

Usher syndrome – 20 Entries

Overzicht gemaakt door Cindy Boer

OMIN code: PS276900/ Human genome version GRCh38

Type	Location*	Chromosome	Basepair (bp) position	Gene size in bp	Protein size amino acids (AA)	Inheritance	Proof	Gene	Other names
Ushersyndrome									
USH2A	1q41	1	216, 173, 946-216, 423, 396	6320	1546	Recessive	Molecular biological evidence	USH2A	USH2A, RP39
USH3A	3q25.1	3	150, 926, 163-150, 972, 999	738	245	Recessive	Molecular biological evidence	CLRN1	CLRN1, USH3A, USH3, RP61
USH2C	5q14.3	5	90, 558, 797-91, 164, 437	19.557	6.306	Recessive	Molecular biological evidence	ADGRV1	ADGRV1, GPR98, MASS1, VLGR1, KIAA0686, FEB4, USH2C
USH3B	5q31.3	5	140, 673, 915-140, 691, 341	1.789	469	Recessive	Molecular biological evidence	HARS	HARS, USH3B, CMT2W
USH2D	9q32	9	114, 402, 080-114, 503, 173	2847	524	Recessive	Molecular biological evidence	WHRN	WHRN, CIP98, KIAA1526, DFNB31, USH2D
USH1K	10p11.21-q21	10	10:34, 200, 000-59, 400, 000	Locus	Locus	Recessive	Identified via linkage DNA region	USH1K	
USH1F	10q21.1	10	53, 821, 110-54, 801, 291	7.032	1.962	Recessive	Molecular biological evidence	PCDH15	PCDH15, DFNB23, USH1F
USH1D	10q22.1	10	71, 396, 962-71, 732, 723	4.814	1.381	Recessive	Molecular biological evidence	CDH23	CDH23, USH1D, DFNB12, PITA5
USH2C	10q24.31	10	101, 007, 683-101, 031, 129	4.108	1.033	Recessive	Molecular biological evidence	PDZD7	PDZD7, DFNB57

USH1C	11p15.1	11	17,493,900-17,544,400	2.159	533	Recessive	Molecular biological evidence	USH1C	USH1C, DFNB18A
USH1B	11q13.5	11	77,128,246-77,215,241	7.483	2215	Recessive	Molecular biological evidence	MYO7A	MYO7A, USH1B, DFNB2, DFNA11
USH1H	15q22-q23	15	58,800,000-72,400,000	Locus	Locus		Identified via linkage DNA region	USH1H	USH1H
USH1J	15q25.1	15	78,104,649-78,131,535	1.511	144	Recessive	Molecular biological evidence	CIB2	CIB2, KIP2
USH4	17q24.2	17	68,291,495-68,420,859	2.599	525	Recessive	Molecular biological evidence	ARSG	ARSG, KIAA1001, USH4
USH1G	17q25.1	17	74,916,083-74,923,255	3.558	461	Recessive	Molecular biological evidence	SANS	SANS, USH1G
USH1E	21q21	21	21:15,000,000-30,200,000	Locus	Locus	Recessive	Identified via linkage DNA region	USH1E	USH1E

- *outdated location indication, is only used by Clinical Genetics*

As soon as new mutations or genes are found, they will be updated on the OMIM website (Online Mendelian Inheritance in Man)

<https://www.omim.org/phenotypicSeries/PS276900>