Oslo universitetssykehus
Avdeling for medisinsk genetikk

Hørselshemning

Genpanel, versjon v01

* Enkelte genomiske regioner har lav eller ingen sekvensdekning ved eksomsekvensering. Dette skyldes at de har stor likhet med andre områder i genomet, slik at spesifikk gjenkjennelse av disse områdene og påvisning av varianter i disse områdene, blir vanskelig og upålitelig. Disse genetiske regionene har vi identifisert ved å benytte USCS segmental duplication hvor områder større enn 1 kb og ≥90% likhet med andre regioner i genomet, gjenkjennes (https://genome.ucsc.edu).

For noen gener ligger alle ekson i områder med segmentale duplikasjoner: **ACTB, ACTG1**

Vi gjør oppmerksom på at ved identifiseringav ekson oppstrøms for startkodon kan eksonnummereringen endres uten at transkript ID endres.

Avdelingens websider har en full oversikt over områder som er affisert av segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ABCC1	51	NM_004996.3		1-31	?Deafness, autosomal dominant 77 OMIM
ABHD12	15868	NM_001042472.2		1-13	Polyneuropathy, hearing loss, ataxia, retinitis pigmentosa, and cataract OMIM
ACOX1	119	NM_004035.6		1-14	Mitchell syndrome OMIM Peroxisomal acyl-CoA oxidase deficiency OMIM
ACTB	132	NM_001101.4	2-6	2-6	?Dystonia, juvenile-onset OMIM Baraitser-Winter syndrome 1 OMIM
ACTG1	144	NM_001614.3	2-6	2-6	Baraitser-Winter syndrome 2 OMIM Deafness, autosomal dominant 20/26 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
AIFM1	8768	NM_004208.3		1-16	Cowchock syndrome OMIM Deafness, X-linked 5 OMIM Spondyloepimetaphyseal dysplasia, X-linked, with hypomyelinating leukodystrophy OMIM
ALMS1	428	NM_015120.4	17-21	1-23	Alstrom syndrome OMIM
ANKH	15492	NM_054027.4		1-12	Chondrocalcinosis 2 OMIM Craniometaphyseal dysplasia OMIM
AP1B1	554	NM_001127.3	2-3, 6	2-23	Keratitis-ichthyosis-deafness syndrome, autosomal recessive OMIM
ARSG	24102	NM_014960.5		2-12	Usher syndrome, type IV OMIM
ATP1A3	801	NM_152296.5		1-23	Alternating hemiplegia of childhood 2 OMIM CAPOS syndrome OMIM Dystonia-12 OMIM
ATP2B2	815	NM_001683.4		2-20	{Deafness, autosomal recessive 12, modifier of} OMIM
ATP6V1B1	853	NM_001692.3		1-14	Distal renal tubular acidosis 2 with progressive sensorineural hearing loss OMIM
ATP6V1B2	854	NM_001693.4		1-14	Deafness, congenital, with onychodystrophy, autosomal dominant OMIM Zimmermann-Laband syndrome 2 OMIM
BCAP31	16695	NM_001139441.1	5-8	2-8	Deafness, dystonia, and cerebral hypomyelination OMIM
BCS1L	1020	NM_004328.5		3-9	Bjornstad syndrome OMIM GRACILE syndrome OMIM Mitochondrial complex III deficiency, nuclear type 1 OMIM
BMP4	1071	NM_001202.6		3-4	Microphthalmia, syndromic 6 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
BSND	16512	NM_057176.2		1-4	Bartter syndrome, type 4a OMIM Sensorineural deafness with mild renal dysfunction OMIM
BTD	1122	NM_000060.3		1-4	Biotinidase deficiency OMIM
C10orf2	1160	NM_021830.5		1-5	Mitochondrial DNA depletion syndrome 7 (hepatocerebral type) OMIM Perrault syndrome 5 OMIM Progressive external ophthalmoplegia with mitochondrial DNA deletions, autosomal dominant 3 OMIM
CABP2	1385	NM_016366.2		1-7	Deafness, autosomal recessive 93 OMIM
