

**Supplementary Table 2. Information on the 22 insertion/deletions identified by whole-exome sequencing**

Chr	Position	Gene	Reference	Substitution	Indel type	Location	Result	Family ID	Linkage	No of UA carriers	No of families with variant	Filter	Removed from PedGenie Analysis
1	112305585	DDX20	TGA	-	deletion	exonic	nonframeshift	15	0	0	2	3	<85% MIP data
2	242163366	ANO7	CA	-	deletion	exonic	frameshift	6	0	0	1	3	
3	130735150	ASTE1	ATC	-	deletion	exonic	nonframeshift	17	0	0	1	1	
4	38774893	TLR10	A	-	deletion	exonic	frameshift	10	0	0	1	2	
4	83350511	HNRPDL	-	G	insertion	exonic	frameshift	14	0	0	1	3	
4	155254559	DCHS2	G	-	deletion	exonic	frameshift	1	0	0	1	2	
6	7373498	CAGE1	A	-	deletion	exonic	frameshift	4	0	0	1	2	
6	31380162	MICA	GCTG	-	deletion	exonic	frameshift	19	1	2	11	1	Artifact
6	32148912	AGER	TG	-	deletion	UTR3		2	0	0	1	3	Failed MIP design
6	32154993	PBX2	TCAG	-	deletion	intronic		11	1	0	2	1	Failed MIP design
6	138539186	PBOV1	-	G	insertion	exonic	frameshift	6	1	3	6	3	
7	1132826	GPER	CT	-	deletion	UTR3		9	0	0	2	2	Failed MIP design
8	98155443	PGCP	T	-	deletion	UTR3		18	0	0	2	2	Failed MIP design
11	3746407	NUP98	GCT	-	deletion	exonic	nonframeshift	8	0	0	1	1	
13	42259275	KIAA0564	TTTGGAGT	-	deletion	exonic	frameshift	7	0	0	1	1	<85% MIP data
13	43462439	EPSTI1	-	TCAGG	insertion	exonic	frameshift	7	0	1	5	1	
14	57672625	EXOC5	GG	-	deletion	UTR3		3	0	0	2	1	Failed MIP design
17	66252964	AMZ2	AGGTG	-	deletion	exonic	frameshift	19	0	0	2	3	<85% MIP data
18	21739568	CABYR	-	T	insertion	exonic	frameshift	4	0	0	1	2	
19	19790878	ZNF101	TA	-	deletion	exonic	frameshift	5	0	0	5	2	
19	46393875	MYPOP	TAG	-	deletion	UTR3		16	1	0	1	1	Failed MIP design
22	30517625	HORMAD2	-	A	insertion	exonic	frameshift	2	0	0	1	3	<85% MIP data