

## *Supplementary Materials*

# **Pathogenic Germline Mutations of DNA Repair Pathway Components in Early-onset Sporadic Colorectal Polyp and Cancer Patients**

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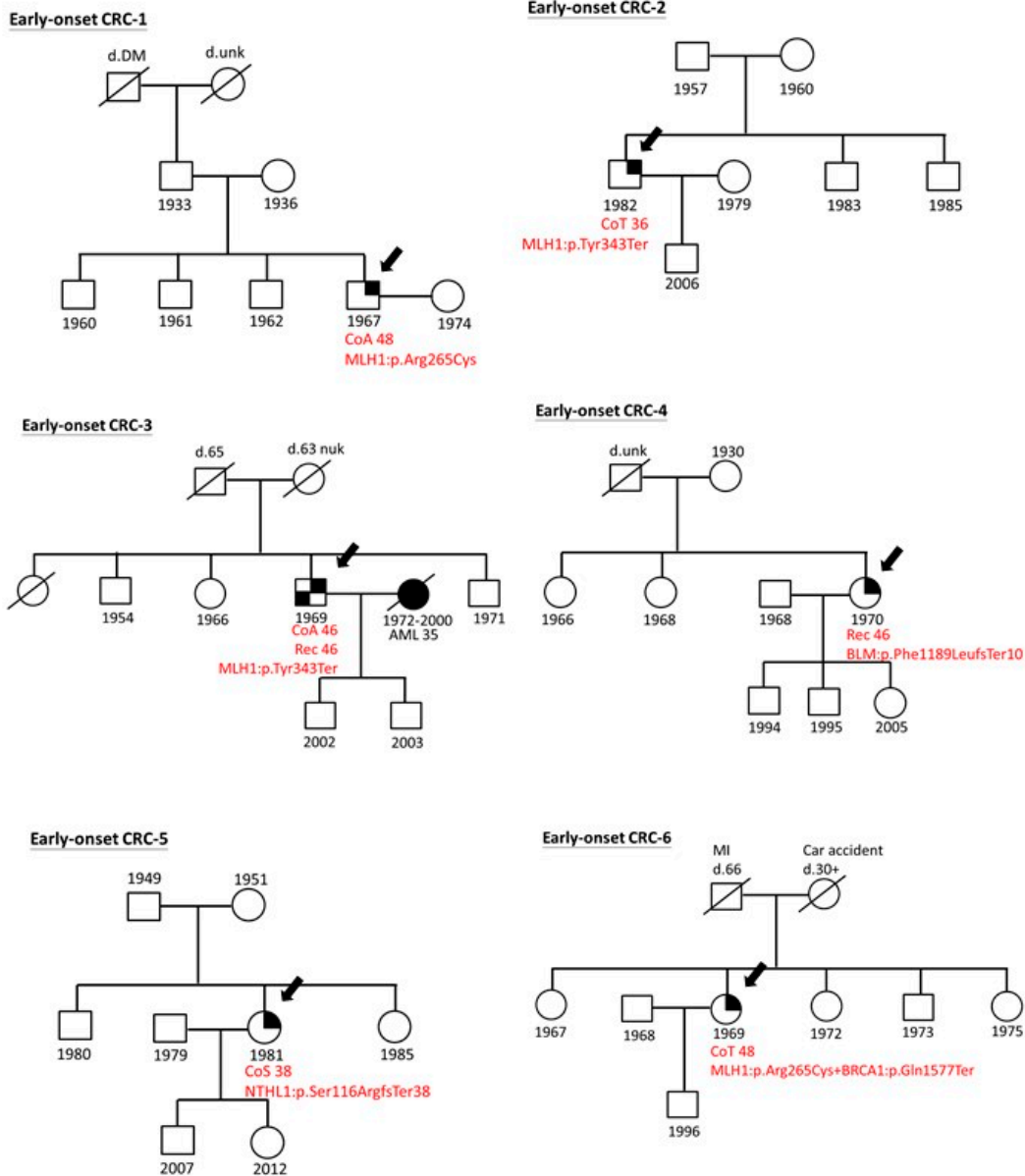
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**Supplementary Table 1:** Detailed clinical and genetic information for the nine mutation carriers

Group	sample ID	Age	Gender	Stage	Gene	Zygoty	Location	NT Change	A.A. Change	Mutation type	ClinVar/VarSome annotation	ExAc_EAS
Normal	2307	39	M		BRCA2	Heterozygous	exon11	c.2652dupA	p.D885Rfs*2	frameshift insertion	Not reported /Pathogenic	
Polyps	1941	41	F		CHEK2	Heterozygous	exon4	c.472delA	p.I158Yfs*9	frameshift deletion	Not reported/ Likely Pathogenic	0.0006
Polyps	1972	49	F		CHEK2	Heterozygous	exon4	c.472delA	p.I158Yfs*9	frameshift deletion	Not reported/ Likely Pathogenic	0.0006
CRC	1657	46	F	IV	BLM	Heterozygous	exon19	c.3566delT	p.F1189Lfs*9	frameshift deletion	Likely pathogenic/ Pathogenic	
CRC	2299	48	M	III	MLH1	Heterozygous	exon10	c.C793T	p.R265C	nonsynonymous SNV	Pathogenic/ Pathogenic	
CRC	2423	46	M	II	MLH1	Heterozygous	exon11	c.C1029A	p.Y343X	stopgain	Not reported/Pathogenic	
CRC	2757	48	F	IV	MLH1	Heterozygous	exon10	c.C793T	p.R265C	nonsynonymous SNV	Pathogenic/ Pathogenic	
CRC					BRCA1	Heterozygous	exon15	c.C4729T	p.Q1577X	stopgain	Pathogenic/ Pathogenic	
CRC	3248	36	M	III	MLH1	Heterozygous	exon11	c.C1029A	p.Y343X	stopgain	Not reported/ Pathogenic	
CRC	SP-0129	38	F	IV	NTHL1	Heterozygous	exon4	c.699_700del	p.S234Rfs*37	frameshift deletion	Not reported/ Pathogenic	

SNV: single-nucleotide variant; MAF in ExAc\_EAS: minor allele frequency in the Exome Aggregation Consortium\_East Asia



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2 **Supplementary Figure 1.** Three-generation pedigree trees of six sporadic early-onset CRC proband  
 3 families. The arrow points out the proband in the family. The number below the square or circle symbol  
 4 stands for the birth year of the family member. The three letters and two-digit number below the  
 5 proband represent for the cancer location and the age with cancer diagnosis. For example: CoT 48  
 6 indicates the patient got the diagnosis at 48 years old and the cancer was located at the Transverse  
 7 colon. A: Ascending; S: Sigmoid; Rec: Rectum. Mutations which the proband carried were denoted in  
 8 the third row.