Diagnostic Yield of Targeted Hearing Loss Gene Panel Sequencing
in a Large German Cohort with a Balanced Age Distribution
from a Single Diagnostic Center: An Eight-Year Study

Supplemental Digital Content 6.
ClinGen Hearing Loss Gene Curation Expert Panel (HL GCEP) classifications for all 35 genes that contributed to the diagnostic yield

Gene	*n*	Disease association	Inheritance pattern	Genetic evidence	Experimental evidence	Total points	Final interpretation	Date of finalization
GJB2	19	Nonsyndromic hearing loss	AR	12	6	18	Definitive	02.03.17
MYO15A	4	Nonsyndromic hearing loss	AR	12	6	18	Definitive	21.11.17
COL11A1	4	Marshall and Stickler syndromes	AD				n.d.	
WFS1	4	Wolfram-like syndrome	AD	10.5	0.5	11	Definitive	17.04.18
SLC26A4	4	Pendred syndrome	AR	12	6	18	Definitive	07.06.17
TMPRSS3	3	Nonsyndromic hearing loss	AR	12	4.5	16.5	Definitive	22.08.17
LOXHD1	3	Nonsyndromic hearing loss	AR	12	2.5	14.5	Definitive	08.05.18
COL4A3	3	Alport syndrome	AR, AD				n.d.	
EDNRB	2	Waardenburg syndrome	AD	5	0	5	Limited	08.05.18
MYO6	2	Nonsyndromic hearing loss	AD	12	3	15	Definitive	20.02.18
USH2A	2	Usher syndrome type 2	AR	12	6	18	Definitive	15.02.17
MYO7A	1	Nonsyndromic hearing loss	AD	9.5	3.5	13	Definitive	19.03.18
TECTA	1	Nonsyndromic hearing loss	AR	12	6	18	Definitive	02.01.18

TECTA	1	Nonsyndromic hearing loss	AD	10.5	5.5	16	Definitive	02.01.18
ACTG1	1	Nonsyndromic hearing loss	AD	12	1	13	Definitive	07.01.19
CDH23	1	Nonsyndromic hearing loss	AR	9	4.5	13.5	Definitive	22.05.18
COCH	1	Nonsyndromic hearing loss	AD	10	3.5	13.5	Definitive	05.01.17
COL2A1	1	Stickler syndrome	AD				n.d.	
COL4A5	1	Alport syndrome	X-linked				n.d.	
COL9A2	1	Stickler syndrome	AR				n.d.	
DIAPH1	1	Hearing loss and macrothrombocytopenia	AD	10	6	16	Definitive	05.01.18
DIAPH3	1	Auditory neuropathy	AD	2	4.5	6.5	Moderate	29.03.17
EYA4	1	Nonsyndromic hearing loss	AD	12	2	14	Definitive	05.01.18
GATA3	1	Hypoparathyroidism, SNHL, and renal dysplasia	AD				n.d.	
KCNQ4	1	Nonsyndromic hearing loss	AD	12	5.5	17.5	Definitive	21.11.17
MANBA	1	Beta-mannosidosis	AR				n.d.	
MARVELD2	1	Nonsyndromic hearing loss	AR	12	5.5	17.5	Definitive	05.01.18
MITF	1	Waardenburg syndrome	AD	11,5	6	17.5	Definitive	21.11.17
OTOF	1	Auditory neuropathy	AR	12	5.5	17.5	Definitive	30.01.17
PAX3	1	Waardenburg syndrome	AD	12	6	18	Definitive	15.11.17
POU4F3	1	Nonsyndromic hearing loss	AD	12	6	18	Definitive	21.11.17
SMPX	1	Nonsyndromic hearing loss	X-linked	12	2	14	Definitive	12.09.17
SOX10	1	Waardenburg syndrome	AD	12	5	17	Definitive	19.06.17

STRC	1	Nonsyndromic hearing loss	AR	12	2.5	14.5	Definitive	19.12.17
TFAP2A	1	Branchiooculofacial syndrome	AD				n.d.	
TMIE	1	Nonsyndromic hearing loss	AR	12	6	18	Definitive	29.09.17

Abbreviations:
AR Autosomal recessive;
AD Autosomal dominant;
n.d. not done
SNHL Sensorineural hearing loss

According to and modified from DiStefano MT, Hemphill SE, Oza AM, Siegert RK, Grant AR, Hughes MY, Cushman BJ, Azaiez H, Booth KT, Chapin A, Duzkale H, Matsunaga T, Shen J, Zhang W, Kenna M, Schimmenti LA, Tekin M, Rehm HL, Tayoun ANA, Amr SS; ClinGen Hearing Loss Clinical Domain Working Group. ClinGen expert clinical validity curation of 164 hearing loss gene-disease pairs. Genet Med. 2019 Oct;21(10):2239-2247. doi: 10.1038/s41436-019-0487-0. Epub 2019 Mar 21. Erratum in: Genet Med. 2019 May 22;: PMID: 30894701; PMCID: PMC7280024.