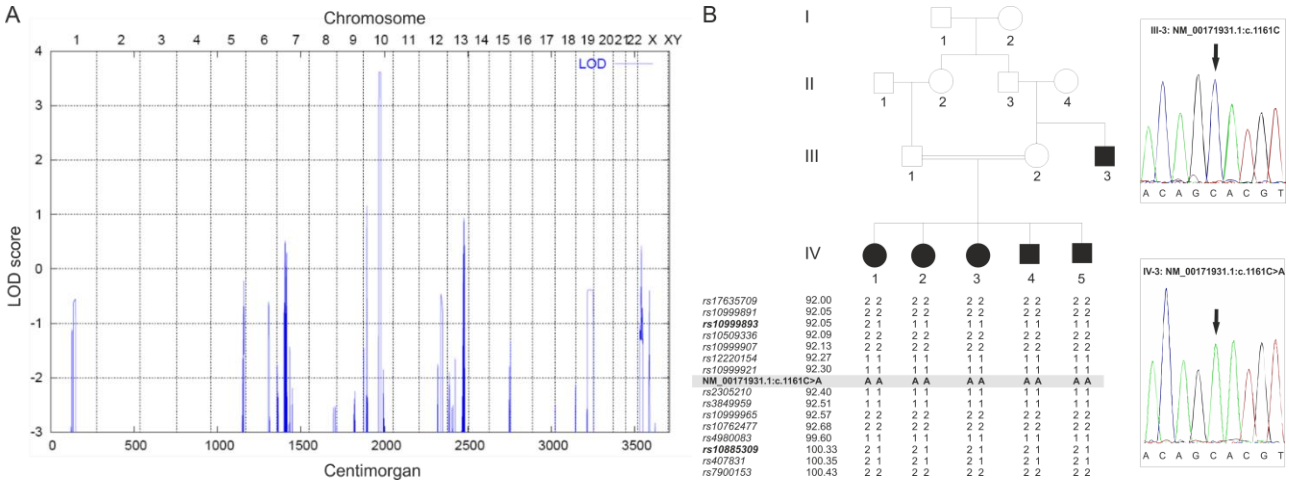
SUPPORTING INFORMATION

Supplementary figures and tables



C

384

H.sapiens	352	EYSVAITELAQVGFALPLFIQVVDKDENLGLN **S** MFEVYLVGNNSHHFIIS	401
P.troglodytes	393	EYSVAITELAQVGFALPLFIQVVDKDENLGLN **S** MFEVYLVGNNSHHFIIS	442
M.mulatta	539	EYSVAITELAQVGFALPLFIQVVDKDENLGLN **S** MFEVYLVGNNSHHFIIS	588
C.lupus	352	EYSVAITELAQVGFALPLFIQVVDKDENLGLN **S** MFEVYLVGNNSQHFIIS	401
B.taurus	352	EYSVAITELAQVGFALPLFIQVVDKDENLGLN **S** MFEVYLVGNNSHHFIIS	401
M.musculus	352	EYSVAITELAQVGFALPLFIQVVDKDENLGLN **S** MFEVYLVGNNSHHFIIS	401
R.norvegicus	352	EYSVAITELAQVGFALPLFIQVVDKDE--GLN **S** MFEVYLVGNNSHHFIIS	399
G.gallus	397	EYSVAIPELAQVGFALPLFIQVQDKDE--GPN **S** VFEVYLVGNNSNHFIIS	444
D.rerio	358	EYRVLITELAQVGFALPLYIQVEDKDE--GVN **S** MFQVFLTGNNSDHFSIS	405
X.tropicalis	353	EYTVSIPELAQLGFALPLYIQVQDRDEVSGLN **S** AFEVYLVGNNSNHFIIS	402

Figure S1 Molecular genetic analysis of family A12. **A** Genome-wide LOD-plot of family A12. For linkage analysis, 20,000 markers with an MAF of 0.15 and an intermarker distance of 100,000 bases were chosen from the GeneChip® Human Mapping 250K SNP Array data set and analyzed with the program ALLEGRO (1), assuming autosomal recessive inheritance, full penetrance, consanguinity, and a disease allele frequency of 0.0001. Chromosome numbers are given on the top of the plot, the genetic distance on the x-axis and the LOD values on the y-axis. **B** Pedigree of family A12 with haplotypes on chromosome 10 and results of the sequencing analysis of *CDH23*. All affected members of the fourth generation are homozygous for a region limited by the proximal and distal SNP markers rs1099893 and rs10885309, respectively (in bold). A novel homozygous missense variant in *CDH23*, NM_00171931.1:c.1161C>A, causing the amino acid change p.(Ser384Arg) was found by targeted sequencing of DNA from individual IV-3. Sanger sequencing of the DNA of all available family members revealed that all affected individuals of the fourth generation are homozygous for this variant, as indicated by the arrow in the representative chromatogram of individual IV-3. In contrast, the affected individual III-3 is homozygous for the wild-type allele. Since he displayed a different phenotype (unilateral total SNHL), a different cause of HL in this patient is likely. **C** Multiple alignment of CDH23 protein sequence around residue 384. The protein sequences originate from the following accession numbers, NP_071407.4 (*Homo sapiens*), XP_507839.3 (*Pan troglodytes*), XP_002805748.1 (*Macaca mulatta*), XP_003434519.1 (*Canis lupus*), NP_001178135.1 (*Bos taurus*), NP_075859.2 (*Mus musculus*), NP_446096.1 (*Rattus norvegicus*), XP_421595.3 (*Gallus gallus*), NP_999974.1 (*Danio rerio*), XP_002939565.2 (*Xenopus tropicalis*).

