Supplementary Figure. Haplotype analysis to investigate a putative founder mutation.

Genotyping results for markers D2S309, D2S2214, D2S116 and D2S2309 are shown for Patients 1-10 as well as the two previously reported cases documented in the literature (denoted RC1[10] and RC2[9]).

		Ρ1	P2	Р3	Ρ4	Ρ5	P6	P7	P8	P9	P10	RC1	RC2
1	1		ullet	•	ullet		•			•		ullet	•
NDUFB3 ►		11	11	11	11	22	13	13	11	11	11	14	11
	D2S309 0.002Mb	11	12	11	11	22	12	12	12	12	11	13	12
	D2S2309 0.07Mb	11	11	12	11	11	11	11	11	11	12	13	11
	D2S2214 0.1Mb	11	11	11	12	11	13	13	11	11	11	14	11
	haplotype	a a	a b	a c	a d	e e	a f	a f	a b	a b	a c	a #	a b