

Table 3. Putative predispositions in familial breast cancer families and probands

Gene	Description	Position	Nucleotide	Amino acid	Type	PolyPhen2*		Cancer-affected member								Frequency
						Score	Prediction	1	2	3	4	6	7	8		
Family 1																
GPRIN1	G protein regulated inducer of neurite outgrowth 1	chr5:176026123	c.T713C	p.L238S	exonic	0.91	D	-	+	+	+	+	+	-	5	
PINK1	PTEN induced putative kinase 1	chr1:20972051	c.960-2A>G		splicing	NA	NA	-	-	+	+	-	-	-	2	
POLK	polymerase (DNA directed) kappa	chr5:74892737	c.A2219G	p.H740R	exonic	0.62	P	-	-	-	+	-	-	-	1	
Family 2																
KAT6B	K(lysine) acetyltransferase 6B	chr10:76789128	c.G4546T	p.D1516Y	exonic	0.95	D	-	+	+	+	+	+		5	
KAT6B	K(lysine) acetyltransferase 6B	chr10:76789311	c.C4729T	p.R1577C	exonic	0.96	D	-	+	+	+	+	+		5	
NOTCH2	Notch 2	chr1:120459167	c.C6178T	p.R2060C	exonic	0.99	D	-	-	+	-	-	+		2	
Family 3																
NANP	N-acetylneuraminic acid phosphatase	chr20:25596725	c.A583G	p.I195V	exonic	0.98	D	+	-	+	-	-			2	
PHKB	phosphorylase kinase, beta	chr16:47628126	c.1204+1G>T		splicing	NA	NA	-	+	-	+	-			2	
Proband																
1	JAKMIP3	Janus kinase and microtubule interacting protein 3	chr10:133955524	c.G1574C	p.G525A	exonic	1.00	D								
2	POLQ	polymerase (DNA directed), theta	chr3:121207798	c.A3980C	p.Q1327P	exonic	1.00	D								
3	DUX2	double homeobox 2	chr10:135494906			splicing	NA	NA								
4	UBE2L3	ubiquitin-conjugating enzyme E2L 3	chr22:21975938	c.G349A	p.E117K	exonic	0.96	D								
5	RAD23B	RAD23 homolog B (S. cerevisiae)	chr9:110087260	c.C1028T	p.P343L	exonic	0.99	D								
7	GATA3	GATA binding protein 3	chr10:8100630	c.C604T	p.R202C	exonic	0.92	D								
8	KAT6B	K(lysine) acetyltransferase 6B	chr10:76744854	c.G2390A	p.S797N	exonic	0.98	D								
9	LIG1	ligase I, DNA, ATP-dependent	chr19:48637322	c.G1525A	p.E509K	exonic	0.95	D								
10	LIG4	ligase IV, DNA, ATP-dependent	chr13:108862463	c.G1154A	p.R385K	exonic	1.00	D								
14	NOTCH2	Notch 2	chr1:120529603	c.G854A	p.R285H	exonic	1.00	D								
15	ABL1	c-abl oncogene 1, non-receptor tyrosine kinase	chr9:133729493	c.G122A	p.G41D	exonic	0.92	D								
16	TNK2	tyrosine kinase, non-receptor, 2	chr3:195596385	c.C1760T	p.P587L	exonic	1.00	D								
17	NFRKB	nuclear factor related to kappaB binding protein	chr11:129755398	c.G611A	p.R204H	exonic	1.00	D								
18	NFKBIZ	nuclear factor of kappa light polypeptide gene enhancer	chr3:101576029			splicing	NA	NA								
19	SMG1	SMG1 phosphatidylinositol 3-kinase-related kinase	chr16:18879624	c.C3083T	p.T1028M	exonic	0.99	D								
20	PRKCQ	protein kinase C, theta	chr10:6528042	c.G855C	p.Q285H	exonic	1.00	D								
21	ADRA2A	adrenoceptor alpha 2A	chr10:112838117	c.C363G	p.C121W	exonic	1.00	D								
22	PPFIA4	protein tyrosine phosphatase, receptor type	chr1:203025582	c.C668T	p.T223M	exonic	0.92	D								

D: Probably damaging (score: 0.909-1); P: Possibly damaging (score: 0.447 - 0.908)