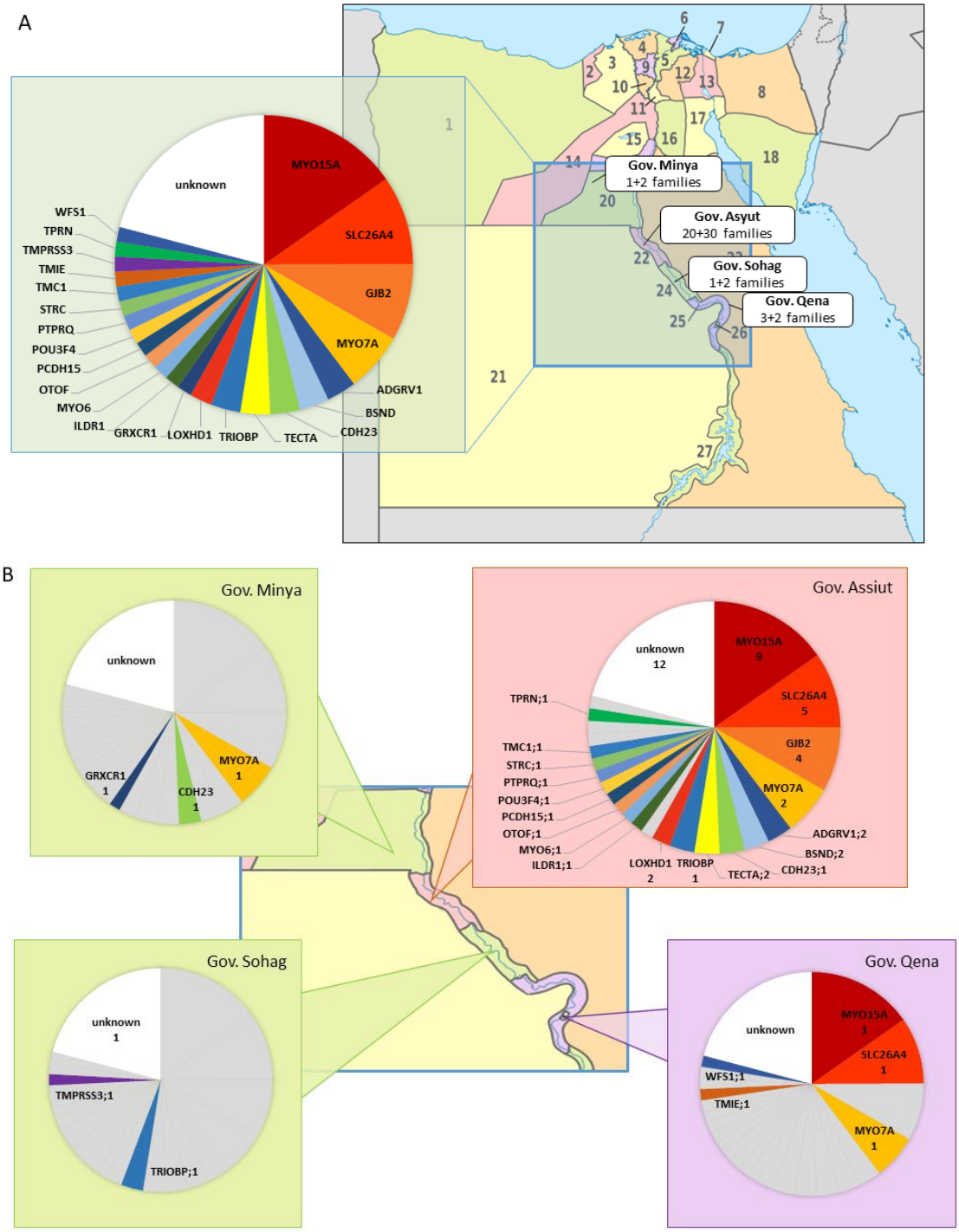


Figure S2 Geographic origin of deafness families and regional distribution of variants detected in HL associated genes. **A** Families originate from four different governorates (Gov.) of Egypt. The total numbers of families are given as a sum of families with A-IDs (first summand) and B-IDs (second summand). The pie chart illustrates the percentages of gene variants associated with SNHL summed up over all governorates (61 families in total). Numbers at the map refer to the following governorates: 1. Matrouh, 2. Alexandria, 3. Beheira, 4. Kafr El Sheikh, 5. Dakahlia, 6. Damietta, 7. Port Said, 8. North Sinai, 9. Gharbia, 10. Monufia, 11. Qalyubia, 12. Sharqia, 13. Ismailia, 14. Giza, 15. Faiyum, 16. Cairo, 17. Suez, 18. South Sinai, 19. Beni Suef, 20. Minya, 21. New Valley, 22. Asyut, 23. Red Sea, 24. Sohag, 25. Qena, 26. Luxor, 27. Aswan. The map was downloaded from https://commons.wikimedia.org/wiki/File:Egypt_-_Administrative_Divisions_-_Nmbrs_-_colored.png.
B Regional contribution to variants in HL associated genes. Genes with variants are shown in colour whereas genes without any variant in this particular governorate are turned to grey. Numbers next to the gene symbol indicate how many families from this governorate are carrying a variant in that gene.

