

Table 1 - PALB2 variants identified in female participants of the kConFab resource

	Nucleotide change	Protein change	rs number	Frequency in cases (n = 747)	%	References
Exonic variants						
Nonsense	c.196C>T	p. Gln66*	rs180177083	1	0.1	Casadei <i>et al.</i> [32]; Wong <i>et al.</i> [33]
	c.3113G>A	p.Trp1038*	rs180177132	7	0.9	Rahman <i>et al.</i> [8]; Casadei <i>et al.</i> [32]; Wong <i>et al.</i> [33]
Frameshift	c.1948_1949insA	p. Glu650fs*13	-	1	0.1	-
	c.2982_2983insT	p. Ala995fs*15	rs180177127	1	0.1	Rahman <i>et al.</i> [8]; Bogdanova <i>et al.</i> [66]
Missense	c.90G>T	p.Lys30Asn	-	1	0.1	-
	c.94C>G	p.Leu32Val	rs151316635	1	0.1	-
	c.596A>G	p.Asp219Gly	rs45594034	1	0.1	Rahman <i>et al.</i> [8]; Hellebrand <i>et al.</i> [34]; Dansonka- Mieskowska <i>et al.</i> [59]
	c.956C>A	p.Ser319Tyr	-	1	0.1	-
	c.1010T>C	p.Leu337Ser	rs45494092	25	3.3	Rahman <i>et al.</i> [8]; Hellebrand <i>et al.</i> [34];
	c.1475G>T	p.Gly492Val	-	1	0.1	-
	c.1676A>G	p.Gln559Arg	rs152451	72	9.6	Rahman <i>et al.</i> [8]; Hellebrand <i>et al.</i> [34]; Garcia <i>et al.</i> [54]; Bogdanova <i>et al.</i> [66]
	c.2014G>C	p.Glu672Gln	rs45532440	51	6.8	Rahman <i>et al.</i> [8]; Hellebrand <i>et al.</i> [34]; Garcia <i>et al.</i> [54]; Bogdanova <i>et al.</i> [66].; Dansonka- Mieskowska <i>et al.</i> [59]

	c.2590C>T	p.Pro864Ser	rs45568339	1	0.1	Rahman <i>et al.</i> [8]; Hellebrand <i>et al.</i> [34]; Garcia <i>et al.</i> [54];
	c.2993G>A	p.Gly998glu	rs45551636	17	2.3	Rahman <i>et al.</i> [8]; Hellebrand <i>et al.</i> [34]; Garcia <i>et al.</i> [54]; Bogdanova <i>et al.</i> [66]
Synonymous	c.1431C>T	p.Thr477Thr	-	1	0.1	-
	c.1470C>T	p.Pro490Pro	rs45612837	1	0.1	Rahman <i>et al.</i> [8]; Bogdanova <i>et al.</i> [66]
	c.1572A>G	p.Ser524Ser	rs45472400	4	0.5	Rahman <i>et al.</i> [8]; Hellebrand <i>et al.</i> [34]; Garcia <i>et al.</i> [54]; Bogdanova <i>et al.</i> [66]
	c.1935G>A	p.Glu645Glu	rs141707455	1	0.1	Hellebrand <i>et al.</i> [34];
	c.2469C>A	p.Leu823Leu	-	1	0.1	-
	c.2823C>A	p.Ile941Ile	-	1	0.1	-
	c.3300T>G	p.Thr1100Thr	rs45516100	45	6	Rahman <i>et al.</i> [8]; Hellebrand <i>et al.</i> [34]; Erkko <i>et al.</i> [53]; Garcia <i>et al.</i> [54]; Bogdanova <i>et al.</i> [66] Dansonka- Mieskowska <i>et al.</i> [59]
	c.3321G>A	p.Leu1107Leu	-	1	0.1	-
Intronic variants	c.-47G>A	-	rs8053188	17	2.3	Hellebrand <i>et al.</i> [34]; Garcia <i>et al.</i> [54];
	c.212-58A>C	-	rs80291632	37	5	Garcia <i>et al.</i> [54]; Dansonka- Mieskowska <i>et al.</i> [59]
	c.1684+41_42insTGA	-	-	2	0.3	-
	c.2834+12C>T	-	-	1	0.1	-