Supplementary Table S1. PCR and sequencing primers

Primers	Sequence 5′ > 3′	Position of primers [a/b]
Primers used for PTT analysis		
RPB_1F_PTT	[c] CTCAGCTGTGAGGAGAAGGA	225-244[b]
RPB_4R	AGAGGTTGCTTCCAGGCTAAGAC	1339-1361[b]
Primers used for PCR and DNA breakpoint analysis		
Deletion of exons 7 - 8		
PB2_i5FWD	AGTGGGTAATGCAGGCAGA	17017-17035[a]
PB2_i9REV	CAGGTTTCAGAGAAAGTTGGGTAA	23443-23466[a]
PB2_i6f_4	AATCTTTCATGGGCCGGGCA	19427-19446[a]
Deletion of exons 9 - 10		
PB2_i7FWD	ACCAAGCATAATTTTTGGCTGC	22228-22249[a]
PB2_i11REV	CTGCTTATGACTTACTGCTCTCAC	32407-32430[a]
PB2_i10r_4	CTGTTAGGTTCCTCTGACAAACAC	26698-26721[a]
Duplication of exons 9 - 11		
PB2_i10FWD	GTTTGTTGGAAGAATGTGATCAGC	32221-32241[a]
PB2_i9REV	TTACCCAACTTTCTCTGAAACCTG	23443-23466[a]
PB2_i11f_11	GGAGACTGAAACTTGAACTTC	36251-36271[a]
Primers used for cDNA analysis		
RPB_1f	AGCTGATCGCGCACTGAGG	61-79[b]
RPB_4R	AGAGGTTGCTTCCAGGCTAAGAC	1339-136[b]
RPB_4F	TCTCCCAGTGACACTCTTGATG	1269-1290[b]
RPB_5R	ACAGAGTCACAGTCACAGGTAG	2593-2614[b]
RPB_5F	TCGACAGTTCAGGCAGCCCAG	2527-2547[b]
RPB_13r	ACATCCAAGATCAGTGGTGCTACC	3909-3932[b]
Primers used for HRM analysis		
PB2_2f	GACTCCACCTTTCCACTTGC	8179-8198[a]
PB2_2r	GAGACAAAAACAGCCCCAGAAA	8378-8399[a]
PB2_3f	AGAAAACGTATTTCTGGGGCTG	8368-8389[a]
PB2_3r	CAATAGCCAAAATATACCTGGGAAATG	8547-8573[a]
PB2_4Bf	CCAGGAGGATTACCTATACAAAGAACA	10149-10175[a]
PB2_4Br	ACTGGTTCTGGAGAATCTGGAAG	10428-10450[a]
PB2_4Cf	GCAAAAATCCTGCTAGATCACCA	10363-10385[a]
PB2_4Cr	CTGCTACCTTTAGGAGGAATGTG	10602-10624[a]
PB2_4Df	AGGGCGACTACAGTTCCTTTA	10542-10562[a]
PB2_4Dr	CTGGTAAGTTATTGTAGGTGAGTTCA	10802-10827[a]
PB2_4Ff	TAGCCTGGAAGCAACCTCTC	10955-10573[a]
PB2_4Fr	TGAACTTGGTTGTCCTGTGC	11218-11237[a]
PB2_5Af	TGTTGGGTTTTGTTACTATTTTGTGAC	15845-15871[a]
PB2_5Ar	GACTCAGTTCCTCTGGAAAAATACA	16161-16185[a]
PB2_13f	TTTGGATATGTAATCTGAATTATATCTTCTTTG	42645-42677[a]
PB2_13r	AGGCCCAATATATCCAGAAAATTG	42920-42943[a]

NCBI reference sequence NG_007406.1 [a] or NM_024675.3 [b]

[c] the sequence 5'-GCTAATACGACTCACTATAGGAACAGACCACCATG-3'contains a T7 RNA polymerase promoter and an initiation codon.

Supplementary Table S2. Identified missense variants in *PALB2* gene not previously reported.

Exon	mRNA change[a]	Protein change	SIFT	PolyPhen	GVGD
3	c.193C>T	p.P65S	tolerated	benign	class C0
4	c.394G>A	p.V132I	tolerated	benign	class C0
4	c.1610C>T	p.S537L	tolerated	benign	class C0
9	c.3181T>C	p.F994S	damaging	probably damaging	class C0
12	c.3228T>A	p.H1076Q	tolerated	benign	class C0

[a]Position in mRNA and protein sequence is according to the NCBI reference sequences NM_024675.3 and NP_078951.2.

Supplementary Table S3. Frequency of mutations in the *BRCA1*, *BRCA2* and *PALB2* genes identified in our high-risk patients.

Group (N)	***BRCA1*** **mutated (%)**	***BRCA2*** **mutated (%)**	***PALB2*** **mutated (%)**	**Negatively tested (%)**
Familial cases (470)				
[a]HBC families (296)	41 (13.9)	20 (6.8)	13 (4.4)	222 (75.0)
[b]HBOC families (158)	57 (36.1)	11 (7.0)	0	90 (57.0)
[c]HOC families (16)	6 (37.5)	3 (18.8)	0	7 (43.8)
Non-familial cases (99)				
Male BC (24)	0	2 (8.3)	1 (4.2)	21 (87.5)
Bilateral BC, 1st at < 50 years (48)	8 (16.7)	1 (2.1)	1 (2.1)	38 (79.2)
Tumor duplicity, BC + OC (27)	6 (22.2)	3 (11.1)	1 (3.7)	17 (63.0)

[a]HBC, hereditary breast cancer; [b]HBOC, hereditary breast and ovarian cancer; [c]HOC, hereditary ovarian cancer.

Note: A *PALB2* mutation analysis was not performed in the carriers of *BRCA1* or *BRCA2* mutation.