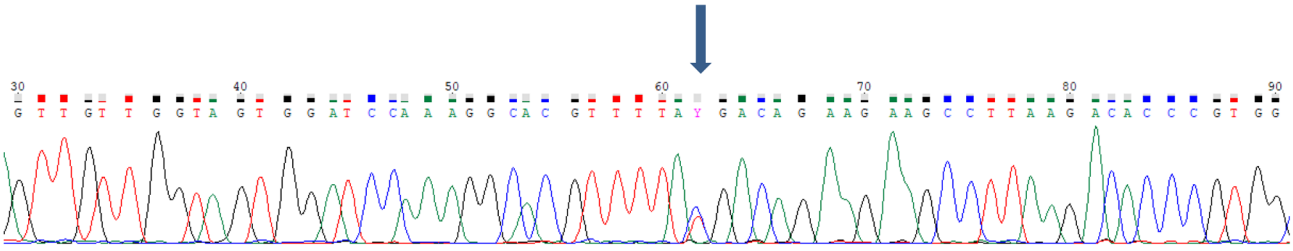
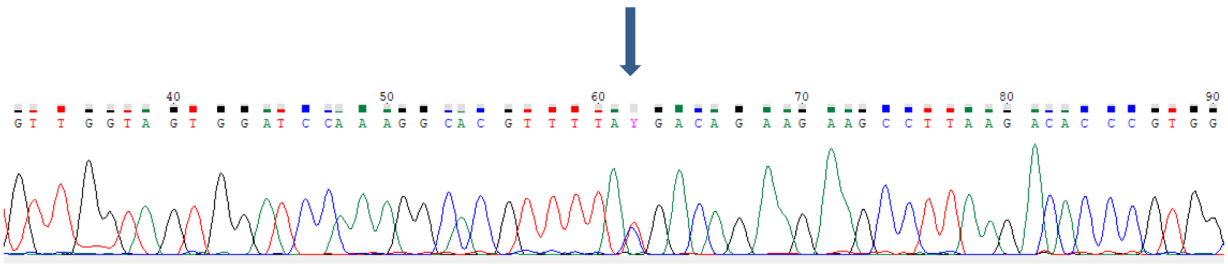
Figure S1. Variant distribution among patients.

