

Table S3. Forty functional Indels in CDH proband

GeneSymbol	Gene ID	Chromosome	Position	Ref	Alt	Novel	SIFT	Phast Cons	Mutation Taster	Endeavour		ToppGene Rank	Total Rank
										NMD Rank	Rank		
<i>AARS2</i>	57505	6	44269192	TC	T	TRUE	frameshift	0.932	FRAMESHIFT	1	53	43	96
<i>ACADVL</i>	37	17	7123231	G	GT	TRUE	utr-5	0.001	FRAMESHIFT	1	17	24	41
<i>ADAMTS18</i>	170692	16	77334227	T	TG	TRUE	frameshift	0.004	FRAMESHIFT	1	10	20	30
<i>AGAP3</i>	116988	7	150831565	C	CG	TRUE	frameshift	1	FRAMESHIFT	1	23	77	100
<i>ANLN</i>	54443	7	36445855	AG	A	TRUE	frameshift	0.022	FRAMESHIFT	1	83	82	165
<i>AQP7</i>	364	9	33395134	A	ACC	TRUE	frameshift	0.053	FRAMESHIFT	1	80	105	185
<i>BZRAP1</i>	9256	17	56386385	T	TTG	TRUE	frameshift	0.673	FRAMESHIFT	1	14	81	95
<i>C14orf21</i>	161424	14	24769875	AT	A	TRUE	frameshift	0.002	FRAMESHIFT	1	67	67	134
<i>C14orf21</i>	161424	14	24769878	GAA	G	TRUE	frameshift	0.002	FRAMESHIFT	1	67	67	134
<i>CPNE1</i>	8904	20	34215237	A	AC	TRUE	frameshift	1	FRAMESHIFT	1	29	86	115
<i>DIAPH1</i>	1729	5	140907260	AG	A	TRUE	frameshift	1	FRAMESHIFT	1	5	94	99
<i>DNAH5</i>	1767	5	13886234	AG	A	TRUE	frameshift	1	FRAMESHIFT	1	18	52	70
<i>FAM135B</i>	51059	8	139165269	A	AC	TRUE	frameshift	0	FRAMESHIFT	1	104	72	176
<i>FMN2</i>	56776	1	240370629	C	CG	TRUE	frameshift	1	FRAMESHIFT	1	58	21	79
<i>GPX7</i>	2882	1	53072454	T	TC	TRUE	frameshift	0.984	FRAMESHIFT	1	2	25	27
<i>HIVEP3</i>	59269	1	42045727	GCTGGT	G	TRUE	frameshift	1	FRAMESHIFT	1	56	65	121
					GAGGT								
<i>HIVEP3</i>	59269	1	42045767	G	GCCA	TRUE	frameshift	1	FRAMESHIFT	1	56	65	121
<i>HLA-DQB1</i>	3120	6	32632649	C	CT	TRUE	frameshift	0.001	FRAMESHIFT	1	72	15	87
<i>HLA-DRB1</i>	3123	6	32552056	AC	A	TRUE	frameshift	0	FRAMESHIFT	1	63	103	166
					GTGCT								
					TGGAG								
					CTCCA								
<i>LAPTM4B</i>	55353	8	98787930	G	GCAGC	TRUE	utr-5	0	FRAMESHIFT	1	73	28	101
<i>LRRC49</i>	54839	15	71276484	CA	C	TRUE	frameshift	0.946	FRAMESHIFT	1	48	62	110
<i>NAALADL2</i>	254827	3	175189446	AG	A	TRUE	frameshift	0.976	FRAMESHIFT	1	100	79	179
<i>NUP54</i>	53371	4	77065320	G	GT	TRUE	frameshift	1	FRAMESHIFT	1	36	96	132
<i>PDE4DIP</i>	9659	1	144923728	GT	G	TRUE	frameshift	1	FRAMESHIFT	1	68	6	74
<i>PDE5A</i>	8654	4	120528327	G	GA	TRUE	frameshift	0.015	FRAMESHIFT	1	57	30	87
<i>PKD1L2</i>	114780	16	81242148	GTT	G	rs150289691	frameshift	1	FRAMESHIFT	1	79	63	142
<i>PLD2</i>	5338	17	4718774	G	GA	TRUE	frameshift	1	FRAMESHIFT	1	27	12	39
<i>PTGR1</i>	22949	9	114355255	CT	C	TRUE	frameshift	0	FRAMESHIFT	1	54	16	70
<i>PTGR1</i>	22949	9	114355259	CA	C	TRUE	frameshift	0.003	FRAMESHIFT	1	54	16	70
<i>PTGR1</i>	22949	9	114355261	CT	C	TRUE	frameshift	0.573	FRAMESHIFT	1	54	16	70
<i>RALGPS2</i>	55103	1	178780467	TAGAG	T	TRUE	frameshift	1	FRAMESHIFT	1	90	88	178

<i>SIRPA</i>	140885	20	1895963	AAC	A	TRUE	frameshift	0.002	FRAMESHIFT	1	34	3 37
<i>SIRPB1</i>	10326	20	1559023	CCA	C	TRUE	frameshift	0.004	FRAMESHIFT	1	55	4 59
<i>SIRPB1</i>	10326	20	1559028	TC	T	TRUE	frameshift	0	FRAMESHIFT	1	55	4 59
<i>STAG2</i>	10735	X	123184970	GC	G	TRUE	frameshift	1	FRAMESHIFT	1	30	102 132
<i>SYT15</i>	83849	10	46965887	T	TG	rs112965082	splice-3	0.911	FRAMESHIFT	1	82	48 130
<i>TMPRSS5</i>	80975	11	113565278	G	GC	TRUE	frameshift	0.934	FRAMESHIFT	1	89	60 149
<i>UBAC1</i>	10422	9	138836942	CCT	C	TRUE	frameshift	0.978	FRAMESHIFT	1	91	70 161
<i>ULK4</i>	54986	3	41996136	T	TC	TRUE	frameshift	1	FRAMESHIFT	1	33	80 113
<i>ZMIZ2</i>	83637	7	44805118	G	GC	TRUE	frameshift	0.337	FRAMESHIFT	1	13	92 105