Ear, Nose, Throat

ENT

Ear ,Nose,Throat (ENT)

Single Gene

Code	Diseases	Gene
sCe4/0001	Deafness, autosomal recessive, type 12	CDH23
sCe4/0002	Marshall syndrome	COL11A1
sCe4/0003	Brown-Vialetto-Van Laere syndrome type 2	SLC52A2
sCe4/0004	Deafness, autosomal recessive	GJB3
sCe4/0005	Deafness, autosomal recessive	SUN1
sCe4/0006	Deafness, autosomal recessive type 15	GIPC3
sCe4/0007	Deafness, autosomal recessive type 16	STRC
sCe4/0008	Deafness, autosomal recessive type 18	USH1C
sCe4/0009	Deafness, autosomal recessive type 1A	GJB2
sCe4/0010	Deafness, autosomal recessive type 1B	GJB6
sCe4/0011	Deafness, autosomal recessive type 2	MYO7A
sCe4/0012	Deafness, autosomal recessive type 22	OTOA
sCe4/0013	Deafness, autosomal recessive type 23	PCDH15
sCe4/0014	Deafness, autosomal recessive type 24	RDX
sCe4/0015	Deafness, autosomal recessive type 25	GRXCR1
sCe4/0016	Deafness, autosomal recessive type 28	TRIOBP
sCe4/0017	Deafness, autosomal recessive type 29	CLDN14
sCe4/0018	Deafness, autosomal recessive type 3	MYO15A
sCe4/0019	Deafness, autosomal recessive type 30	MYO3A
sCe4/0020	Deafness, autosomal recessive type 31	WHRN
sCe4/0021	Deafness, autosomal recessive type 35	ESRRB
sCe4/0022	Deafness, autosomal recessive type 36	ESPN
sCe4/0023	Deafness, autosomal recessive type 39	HGF
sCe4/0024	Deafness, autosomal recessive type 4	FOXI1
sCe4/0025	Deafness, autosomal recessive type 42	ILDR1

sCe4/0026	Deafness, autosomal recessive type 48	CIB2
sCe4/0027	Deafness, autosomal recessive type 49	MARVELD2
sCe4/0028	Deafness, autosomal recessive type 53	COL11A2
sCe4/0029	Deafness, autosomal recessive type 59	DFNB59
sCe4/0030	Deafness, autosomal recessive type 6	TMIE
sCe4/0031	Deafness, autosomal recessive type 61	SLC26A5
sCe4/0032	Deafness, autosomal recessive type 63	LRTOMT
sCe4/0033	Deafness, autosomal recessive type 66	DCDC2
sCe4/0034	Deafness, autosomal recessive type 67	LHFPL5
sCe4/0035	Deafness, autosomal recessive type 7	TMC1
sCe4/0036	Deafness, autosomal recessive type 70	PNPT1
sCe4/0037	Deafness, autosomal recessive type 74	MSRB3
sCe4/0038	Deafness, autosomal recessive type 76	SYNE4
sCe4/0039	Deafness, autosomal recessive type 77	LOXHD1
sCe4/0040	Deafness, autosomal recessive type 79	TPRN
sCe4/0041	Deafness, autosomal recessive type 8/10	TMPRSS3
sCe4/0042	Deafness, autosomal recessive type 84	PTPRQ
sCe4/0043	Deafness, autosomal recessive type 86	TBC1D24
sCe4/0044	Deafness, autosomal recessive type 89	KARS
sCe4/0045	Deafness, autosomal recessive type 9	OTOF
sCe4/0046	Deafness, autosomal recessive type 91	SERPINB6
sCe4/0047	Deafness, autosomal recessive type 93	CABP2
sCe4/0048	Deafness with keratopachydermia and constrictions of fingers and toes	GJB2
sCe4/0049	Deafness, autosomal dominant type 1	DIAPH1
sCe4/0050	Deafness, autosomal dominant type 10	EYA4
sCe4/0051	Deafness, autosomal dominant type 11	MYO7A
sCe4/0052	Deafness, autosomal dominant type 12	TECTA
sCe4/0053	Deafness, autosomal dominant type 13	COL11A2
sCe4/0054	Deafness, autosomal dominant type 15	POU4F3

sCe4/0055	Deafness, autosomal dominant type 17	MYH9
sCe4/0056	Deafness, autosomal dominant type 20	ACTG1
sCe4/0057	Deafness, autosomal dominant type 22	MYO6
sCe4/0058	Deafness, autosomal dominant type 23	SIX1
sCe4/0059	Deafness, autosomal dominant type 25	SLC17A8
sCe4/0060	Deafness, autosomal dominant type 28	GRHL2
sCe4/0061	Deafness, autosomal dominant type 2A	KCNQ4
sCe4/0062	Deafness, autosomal dominant type 2B	GJB3
sCe4/0063	Deafness, autosomal dominant type 36	TMC1
sCe4/0064	Deafness, autosomal dominant type 39, with dentinogenesis type 1	DSPP
sCe4/0065	Deafness, autosomal dominant type 3A	GJB2
sCe4/0066	Deafness, autosomal dominant type 3B	GJB6
sCe4/0067	Deafness, autosomal dominant type 4	MYH14
sCe4/0068	Deafness, autosomal dominant type 40	CRYM
sCe4/0069	Deafness, autosomal dominant type 44	CCDC50
sCe4/0070	Deafness, autosomal dominant type 48	MYO1A
sCe4/0071	Deafness, autosomal dominant type 4B	CEACAM16
sCe4/0072	Deafness, autosomal dominant type 5	DFNA5
sCe4/0073	Deafness, autosomal dominant type 50	MIR96
sCe4/0074	Deafness, autosomal dominant type 52	POU4F3
sCe4/0075	Deafness, autosomal dominant type 6	WFS1
sCe4/0076	Deafness, autosomal dominant type 64	DIABLO
sCe4/0077	Deafness, autosomal dominant type 65	TBC1D24
sCe4/0078	Deafness, autosomal dominant type 9	COCH
sCe4/0079	Keratitis ichthyosis deafness syndrome autosomal dominant	GJB2
sCe4/0080	Keratoderma, palmoplantar, with deafness	GJB2
sCe4/0081	Knuckle pads and leukonychia sensorineural deafness	GJB2
sCe4/0082	Deafness, dystonia, and cerebral hypomyelination, X-linked	BCAP31
sCe4/0083	Deafness, X-linked type 1	PRPS1