CACNA1D	1391	NM_000720.4		1-49	Sinoatrial node dysfunction and deafness OMIM
CCDC50	18111	NM_178335.2		1-12	?Deafness, autosomal dominant 44 OMIM
CDC14A	1718	NM_033312.2		1-15	Deafness, autosomal recessive 32, with or without immotile sperm OMIM
CDC6	1744	NM_001254.3		2-12	?Meier-Gorlin syndrome 5 OMIM
CDH23	13733	NM_022124.5		2-68	Deafness, autosomal recessive 12 OMIM Usher syndrome, type 1D OMIM Usher syndrome, type 1D/F digenic OMIM
CDK5RAP2	18672	NM_018249.6		1-38	Microcephaly 3, primary, autosomal recessive OMIM
CDK9	1780	NM_001261.4		1-7	Multiple malformation syndrome with retinal dystrophy, mimicing CHARGE PubMed
CDT1	24576	NM_030928.3		1-10	Meier-Gorlin syndrome 4 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
CEACAM16	31948	NM_001039213.3		2-7	Deafness, autosomal dominant 4B OMIM Deafness, autosomal recessive 113 OMIM
CEP250	1859	NM_007186.5		4-35	Cone-rod dystrophy and hearing loss 2 OMIM
CEP78	25740	NM_001098802.1		1-16	Cone-rod dystrophy and hearing loss OMIM
CHD7	20626	NM_017780.3		2-38	CHARGE syndrome OMIM Hypogonadotropic hypogonadism 5 with or without anosmia OMIM
CHSY1	17198	NM_014918.4		1-3	Temtamy preaxial brachydactyly syndrome OMIM
CIB2	24579	NM_006383.3		1-6	Deafness, autosomal recessive 48 OMIM Usher syndrome, type IJ OMIM
CISD2	24212	NM_001008388.4	3	1-3	Wolfram syndrome 2 OMIM
CLDN14	2035	NM_144492.2		3	Deafness, autosomal recessive 29 OMIM
CLDN9	2051	NM_020982.3		1	?Deafness, autosomal recessive 116 OMIM
CLIC5	13517	NM_001114086.2		1-6	?Deafness, autosomal recessive 103 OMIM
CLPP	2084	NM_006012.2		1-6	Perrault syndrome 3 OMIM
CLRN1	12605	NM_174878.2		1-3	Usher syndrome, type 3A OMIM
CLRN2	33939	NM_001079827.2		1-3	?Deafness, autosomal recessive 117 OMIM
COCH	2180	NM_004086.3		2-12	?Deafness, autosomal recessive 110 OMIM Deafness, autosomal dominant 9 OMIM
COG4	18620	NM_015386.2		1-19	Saul-Wilson syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
COL11A1	2186	NM_001854.3		1-67	Deafness, autosomal dominant 37 OMIM Fibrochondrogenesis 1 OMIM Marshall syndrome OMIM Stickler syndrome, type II OMIM
COL11A2	2187	NM_080680.2		1-66	Deafness, autosomal dominant 13 OMIM Deafness, autosomal recessive 53 OMIM Otospondylomegaepiphyseal dysplasia, autosomal dominant OMIM Otospondylomegaepiphyseal dysplasia, autosomal recessive OMIM
COL2A1	2200	NM_001844.5		1-54	?Epiphyseal dysplasia, multiple, with myopia and deafness OMIM Kniest dysplasia OMIM SED congenita OMIM Spondyloperipheral dysplasia OMIM Stickler sydrome, type I, nonsyndromic ocular OMIM Stickler syndrome, type I OMIM
COL4A3	2204	NM_000091.4		1-52	Alport syndrome 2, autosomal recessive OMIM Alport syndrome 3, autosomal dominant OMIM
COL4A4	2206	NM_000092.5		2-48	Alport syndrome 2, autosomal recessive OMIM
COL4A5	2207	NM_000495.4		1-51	Alport syndrome 1, X-linked OMIM
COL4A6	2208	NM_001847.4		1-45	?Deafness, X-linked 6 OMIM
COL9A1	2217	NM_001851.5		1-38	Stickler syndrome, type IV OMIM PubMed
COL9A2	2218	NM_001852.4		1-32	?Stickler syndrome, type V OMIM
COL9A3	2219	NM_001853.3		1-32	Epiphyseal dysplasia, multiple, 3, with or without myopathy OMIM

