Appendix 2. Presumed non-pathogenic alterations found in *PCDH15*.

Nucleotide change	Amino acid change	Exon/intron	Allele frequency	SNP
Exonic variants				
c.55T>G	p.S19A	2	11/38	rs11004439
c.521A>G	p.N174S	6	1/38	NV
c.546A>G	p.G182G	6	3/38	NV
c.1138G>A	p.G380S	11	3/38	rs10825269
c.1263T>C	p.T421T	11	3/38	rs7921598
c.1304A>C	p.D435A	11	13/38	rs4935502
c.2786G>A	p.R929Q	21	15/38	rs2135720
c.3817C>A	p.R1273S	29	1/38	rs111033363
c.4581C>A	p.P1527P	33	7/38	NV
c.4832A>C*	p.E1611A*	35	3/38	NV
c.4961A>C**	p.Q1654P**	36	3/38	NV
Intronic variants				
c.92–52T>G	None	2	15/38	rs10825347
c.157+3A>G	None	3	3/38	rs41274636
c.319–31T>C	None	4	9/38	rs11594958
c.595–101G>A	None	6	2/38	NV
c.705+93C>T	None	7	27/38	rs857395
c.706–8C>T	None	7	27/38	rs10740579

c.875+56T>G	None	8	21/38	rs10763098
c.877–17C>T	None	8	4/38	NV
c.986–81C>T	None	9	21/38	—
c.1098+107G>A	None	10	21/38	rs10825279
c.1591–68G>A	None	13	1/38	rs41274632
c.1997+132T>G	None	16	1/38	NV
c.2220+47T>C	None	18	1/38	rs3812657
c.2751+37C>T	None	20	26/38	rs3812658
c.2751+43C>G	None	20	2/38	rs2660169
c.2752–75C>G	None	20	2/38	NV
c.2752–52A>G	None	20	1/38	NV
c.2868+69G>A	None	21	4/38	rs11003980
c.2868+106A>G	None	21	1/38	NV
c.2869–109T>C	None	21	29/38	rs2456699
c.3010–48G>A	None	22	24/38	rs2593107
c.3374–72A>G	None	25	29/38	rs2593124
c.3374–68T>G	None	25	29/38	rs10740559
c.3717+35T>C	None	27	16/38	rs10825135
c.3984–20C>T	None	29	5/38	rs7089209
c.4203–151C>A	None	30	25/38	NV
c.4203–146G>A	None	30	1/38	NV

NV: Variants not previously described. *: Nomenclature based on the protocadherin-15-CD2 (EU718480). **: Nomenclature based on the protocadherin-15-CD3 (EU718482). Variant c.986–81C>T has been previously described [11], but no SNP number has been assigned.