Variant	Clinical Significance	Personal History of Cancer	Family History of Cancer	Consanguineous Marriage Status	File name Forward (F)	File name Reverse (R)	Patient
NM_007194(CHEK2):c.1427C>T, p.(Thr476Met)	Likely pathogenic	Breast	Breast, colon	Yes	GA3130_3150_CHEK2X13_F_2017-06-14_003.ab1	GA3130_3150_CHEK2X13_R_2017-06-14_004.ab1	P1
NM_007194(CHEK2):c.1427C>T, p.(Thr476Met)	Likely pathogenic	Breast	Breast, colorectal, kidney	Yes	GA3130_8452_CHEK2X13_F_2017-12-18_001.ab1	GA3130_8452_CHEK2X13_R_2017-12-18_002.ab1	P2
NM_001128425(MUTYH):c.1171C>T, p.(Gln391*)	Pathogenic (monoallelic)	Breast	Breast	Yes	GA3130_2823_MYHX11-12_F_2018-05-15_003.ab1	GA3130_2823_MYHX11-12_R_2018-05-15_004.ab1	P3
NM_007294(BRCA1):c.4035delA, p.(Glu1346Lysfs*20)	Pathogenic	Breast	Breast, lung	Yes	4591_1X11.9_F.ab1	4591_1X11.9_R.ab1	P4
NM_000059(BRCA2):c.9682delA, p.(Ser3228Valfs*21)	Pathogenic	Breast	Breast, ovarian	No	GA3130_2269_2X27.1_F_2017-05-19_003.ab1	GA3130_2269_2X27.1_R_2017-05-19_004.ab1	P5
NM_000059(BRCA2):c.8087T>A, p.(Leu2696*)	Pathogenic	Breast	Breast, prostate, lung, gastric	No	GA3130_4517_BR2X18_F_2017-07-21_003.ab1	GA3130_4517_BR2X18_R_2017-07-21_004.ab1	P6
NM_024675(PALB2):c.3271C>T, p.(Gln1091*)	Pathogenic	Breast	Breast	No	GA3130_7264 NEW_PALB2X12_F_2017-11-10_003.ab1	GA3130_7264 NEW_PALB2X12_R_2017-11-10_004.ab1	P7
NM_000059(BRCA2):c.5557dupT, p.(Cys1853Leufs*5)	Pathogenic	Breast	Breast	No	GA3130_9697_2X11K_F_2018-01-15_003.ab1	GA3130_9697_2X11K_R_2018-01-15_004.ab1	P8
NM_007294(BRCA1):c.5266dupC, p.(Gln1756Profs*74)	Pathogenic	Breast	Breast, gastric, skin, lung	No	GA3130_0784_1X20_F_2018-03-06_001.ab1	GA3130_0784_1X20_R_2018-03-06_002.ab1	P9
NM_058216(RAD51C):c.904+5G>T	Likely pathogenic	Breast, Ovarian	Breast, ovarian, prostate	No	GA3130_1175_RAD51CX6_F_2018-03-22_003.ab1	GA3130_1175_RAD51CX6_R_2018-03-22_004.ab1	P10
NM_000051(ATM):c.6527delT, p.(Leu2176Cysfs*59)	Pathogenic	Breast	Breast, lung, uterine	No	GA3130_1765_ATMX45_F_2018-04-24_003.ab1	GA3130_1765_ATMX45_R_2018-04-24_004.ab1	P11
NM_007294(BRCA1):c.3700_3704delGTAAA, p.(Val1234Glnfs*8)	Pathogenic	Breast	Breast, skin, melanoma, uterine	No	GA3130_2091_1X11.8_F_2018-04-23_003.ab1	GA3130_2091_1X11.8_R_2018-04-23_004.ab1	P12
NM_005732(RAD50):c.326_329delCAGA, p.(Thr109Asnfs*20)	Pathogenic				GA3130_2091_RAD50X3_F_2018-04-23_001.ab1	GA3130_2091_RAD50X3_R_2018-04-23_002.ab1	
NM_002485(NBN):c.657_661delACAAA, p.(Lys219Asnfs*16)	Pathogenic	Breast	lymphoma	No	4618_NBNX6_F.ab1	4618_NBNX6_R.ab1	P13
NM_007194(CHEK2):c.1427C>T, p.(Thr476Met)	Likely pathogenic	Breast	None	No	GA3130_2397_CHEK2X13_F_2018-04-26_003.ab1	GA3130_2397_CHEK2X13_R_2018-04-26_004.ab1	P14
NM_001128425(MUTYH):c.884C>T, p.(Pro295Leu)	Pathogenic (monoallelic)	None	Pancreatic	NA	GA3130_8355_MUTYHX10_F_2017-12-04_003.ab1	GA3130_8355_MUTYHX10_R_2017-12-04_004.ab1	P15
NM_000059(BRCA2):c.4936_4939delGAAA, p.Glu1646Glnfs*23	Pathogenic	None	Breast, ovarian	NA	GA3130_8940_2X11J_F_2017-12-13_003.ab1	GA3130_8940_2X11J_R_2017-12-13_004.ab1	P16
NM_000179(MSH6):c.2764C>T, p.(Arg922*)	Pathogenic	None	Breast, intestine	NA	GA3130_2683_6X4.5_F_2018-05-09_003.ab1	GA3130_2683_6X4.6_R_2018-05-09_004.ab1	P17
NM_007194(CHEK2):c.793-1G>A	Pathogenic	None	Breast, ovarian, lung	NA	5226_CHEK2X7_F.ab1	5226_CHEK2X7_R.ab1	P18