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CRYM	2418	NM_001888.5		3-10	Deafness, autosomal dominant 40 OMIM
DCAF17	25784	NM_025000.3		1-14	Woodhouse-Sakati syndrome OMIM
DFNA5	2810	NM_004403.2		2-10	Deafness, autosomal dominant 5 OMIM
DFNB31	16361	NM_001173425.1		1-12	Deafness, autosomal recessive 31 OMIM Usher syndrome, type 2D OMIM
DFNB59	29502	NM_001042702.3		2-7	Deafness, autosomal recessive 59 OMIM
DHODH	2867	NM_001361.4		1-9	Miller syndrome OMIM
DIABLO	21528	NM_019887.5		2-7	Deafness, autosomal dominant 64 OMIM
DIAPH1	2876	NM_005219.4		1-28	Deafness, autosomal dominant 1, with or without thrombocytopenia OMIM Seizures, cortical blindness, microcephaly syndrome OMIM
DIAPH3	15480	NM_001042517.1		1-28	Auditory neuropathy, autosomal dominant, 1 OMIM
DMXL2	2938	NM_001174116.2		1-43	?Deafness, autosomal dominant 71 OMIM ?Polyendocrine-polyneuropathy syndrome OMIM Developmental and epileptic encephalopathy 81 OMIM
DNMT1	2976	NM_001130823.1		1-41	Cerebellar ataxia, deafness, and narcolepsy, autosomal dominant OMIM Neuropathy, hereditary sensory, type IE OMIM
DSPP	3054	NM_014208.3		2-5	Deafness, autosomal dominant 39, with dentinogenesis OMIM
EDN3	3178	NM_207034.2		1-5	Waardenburg syndrome, type 4B OMIM PubMed

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
EDNRA	3179	NM_001957.3		2-8	Mandibulofacial dysostosis with alopecia OMIM
EDNRB	3180	NM_000115.5		2-8	ABCD syndrome OMIM Waardenburg syndrome, type 4A OMIM PubMed
EFTUD2	30858	NM_004247.4		2-28	Mandibulofacial dysostosis, Guion-Almeida type OMIM
EIF3F	3275	NM_003754.2		1-8	Mental retardation, autosomal recessive 67 OMIM
EIF4A3	18683	NM_014740.3		1-12	Robin sequence with cleft mandible and limb anomalies OMIM
ELMOD3	26158	NM_001135022.1		4-14	?Deafness, autosomal recessive 88 OMIM
EPS8	3420	NM_004447.5		2-21	?Deafness, autosomal recessive 102 OMIM
EPS8L2	21296	NM_022772.3		2-21	Deafness autosomal recessive 106 OMIM
ERAL1	3424	NM_005702.3		1-10	Perrault syndrome 6 OMIM
ESPN	13281	NM_031475.2	2-12	1-13	?Usher syndrome, type 1M OMIM Deafness, autosomal recessive 36 OMIM Deafness, neurosensory, without vestibular involvement, autosomal dominant OMIM
ESRP1	25966	NM_017697.3		1-15	?Deafness, autosomal recessive 109 OMIM
ESRRB	3473	NM_004452.3		4-11	Deafness, autosomal recessive 35 OMIM
EYA1	3519	NM_000503.5		3-18	?Otofaciocervical syndrome OMIM Anterior segment anomalies with or without cataract OMIM Branchiootic syndrome 1 OMIM Branchiootorenal syndrome 1, with or without cataracts OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
EYA4	3522	NM_004100.4		2-20	Deafness, autosomal dominant 10 OMIM
FAM65B	13872	NM_001286445.3		1-22	?Deafness, autosomal recessive 104 OMIM
FDXR	3642	NM_024417.4		1-12	Auditory neuropathy and optic atrophy OMIM
FGF10	3666	NM_004465.1		1-3	Aplasia of lacrimal and salivary glands OMIM LADD syndrome OMIM
FGF3	3681	NM_005247.4		1-3	Deafness, congenital with inner ear agenesis, microtia, and microdontia OMIM
FGFR2	3689	NM_000141.4		2-18	Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis OMIM Apert syndrome OMIM Beare-Stevenson cutis gyrata syndrome OMIM Bent bone dysplasia syndrome OMIM Craniofacial-skeletal-dermatologic dysplasia OMIM Crouzon syndrome OMIM Jackson-Weiss syndrome OMIM LADD syndrome OMIM Pfeiffer syndrome OMIM Saethre-Chotzen syndrome OMIM
FGFR3	3690	NM_000142.4		2-18	Achondroplasia OMIM CATSHL syndrome OMIM Crouzon syndrome with acanthosis nigricans OMIM Hypochondroplasia OMIM LADD syndrome OMIM Muenke syndrome OMIM SADDAN OMIM Thanatophoric dysplasia, type I OMIM Thanatophoric dysplasia, type II OMIM