Table S1. Gene lists for panel designs with RainDance and Agilent SureSelect

Gene panel	Enrichment method	Genes
GP1	RainDance	*CDH23, DIAPH1, MYO15A, MYO7A, OTOF, POU4F3, RNF135, SLC26A4, TCOF1, TECTA, TMC1, TMPRSS3*
GP2	SureSelect	*ACTB, ACTG1, ADGRV1, ATP6V1B1, BSND, CCDC50, CDH23, CEACAM16, CLDN14, CLRN1, COCH, COL11A2, CRYM, DFNA5, DFNB59, DIABLO, DIAPH1, DIAPH3, DSPP, ESPN, ESRRB, EYA1, EYA4, FGF3, FOXI1, GATA3, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, HGF, ILDR1, JAG1, KCNJ10, KCNQ4, LHFPL5, LHX3, LOXHD1, LRTOMT, MARVELD2, MIR182, MIR183, MIR96, MITF, MSRB3, MTRNR1, MTTS1, MYH14, MYH9, MYO15A, MYO1A, MYO3A, MYO6, MYO7A, OTOA, OTOF, OTOG, PAX3, PCDH15, PDZD7, PMP22, POU3F4, POU4F3, PRPS1, PTPRQ, RDX, SERPINB6, SIX1, SIX5, SLC17A8, SLC26A4, SLC26A5, SLC4A11, SMPX, SOX10, SOX2, STRC, TBL1X, TECTA, TIMM8A, TJP2, TMC1, TMIE, TMPRSS3, TPRN, TRIOBP, USH1C, USH1G, USH2A, WHRN, WFS1*

