OtoSCOPE Version 5 Overview

AUTOSOMAL RECESSIVE GENES*				
Gene	**Loci**	**# of Exons[#]**	**# of BP**	
ATP2B2	-	25	9842	
CABP2	DFNB93	7	961	x
CATSPER2	-	13	2844	x
CDH23	DFNB12/USH1D	69	14138	
CIB2	DFNB48/USH1J	6	1,953	X
CLDN14	DFNB29	3	2335	x
COL11A2	DFNB53/DFNA13	66	6696	
CRYL1	DFNB1	8	840	x
ESPN	DFNB36	13	3606	X
ESSRB	DFNB35	12	885	X
GIPC3	DFNB15/DFNB95	6	1212	x
GJB2	DFNB1/DFNA3	2	2331	x
GJB3	DFNA2	2	2352	x
GJB6	DFNB1/DFNA3	5	2514	x
GPSM2	DFNB82	16	3310	x
GRXCR1	DFNB25	4	991	x
HGF	DFNB39	18	4284	x
ILDR1	DFNB42	7	2659	
LHFPL5	DFNB66/67	4	2147	x
LOXHD1	DFNB77	42	8177	x
LRTOMT	DFNB63	6	5518	
MARVELD2	DFNB49	7	3183	
MYO3A	DFNB30	35	7678	
MYO6	DFNB37/DFNA22	35	8662	
MYO7A	DFNB2/DFNA11/USH1B	49	8907	
MYO15A	DFNB3	65	11869	
MSRB3	DFNB74	9	4666	
OTOA	DFNB22	28	113384	
OTOF	DFNB9	47	7449	
OTOG	DFNB18	22	4992	
OTOGL	DFNB84	58	8231	
PCDH15	DFNB23/USH1F	35	13861	
PJVK	DFNB59	7	1531	
PNPT1	DFNB70	28	4579	
PTPRQ	DFNB84	63	6972	x
RDX	DFNB24	14	5711	x
SERPINB6	DFNB91	8	7603	
SLC26A4	DFNB4/PDS	21	4928	x
SLC26A5	DFNB61	20	2689	x
STRC	DFNB16	29	50189	
TECTA	DFNB21/DFNA8/DFNA12	24	6468	
TMC1	DFNB7/DFNB11/DFNA36	24	3520	
TMIE	DFNB6	4	1861	
TMPRSS3	DFNB8/DFNB10	13	4461	
TPRN	DFNB79	4	3225	
TRIOBP	DFNB28	24	13023	
TSPEAR	DFNB98	13	4029	
USH1C	DFNB18/USH1C	27	3508	
WHRN	DFNB31/USH2D	12	4388	
Totals	**49**	**1014**	**17943**	

AUTOSOMAL DOMINANT GENES*			
Gene	**Loci**	**# of Exons[#]**	**# of BP**
ACTG1	DFNA20/26	6	2287
CCDC50	DFNA44	12	8948
CEACAM16	DFNA4	7	1692
COCH	DFNA9	12	3155
CRYM	-	9	1803
DIABLO	DFNA64	7	2348
DFNA5	DFNA5	10	3349
DIAPH1	DFNA1	28	6982
DSPP	DFNA39	5	4331
EYA4	DFNA10	20	7932
GRHL2	DFNA28	16	5863
KCNQ4	DFNA2	14	2335
MYH14	DFNA4	42	8630
MYH9	DFNA17	41	6431
MYO1A	DFNA48	28	3621
POU4F3	DFNA15	2	1182
SLC17A8	DFNA25	12	3983
TJP2	DFNA51	23	4611
WFS1	DFNA6/DFNA14	8	4140
P2RX2	DFNA41	10	1932
Totals	**20**	**312**	**85555**

MICRORNAS			
miR-96	-	1	78
miR-182	-	1	110
miR-183	-	1	110
Totals	**3**	**3**	**298**

MITOCHONDRIAL			
MTRNR1	-	1	967
MTTS1	-	1	71
Totals	**2**	**2**	**1038**

USHER SYNDROME GENES*			
CLRN1	USH3	4	3180
GPR98	USH2C	90	19682
PDZD7	USH2A modifier	18	5071
USH1G	USH1G	3	3561
USH2A	USH2A	72	20185
Totals	**5**	**187**	**51679**

PENDRED SYNDROME GENES*			
FOXI1	-	2	2296
KCNJ10	-	2	5306
Totals	**2**	**4**	**7602**

BOR SYNDROME GENES			
EYA1	BOR1	17	4878
SIX1	BOS3	2	2687
Totals	**2**	**17**	**4878**

X-LINKED GENES			
POU3F4	DFNX3	1	1507
PRPS1	DFNX2	7	2156
SMPX	DFNX4	6	1037
Totals	**3**	**14**	**4700**

X linked recessive auditory and peripheral neuropathy			
AIFM1	AUNX1	16	3253
Totals	**1**	**16**	**3253**

Alström syndrome			
ALMS1	ALMS1	23	13630
Totals	**1**	**23**	**13630**

Jervell and Lange Nielsen			
KCNQ1	JLNS1	16	3617
Totals	**1**	**16**	**3617**

Sinoatrial node disfunction and deafness			
CACNA1D	-	49	8304
Totals	**1**	**49**	**8304**

GRAND TOTALS	
Number of genes and microRNAs included	**90**
Number of exons targeted	**1,553**
Number of BP targeted	**202,497**
Number of unique capture regions	**14,455**

*Those genes that cause both ARNSHL and ADNHSL, Usher syndrome or Pendred Syndrome are included under ARNSHL

[#] Number of exons is based on isoform with greatest number of exons

8 overlap between AR and AD (COLL1A2, GJB2, GJB3, GJB6, MYO6, MYO7A, TECTA, TMC1)