

Genliste Schwerhörigkeit-Panel (Fett markierte Gene sind in unterschiedlichen Standard-Panels (<25kb) enthalten; siehe Begutachtungsauftrag Panel-Diagnostik)

	Gensymbol	Genname	Erkrankung(en)	OMIM	Gengröße (kb)
1	ACTG1	<i>Actin Gamma-1</i>	DFNA20/26 Baraitser-Winter-Syndrom 2, AD	102560	1,1
2	<i>CDH23</i>	<i>Cadherin 23</i>	DFNB12 Usher-Syndrom 1D (AR), 1D/F (AR, digenisch)	605516	10,1
3	<i>CHD7</i>	<i>Chromodomain helicase DNA-binding protein 7</i>	CHARGE-Syndrom, AD	608892	9,0
4	<i>COCH</i>	<i>Cochlin</i>	DFNA9	603196	1,7
5	<i>COL11A2</i>	<i>Collagen type XI alpha-2</i>	DFNA13 DFNB53 Stickler-Syndrom Typ III, AD	120290	5,2
6	<i>COL4A5</i>	<i>Collagen type IV alpha-5</i>	Alport-Syndrom, X-linked dominant	303630	5,1
7	<i>DFNA5</i>	<i>DFNA5 gene</i>	DFNA5	608798	1,5
8	<i>DIAPH1</i>	<i>Diaphanous, Drosophila, Homolog of-1</i>	DFNA1	602121	3,8
9	EYA1	<i>Eyes absent-1</i>	Branchio-oto-(renales-)Syndrom 1, AD	601653	1,8
10	<i>FOXI1</i>	<i>Forkhead Box I1</i>	DFNB4 / Erweitertes vestibuläres Aquädukt (EVA), teilweise digenisch s. <i>SLC26A4</i> und <i>KCNJ10</i>	601093	2,7
11	<i>GJB2</i>	<i>Gap-junction protein beta-2</i>	DFNA3A DFNB1A, auch digenisch <i>GJB3</i> und <i>GJB6</i>	121011	0,7
12	<i>GJB3</i>	<i>Gap-junction protein beta-3</i>	DFNA2B DFNB1A, digenisch	603324	0,8
13	<i>GJB6</i>	<i>Gap-junction protein beta-6</i>	DFNA3B DFNB1B DFNB1A, digenisch	604418	0,8
14	<i>GPR98 (ADGRV1)</i>	G Protein-coupled receptor 98	Usher-Syndrom 2C, AR auch digenisch	602851	18,9
15	KCNQ4	<i>Potassium channel, voltage-gated, KQT-like subfamily, member 4</i>	DFNA2A	603537	2,1

	Gensymbol	Genname	Erkrankung(en)	OMIM	Gengröße (kb)
16	<i>MITF</i>	<i>Microphthalmia-associated transcription factor</i>	Waardenburg-Syndrom 2A, auch digenisch Tietz-Albinismus-Schwerhörigkeit-Syndrom, AD	156845	1,3
17	MYH14	<i>Myosin, heavy chain 14, non-muscle</i>	DFNA4A	608568	6,0
18	<i>MYO6</i>	<i>Myosin VI</i>	DFNA22 DFNB37	600970	3,9
19	MYO7A	<i>Myosin VIIA</i>	DFNA11 DFNB2 Usher-Syndrom 1B, AR	276903	6,6
20	MYO15A	<i>Myosin XVA</i>	DFNB3	602666	10,6
21	<i>OTOA</i>	<i>Otoancorin</i>	DFNB22	607038	3,4
22	OTOF	<i>Otoferlin</i>	DFNB9 / Auditorische Neuropathie	603681	6,0
23	PAX3	<i>Paired box gene 3</i>	Craniofacial-deafness-hand-syndrome, AD Waardenburg-Syndrom 1, AD Waardenburg-Syndrom 3, AD/AR	606597	1,4
24	<i>PCDH15</i>	<i>Protocadherin 15</i>	DFNB23 Usher-Syndrom 1F (AR), 1D/F (AR, digenisch)	605514	5,9
25	<i>PDZD7</i>	<i>PDZ domain-containing 7</i>	Usher-Syndrom 2C, AR digenisch	612971	3,1
26	<i>POU3F4</i>	<i>POU domain, class 3, transcription factor 4</i>	DFNX2	300039	1,1
27	<i>SIX5</i>	<i>Sine oculis homeobox, drosophila, homolog of 5</i>	Branchio-oto-renales-Syndrom 2, AD	600963	2,2
28	SLC26A4	<i>Solute carrier family 26, member 4</i>	Pendred-Syndrom, AR DFNB4	605646	2,3
29	STRC	<i>Stereocilin</i>	DFNB16	606440	5,3
30	<i>TECTA</i>	<i>Tectorin, alpha</i>	DFNA8/12 DFNB21	602574	6,5
31	<i>TMC1</i>	<i>Transmembrane channel-like protein 1</i>	DFNA36 DFNB7	606706	2,3

	Gensymbol	Gename	Erkrankung(en)	OMIM	Gengröße (kb)
32	<i>TMIE</i>	<i>Transmembrane inner ear-expressed gene</i>	DFNB6	607237	0,5
33	<i>TMPRSS3</i>	<i>Transmembrane protease, serine 3</i>	DFNB8/10	605511	1,4
34	<i>USH1C</i>	<i>USH1C gene</i>	DFNB18A Usher-Syndrom 1C, AR	605242	2,7
35	<i>USH1G</i>	<i>USH1G gene</i>	Usher-Syndrom 1G, AR	607696	1,4
36	<i>USH2A</i>	<i>USH2A gene</i>	Usher-Syndrom 2A, AR	608400	15,6
37	<i>WFS1</i>	<i>WFS1 gene</i>	DFNA6/14/38 Wolfram-Syndrom, AR <i>Wolfram-like syndrome, AD</i>	606201	2,7

DFNA ***Deafness autosomal dominant***

DFNB ***Deafness autosomal recessive***

DFNX ***Deafness X-chromosomal***