Table S2 Phenotypic and genotypic spectrum of families from Southern Egypt

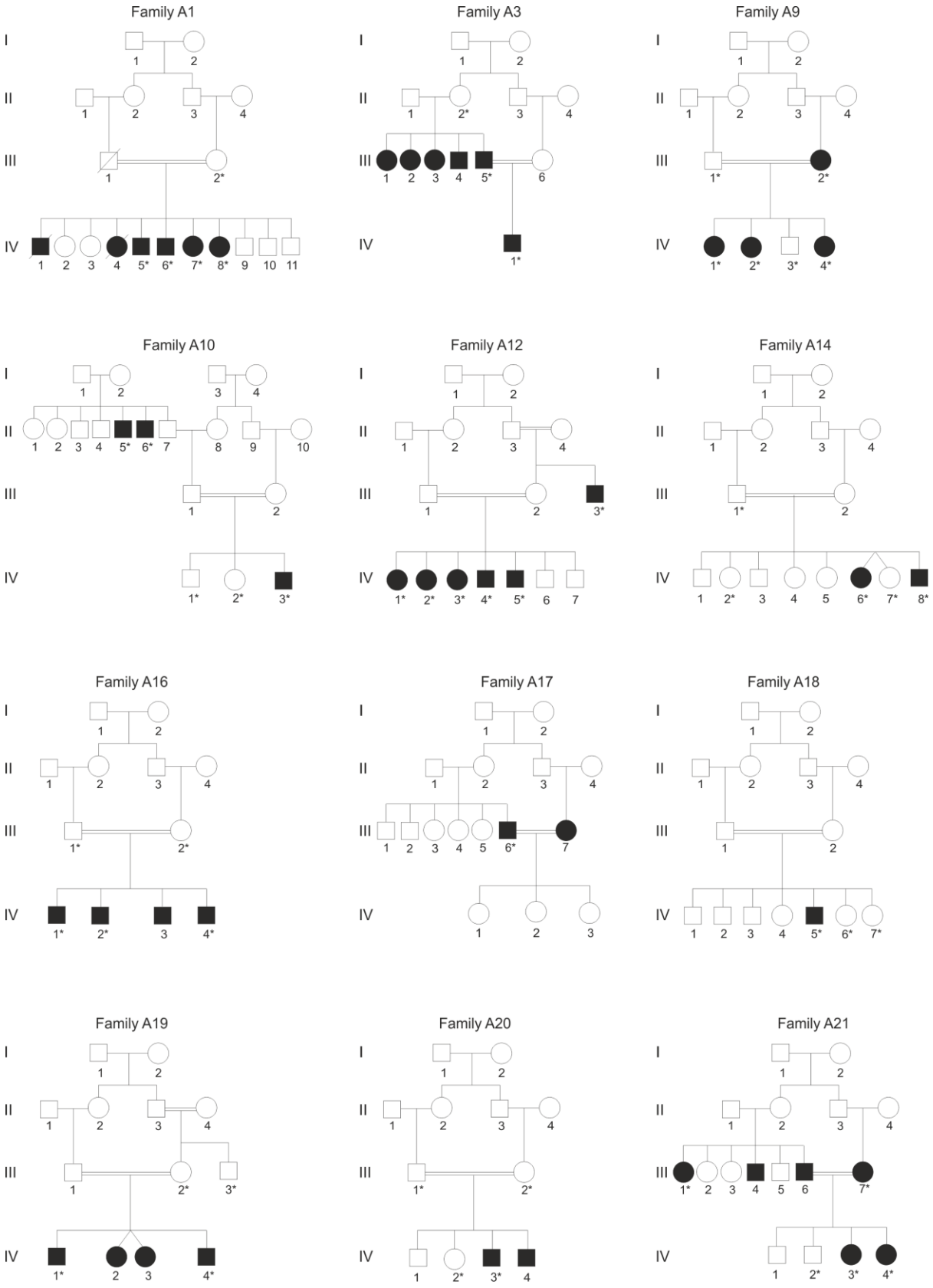
Family / Case No.	Pheno-type Right / left ear	MoI	Gene name	Genotype	Amino acid change	Predicted Variant Effect	GP	Ref.
A10/ II-5	pSNHL/ pSNHL	AR	*ADGRV1*	NM_032119.3:c.3508del NM_032119.3:c.8809_8816del	p.(Tyr1170Metfs*36) p.(Phe2937Glnfs*14)	Frameshift Frameshift	2	
B48/ IV-3	msSNHL/ msSNHL	AR	*ADGRV1*	NM_032119.3: c.6500A>G NM_032119.3: c.7839_7840del	p.(Tyr2167Cys) p.(Gly2615Glnfs*18)	Missense Frameshift	2	
A38/ IV-3	mSNHL/ pSNHL	AR	*BSND*	NM_057176.2:c.107C>A NM_057176.2:c.107C>A	p.(Thr36Asn) p.(Thr36Asn)	Missense Missense	2	
B91/ IV-5	spSNHL/ spSNHL	AR	*BSND*	NM_057176.2:c.107C>A NM_057176.2:c.107C>A	p.(Thr36Asn) p.(Thr36Asn)	Missense Missense	2	
A12/ IV-3	pSNHL/ pSNHL	AR	*CDH23*	NM_001171931.1:c.1152C>A NM_001171931.1:c.1152C>A	p.(Ser384Arg) p.(Ser384Arg)	Missense Missense	1	
B57/ IV-2	spSNHL/ spSNHL	AR	*CDH23*	NM_001171931.1:c.2595del NM_001171931.1:c.2595del	p.(Arg865Serfs*4) p.(Arg865Serfs*4)	Frameshift Frameshift	2	
B33/ IV-7	pSNHL/ pSNHL	AR	*GJB2*	NM_004004.5:c.35del NM_004004.5:c.35del	p.Gly12Valfs*2 p.Gly12Valfs*2	Frameshift Frameshift	2	(2), (3)
B78/ IV-7	spSNHL/ spSNHL	AR	*GJB2*	NM_004004.5:c.35del NM_004004.5:c.35del	p.Gly12Valfs*2 p.Gly12Valfs*2	Frameshift Frameshift	2	(2), (3)
B85/ IV-3	spSNHL/ spSNHL	AR	*GJB2*	NM_004004.5:c.35del NM_004004.5:c.35del	p.Gly12Valfs*2 p.Gly12Valfs*2	Frameshift Frameshift	2	(2), (3)
B97/ IV-4	ntSNHL/ ntSNHL	AR	*GJB2*	NM_004004.5:c.35del NM_004004.5:c.35del	p.Gly12Valfs*2 p.Gly12Valfs*2	Frameshift Frameshift	2	(2), (3)
B75/ IV-8	spSNHL/ spSNHL	AR	*GRXCR1*	NM_001080476.2:c.568C>T NM_001080476.2:c.568C>T	p.(Arg190*) p.(Arg190*)	Stop gained Stop gained	2	(4)
A28/ IV-3	pSNHL/ pSNHL	AR	*ILDR1*	NM_175924.3:c.357_361del NM_175924.3:c.357_361del	p.(Arg120Aspfs*13) p.(Arg120Aspfs*13)	Frameshift Frameshift	2	
A9/ IV-1	pSNHL/ pSNHL	AR	*LOXHD1*	NG_016646.2(NM_144612.6):c.3350+1G>A NM_144612.6:c.3727C>T	 p.(Arg1243Trp)	Splice donor Missense	2	
B82/ IV-3	pSNHL/ msSNHL	AR	*LOXHD1*	NM_144612.6: c.4465G>C NM_144612.6: c.4465G>C	p.(Gly1489Arg) p.(Gly1489Arg)	Missense Missense	2	
