

Table S1. Estimated allele frequencies of the microsatellite markers D22S1163, D22S689, D22S275 and D22S1150 flanking the *CHEK2* gene used for haplotype analysis.

D22S1163		D22S689		D22S275		D22S1150	
Allele	Frequency	Allele	Frequency	Allele	Frequency	Allele	Frequency
141	0	202	0.067	158	0	213	0.019
143	0	206	0.019	160	0.028	215	0.057
145	0	210	0.134	162	0.163	217	0.038
147	0.201	214	0.201	164	0.144	219	0.192
149	0.269	218	0.211	166	0.201	221	0.038
151	0.230	222	0.048	168	0.259	223	0.451
153	0.115	226	0.019	170	0.096	225	0.028
155	0			172	0.009	227	0.019
157	0.105			174	0.076	229	0.038
159	0.076			176	0	231	0.028
161	0			178	0	233	0.057
163	0			180	0	235	0

Table S2. Haplotype analysis for microsatellite markers D22S1163, D22S689, D22S275 and D22S1150 in *CHEK2* c.499G>A carriers and non-carriers.

Sample ID	D22S1163	D22S689	D22S275	D22S1150
F667	143/159	202/210	168/168	223/231
F1326	143/149	210/210	168/168	223/223
F2050	143/151	210/218	168/168	215/223
F2050a	143/151	210/218	168/170	219/223
F2276	N.A.	N.A.	N.A.	N.A.
F2469	143/159	210/214	166/168	223/233
F2469b	143/143	206/210	168/168	223/231
F2605	143/151	210/218	162/168	223/229
F2774	143/147	210/214	164/168	223/235
F3445	143/153	210/214	168/168	223/233
F3754	N.A.	N.A.	N.A.	N.A.
Common allele between carriers	143	210	168	223
Non-carriers				
F667a	141/143	202/222	166/168	219/231
F667b	141/159	210/222	168/168	225/231
F667c	151/159	202/206	160/168	231/233
F2469a	157/159	214/222	162/166	215/233
F2605a	151/157	218/222	162/162	219/229
F2605b	151/159	202/218	162/166	219/229
F2774a	143/151	214/214	166/166	225/235
F3445a	149/153	214/218	166/166	219/233
F3445b	151/153	206/214	166/168	219/233

*Alleles segregating with the disease are highlighted in red.

Table S3. Haplotype analysis for microsatellite markers D22S1163, D22S689, D22S275 and D22S1150 in *CHEK2* c.549G>C carriers and non-carriers

Sample ID	D22S1163	D22S689	D22S275	D22S1150
F459	143/149	214/218	160/166	221/221
F459a	143/147	206/218	166/172	221/221
F498	143/159	214/218	166/170	213/221
F498a	143/149	214/218	166/170	219/221
F1018	143/149	210/218	164/166	221/233
F1327	143/143	218/222	166/166	215/221
Common allele between carriers	143	218	166	221
Non-carriers				
459b	149/159	210/218	160/162	223/223
459c	149/151	214/214	160/168	223/223
498b	151/157	214/214	170/170	213/219

*Alleles segregating with the disease are highlighted in red.

Table S4. Haplotype analysis for microsatellite markers D22S1163, D22S689, D22S275 and D22S1150 in *CHEK2* c.592+3A>T carriers and non-carriers

Sample ID	D22S1163	D22S689	D22S275	D22S1150
F1313	149/149	206/210	162/164	219/233
F2482	149/149	206/210	162/170	219/219
F3247	149/157	206/206	162/166	219/223
F3710	149/151	206/206	162/170	219/219
F3710a	149/151	206/206	162/162	219/219
Common allele between carriers	149	206	162	219
Non-carriers				
2482a	143/149	210/218	170/166	219/219
2482b	151/153	210/218	162/168	219/233
2482c	149/147	210/210	162/170	219/223

*Alleles segregating with the disease are highlighted in red.

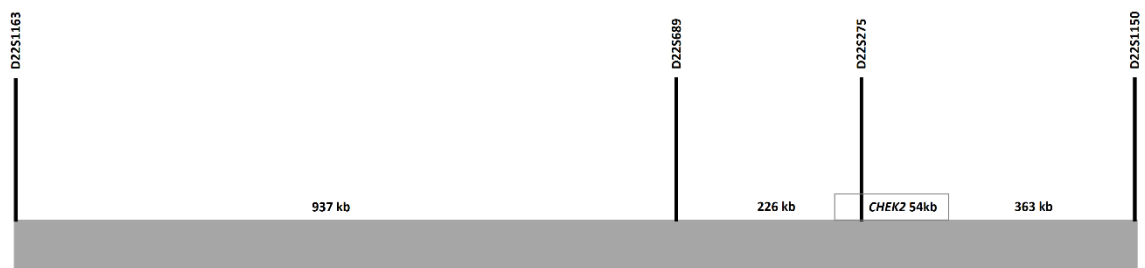


Figure S1. Physical map showing the microsatellite markers used for haplotype analysis relative to *CHEK2*, on chromosome 22.