

Table S4. The genotypes and the allelic frequencies of analyzed SNPs in patients homozygous for c.919-2A>G and in the control sample.

SNPs (distance from c.919-2A>G)	Genotypes	Homozygotes for c.919- 2A>G (n=23)	Controls (n=23)	Alleles	Homozygotes for c.919-2A>G		Controls		<i>p</i>	δ
					Number of alleles	Frequency of alleles	Number of alleles	Frequency of alleles		
rs2248464 (20276)	AA	23	4	A	46	1.0	26	0.2063	0.1697 x 10⁻²²	1.0
	AT	0	18							
	TT	0	41	T	0	0.0	100	0.7937	0.1687 x 10⁻²²	-3.8462
	Total	23	63							
rs2248465 (20270)	CC	23	9	C	46	1.0	40	0.3175	0.3332 x 10⁻¹⁷	1.0
	CT	0	22							
	TT	0	32	T	0	0.0	86	0.6825	0.3300 x 10⁻¹⁷	-2.1500
	Total	23	63							
rs3801943 (2232)	TT	23	45	T	46	1.0	107	0.8492	1.0	1.0
	TA	0	17							
	AA	0	1	A	0	0.0	19	0.1508	0.1818 x 10⁻²	-0.1776
	Total	23	63							
rs2712212 * (2183)	AA	23	28	A	46	1.0	84	0.6667	1.0	1.0
	AG	0	28							
	GG	0	7	G	0	0.0	42	0.3333	0.2168 x 10⁻⁶	-0.5000
	Total	23	63							
c.919-2A>G										
rs2395911 * (215)	GG	23	9	G	46	1.0	44	0.3492	0.6282x 10⁻¹⁶	1.0
	GT	0	26							
	TT	0	28	T	0	0.0	82	0.6508	0.6211 x 10⁻¹⁶	-1.8636
	Total	23	63							
rs2712211 * (2024)	CC	0	26	C	0	0.0	78	0.6190	0.9734 x 10⁻¹⁵	-1.6250
	CG	0	26							
	GG	23	11	G	46	1.0	48	0.3810	1.0	1.0
	Total	23	63							
rs3801940 * (2318)	CC	23	14	C	46	1.0	56	0.4444	1.0	1.0
	CG	0	28							
	GG	0	21	G	0	0.0	70	0.5556	0.1503 x 10⁻¹²	-1.2500
	Total	23	63							
rs2072064 (10615)	TT	0	25	T	0	0.0	75	0.5952	0.6892 x 10⁻¹⁴	1.4706
	TA	0	25							
	AA	23	13	A	46	1.0	51	0.4048	1.0	1.0
	Total	23	63							
rs2072065 (10763)	CC	23	16	C	46	1.0	64	0.5079	1.0	1.0
	CT	0	32							
	TT	0	15	T	0	0.0	62	0.4921	0.1391 x 10⁻¹⁰	-0.9688
	Total	23	63							

* – rs2712212, rs2395911, rs2712211, and rs3801940 were included for comparative analysis with the data from the study by Wu et al. [12]. The maximum indices of linkage disequilibrium (δ) and statistically significant ($p < 0.05$) differences in allele frequencies between the c.919-2A>G homozygotes and the control sample are in bold.