B82/ IV-6	spSNHL/ spSNHL	AR	*MYO15A*	NM_016239.3:c.4310A>G NM_016239.3:c.4310A>G	p.(Tyr1437Cys) p.(Tyr1437Cys)	Missense Missense	2	(4)
A14/ IV-6	pSNHL/ pSNHL	AR	*MYO15A*	NM_016239.3:c.5330C>A NM_016239.3:c.5330C>A	p.(Ser1777*) p.(Ser1777*)	Stop gained Stop gained	1	
A36/ IV-4	pSNHL/ pSNHL	AR	*MYO15A*	NM_016239.3:c.6100C>T NM_016239.3:c.6100C>T	p.(Gln2034*) p.(Gln2034*)	Stop gained Stop gained	1	
A50/ IV-2	pSNHL/ pSNHL	AR	*MYO15A*	NM_016239.3:c.6340G>A NM_016239.3:c.6340G>A	p.(Val2114Met) p.(Val2114Met)	Missense Missense	1	(5)
B99/ IV-3	pSNHL/ spSNHL	AR	*MYO15A*	NM_016239.3:c.6340G>A NM_016239.3:c.6340G>A	p.(Val2114Met) p.(Val2114Met)	Missense missense	2	(5)
A40/ IV-3	pSNHL/ pSNHL	AR	*MYO15A*	NG_011634.2(NM_016239.3):c.8601+2T>G NG_011634.2(NM_016239.3):c.8601+2T>G	 	Splice donor Splice donor	1	(6)
B27/ IV-7	spSNHL/ spSNHL	AR	*MYO15A*	NG_011634.2(NM_016239.3):c.8601+2T>G NG_011634.2(NM_016239.3):c.8601+2T>G	 	Splice donor splice donor	2	(6)
B84/ IV-6	ntSNHL/ ntSNHL	AR	*MYO15A*	NG_011634.2(NM_016239.3):c.8601+2T>G NG_011634.2(NM_016239.3):c.8601+2T>G	 	Splice donor Splice donor	2	(6)
A20/ IV-3	pSNHL/ pSNHL	AR	*MYO15A*	NM_016239.3:c.8899dup NM_016239.3:c.8899dup	p.(Arg2967Profs*33) p.(Arg2967Profs*33)	Frameshift Frameshift	1	
A41/ IV-7	pSNHL/ pSNHL	AR	*MYO15A*	NM_016239.3:c.8899C>T NM_016239.3:c.8899C>T	p.(Arg2967*) p.(Arg2967*)	Stop gained Stop gained	1	
B92/ IV-4	ntSNHL/ ntSNHL	AR	*MYO6*	NM_004999.3:c.2302C>T NM_004999.3:c.2302C>T	p.(Gln768*) p.(Gln768*)	Stop gained Stop gained	2	
B89/ IV-4	spSNHL/ spSNHL	AR	*MYO7A*	NM_000260.3:c.3659C>T NM_000260.3:c.3659C>T NM_000260.3:c.5581C>T NM_000260.3:c.5581C>T	p.(Pro1220Leu) p.(Pro1220Leu) p.Arg1861* p.Arg1861*	Missense Missense Stop gained Stop gained	2 2	(7) (8), (9)
B902/ IV-6	spSNHL/ spSNHL	AR	*MYO7A*	NM_000260.3:c.3997C>T NM_000260.3:c.3997C>T	p.(Gln1333*) p.(Gln1333*)	Stop gained Stop gained	2	
A1/ IV-7	pSNHL/ pSNHL	AR	*MYO7A*	NM_000260.3:c.4111G>A/ NM_000260.3:c.4111G>A	p.(Val1371Met) p.(Val1371Met)	Missense Missense	1	
B79/ IV-1	pSNHL/ pSNHL	AR	*MYO7A*	NM_000260.3:c.5501G>C NM_000260.3:c.5501G>C	p.(Trp1834Ser) p.(Trp1834Ser)	Missense Missense	2	
B901/ IV-4	spSNHL/ spSNHL	AR	*OTOF*	NM_194248.2:c.1492C>T NM_194248.2:c.1492C>T	p.(Gln498*) p.(Gln498*)	Stop gained Stop gained	2	