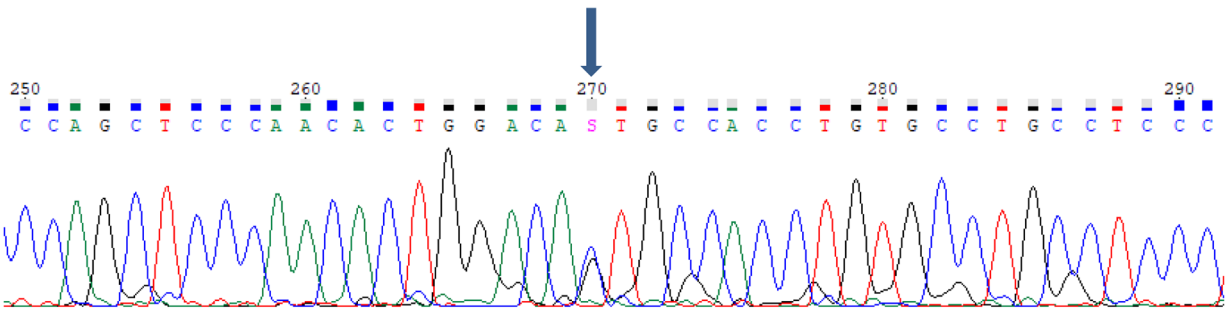
Figure S2. Sanger Sequencing analysis of genomic DNA from probands P1 and 2 carrying the variant CHEK2:c.1427C>T, p.(Thr476Met), P3 carrying the variant MUTYH:c.1171C>T, p.(Gln391*)and P4 carrying the variant BRCA1:c.4035delA, p.(Glu1346Lysfs*20).



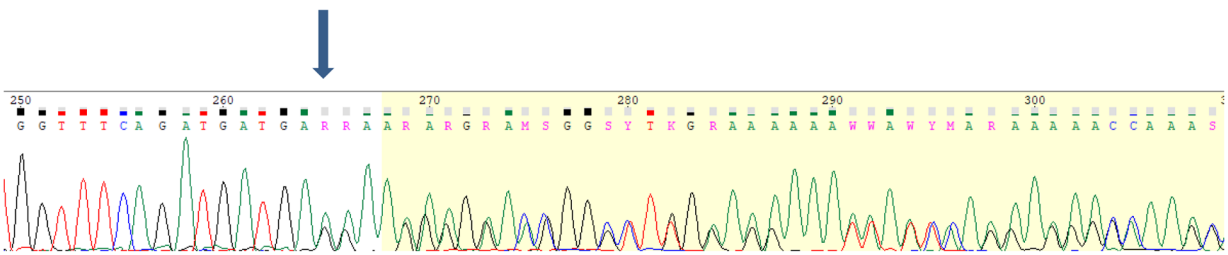
P1. CHEK2:c.1427C>T, p.(Thr476Met)



P2. CHEK2: c.1427C>T, p.(Thr476Met)

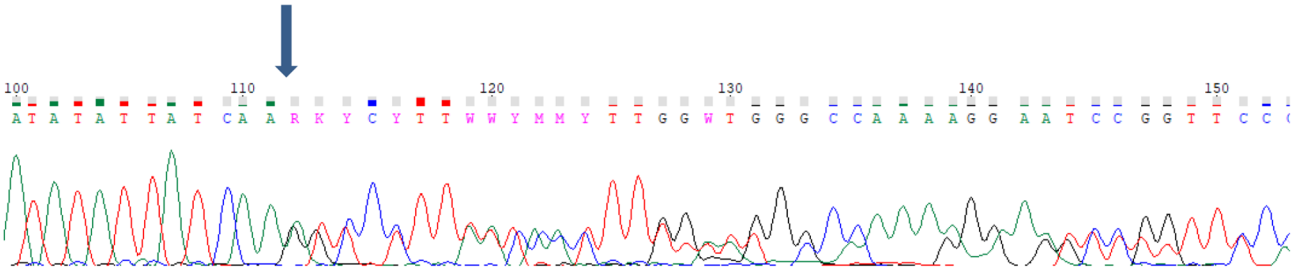


P3. MUTYH:c.1171C>T, p.(Gln391*)

