Supplementary Table 5: Predicted deleterious truncating mutations identified in 11 candidate genes

cDNA	Protein	Mutation type	Exon/intron	cases	controls
APEX1					
c.182_183delCA	p.T61fs	frameshift	Exon 3	0	1
c.457C>T	p.Q153*	nonsense	Exon 5	1	0
c.872dupT	p.L291fs	frameshift	Exon 5	2	2
All				**3**	**3**
APLF					
c.22C>T	p.Q8*	nonsense	Exon 1	0	1
c.64G>T	p.E22*	nonsense	Exon 1	0	1
c.157C>T	p.R53*	nonsense	Exon 2	0	1
c.168+5A>G	In frame 24 aa del	splicing	Intron2	0	1
c.202G>T	p.E68*	nonsense	Exon 3	1	0
c.293delT	p.F98fs	frameshift	Exon 3	1	0
c.342-1G>A	Stop at aa 117	splicing	Intron 3	1	0
c.688C>T	p.Q230*	nonsense	Exon 6	0	1
c.703C>T	p.Q235*	nonsense	Exon 6	0	1
c.1160+1G>C	Stop at aa 278	splicing	Intron 7	0	2
c.1287-2A>G	Stop at aa 435	splicing	Intron 8	4	2
c.1288A>T	p.K430*	nonsense	Exon 9	1	0
c.1333+2T>C	Stop at aa 435	splicing	Intron9	1	0
c.1477dupG	p.E493fs	frameshift	Exon 10	0	3
c.1492G>T	p.E498*	nonsense	Exon 10	0	3
c.1528delA	p.R510fs	frameshift	Exon 10	84	111
All				**93**	**127**
APTX					
c.133G>A	p.V45I / no start codon	splicing	Exon 3	1	0
c.229C>T	p.Q77*	nonsense	Exon 5	1	0
c.347delA	p.K116fs	frameshift	Exon 5	0	1
c.771-5A>G	Stop at aa 267	splicing	Intron 7	1	0
c.837G>A	p.W279*	nonsense	Exon 8	2	2
c.874+2T>C	Stop at aa 267	splicing	Intron 8	0	1
c.916C>T	p.R306*	nonsense	Exon 9	1	0
All				**6**	**4**
EME1					
c.1386-2A>G	Stop at aa 488	splicing	Intron 7	3	1
c.1393C>T	p.R465*	nonsense	Exon 8	0	1
c.1720C>T	p.Q574*	nonsense	Exon 9	0	1
c.1724delC	p.P575fs	frameshift	Exon 9	1	0
All				**4**	**3**
FANCL					
c.170G>A	p.W57*	nonsense	Exon 3	1	0
c.211C>T	p.Q71*	nonsense	Exon 3	0	1

c.335C>A	p.S112*	nonsense	Exon 5	0	1
c.378delT	p.L126fs	frameshift	Exon 6	1	0
c.707-2A>G	Stop at aa 274	splicing	Intron 9	1	0
c.1111_1114dupATTA	p.T372fs	frameshift	Exon 14	23	27
All				**26**	**29**
MAD2L2					
c.594G>T	p.K198N	splicing	Exon 8	1	0
All				**1**	**0**
PARP2					
c.44G>A	p.R15K / no start codon	splicing	Exon 1	1	0
c.364-6A>G	Stop at aa 196	splicing	Intron4	1	0
c.941+1G>A	Stop at aa 274	splicing	Intron9	0	1
c.991dupA	p.I331fs	frameshift	Exon 10	0	1
c.1049_1050delAG	p.E350fs	frameshift	Exon 11	0	2
c.1269-13G>A	Stop at aa 446	splicing	Intron12	1	0
c.1330C>T	p.R444*	nonsense	Exon 13	1	0
c.1369-2A>G	In frame 33 aa del	splicing	Intron 13	1	0
c.1480C>T	p.Q494*	nonsense	Exon 15	0	1
All				**5**	**5**
PARP3					
c.334-1G>T	In frame 63 aa del	splicing	Intron 3	0	1
c.523-1G>A	Stop at aa 176	splicing	Intron 4	0	3
c.656-2A>G	Stop at aa 234	splicing	Intron 5	1	0
c.970C>T	p.R324*	nonsense	Exon 7	1	0
c.1039C>T	p.Q347*	nonsense	Exon 8	6	8
c.1297+4A>G	Stop at aa 377	splicing	Intron 9	0	1
All				**8**	**13**
POLN					
c.133+5_133+8delGTAA	No start codon	splicing	Intron 1	0	2
c.213+1G>T	Stop at aa 47	splicing	Intron 2	0	1
c.628C>T	p.Q210*	nonsense	Exon 3	1	0
c.714G>A	p.Q238Q / In frame 168 aa del	splicing	Exon 3	1	0
c.767_768delAG	p.E256fs	frameshift	Exon 4	1	0
c.1179+1G>A	Stop at aa 346	splicing	Intron 6	3	2
c.1248+1G>T	In frame 23 aa del	splicing	Intron 7	1	1
c.1375-2A>G	In frame 28 aa del	splicing	Intron 9	1	0
c.1457A>T	p.E486V	splicing	Exon 10	1	1
c.1795G>T	p.E599*	nonsense	Exon 16	1	0
c.2065G>C	p.G689R / Stop at aa 724	splicing	Exon 18	1	0

c.2308+1G>A	In frame 37 aa del	splicing	Intron 20	1	1
c.2387+1G>A	Stop at aa 818	splicing	Intron 21	1	0
c.2455+1G>A	Stop at aa 849	splicing	Intron 22	0	1
c.2456-6C>G	Stop at aa 851	splicing	Intron 22	0	1
c.2509delC	p.Q837fs	frameshift	Exon 23	11	19
c.2643delT	p.P881fs	frameshift	Exon 24	5	1
All				**29**	**30**
RAD54L					
c.407+1G>A	Stop at aa 116	splicing	Intron5	1	0
c.678G>A	p.W226*	nonsense	Exon 7	0	1
c.748G>T	p.E250*	nonsense	Exon 7	0	1
c.767-2A>G	Stop at aa 263	splicing	Intron7	1	0
c.1093C>T	p.R365*	nonsense	Exon 10	1	1
c.1525C>T	p.R509*	nonsense	Exon 14	0	2
c.1610+1G>A	Stop at aa 515	splicing	Intron14	2	1
c.1897C>T	p.Q633*	nonsense	Exon 17	1	0
c.2033G>C	p.R678T / Stop at aa 640	splicing	Exon 17	1	0
c.2139G>A	p.W713*	nonsense	Exon 18	1	0
All				**8**	**6**
SMUG1					
c.7C>T	p.Q3*	nonsense	Exon 3	3	2
c.143delC	p.S48fs	frameshift	Exon 3	0	1
c.370C>T	p.R124*	nonsense	Exon 4	1	0
All				**4**	**3**

Protein indicates known protein changes and for splicing variants it shows that predictions of the position of the premature stop or whether the loss of the closest exon would be predicted to cause an in-frame deletion. For some missense changes that may also affect splicing missense change and effect of splicing is shown