

Table 3 - Family history of probands identified as *PALB2* mutation carriers and the histopathology features of *PALB2*-associated tumours

Mutation	Incidences of breast cancers		Individuals affected with breast cancer	Age at dx	Carrier status	Histological grade	Histological type (1°)	ER	PR	HER2	Types of cancers in family (age of dx)
a) <i>PALB2</i> c.3113G>A	Pedigree A	5	Proband	79, 81	+						Biliary tract (65) Cervix (51) Colon (60) Melanoma (56) - Oesophagus (94) Ovary (uk) Ovary (56) Stomach (77) Prostate (77) -
			Paternal niece	32	+						
			Paternal cousin	59	-						
			Paternal aunt	60							
	Pedigree B	6	Proband	63	+	3	IDC	pos	neg	pos	Bowel (50) Bowel (69) Colon (59) - Endometrium (63) Leukaemia (57) Leukaemia (76) Liver (67) + Lymph nodes (80) Pancreas (65)
			Paternal aunt	49, 52							
			Paternal cousin	54	-						
			Paternal cousin	62							
			Paternal grandmother	52							
	Pedigree C	6	Proband	48, 51	+	1	Tubular	pos	pos	neg	Bowel (45) Bowel (59) Cervix (65) Hematopoietic system (40) Lung (68) Melanoma (69) Prostate (74) Ovary (40) Ovary (55) Ovary (58) Unknown primary (77) Unknown primary (85)
			Sister	49	+	3	IDC	pos	neg		
			Paternal aunt	49							
			Maternal aunt	53							
			Maternal cousin	34							
Pedigree D	6	Proband	46	+	2	IDC*				Leukaemia (95)	

			Sister	47	+	1	IDC				Prostate (84)
			Paternal cousin	45	+						
			Paternal cousin	46							
			Paternal aunt	48							
			Paternal aunt	65							
	Pedigree E	7	Proband	54	+	3	IDC	neg	neg	pos	Cervical lymph nodes (77) Colon (70) + Lung (41) Lung (45) Prostate (74)
			Sister	48							
			Maternal cousin	41	+						
			Maternal cousin	37							
			Maternal cousin	41	+	2	Pleomorphic lobular	pos	pos		
			Maternal aunt	42, 63							
	Pedigree F	2	Proband	32, 35	+	3	IDC	pos	pos	pos	Ovary (56) - Prostate (68) Testis (43) -
	Pedigree G	3	Proband	40	+	2	Lobular classical				
			Sister	55, 57							
	Pedigree H	5	Proband	49	+	2	IDC	pos	neg	pos	
			Sister	61	+	2	IDC	pos	pos		
			Mother	68	-						
			Maternal grandmother	64, 67							
b) <i>PALB2</i> c.196C>T	Pedigree I	9	Proband	42	+	3	IDC	pos	pos		Hodgkin's disease (40) - Lung (40) Lung (52) Melanoma (uk) Stomach (uk) Prostate (66) + Prostate (70) Prostate (uk) Uterus (uk)
			Mother	60	-						
			Paternal cousin	42							
			Paternal cousin	36							
			Paternal great-grand mother	90							
			Paternal grand aunt	55, 66							
			Paternal grand aunt	31							
			Paternal grand aunt	73							
c) <i>PALB2</i> c.1948_1949insA	Pedigree J	8	Proband	42	+	2	Pleomorphic lobular	pos	pos	pos	Bone (42) Endometrium (30)

			Sister	50							Endometrium (43)
			Maternal cousin	47							Lung (46)
			Maternal cousin	50							Melanoma (58)
			Maternal aunt	60							Prostate (69)
			Maternal grand aunt	70							Stomach (63)
			Paternal cousin	42							Thyroid (54)
			Sister in law	52							
d) <i>PALB2</i> c.2982_2983insT	Pedigree K	5	Proband	45, 54	+	3	IDC				
			Sister	47	+	2-3	IDC	pos	neg	pos	Lymphoma (62)
			Maternal aunt	42, 44							Pancreas (81)
											Sarcoma (59)

Histopathology and carrier status information were presented where available. Dx: Diagnosis. ER: Estrogen receptor. PR: Progesterone receptor. HER2: Human epidermal growth factor-2. IDC: Invasive ductal carcinoma. Pos: Positive. Neg: Negative. +: Genotyped and identified to be a carrier of the respective *PALB2* mutation. -: Genotyped and found to not be a carrier of a *PALB2* mutation. Uk: unknown. *Secondary histological type is lobular carcinoma.