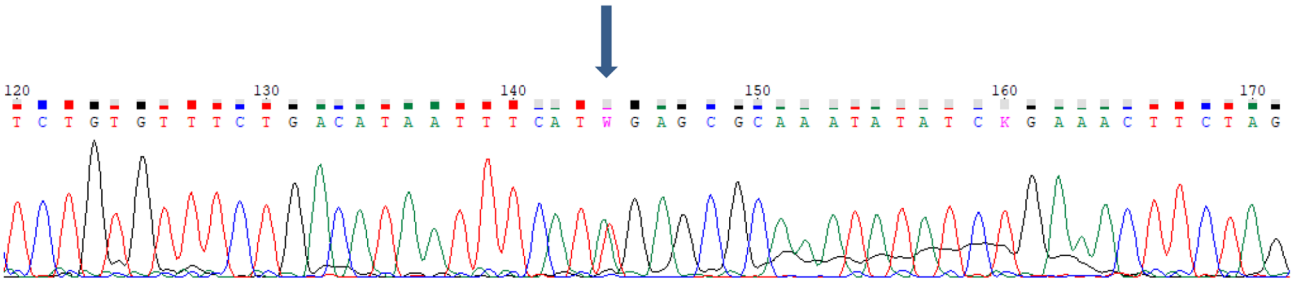


P4. BRCA1:c.4035delA, p.(Glu1346Lysfs*20)

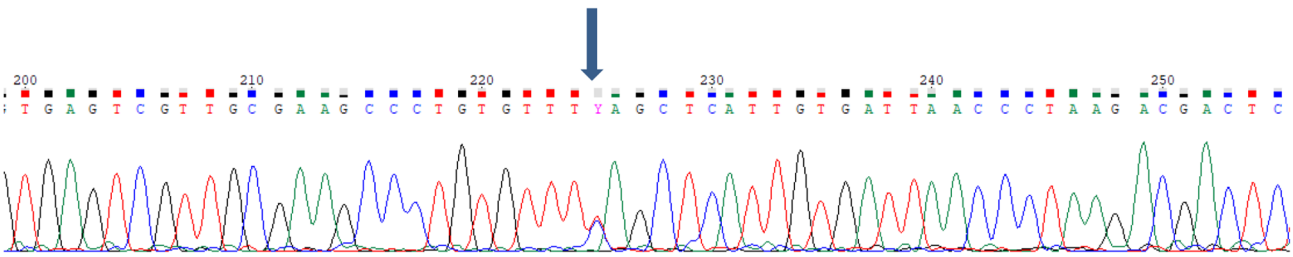
Figure S3. Sanger Sequencing analysis of genomic DNA from proband P5 carrying the variant BRCA2:c.9682delA, p.(Ser3228Valfs*21), P6 carrying the variant BRCA2:c.8087T>A,p.(Leu2696*), P7 carrying the variant PALB2:c.3271C>T, p.(Gln1091*) and P8 carrying the variant BRCA2:c.5557dupT,p.(Cys1853Leufs*5).



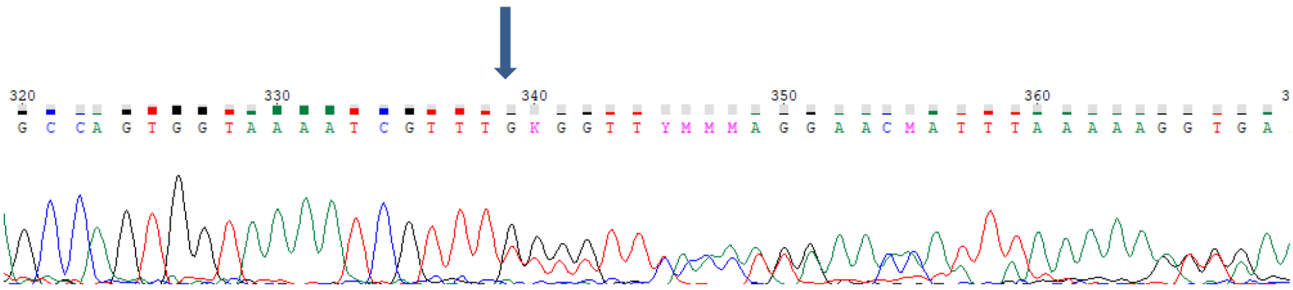
P5. BRCA2:c.9682delA, p.(Ser3228Valfs*21)



