Supplementary 2: Details of pathogenic/likely pathogenic/ increased risk allele germline mutations

Classification	Genes	Exon/ Intron	Nucleotide Changes	Amino Acid Changes	Variant Type
Increased Risk Allele	APC	Exon 16	c.3920T>A	(p.Ile1307Lys)	Missense
Increased Risk Allele	APC	Exon 16	c.3920T>A	(p.Ile1307Lys)	Missense
Likely Pathogenic	PMS2	Intron 1	c.23+1G>A	(Splice donor)	Splice Site Variant
Pathogenic	TP53	Exon 5	c.473G>A	(p.Arg158His)	Missense
Pathogenic	PALB2	Exon 5	c.2328del	(p.Phe776Leufs*75)	Deletion
Pathogenic	RECQL4	Exon 5	c.1015dup	(p.Leu339Profs*11)	Duplication
Pathogenic	RET	Exon 14	c.2410G>A	(p.Val804Met)	Missense
Pathogenic	TP53	Exon 5	c.541C>T	(p.Arg181Cys)	Missense
Likely Pathogenic	BRIP1	Exon 18	copy number = 3	Duplication	Duplication
Pathogenic	BRCA2	Exon 11	c.2254_2257del	(p.Asp752Phefs*19)	Deletion
Pathogenic	BRCA2	Exon 11	c.4222_4223del	(p.Gln1408Valfs*5)	Deletion
Pathogenic	CHEK2	Exon 4	c.470T>C	(p.Ile157Thr)	Missense
Pathogenic	BRCA2	Exon 11	c.2254_2257del	(p.Asp752Phefs*19)	Deletion
Pathogenic	BRCA2	Exons 5-11	copy number = 3	Duplication	Duplication
Likely Pathogenic	CHEK2	Intron 7	c.846+1G>C	(Splice donor)	Splice Site Variant
Pathogenic	MLH1	Exon 16	c.1837_1840del	(p.Glu613Phefs*2)	Deletion