

Supplementary Materials

Legends

Table S1. Description of germline pathogenic variants identified in the study.

Figure S1. Impact of the presence of pathogenic variants on the EFS in patients with early onset breast cancer.

Supplementary Table S1. Description of germline pathogenic variants identified in the study

Patient ID	Gene	DNA variation	Predicted Protein consequence
F3228	ATM	c.5979_5983del	p.(Ser1993ArgfsTer23)
F3068	ATM	c.8122G>A	p.(Asp2708Asn)
F124	BAP1	c.1203T>G	p.(Tyr401Ter)
F2394	BRCA1	g.(41267773_41276097)_(41278204_41282088)del	p.(?)
F1472	BRCA1	c.302-2A>G	p.?
F1238	BRCA1	c.1059G>A	p.(Trp353Ter)
F315	BRCA1	c.1504_1508del	p.(Leu502AlafsTer2)
F1235	BRCA1	c.1521_1531del	p.(Thr509SerfsTer3)
F130	BRCA1	c.1885A>T	p.(Arg629Ter)
F2999	BRCA1	c.2019del	p.(Glu673AspfsTer28)
F3300	BRCA1	c.2269del	p.(Val757PhefsTer8)
F796	BRCA1	c.2963C>A	p.(Ser988Ter)
F3961	BRCA1	c.3157dup	p.(Glu1053GlyfsTer7)
F483	BRCA1	c.3375_3376del	p.(Pro1126IlefsTer6)
F1144	BRCA1	c.3375_3376del	p.(Pro1126IlefsTer6)
F230	BRCA1	c.3607C>T	p.(Arg1203Ter)
F430a	BRCA1	c.3607C>T	p.(Arg1203Ter)
F1789	BRCA1	c.3607C>T	p.(Arg1203Ter)
F820	BRCA1	c.3624del	p.(Lys1208AsnfsTer1)
F558	BRCA1	c.3700_3704del	p.(Val1234GlnfsTer8)
F3597	BRCA1	c.3700_3704del	p.(Val1234GlnfsTer8)
F3448	BRCA1	c.4096+1G>A	p.Ala224_Leu1365del
F1675	BRCA1	c.4327C>T	p.(Arg1443Ter)
F2099	BRCA1	c.4327C>T	p.(Arg1443Ter)
F678	BRCA1	c.4576G>T	p.(Glu1526Ter)
F3790	BRCA1	c.5098A>C	p.(Thr1700Pro)
F470	BRCA1	c.5152+1G>A	p.Asp1692_Trp1718del
F276	BRCA1	c.5212G>A	p.(Gly1738Arg)
F707a	BRCA1	c.5212G>A	p.(Gly1738Arg)
F739	BRCA1	c.5212G>A	p.(Gly1738Arg)
F1539	BRCA1	c.5212G>A	p.(Gly1738Arg)
F1652	BRCA1	c.5212G>A	p.(Gly1738Arg)
F3658	BRCA1	c.5212G>A	p.(Gly1738Arg)
F3800	BRCA1	c.5212G>A	p.(Gly1738Arg)
F1420	BRCA1	c.5246C>G	p.(Pro1749Arg)
F298	BRCA1	c.5251C>T	p.(Arg1751Ter)
F1198	BRCA1	c.5251C>T	p.(Arg1751Ter)
F1687	BRCA1	c.5251C>T	p.(Arg1751Ter)
F2229a	BRCA1	c.5251C>T	p.(Arg1751Ter)
F2754	BRCA1	c.5251C>T	p.(Arg1751Ter)
F3450	BRCA1	c.5251C>T	p.(Arg1751Ter)
F582	BRCA1	c.5256_5277+3179del	p.Ala1752_Lys1759del
F3227	BRCA1	c.5256_5277+3179del	p.Ala1752_Lys1759del
F371	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)