P6. BRCA2:c.8087T>A, p.(Leu2696*)

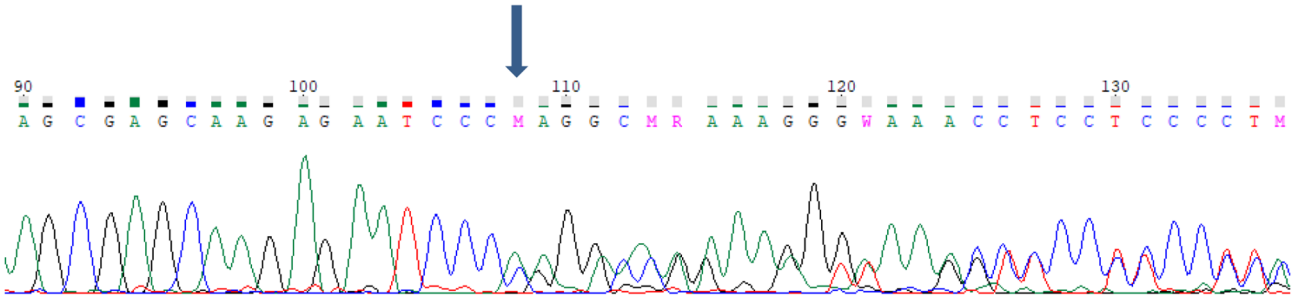


P7. PALB2:c.3271C>T, p.(Gln1091*)

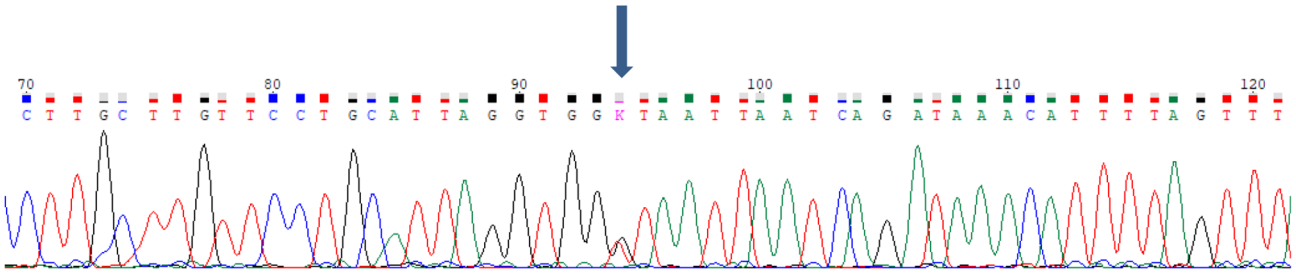


P8. BRCA2:c.5557dupT, p.(Cys1853Leufs*5)

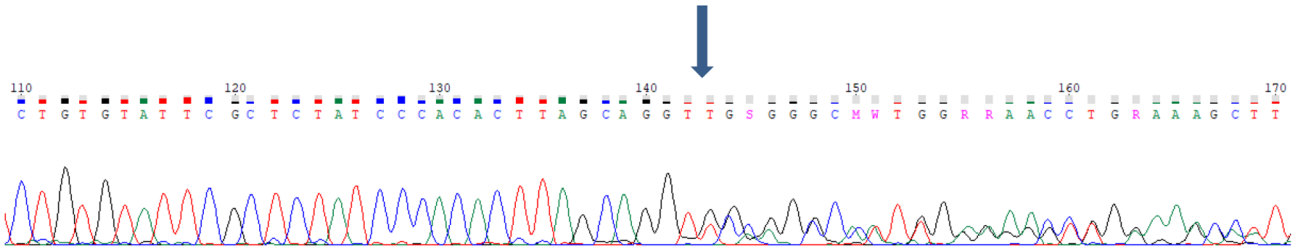
Figure S4. Sanger Sequencing analysis of genomic DNA from proband P9 carrying the variant BRCA1:c.5266dupC, p.(Gln1756Profs*74)], P10 carrying the variant RAD51C:c.904+5G>T (indicated by the arrow), P11 carrying the variant ATM:c.6527delT, p.(Leu2176Cysfs*59) and P12 carrying the variant BRCA1:c.3700_3704delGTAAA, p.(Val1234Glnfs*8).



