

Supplementary Table. Clinical and instrumental features of the three patients.

Patient n.	1	2	3
Sex	Male	Male	Female
Ethnic origin	Italian	Italian	Turkish
Parental consanguinity	No	No	Yes (second grade cousins)
Unaffected sibs	No	No	No
Affected sibs	Yes (patient 2 and spontaneous abortion)	Yes (patient 1 and spontaneous abortion)	No
Family history	Positive for Gaucher disease in paternal line	Positive for Gaucher disease in paternal line	Negative
Age at evaluation	14 years	8 years	11 years
Psychomotor development	Delayed: walk unaided at 6 years of age; no acquisition of expressive language	Delayed: walk unaided at 3 years of age, no acquisition of expressive language	Delayed
Intelligence Quotient (WISC III-R)	Moderate intellectual disability (PIQ= 46). Only performance skills	Moderate intellectual disability (PIQ= 41). Only performance skills could be	Severe intellectual disability

	could be evaluated	evaluated	
Behavior, social skills	Good social skills	Good social skills	Poor social skills
Learning	Able to read and write words in capital letters printed	None acquisition	None acquisition
Language	Absent expressive language. Learning sign language. Oro-motor dyspraxia	Absent expressive language. Learning sign language. Oro-motor dyspraxia	Poor expressive language and social skills
Neurological findings	Diffuse hypotonia, hand dysmetria, and gait ataxia	Diffuse hypotonia, hand dysmetria, and gait ataxia	Diffuse hypotonia and clumsiness.
Neuro-ophthalmological findings	Reduced visual acuity, nystagmus, oculomotor apraxia. Fundus oculi: bilateral optic nerve coloboma	Oculomotor apraxia, nystagmus Fundus oculi: coloboma in left eye	Severe visual impairment and roving nystagmus (Leber amaurosis)
Electroretinogram	amplitude reduction (early stage)	Normal	Response was absent
Brainstem auditory evoked	Normal	Normal	Normal

potentials			
EEG	Normal	Normal	/
Dimorphisms and extra-brain malformations	No	No	Facial dysmorphisms
Liver	Mild hepato-splenomegaly; congenital hepatic fibrosis; mild portal hypertension	Hepatosplenomegaly; congenital hepatic fibrosis and portal hypertension, treated with specific drugs	Normal
Kidney	Congenital unilateral polycystic kidney (right) with progressive atrophy and hypertrophy of contralateral kidney	Normal	Normal

Abbreviations: PIQ= performance Intellectual Quotient; WISC-III= ; Wechsler Intelligence Scale III-Revised.