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FITM2	16135	NM_001080472.4		1-2	Siddiqi syndrome OMIM PubMed
FOXF2	3810	NM_001452.1		1-2	cochlea anomali, sensorinevralt hoerselstap PubMed
FOXI1	3815	NM_012188.4		1-2	Enlarged vestibular aqueduct OMIM
FRAS1	19185	NM_025074.6		1-74	Fraser syndrome 1 OMIM
FREM2	25396	NM_207361.6		1-24	Cryptophthalmos, unilateral or bilateral, isolated OMIM Fraser syndrome 2 OMIM
GATA3	4172	NM_001002295.2		2-6	Hypoparathyroidism, sensorineural deafness, and renal dysplasia OMIM
GDF6	4221	NM_001001557.3		1-2	Klippel-Feil syndrome 1, autosomal dominant OMIM Microphthalmia with coloboma 6, digenic OMIM Multiple synostoses syndrome 4 OMIM
GGPS1	4249	NM_001037277.1		2-4	Muskeldystrofi, hoerselstap og ovarial insuffisiens PubMed
GIPC3	18183	NM_133261.2		1-6	Deafness, autosomal recessive 15 OMIM
GJB2	4284	NM_004004.6		2	Bart-Pumphrey syndrome OMIM Deafness, autosomal dominant 3A OMIM Deafness, autosomal recessive 1A OMIM Hystrix-like ichthyosis with deafness OMIM Keratitis-ichthyosis-deafness syndrome OMIM Keratoderma, palmoplantar, with deafness OMIM Vohwinkel syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
GJB3	4285	NM_024009.2		2	Deafness, autosomal dominant 2B OMIM Deafness, autosomal dominant, with peripheral neuropathy Deafness, autosomal recessive Deafness, digenic, GJB2/GJB3 OMIM
GJB6	4288	NM_006783.4		3	Deafness, autosomal dominant 3B OMIM Deafness, autosomal recessive 1B OMIM Deafness, digenic GJB2/GJB6 OMIM
GNAI3	4387	NM_006496.3		1-8	Auriculocondylar syndrome 1 OMIM
GPR98	17416	NM_032119.3		1-90	Usher syndrome, type 2C OMIM Usher syndrome, type 2C, GPR98/PDZD7 digenic OMIM
GPSM2	29501	NM_013296.4		2-15	Chudley-McCullough syndrome OMIM
GRHL2	2799	NM_024915.4		1-16	Deafness, autosomal dominant 28 OMIM Ectodermal dysplasia/short stature syndrome OMIM
GRIP1	18708	NM_021150.4		1-24	Fraser syndrome 3 OMIM
GRXCR1	31673	NM_001080476.2		1-4	Deafness, autosomal recessive 25 OMIM
GRXCR2	33862	NM_001080516.1		1-3	?Deafness, autosomal recessive 101 OMIM
GSC	4612	NM_173849.2		1-3	Short stature, auditory canal atresia, mandibular hypoplasia, skeletal abnormalities OMIM
HAAO	4796	NM_012205.3		1-10	Vertebral, cardiac, renal, and limb defects syndrome 1 OMIM
HARS	4816	NM_002109.5		1-13	Usher syndrome type 3B OMIM

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HARS2	4817	NM_012208.3		1-13	Perrault syndrome 2 OMIM
HGF	4893	NM_000601.5		1-18	Deafness, autosomal recessive 39 OMIM
HMX1	5017	NM_018942.2		1-2	Oculoauricular syndrome OMIM
HOMER2	17513	NM_004839.4		1-9	?Deafness, autosomal dominant 68 OMIM
HOXA2	5103	NM_006735.3		1-2	?Microtia, hearing impairment, and cleft palate (AR) OMIM Microtia with or without hearing impairment (AD) OMIM
HOXB1	5111	NM_002144.4		1-2	Facial paresis, hereditary congenital, 3 OMIM
HSD17B4	5213	NM_000414.3		1-24	D-bifunctional protein deficiency OMIM Perrault syndrome 1 OMIM
HSPA9	5244	NM_004134.6		1-17	Even-plus syndrome OMIM
ILDR1	28741	NM_001199799.2		1-8	Deafness, autosomal recessive 42 OMIM
KARS	6215	NM_001130089.1		2-15	Deafness, autosomal recessive 89 OMIM Deafness, congenital, and adult-onset progressive leukoencephalopathy OMIM Leukoencephalopathy, progressive, infantile-onset, with or without deafness OMIM
KCNE1	6240	NM_000219.5		4	Jervell and Lange-Nielsen syndrome 2 OMIM
KCNJ10	6256	NM_002241.4		2	Enlarged vestibular aqueduct, digenic OMIM SESAME syndrome OMIM
KCNQ1	6294	NM_000218.2		1-16	Jervell and Lange-Nielsen syndrome OMIM