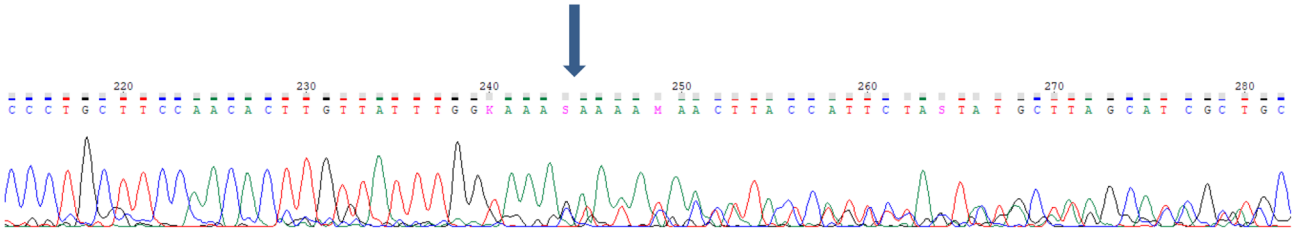
P9. BRCA1:c.5266dupC, p.(Gln1756Profs*74)



P10. RAD51C:c.904+5G>T

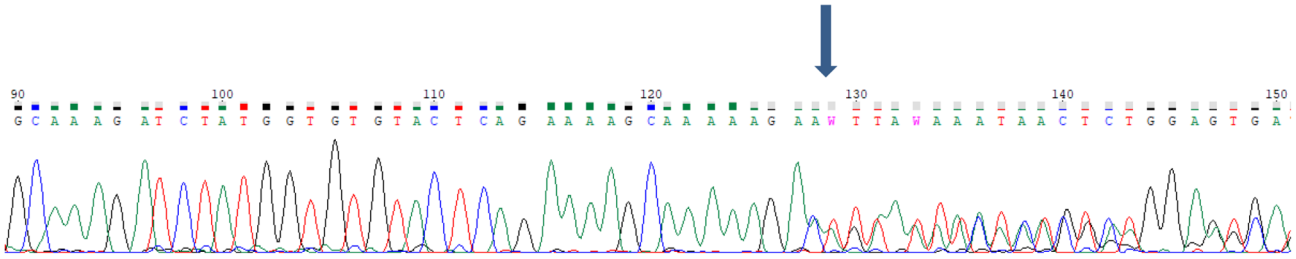


P11. ATM:c.6527delT, p.(Leu2176Cysfs*59)

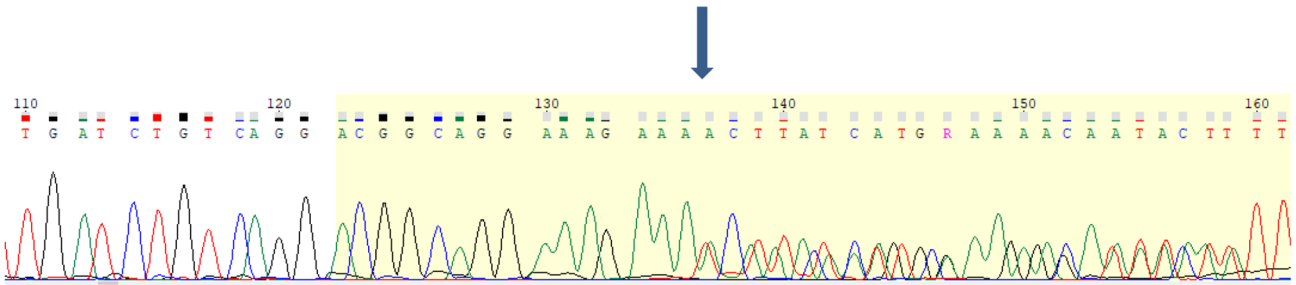


P12. BRCA1:c.3700_3704delGTAAA, p.(Val1234Glnfs*8)