F655	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)
F777	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)
F933	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)
F1792	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)
F1862	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)
F2590	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)
F2714	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)
F3373a	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)
F3432	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)
F3442	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)
F3705	BRCA1	c.5266dup	p.(Gln1756ProfsTer74)
F1343	BRCA1	c.5278-1G>T	p.Ile1760_Asp1778del
F062	BRCA1	c.5407-754_5592+8273del	p.(Gly1803_Tyr1863del)
F416	BRCA1	c.5407-754_5592+8273del	p.(Gly1803_Tyr1863del)
F686	BRCA1	c.5407-754_5592+8273del	p.(Gly1803_Tyr1863del)
F851	BRCA1	c.5407-754_5592+8273del	p.(Gly1803_Tyr1863del)
F976	BRCA1	c.5407-754_5592+8273del	p.(Gly1803_Tyr1863del)
F2740	BRCA1	c.5407-754_5592+8273del	p.(Gly1803_Tyr1863del)
F2887	BRCA1	c.5407-754_5592+8273del	p.(Gly1803_Tyr1863del)
F2911	BRCA1	c.5407-754_5592+8273del	p.(Gly1803_Tyr1863del)
F3222	BRCA1	c.5407-754_5592+8273del	p.(Gly1803_Tyr1863del)
F500	BRCA1	c.5431C>T	p.(Gln1811Ter)
F757	BRCA1	c.5431C>T	p.(Gln1811Ter)
F178	BRCA1	c.5467G>A	p.(Gly1803_Ala1813del)
F240	BRCA1	g.79398_83825del	p.0
F332	BRCA1	g.79398_83825del	p.0
F382	BRCA1	g.79398_83825del	p.0
F567	BRCA1	g.79398_83825del	p.0
F736	BRCA1	g.79398_83825del	p.0
F1059	BRCA1	g.79398_83825del	p.0
F782	BRCA1	c.5492del	p.(Pro1831Leufs)
F1460	BRCA1	c.5497G>A	p.(Val1833Met)
F3367	BRCA1	c.5497G>A	p.(Val1833Met)
F135	BRCA2	c.4409_4410del	p.(Ile1470LysfsTer11)
F967	BRCA2	c.4415_4418del	p.(Lys1472ThrfsTer6)
F2264	BRCA2	c.5645C>A	p.(Ser1882Ter)
F3508	BRCA2	c.6490C>T	p.(Gln2164Ter)
F903	BRCA2	c.6842-2675_7008-5558del	p.(Gly2281_Arg2336del)
F943a	BRCA2	c.6842-2675_7008-5558del	p.(Gly2281_Arg2336del)
F852	BRCA2	c.6941del	p.(Thr2314LysfsTer2)
F465	BRCA2	c.8993_9024del	p.(Ser2998IlefsTer9)
F2420	BRCA2	c.9246_9247insA	p.(Thr3085AsnfsTer26)
F3873	BRCA2	c.9018C>A	p.(Tyr3006Ter)
F160	BRCA2	c.9382 C>T	p.(Arg3128Ter)
F3252	BRIP1	c.984_990del	p.(Phe328LeufsTer8)
F1473	BRIP1	c.2947del	p.(Ile983LeufsTer2)
F1484	CHEK2	c.549G>C	p.(Leu183Phe)
F2839	CHEK2	c.1188del	p.(Val397PhefsTer17)

F3710a	<i>CHEK2</i>	c.592+3A>T	p.(Val148PhefsTer7)
F1487	<i>NBN</i>	c.2140C>T	p.(Arg714Ter)
F3449	<i>NBN</i>	c.2140C>T	p.(Arg714Ter)
F924	<i>PALB2</i>	c.1591_1600del	p.(Pro532AlafsTer26)
F975	<i>PTEN</i>	c.424del	p.(Arg142GlyfsTer5)
F2615	<i>RAD51C</i>	c.577C>T	p.(Arg193Ter)
F1686	<i>TP53</i>	g.(7579941_7590694)_(7590868_?)del	p.(?)
F920	<i>TP53</i>	c.324_327dup	p.(Arg110PhefsTer40)
F3685	<i>TP53</i>	c.329_330insGTTTCCG	p.(Leu111PhefsTer40)
F329	<i>TP53</i>	c.375G>A	p.(Ser33_Thr125del)
F1995	<i>TP53</i>	c.559+2T>C	p.?
F2533	<i>TP53</i>	c.560G>T	p.(Gly187Val)
F3364	<i>TP53</i>	c.570del	p.(Pro191LeufsTer56)
F1966	<i>TP53</i>	c.578A>T	p.(His193Leu)
F1212	<i>TP53</i>	c.637C>T	p.(Arg213Ter)
F3293	<i>TP53</i>	c.658T>C	p.(Tyr220His)
F747	<i>TP53</i>	c.743G>A	p.(Arg248Gln)
F1049	<i>TP53</i>	c.825T>A	p.(Cys275Ter)

Figure S1. Impact of the presence of pathogenic variants on the EFS in patients with early onset breast cancer.

