal tension, susceptibility to	-
		Optic atrophy plus syndrome	AD
		Behr syndrome	AR
OSBPL2	606731	Deafness, autosomal dominant 67	AD
OTOA	607038	Deafness, autosomal recessive 22	AR
OTOF	603681	Deafness, autosomal recessive 9	AR
		Auditory neuropathy, autosomal recessive, 1	AR
OTOG	604487	Deafness, autosomal recessive 18B	AR
OTOGL	614925	Deafness, autosomal recessive 84B	AR
P2RX2	600844	Deafness, autosomal dominant 41	AD
PAX3	606597	Waardenburg syndrome, type 3	AD, AR

		Rhabdomyosarcoma 2, alveolar	SM
		Craniofacial-deafness-hand syndrome	AD
		Waardenburg syndrome, type 1	AD
PCDH15	605514	Usher syndrome, type 1D/F digenic	AR, DR
		Deafness, autosomal recessive 23	AR
		Usher syndrome, type 1F	AR
PDZD7	612971	Retinal disease in Usher syndrome type IIA, modifier of	AR
		Usher syndrome, type IIC, GPR98/PDZD7 digenic	AR, DD
		Deafness, autosomal recessive 57	AR
PEX1	602136	Peroxisome biogenesis disorder 1A (Zellweger)	AR
		Peroxisome biogenesis disorder 1B (NALD/IRD)	AR
		Heimler syndrome 1	AR
PEX26	608666	Peroxisome biogenesis disorder 7A (Zellweger)	AR
		Peroxisome biogenesis disorder 7B	AR
PEX6	-	Peroxisome biogenesis disorder 4B	AD, AR
		Peroxisome biogenesis disorder 4A (Zellweger)	AR
		Heimler syndrome 2	AR
PJVK	610219	Deafness, autosomal recessive 59	AR
PMP22	601097	Neuropathy, recurrent, with pressure palsies	AD
		Dejerine-Sottas disease	AD, AR
		Roussy-Levy syndrome	AD

		?Neuropathy, inflammatory demyelinating	?AD
		Charcot-Marie-Tooth disease, type 1E	AD
		Charcot-Marie-Tooth disease, type 1A	AD
PNPT1	610316	Deafness, autosomal recessive 70	AR
		Combined oxidative phosphorylation deficiency 13	AR
		Spinocerebellar ataxia 25	-
POLR1C	610060	Leukodystrophy, hypomyelinating, 11	AR
		Treacher Collins syndrome 3	AR
POLR1D	613715	Treacher Collins syndrome 2	AD, AR
POU3F4	300039	Deafness, X-linked 2	XLR
POU4F3	602460	Deafness, autosomal dominant 15	AD
PRPS1	311850	Charcot-Marie-Tooth disease, X-linked recessive, 5	XLR
		Deafness, X-linked 1	XL
		Phosphoribosylpyrophosphate synthetase superactivity	XLR
		Arts syndrome	XLR
		Gout, PRPS-related	XLR
RDX	179410	Deafness, autosomal recessive 24	AR
RMND1	614917	Combined oxidative phosphorylation deficiency 11	AR
ROR1	602336	Deafness, autosomal recessive 108	AR
RPS6KA3	300075	Coffin-Lowry syndrome	XLD
		Mental retardation, X-linked 19	XLD

S1PR2	605111	Deafness, autosomal recessive 68	AR
SALL1	602218	Townes-Brocks branchiootorenal-like syndrome	AD
		Townes-Brocks syndrome 1	AD
SALL4	607343	IVIC syndrome	AD
		Duane-radial ray syndrome	AD
SEMA3E	608166	CHARGE syndrome	AD
SERPINB6	173321	Deafness, autosomal recessive 91	AR
SIX1	601205	Deafness, autosomal dominant 23	AD
		Branchiootic syndrome 3	AD
SIX5	600963	Branchiootorenal syndrome 2	-
SLC12A1	600839	Bartter syndrome, type 1	AR
SLC17A8	607557	Deafness, autosomal dominant 25	AD
SLC19A2	603941	Thiamine-responsive megaloblastic anemia syndrome	AR
SLC26A4	605646	Pendred syndrome	AR
		Deafness, autosomal recessive 4, with enlarged vestibular aqueduct	AR
SLC26A5	604943	Deafness, autosomal recessive 61	AR
SLC29A3	612373	Histiocytosis-lymphadenopathy plus syndrome	AR
SLC33A1	603690	Congenital cataracts, hearing loss, and neurodegeneration	AR
		Spastic paraplegia 42, autosomal dominant	AD
SLC44A4	606107	Deafness, autosomal dominant 72	AD
SLC52A2	607882	Brown-Vialetto-Van Laere syndrome 2	AR