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KCNQ4	6298	NM_004700.3		1-14	Deafness, autosomal dominant 2A OMIM
KDM6A	12637	NM_021140.3		1-29	Kabuki syndrome 2 OMIM
KIT	6342	NM_000222.2		1-21	Piebaldism OMIM
KITLG	6343	NM_000899.5		1-9	Deafness, autosomal dominant 69, unilateral or asymmetric OMIM
KMT2D	7133	NM_003482.3		1-54	Kabuki syndrome 1 OMIM
LARS2	17095	NM_015340.3		3-22	Hydrops, lactic acidosis, and sideroblastic anemia OMIM Perrault syndrome 4 OMIM
LHFPL5	21253	NM_182548.3		1-3	Deafness, autosomal recessive 67 OMIM
LHX3	6595	NM_014564.3		1-6	Pituitary hormone deficiency, combined, 3 OMIM
LMX1A	6653	NM_177398.4		2-9	Deafness, autosomal dominant 7 OMIM
LOXHD1	26521	NM_144612.6		1-40	Deafness, autosomal recessive 77 OMIM
LRP2	6694	NM_004525.2		1-79	Donnai-Barrow syndrome OMIM
LRTOMT	25033	NM_001145308.4		3-7	Deafness, autosomal recessive 63 OMIM
MAN2B1	6826	NM_000528.4		1-24	Mannosidosis, alpha-, types I and II OMIM
MANBA	6831	NM_005908.3		1-17	Mannosidosis, beta OMIM
MARVELD2	26401	NM_001038603.2		2-7	Deafness, autosomal recessive 49 OMIM
MASP1	6901	NM_139125.3		1-11	3MC syndrome 1 OMIM

