Family	Index and family members	Exon	Nucleotide mutation	Allele state	Protein alteration	Frequency, Prediction, Conservation and Remark
1	**II.1**	7	c.2401G>T	Homo	p.(Glu801*)	NMD or truncated protein
	Unaff. father I.1	7	c.2401G>T	Het	p.(Glu801*)	
	Unaff. mother I.2	7	c.2401G>T	Het	p.(Glu801*)	
	Aff. brother II.2	7	c.2401G>T	Homo	p.(Glu801*)	
2	**II.4**	2	c.1691_1693delTCT	Het	p.(Phe564del)	Deletion localized in a transmembrane domain
		10	c.3291_3294delATCT	Het	p.(Val1099Glufs*31)	ExAC: ALL:0.00084% - SAS:0.0063%
						Longer protein
	Unaff. mother I.2	2	no			
		10	c.3291_3294delATCT	Het	p.(Val1099Glufs*31)	
	Unaff. sister II.3	2	no			
		10	c.3291_3294delATCT	Het	p.(Val1099Glufs*31)	
3	**IV.2**	9	c.2968A>C	Homo	p.(Ser990Arg)	Polyphen-2: probably damaging with a score of 1.000 (sensitivity: 0.00; specificity: 1.00)
	Unaff. mother III.1	9	c.2968A>C	Het	p.(Ser990Arg)	SIFT: Deleterious (score: 0.03, median: 3.33)
	Unaff. brother IV.1		no			MutationTaster: disease causing (p-value: 1)
						UCSC: highly conserved
						Substitution localized in a transmembrane domain Ser990 is homologous to Ser552 present in NCKX2 which appears to be critical for cation binding and transport