(continues)

Table S2 (continued)

Family / Case No.	Pheno-type Right / left ear	MoI	Gene name	Genotype	Amino acid change	Predicted Variant Effect	GP	Ref.
B104/ IV-5	spSNHL/ spSNHL	AR	*PCDH15*	NM_001142769.1:c.4542dup NM_001142769.1:c.4542dup	p.Pro1515Thrfs*4 p.Pro1515Thrfs*4	Frameshift Frameshift	2	(10)
B60/ III-7	spSNHL/ spSNHL	XL	*POU3F4*	NM_000307.4:c.346dup	p.Ala116Glyfs*77	Frameshift	2	
A16/ IV-1	sSNHL/ sSNHL	AR	*PTPRQ*	NG_034052.1(NM_001145026.1):c. 6193-2A>C NG_034052.1(NM_001145026.1):c. 6193-2A>C		Splice acceptor Splice acceptor	2	
B41/ IV-7	sSNHL/ sSNHL	AR	*SLC26A4*	NG_008489.1(NM_000441.1):c.164+1del NG_008489.1(NM_000441.1):c.164+1del		Splice donor Splice donor	2	(11), (12)
B94/ IV-7	spSNHL/ spSNHL	AR	*SLC26A4*	NM_000441.1:c.346G>A NM_000441.1:c.346G>A	p.(Gly116Ser) p.(Gly116Ser)	Missense Missense	2	(13)
B29/ IV-4	spSNHL/ spSNHL	AR	*SLC26A4*	NM_000441.1:c.691G>A NM_000441.1:c.691G>A	p.(Val231Met) p.(Val231Met)	Missense Missense	2	(14)
A542/ IV-2	pSNHL/ sSNHL	AR	*SLC26A4*	NM_000441.1:c.1198del NM_000441.1:c.1198del	p.(Cys400Valfs*32) p.(Cys400Valfs*32)	Frameshift Frameshift	2	(15)
B43/ IV-2	ntSNHL/ pSNHL	AR	*SLC26A4*	NM_000441.1:c.1198del NM_000441.1:c.1198del	p.(Cys400Valfs*32) p.(Cys400Valfs*32)	Frameshift Frameshift	2	(15)
B65/ IV-3	spSNHL/ spSNHL	AR	*SLC26A4*	NM_000441.1:c.1198del NM_000441.1:c.1198del	p.(Cys400Valfs*32) p.(Cys400Valfs*32)	Frameshift Frameshift	2	(15)
B103/ IV-4	mSNHL/ msSNHL	AR	*STRC*	NM_153700.2:c.3851T>A NM_153700.2:c.3851T>A	p.(Val1284Glu) p.(Val1284Glu)	Missense Missense	2	
A19/ IV-1	sSNHL/ sSNHL	AR	*TECTA*	NM_005422.2:c.5870_5884del NM_005422.2:c.5870_5884del	p.(Asp1957_Val1961del) p.(Asp1957_Val1961del)	In-frame deletion	1	
B23/ III-1	msSNHL/ msSNHL	AD	*TECTA*	NG_011633.1(NM_005422.2):c.[6156_6162+3del;6162+4A>G]		Deletion in splice region	2	
B54/ IV-6	spSNHL/ spSNHL	AR	*TMC1*	NM_138691.2:c.420delA NM_138691.2:c.420delA	p.(Lys140Asnfs*8) p.(Lys140Asnfs*8)	Frameshift Frameshift	2	
A29/ IV-1	sSNHL/ sSNHL	AR	*TMIE*	NM_147196.2:c.247C>T NM_147196.2:c.247C>T	p.(Pro83Ser) p.(Pro83Ser)	Missense Missense	2	
B105/ IV-4	spSNHL/ spSNHL	AR	*TMPRSS3*	NM_024022.2:c.1029G>C NM_024022.2:c.1029G>C	p.(Trp343Cys) p.(Trp343Cys)	Missense Missense	2	
A44/ IV-5	sSNHL/ sSNHL	AR	*TPRN*	NM_001128228.2:c.440_444dup NM_001128228.2:c.440_444dup	p.Arg149Alafs*303 p.Arg149Alafs*303	Frameshift Frameshift	2	
A21/ IV-4	pSNHL/ pSNHL	AR	*TRIOBP*	NM_001039141.2:c.1039C>T NM_001039141.2:c.1039C>T	p.(Arg347*) p.(Arg347*)	Stop gained Stop gained	2	(16)
A39/ IV-1	pSNHL/ pSNHL	AR	*TRIOBP*	NM_001039141.2:c.4984dup NM_001039141.2:c.4984dup	p.(Thr1662Asnfs*48) p.(Thr1662Asnfs*48)	Frameshift Frameshift	2	
B100/ IV-7	ntSNHL/ ntSNHL	AR	*WFS1*	NM_006005.3:c.972C>G NM_006005.3:c.972C>G	p.(Ile324Met) p.(Ile324Met)	Missense Missense	2	