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MET	7029	NM_001127500.2		2-21	?Deafness, autosomal recessive 97 OMIM
MGP	7060	NM_000900.3		1-4	Keutel syndrome OMIM
MITF	7105	NM_000248.3		1-9	COMMAD syndrome OMIM Tietz albinism-deafness syndrome OMIM Waardenburg syndrome, type 2A OMIM
MN1	7180	NM_002430.2		1-2	CEBALID syndrome OMIM
MORC2	23573	NM_014941.2		5-27	Charcot-Marie-Tooth disease, axonal, type 2Z OMIM Developmental delay, impaired growth, dysmorphic facies, and axonal neuropathy OMIM
MPZL2	3496	NM_005797.4		1-5	Deafness, autosomal recessive 111 OMIM
MSRB3	27375	NM_198080.3		1-6	Deafness, autosomal recessive 74 OMIM
MYH14	23212	NM_024729.3		2-41	?Peripheral neuropathy, myopathy, hoarseness, and hearing loss OMIM Deafness, autosomal dominant 4A OMIM
MYH9	7579	NM_002473.5		2-41	Deafness, autosomal dominant 17 OMIM Macrothrombocytopenia and granulocyte inclusions with or without nephritis or sensorineural hearing loss OMIM
MYO15A	7594	NM_016239.3		2-65	Deafness, autosomal recessive 3 OMIM
MYO3A	7601	NM_017433.5		3-35	Deafness, autosomal recessive 30 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
MYO6	7605	NM_004999.4		2-35	Deafness, autosomal dominant 22 OMIM Deafness, autosomal dominant 22, with hypertrophic cardiomyopathy OMIM Deafness, autosomal recessive 37 OMIM
MYO7A	7606	NM_000260.3		2-49	Deafness, autosomal dominant 11 OMIM Deafness, autosomal recessive 2 OMIM Usher syndrome, type 1B OMIM
NARS2	26274	NM_024678.5		1-14	?Deafness, autosomal recessive 94 OMIM Combined oxidative phosphorylation deficiency 24 OMIM
NDRG1	7679	NM_006096.4		2-16	Charcot-Marie-Tooth disease, type 4D OMIM
OFD1	2567	NM_003611.3		1-23	?Retinitis pigmentosa 23 OMIM Joubert syndrome 10 OMIM Orofaciodigital syndrome I OMIM Simpson-Golabi-Behmel syndrome, type 2 OMIM
OPA1	8140	NM_015560.2		1-28	?Mitochondrial DNA depletion syndrome 14 (encephalocardiomyopathic type) OMIM Optic atrophy plus syndrome OMIM
ORC1	8487	NM_004153.3		2-17	Meier-Gorlin syndrome 1 OMIM
ORC4	8490	NM_002552.5		2-14	Meier-Gorlin syndrome 2 OMIM
ORC6	17151	NM_014321.3		1-7	Meier-Gorlin syndrome 3 OMIM
OSBPL2	15761	NM_144498.3		2-14	Deafness, autosomal dominant 67 OMIM
OTOA	16378	NM_144672.3	20-28	1-28	Deafness, autosomal recessive 22 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
OTOF	8515	NM_194248.2		1-46	Auditory neuropathy, autosomal recessive, 1 OMIM Deafness, autosomal recessive 9 OMIM
OTOG	8516	NM_001277269.1		1-55	Deafness, autosomal recessive 18B OMIM
OTOGL	26901	NM_173591.3		1-58	Deafness, autosomal recessive 84B OMIM
OTX2	8522	NM_001270524.1		2-4	Microphthalmia, syndromic 5 OMIM
P2RX2	15459	NM_174873.2		1-12	Deafness, autosomal dominant 41 OMIM
PAX1	8615	NM_006192.5		1-5	Otofaciocervical syndrome 2 OMIM
PAX2	8616	NM_003987.4		1-11	Papillorenal syndrome OMIM
PAX3	8617	NM_181457.3		1-8	Craniofacial-deafness-hand syndrome OMIM Waardenburg syndrome, type 1 OMIM Waardenburg syndrome, type 3 OMIM
PBX1	8632	NM_002585.3		1-9	Congenital anomalies of kidney and urinary tract syndrome with or without hearing loss, abnormal ears, or developmental delay OMIM
PCDH15	14674	NM_033056.3		2-33	Deafness, autosomal recessive 23 OMIM Usher syndrome, type 1D/F digenic OMIM Usher syndrome, type 1F OMIM
PDSS1	17759	NM_014317.3		1-12	Coenzyme Q10 deficiency, primary, 2 OMIM
PDZD7	26257	NM_024895.4		2-10	{Retinal disease in Usher syndrome type IIA, modifier of} OMIM Deafness, autosomal recessive 57 OMIM Usher syndrome, type IIC, GPR98/PDZD7 digenic OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
PEX1	8850	NM_000466.2		1-24	Heimler syndrome 1 OMIM Peroxisome biogenesis disorder 1A (Zellweger) OMIM Peroxisome biogenesis disorder 1B (NALD/IRD) OMIM
PEX26	22965	NM_017929.6		2-6	Peroxisome biogenesis disorder 7A (Zellweger) OMIM Peroxisome biogenesis disorder 7B OMIM
PEX6	8859	NM_000287.3		1-17	Heimler syndrome 2 OMIM Peroxisome biogenesis disorder 4A (Zellweger) OMIM Peroxisome biogenesis disorder 4B OMIM
PISD	8999	NM_014338.3		3-9	Liberfarb syndrome OMIM
PLCB4	9059	NM_000933.3		1-36	Auriculocondylar syndrome 2 OMIM
PLOD3	9083	NM_001084.4		1-19	Lysyl hydroxylase 3 deficiency OMIM
PLS1	9090	NM_002670.2		2-16	Deafness, autosomal dominant 76 OMIM
PMP22	9118	NM_000304.4		2-5	Charcot-Marie-Tooth disease, type 1E OMIM
PNPT1	23166	NM_033109.4	28	1-28	Combined oxidative phosphorylation deficiency 13 OMIM Deafness, autosomal recessive 70 OMIM
POLD1	9175	NM_002691.4		2-27	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome OMIM
POLR1A	17264	NM_015425.3		1-34	Acrofacial dysostosis, Cincinnati type OMIM
POLR1C	20194	NM_203290.3		1-9	Treacher Collins syndrome 3 OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
POLR1D	20422	NM_015972.3		1-2	Treacher Collins syndrome 2 OMIM
POU3F4	9217	NM_000307.4		1	Deafness, X-linked 2 OMIM
POU4F3	9220	NM_002700.2		1-2	Deafness, autosomal dominant 15 OMIM
PPIP5K2	29035	NM_001281471.1		2-33	Deafness, autosomal recessive 100 OMIM
PRPS1	9462	NM_002764.4	7	1-7	Arts syndrome OMIM Charcot-Marie-Tooth disease, X-linked recessive, 5 OMIM Deafness, X-linked 1 OMIM Gout, PRPS-related OMIM Phosphoribosylpyrophosphate synthetase superactivity OMIM
PTPRQ	9679	NM_001145026.2		1-42	Deafness, autosomal dominant 73 OMIM Deafness, autosomal recessive 84A OMIM
RDX	9944	NM_002906.3	14	2-14	Deafness, autosomal recessive 24 OMIM
RFT1	30220	NM_052859.3		1-13	Congenital disorder of glycosylation, type In OMIM
RMND1	21176	NM_017909.4	2	2-12	Combined oxidative phosphorylation deficiency 11 OMIM
ROR1	10256	NM_005012.3		1-9	?Deafness, autosomal recessive 108 OMIM
RPS28	10418	NM_001031.5		1-3	Diamond Blackfan anemia 15 with mandibulofacial dysostosis OMIM
RPS6KA3	10432	NM_004586.3		1-22	Coffin-Lowry syndrome OMIM
S1PR2	3169	NM_004230.3		2	Deafness, autosomal recessive 68 OMIM