sCe4/0084	Deafness, X-linked type 2	POU3F4
sCe4/0085	Deafness, X-linked type 4	SMPX
sCe4/0086	Deafness, X-linked type 5	AIFM1
sCe4/0087	Deafness, X-linked type 6	COL4A6
sCe4/0088	Alport syndrome, autosomal recessive	COL4A4
sCe4/0089	Alport syndrome, autosomal recessive	COL4A3
sCe4/0090	Alport syndrome, X-Linked	COL4A5
sCe4/0091	Auditory neuropathy, autosomal dominant	DIAPH3
sCe4/0092	Auriculocondylar syndrome type 2	PLCB4
sCe4/0093	Branchiootic syndrome type 1	EYA1
sCe4/0094	Brown-Vialetto-Van Laere syndrome 1	SLC52A3
sCe4/0095	Ciliogenesis related disorder	PTPN23
sCe4/0096	Deafness and male infertility, CATSPER2 related	CATSPER2
sCe4/0097	Deafness, congenital with inner ear agenesis, microtia, and microdontia	FGF3
sCe4/0098	Deafness, nonsyndromic, sensorineural, mitochondrial	MT-RNR1
sCe4/0099	Fazio-Londe disease	SLC52A3
sCe4/0100	Hearing loss, MAP1A related	MAP1A
sCe4/0101	Hearing loss, MYH7B related	MYH7B
sCe4/0102	Heimler syndrome type 1	PEX1
sCe4/0103	Mitochondrial modifier of deafness	TRMU
sCe4/0104	Opticoacoustic nerve atrophy with dementia	TIMM8A
sCe4/0105	Otopaladigital syndrome type 1	FLNA
sCe4/0106	Otopaladigital syndrome type 2	FLNA
sCe4/0107	Pendred syndrome	SLC26A4
sCe4/0108	Pneumothorax, primary spontaneous	FLCN
sCe4/0109	Primary ciliary dyskinesia type 1	DNAI1
sCe4/0110	Primary ciliary dyskinesia type 10	DNAAF2
sCe4/0111	Primary ciliary dyskinesia type 11	RSPH4A
sCe4/0112	Primary ciliary dyskinesia type 12	RSPH9

sCe4/0113	Primary ciliary dyskinesia type 13	DNAAF1
sCe4/0114	Primary ciliary dyskinesia type 14	CCDC39
sCe4/0115	Primary ciliary dyskinesia type 15	CCDC40
sCe4/0116	Primary ciliary dyskinesia type 16	DNAL1
sCe4/0117	Primary ciliary dyskinesia type 17	CCDC103
sCe4/0118	Primary ciliary dyskinesia type 18	DNAAF5
sCe4/0119	Primary ciliary dyskinesia type 19	LRRC6
sCe4/0120	Primary ciliary dyskinesia type 2	DNAAF3
sCe4/0121	Primary ciliary dyskinesia type 20	CCDC114
sCe4/0122	Primary ciliary dyskinesia type 23	ARMC4
sCe4/0123	Primary ciliary dyskinesia type 24	RSPH1
sCe4/0124	Primary ciliary dyskinesia type 25	DYX1C1
sCe4/0125	Primary ciliary dyskinesia type 26	C21orf59
sCe4/0126	Primary ciliary dyskinesia type 27	CCDC65
sCe4/0127	Primary ciliary dyskinesia type 28	SPAG1
sCe4/0128	Primary ciliary dyskinesia type 29	CCNO
sCe4/0129	Primary ciliary dyskinesia type 3	DNAH5
sCe4/0130	Primary ciliary dyskinesia type 5	HYDIN
sCe4/0131	Primary ciliary dyskinesia type 6	NME8
sCe4/0132	Primary ciliary dyskinesia type 7	DNAH11
sCe4/0133	Primary ciliary dyskinesia type 9	DNAI2
sCe4/0134	Primary ciliary dyskinesia, DNAH9 related	DNAH9
sCe4/0135	Progressive hearing loss	P2RX2
sCe4/0136	Pulmonary fibrosis, idiopathic	SFTPA2
sCe4/0137	Sinoatrial node dysfunction and deafness	CACNA1D
sCe4/0138	Tietz albinism-deafness syndrome	MITF
sCe4/0139	Wolfram syndrome type 1	WFS1
sCe4/0140	Wolfram syndrome type 2	CISD2
sCe4/0141	Wolfram-like syndrome, autosomal dominant	WFS1

Panel

Code	product name	TAT-day
	Clinical Exome Whole exome Solo	
pCe4/0001	Alport syndrome panel	25
pCe4/0002	Ciliary (primary) dyskinesia panel	25
pCe4/0003	Deafness, non-syndromic sensorineural autosomal dominant panel	25
pCe4/0004	Deafness, non-syndromic sensorineural autosomal recessive panel	25