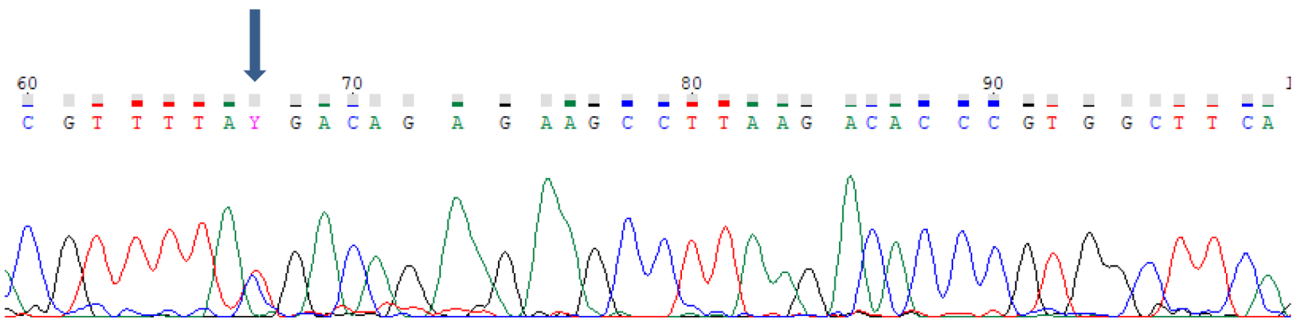
Figure S5. Sanger Sequencing analysis of genomic DNA from the proband P12 carrying the variant RAD50:c.326_329delCAGA, p.(Thr109Asnfs*20), P13 carrying the variant NBN:c.657_661delACAAA, p.(Lys219Asnfs*16) and P14 carrying the variant CHEK2:c.1427C>T, p.(Thr476Met).



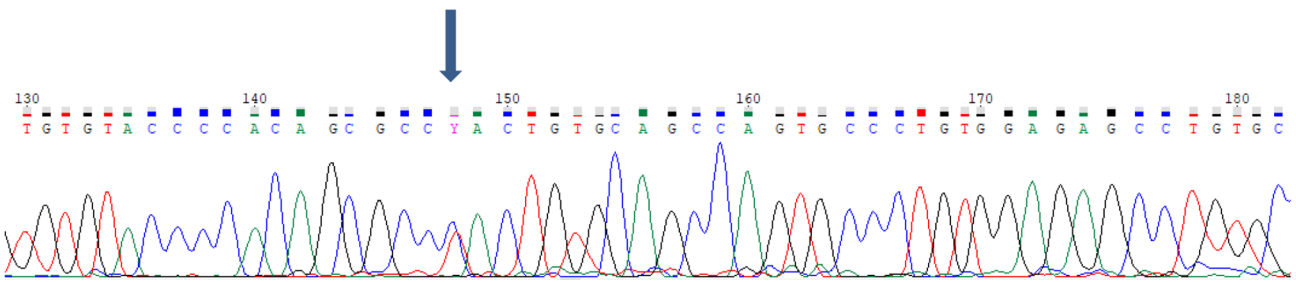
P12. RAD50:c.326_329delCAGA, p.(Thr109Asnfs*20)



P13. NBN:c.657_661delACAAA, p.(Lys219Asnfs*16)