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SALL1	10524	NM_002968.2	2-3	1-3	Townes-Brocks branchiootorenal-like syndrome OMIM Townes-Brocks syndrome 1 OMIM
SALL4	15924	NM_020436.3		1-4	Duane-radial ray syndrome OMIM IVIC syndrome OMIM
SERAC1	21061	NM_032861.4		2-17	3-methylglutaconic aciduria with deafness, encephalopathy, and Leigh-like syndrome OMIM
SERPINB6	8950	NM_004568.5		2-7	?Deafness, autosomal recessive 91 OMIM
SF3B4	10771	NM_005850.4		1-6	Acrofacial dysostosis 1, Nager type OMIM
SGPL1	10817	NM_003901.4		2-15	Nephrotic syndrome, type 14 OMIM
SIX1	10887	NM_005982.4		1-2	Branchiootic syndrome 3 OMIM Deafness, autosomal dominant 23 OMIM
SIX5	10891	NM_175875.4		1-3	Branchiootorenal syndrome 2 OMIM
SLC12A2	10911	NM_001046.3		1-27	Deafness, autosomal dominant 78 OMIM Delpire-McNeill syndrome OMIM Kilquist syndrome OMIM
SLC17A8	20151	NM_139319.2		1-12	Deafness, autosomal dominant 25 OMIM
SLC19A2	10938	NM_006996.2		1-6	Thiamine-responsive megaloblastic anemia syndrome OMIM
SLC26A4	8818	NM_000441.1		2-21	Deafness, autosomal recessive 4, with enlarged vestibular aqueduct OMIM Pendred syndrome OMIM
SLC26A5	9359	NM_198999.2		3-20	?Deafness, autosomal recessive 61 OMIM
SLC29A3	23096	NM_018344.6		1-6	Histiocytosis-lymphadenopathy plus syndrome OMIM

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SLC33A1	95	NM_004733.3	6	1-6	Congenital cataracts, hearing loss, and neurodegeneration OMIM
SLC4A11	16438	NM_032034.3		1-19	Corneal endothelial dystrophy and perceptive deafness OMIM Corneal endothelial dystrophy, autosomal recessive OMIM
SLC52A2	30224	NM_024531.5		2-5	Brown-Vialetto-Van Laere syndrome 2 OMIM
SLC52A3	16187	NM_033409.4		2-5	?Fazio-Londe disease OMIM Brown-Vialetto-Van Laere syndrome 1 OMIM
SLC9A1	11071	NM_003047.5		1-12	Lichtenstein-Knorr syndrome OMIM
SLITRK6	23503	NM_032229.3		2	Deafness and myopia OMIM
SMAD4	6770	NM_005359.5		2-12	Myhre syndrome OMIM
SMPX	11122	NM_014332.2		2-4	Deafness, X-linked 4 OMIM
SNAI2	11094	NM_003068.4		1-3	Piebaldism OMIM Waardenburg syndrome, type 2D OMIM
SOX10	11190	NM_006941.3		2-4	PCWH syndrome OMIM Waardenburg syndrome, type 2E, with or without neurologic involvement OMIM Waardenburg syndrome, type 4C OMIM
SOX2	11195	NM_003106.4	1	1	Microphthalmia, syndromic 3 OMIM Optic nerve hypoplasia and abnormalities of the central nervous system OMIM
SPATA5	18119	NM_145207.2		1-16	Epilepsy, hearing loss, and mental retardation syndrome OMIM
SPATC1L	1298	NM_001142854.2		2-5	Hoerselstap PubMed

