

Table S2. List of variants found in Discovery WES1 and tNGS studies for recruited patients. Previously described variants in F1 and F2 are also shown (N=76). No variants were found in the panel genes *KCNE2*, *KCNQ1* and *CCKBR2*. Minor allele frequency (MAF) is annotated from European population in Exome Variant server from Exome Sequencing Project (ESP) database and 1000 Genomes project. Sift and Polyphen scores were considered for damage prediction annotation (D: Damaging; PD: Provably Damaging; T: Tolerated)

Gene	position	cDNA	Protein	dbSNP	ESP MAF	1000 Genomes		SIFT	Polyphen	
						MAF	Total			
<i>SLC26A7</i>	91334295	c.643A>G	p.Ile215Val	rs16912250	0.03047	0.0206	4*	2	T	PD
	91363504	c.1454A>T	p.Glu485Val	rs140569478	0.0	0.0024	3	0	D	PD
	91343370	c.1045C>T	p.Leu349Phe	rs113085951	0.001	0.004	1	0	D	D
	91343371	c.1363C>T	p.Val455Met	ND	.	.	1	0	.	.
	91393793	c.1777-4C>T	Unknown frameshift	rs16912805	0.0579	0.0606	2	1	.	.
<i>SLC26A9</i>	205915098	c.2458G>T	p.Ala820Ser	rs34309781	0.004312	0.014	2	0	T	T
	205921757	c.1864G>C	p.Val622Leu	M0911223	0.0002271	0.001	1	0	D	PD
	205915034	c.2522G>A	p.Gly841Glu	ND	0	0	1	0	.	.
	205923608	c.1502A>G	p.Asn501Ser	rs148928330	0.0	0.0008	2	0	T	PD
	205931898	c.514G>A	p.Val172Met	rs146704092	0.00227	0.0036	4*	1	D	PD
	205915010	c.2546G>A	p.Arg849Gln	rs140760240	0.001135	0.0076	3*	0	T	T
	205914894	c.2662T>C	p.Ter888GlnxtTer2	ND	0	0	1	0	.	.
	205918866	c.2230G>A	p.Val744Met	rs3811428,CM128010	0.001628	0.0036	1	0	D	T
	205918853	c.2243A>G	p.His748Arg	rs16856462	0.0	0.0763	1	0	T	PD
	<i>SLC4A2</i>	151070273	c.1376C>T	p.Ala459Val	rs140685664	0.001163	0.001	3	0	T
151064649		c.341C>T	p.Pro114Leu	rs200121811,COSM3879367	0	0	1	0	D	PD
151064613		c.305C>T	p.Pro102Leu	rs146857685	0.0004	0.001	1	0	T	T
151064628		c.320G>A	p.Arg107Gln	rs145741704	0.0015	0.0013	1	0	T	D
<i>PTH1R</i>	46903510	c.1636G>A	p.Glu546Lys	rs77048718	0.004085	0.0086	5**	2	PD	T

	46895873	c.313+4C>T	Unknown frameshift	rs200207404	0.001047	0.0004	1	0	.	.
	46898372	c.544-6G>C	Unknown frameshift	rs764927778 rs749330849,C	0	0	1	0	.	.
	46903558	c.1684G>A	p.Gly562Arg	OSM730733		1		0	D	D
<i>ATP4A</i>	35560431	c.719C>A	p.Pro240His	rs139075511,C OSM3692657	0.002724	0.0042	3*	1	D	PD
	35558357	c.1500+5G>T	Unknown frameshift	rs145047297	0.022	0.0066	5	0	.	.
	35555680	c.2002C>T	p.Arg668Cys		0.0002	0	2	0	.	.
	35555799	c.1883C>A	p.Thr628Lys	ND	0	0	1	0	.	.
	35555558	c.2039A>T G	p.Gln680Leu A	rs61729956 c.2107 C>T	0.0004539	0.0016	2**	0	D	PD
				rs61744730,CO				0	.	.
<i>PTH2R</i>	208442379	c.427C>T	p.Arg143Cys	SM2157069	0.003404	0.0098	3	1	D	D
	208489046	c.1112delT	p.Val371Ala sTer54	rs760717672	0	0	1	0	.	.
	.	.	p.Ile194Met	rs143633380	0	0	1*	0	D	D
<i>SLC9A2</i>	102704665	c.1967G>A	p.Arg656Gln	rs115244050	0.002674	0.0004	1	0	PD	T
<i>KCNQ1</i>	2847914	c.1942G>A	p.Val648Ile	rs34150427	0.0002367	0.0074	1	0	T	T
	2583448	c.935C>T	p.Thr312Ile	rs120074182,C M960901				1	D	D
<i>SLC9A4</i>	102478928	c.346G>A	p.Gly116Ser	rs17027275	0.0001163	0.0088	1	0	D	PD

* Variants found in the Discovery WES1 study

** Variants found in F2 (Calvete et al. 2017)

*** Variant found F1 (Calvete et al. 2019)