SLC52A3	613350	Fazio-Londe disease	AR
		Brown-Vialetto-Van Laere syndrome 1	AR
SLITRK6	609681	Deafness and myopia	AR
SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	AD
		Pancreatic cancer, somatic	-
		Myhre syndrome	AD
		Polyposis, juvenile intestinal	AD
SMPX	300226	Myopathy, distal, 7, adult-onset, X-linked	XLR
		Deafness, X-linked 4	XLD
SNAI2	602150	Waardenburg syndrome, type 2D	AR
		Piebaldism	AD
SOX10	602229	Waardenburg syndrome, type 4C	AD
		PCWH syndrome	AD
		Waardenburg syndrome, type 2E, with or without neurologic involvement	AD
SOX2	184429	Microphthalmia, syndromic 3	AD
		Optic nerve hypoplasia and abnormalities of the central nervous system	AD
SPATA5	613940	Epilepsy, hearing loss, and mental retardation syndrome	AR
STRC	606440	Deafness, autosomal recessive 16	AR
SUCLA2	603921	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria)	AR
SUCLG1	611224	Mitochondrial DNA depletion syndrome 9 (encephalomyopathic type with methylmalonic aciduria)	AR
SYNE4	615535	Deafness, autosomal recessive 76	AR

TBC1D24	613577	Deafness, autosomal dominant 65	AD
		Deafness, autosomal recessive 86	AR
		Myoclonic epilepsy, infantile, familial	AR
		Developmental and epileptic encephalopathy 16	AR
		DOORS syndrome	AR
		Epilepsy, rolandic, with proxysmal exercise-induce dystonia and writer's cramp	AR
TBX1	602054	Velocardiofacial syndrome	AD
		Tetralogy of Fallot	AD
		DiGeorge syndrome	AD
		Conotruncal anomaly face syndrome	-
TCOF1	606847	Treacher Collins syndrome 1	AD
TECTA	602574	Deafness, autosomal dominant 8/12	AD
		Deafness, autosomal recessive 21	AR
TFAP2A	107580	Branchiooculofacial syndrome	AD
TIMM8A	300356	Mohr-Tranebjaerg syndrome	XLR
TJP2	607709	Hypercholanemia, familial	AR
		Cholestasis, progressive familial intrahepatic 4	AR
TMC1	606706	Deafness, autosomal dominant 36	AD
		Deafness, autosomal recessive 7	AR
TMIE	607237	Deafness, autosomal recessive 6	AR
TMPRSS3	605511	Deafness, autosomal recessive 8/10	AR

TNC	187380	Deafness, autosomal dominant 56	AD
TPRN	613354	Deafness, autosomal recessive 79	AR
TRIOBP	609761	Deafness, autosomal recessive 28	AR
TRMU	610230	Liver failure, transient infantile	AR
		Deafness, mitochondrial, modifier of	Mitochondrial
TSPEAR	612920	Deafness, autosomal recessive 98	AR
		Ectodermal dysplasia 14, hair/tooth type with or without hypohidrosis	AR
		Tooth agenesis, selective, 10	-
TWNK	606075	Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3	AD
		Perrault syndrome 5	AR
		Mitochondrial DNA depletion syndrome 7 (hepatocerebral type)	AR
TYR	606933	[Skin/hair/eye pigmentation 3, light/dark/freckling skin]	AD
		Waardenburg syndrome/albinism, digenic	-
		[Skin/hair/eye pigmentation 3, blue/green eyes]	AD
		Melanoma, cutaneous malignant, susceptibility to, 8	AD
		Albinism, oculocutaneous, type IA	AR
		Albinism, oculocutaneous, type IB	AR
USH1C	605242	Usher syndrome, type 1C	AR
		Deafness, autosomal recessive 18A	AR
USH1G	607696	Usher syndrome, type 1G	AR
USH2A	608400	Usher syndrome, type 2A	AR

		Retinitis pigmentosa 39	-
VCAN	118661	Wagner syndrome 1	AD
WBP2	606962	Deafness, autosomal recessive 107	AR
WFS1	606201	Cataract 41	AD
		Wolfram-like syndrome, autosomal dominant	AD
		Wolfram syndrome 1	AR
		Diabetes mellitus, noninsulin-dependent, association with	AD
		Deafness, autosomal dominant 6/14/38	AD
WHRN	607928	Deafness, autosomal recessive 31	AR
		Usher syndrome, type 2D	AR