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SPNS2	26992	NM_001124758.2		1-12	?Deafness, autosomal recessive 115 OMIM
SPTBN4	14896	NM_020971.2		2-36	Neurodevelopmental disorder with hypotonia, neuropathy, and deafness OMIM
STAG2	11355	NM_001042749.2		3-35	Holoprosencephaly 13, X-linked OMIM Mullegama-Klein-Martinez syndrome OMIM
SUCLA2	11448	NM_003850.2		1-11	Mitochondrial DNA depletion syndrome 5 (encephalomyopathic with or without methylmalonic aciduria) OMIM
SYNE4	26703	NM_001039876.2		1-8	Deafness, autosomal recessive 76 OMIM
TBC1D24	29203	NM_001199107.1		2-8	Deafness, autosomal dominant 65 OMIM Deafness, autosomal recessive 86 OMIM DOORS syndrome OMIM
TBL1X	11585	NM_005647.4		4-18	Hypothyroidism, congenital, nongoitrous, 8 OMIM
TCOF1	11654	NM_001135243.1		1-26	Treacher Collins syndrome 1 OMIM
TECTA	11720	NM_005422.2		1-23	Deafness, autosomal dominant 8/12 OMIM Deafness, autosomal recessive 21 OMIM
TFAP2A	11742	NM_003220.2		1-7	Branchiooculofacial syndrome OMIM
THOC1	19070	NM_005131.3		1-21	Non-syndromic late-onset hearing loss PubMed
TIMM8A	11817	NM_004085.3	2	1-2	Mohr-Tranebjaerg syndrome OMIM
TMC1	16513	NM_138691.2		5-24	Deafness, autosomal dominant 36 OMIM Deafness, autosomal recessive 7 OMIM
TMEM132E	26991	NM_207313.2		1-10	Deafness, autosomal recessive 99 OMIM

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TMIE	30800	NM_147196.2		1-4	Deafness, autosomal recessive 6 OMIM
TMPRSS3	11877	NM_024022.2		2-13	Deafness, autosomal recessive 8/10 OMIM
TMTC2	25440	NM_152588.3		1-12	sensorinevralt hoerselstap PubMed
TNC	5318	NM_002160.3		2-28	Deafness, autosomal dominant 56 OMIM
TOP2B	11990	NM_001068.3		1-36	autosomal dominant arvelig hoerselstap PubMed
TPRN	26894	NM_001128228.3		1-4	Deafness, autosomal recessive 79 OMIM
TRIOBP	17009	NM_001039141.2		3-23	Deafness, autosomal recessive 28 OMIM
TRPV4	18083	NM_021625.5		2-16	Hereditary motor and sensory neuropathy, type IIc OMIM
TSHZ1	10669	NM_005786.5		2	Aural atresia, congenital OMIM
TUBB4B	20771	NM_006088.5		1-4	Leber congenital amaurosis with early-onset deafness OMIM
TXNL4A	30551	NM_006701.4		1-3	Burn-McKeown syndrome OMIM
USH1C	12597	NM_005709.3		1-21	Deafness, autosomal recessive 18A OMIM Usher syndrome, type 1C OMIM
USH1G	16356	NM_173477.5		1-3	Usher syndrome, type 1G OMIM
USH2A	12601	NM_206933.2		2-72	Usher syndrome, type 2A OMIM
VPS33B	12712	NM_018668.4		1-23	Arthrogryposis, renal dysfunction, and cholestasis 1 OMIM
WBP2	12738	NM_012478.3		1-8	Deafness, autosomal recessive 107 OMIM

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WFS1	12762	NM_006005.3		2-8	Deafness, autosomal dominant 6/14/38 OMIM Wolfram syndrome 1 OMIM Wolfram-like syndrome, autosomal dominant OMIM
XYLT2	15517	NM_022167.4		1-11	Spondyloocular syndrome OMIM
YARS	12840	NM_003680.3		1-13	Charcot-Marie-Tooth disease, dominant intermediate C OMIM