Abbreviations: MoI – mode of inheritance, GP - gene panel (1 = RainDance enrichment; 2 = SureSelect enrichment), SNHL - sensorineural hearing loss, mSNHL – moderately SNHL, msSNHL – moderately to severe SNHL, ntSNHL – near total SNHL, pSNHL – profound SNHL, spSNHL – severe to profound SNHL, sSNHL - severe SNHL, AR – autosomal recessive, AD – autosomal dominant, XL – X linked.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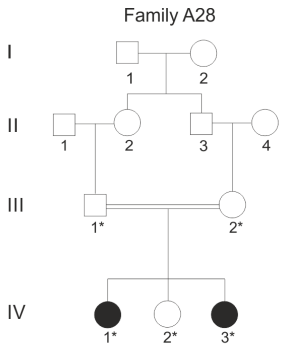
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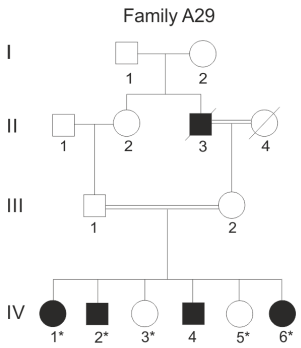
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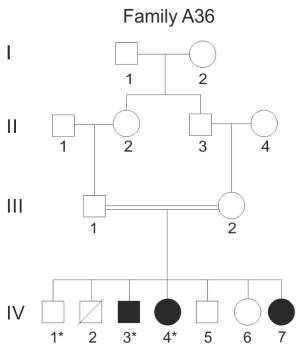
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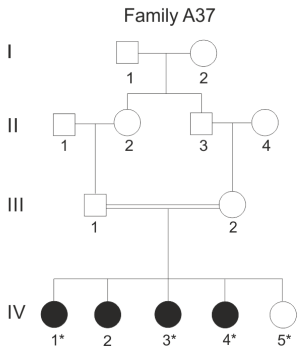
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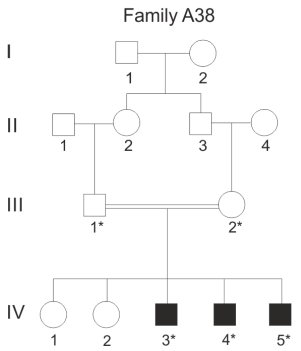
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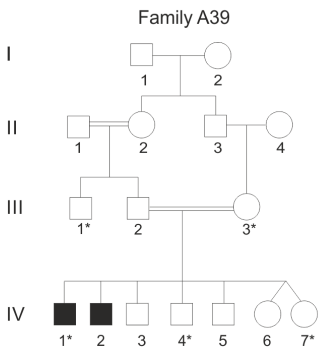
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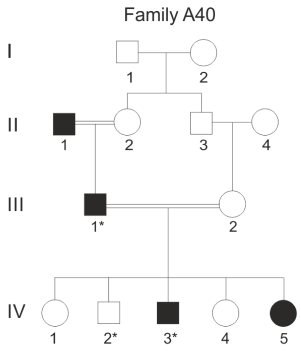
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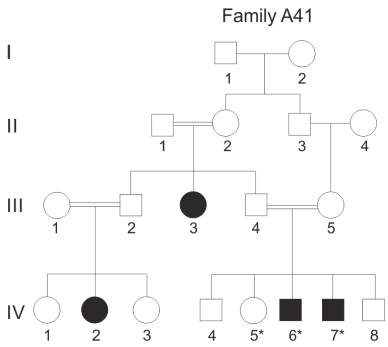
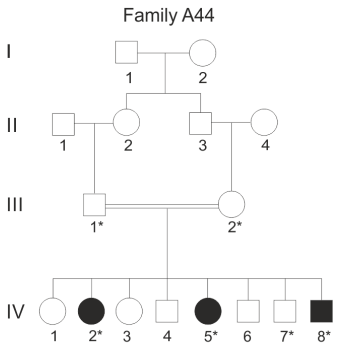
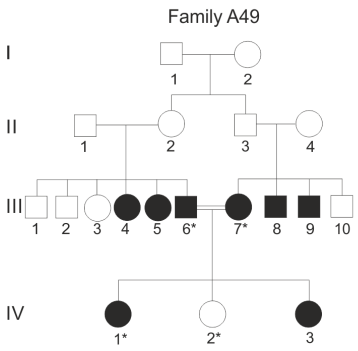
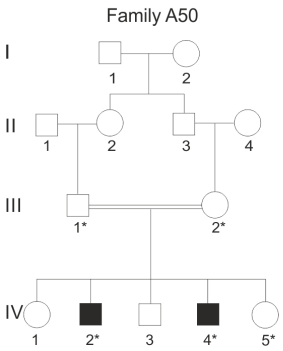
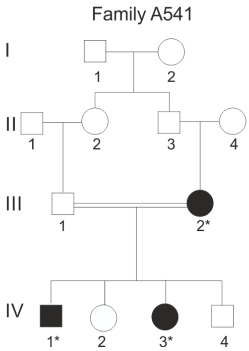
IV
1 2* 3* 4 5

