

Supplementary Table S1. Clinical and diagnostic evaluations. ERG: electroretinogram. VEP: visual evoked potential. OCT: optical coherence tomography. MRI: magnetic resonance imaging.

Diagnostic exams	N=60	Results	n	% of performed	
ERG	47 (80%)	Normal	42	89	
		Not interpretable due to scarce collaboration	5	11	
VEPs	53 (88%)	Normal	15	28	
		Aspecific alterations	38	72	
OCT	0	/	/	/	
Brain MRI	24 (40%)	Normal	24	100	
Genetic testing	23 (38%)	Negative	12	52	
		Ongoing	5	22	
		FRMD7	6	26	
Ophthalmologic follow-up	60 (100)	Anterior segment	Normal	60	100
		Eye lens	Normal	60	100
		Fundus oculi	Normal	60	100
Neuropediatric follow-up	60 (100)	Normal	60	100	

Supplementary Table S2. Cognitive test performed and results according to scoring rules of each test. A total of 27 patients (17 M, 10 F, mean age: 9.5 y, range: 4.3-17.8 y) underwent the cognitive assessment as explained in the methods section, according to their age. Wechsler preschool and primary scale of intelligence, fourth edition (WPPSI-IV); verbal comprehension index (VCI); visual-spatial index (VSI); fluid reasoning index (FRI); working memory index (WMI); processing speed index (PSI); and full-scale IQ (FSIQ). Wechsler preschool and primary scale of intelligence, third edition: verbal comprehension index (VCI); performance index (PI); processing speed index (PSI); and total intelligence quotient (TIQ). Wechsler intelligence scale for children (WISC-IV) and Wechsler adult intelligence scale (WAIS-IV): verbal comprehension index (VCI); perceptual reasoning index (PRI); working memory index (WMI); processing speed index (PSI); and intelligence quotient (IQ).

IQ test (n=27)	Index	Performed n (%)	Not interpretable (%)	Index <85 (% of performed)	Index ≥85 (% of performed)
WPPSI-IV (n=2)	VCI	2 (100)	0	0	2 (100)
	VSI	2 (100)	0	0	2 (100)
	FRI	2 (100)	0	0	2 (100)
	WMI	2 (100)	0	0	2 (100)
	PSI	2 (100)	0	0	2 (100)
	FSIQ	2 (100)	0	0	2 (100)
WPPSI-III (n=5)	VCI	5 (100)	0	0	5 (100)
	PRI	5 (100)	0	0	5 (100)
	PSI	4 (80)	1 (20)	0	4 (100)
	IQ	5 (100)	0	0	5 (100)
WISC-IV (n=18)	VCI	18 (100))	0	0	18 (100)
	PRI	18 (100)	0	0	18 (100)
	WMI	17 (94)	1 (6)	1 (6)	16 (94)
	PSI	18 (100)	0	5 (28)	13 (72)
	IQ	15 (83)	3 (17)	1 (7)	14 (93)
WAIS-IV (n=2)	VCI	2(100)	0	0	2 (100)
	PRI	2 (100)	0	0	2 (100)
	WMI	2 (100)	0	0	2 (100)
	PSI	2 (100)	0	0	2 (100)
	IQ	2 (100)	0	0	2 (100)