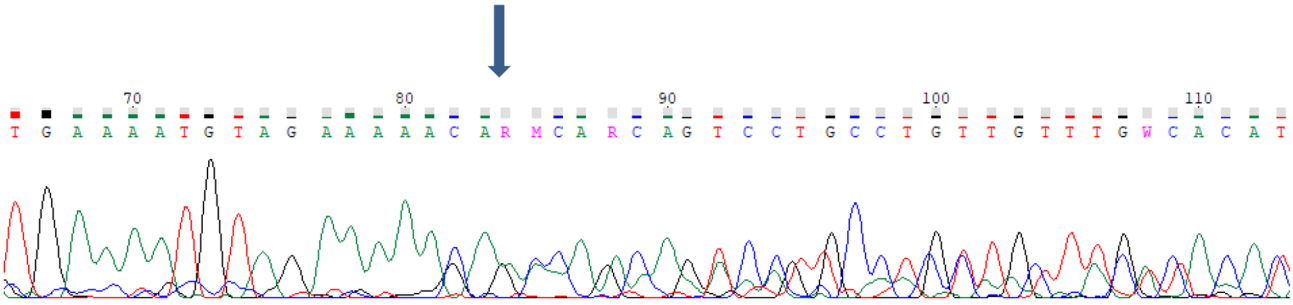


P14. CHEK2:c.1427C>T, p.(Thr476Met)

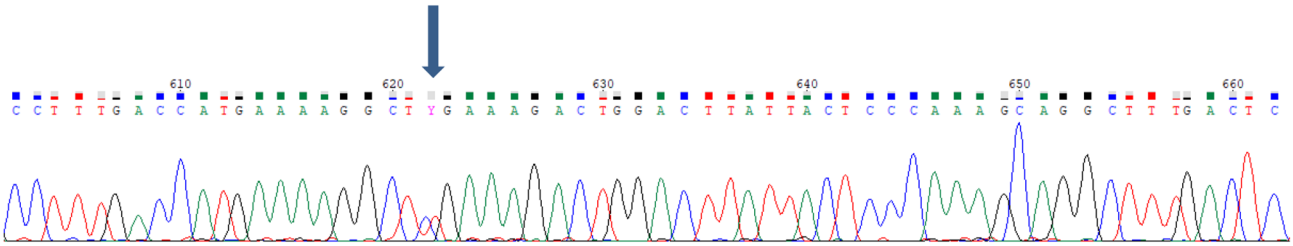


P15. MUTYH:c.884C>T, p.(Pro295Leu)

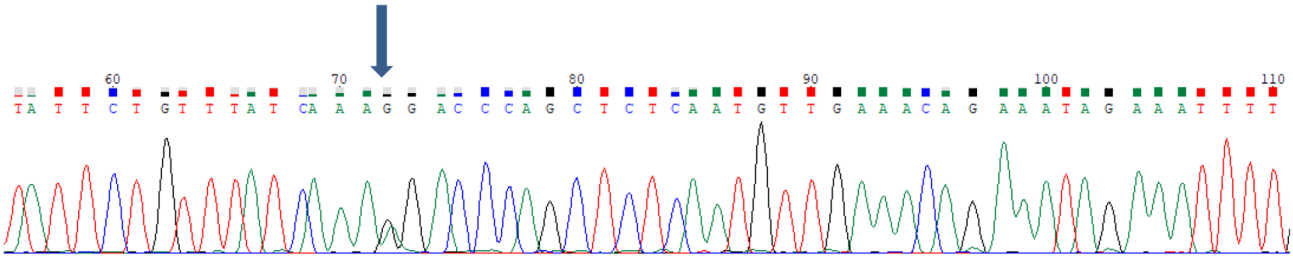
Figure S6. Sanger Sequencing analysis of genomic DNA from proband P15 carrying the variant MUTYH:c.884C>T, p.(Pro295Leu), P16 carrying the variant BRCA2:c.4936_4939delGAAA, p.(Glu1646Glnfs*23), P17 carrying the variant MSH6:c.2764C>T, p.(Arg922*) and P18 carrying the variant CHEK2:c.793-1G>A.



P16. BRCA2:c.4936_4939delGAAA, p.Glu1646Glnfs*23



P17. MSH6:c.2764C>T, p.(Arg922*)



P18. CHEK2:c.793-1G>A