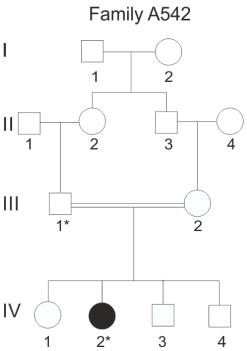

Family A41
I
1 2
II
1 2 3 4
III
1 2 3 4 5
IV
1 2 3 4 5* 6* 7* 8

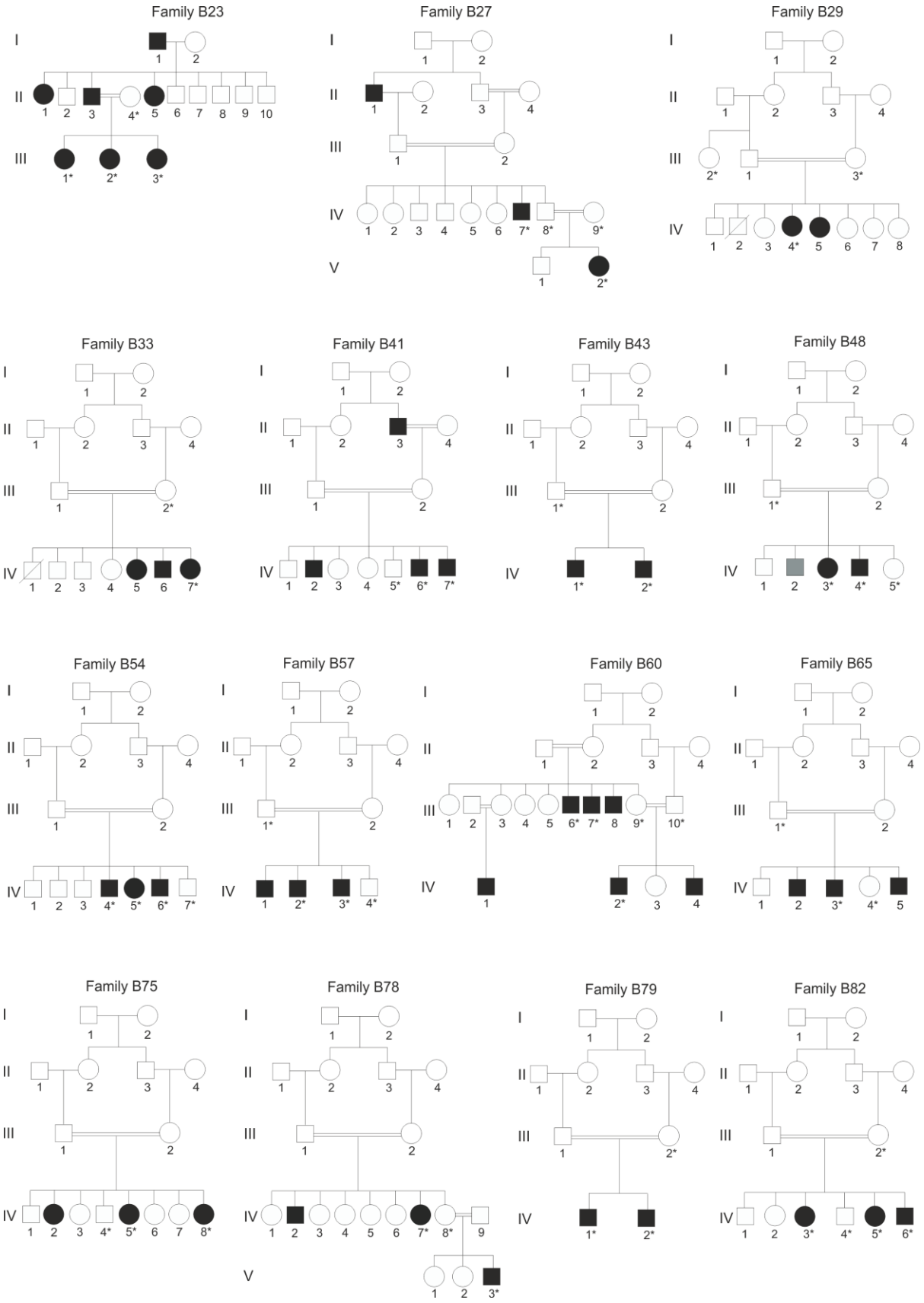

Family A44
I
1 2
II
1 2 3 4
III
1* 2*
IV
1 2* 3 4 5* 6 7* 8*


Family A49
I
1 2
II
1 2 3 4
III
1 2 3 4 5 6* 7* 8 9 10
IV
1* 2* 3


Family A50
I
1 2
II
1 2 3 4
III
1* 2*
IV
1 2* 3 4* 5*


Family A541
I
1 2
II
1 2 3 4
III
1 2*
IV
1* 2 3* 4


Family A542
I
1 2
II
1 2 3 4
III
1* 2
IV
1 2* 3 4


Family B23
Family B27
Family B29
Family B33
Family B41
Family B43
Family B48
Family B54
Family B57
Family B60
Family B65
Family B75
Family B78
Family B79
Family B82

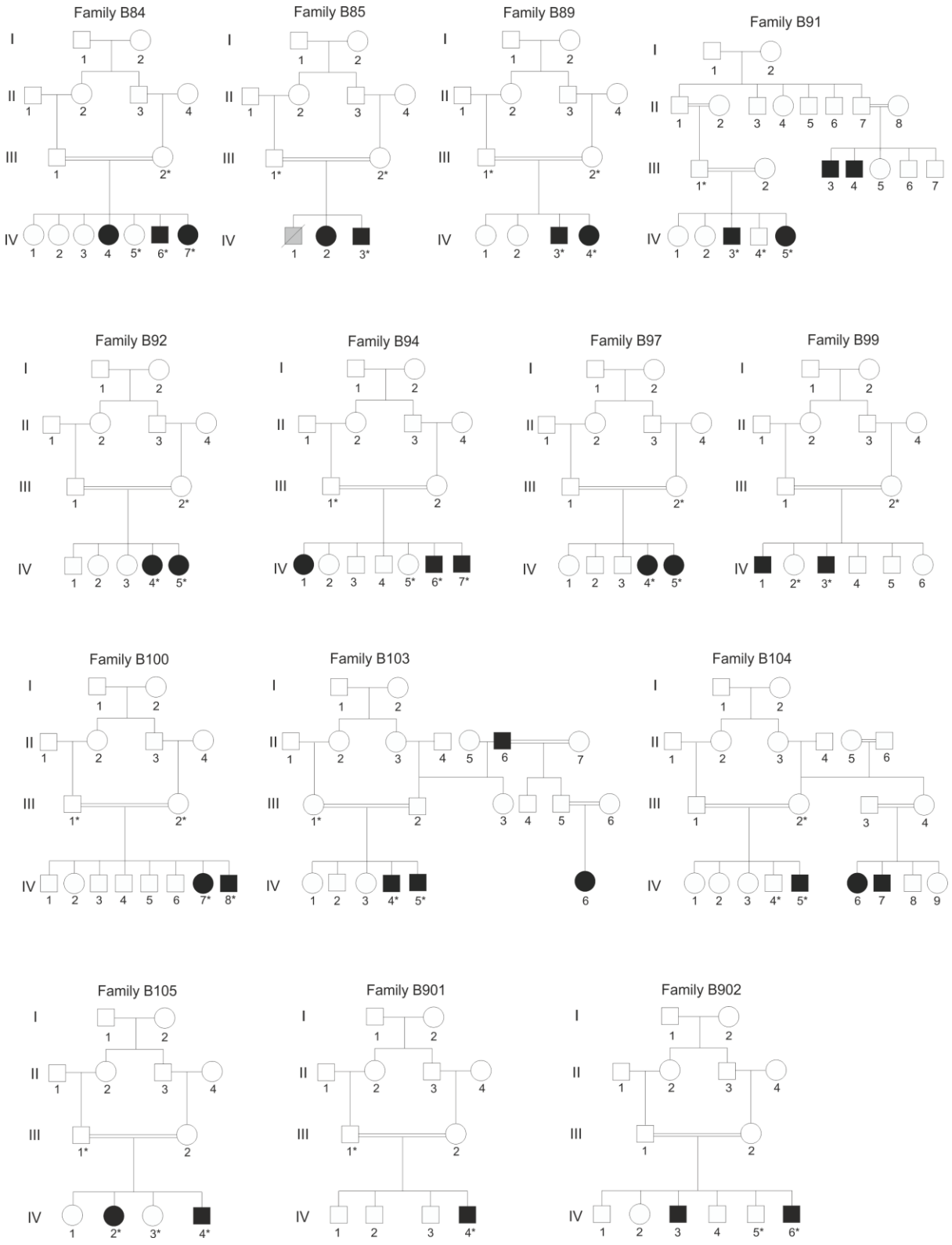


Figure S3 Pedigrees of Egyptian families with ARNSHL. Families with A-ID numbers belong to the first set of families. They were all prescreened for *GJB2* variants and included into genome-wide linkage analysis. Families with B-ID numbers belong to the second set which was not prescreened for *GJB2* variants. Of the 36 families representing the second set, only 29 families are shown. These were subjected to segregation analysis of candidate variants. Asterisks indicate availability of DNA samples. Grey symbols indicate that the phenotype is not known.

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