Table S1: Pathogenic variants within *USH2A* and *ADGRV1* genes identified in Italian cohorts.

Data are aligned to the human genome reference build 37 (GRCh37). *USH2A* and *ADGRV1* gene variants were extracted considering a Minor Allele Frequency <0.05. Functional annotatation to determine the variant consequence was performed with bcftools' plug in "Split-VEP". Pathogenicity of the extracted variants was checked in HGMD®. Frequency of the extracted variants was checked in Non-Finnish Europeans in the GnomAD database. **A)** List of the extracted pathogenic variants within *USH2A* gene, **B)** List of the extracted pathogenic variants within *ADGRV1* gene.

Cohort = name of the considered Italian cohort (FVG = Friuli-Venezia Giulia, VBI = Val Borbera, CAR= Carlatino), Chr = chromosome, Pos = genomic position, rsID = unique identifier for a specific variant, Ref = reference allele, Alt = alternative allele, Freq = frequency of the reference allele, N carriers = number of pathogenic variant carriers, Freq gnomAD_NFE = frequency of the reference allele in Non-Finnish Europeans in the GnomAD database. NA= not available data. The values in brackets represent the total number of individuals for each cohort.

A)

Frequency of *USH2A* pathogenic variants in Italian cohorts

Cohort	Chr	Pos	rsID	Ref allele	Alt allele	Freq	Consequence	HGMD *in-silico* pathogenicity prediction	N carriers	Freq gnomAD_ NFE
FVG (n=663)	1	215628969	rs111033402	A	G	0.0015083	Missense	Damaging	2	0.0004836
	1	215680331	rs754979740	T	C	0.000754148	Missense	Damaging	1	0.00004418
	1	215728232	rs111033364	C	T	0.000754148	Stop gained	Damaging	1	0.0001939
	1	215728281	rs146264950	C	T	0.000754148	Missense	Damaging	1	0.0008272
	1	215879002	rs111033533	C	T	0.000754148	Missense	Damaging	1	0.0006937
	1	216000489	rs55958016	C	T	0.000754148	Stop gained	Damaging	1	0.00000000
	1	216078145	rs886039867	A	G	0.00226244	Missense	Damaging	3	NA
	1	216289285	rs146824138	C	T	0.0015083	Missense	Damaging	2	0.001109
	1	216325393	rs780308389	G	A	0.000754148	Missense	Damaging	1	0.00001763
	1	216422149	rs369806765	C	T	0.00678733	Missense	Damaging	9	0.000008816

VBI (n=424)	1	216198494	rs111033524	C	A	0.00117925	Missense	Damaging	1	0.0001412
	1	216247118	rs80338902	C	A	0.00117925	Missense	Damaging	1	0.001405
	1	216325540	rs371777049	C	T	0.00117925	Missense	Damaging	1	0.00006195
CAR (n=124)	1	215650687	rs727504867	G	A	0.00806452	Stop gained	Damaging	2	0.00000000
	1	216073264	rs111033409	C	T	0.00806452	Missense	Damaging	2	0.0001679
	1	216247118	rs80338902	C	A	0.016129	Missense	Damaging	4	0.001405

B)

Frequency of *ADGRV1* pathogenic variants in Italian cohorts

Cohort	Chr	Pos	rsID	Ref allele	Alt allele	Freq	Consequence	HGMD *in-silico* pathogenicity prediction	N carriers	Freq gnomAD_ NFE
FVG (n=663)	5	90627592	rs755371825	C	A	0.0015083	Missense	Damaging	2	0.00005314
	5	90684054	rs41308846	G	A	0.0015083	Missense	Damaging	20	0.007248
	5	91153342	rs41311625	T	G	0.000754148	Missense	Damaging	1	0.0004636
VBI (n=424)	5	90684054	rs111033524	G	A	0.00235849	Missense	Damaging	2	0.007248
	5	90705414	rs80338902	G	A	0.00117925	Missense	Damaging	1	0.00001779
	5	90763453	rs371777049	C	A	0.00117925	Missense	Damaging	1	0.00559
	5	90848725	rs201073459	G	A	0.00117925	Missense	Damaging	1	0.0001819
CAR (n=124)	5	90629222	rs61744480	A	C	0.00806452	Missense	Damaging	2	0.002695
	5	90684054	rs41308846	G	A	0.0120968	Missense	Damaging	1	0.007248