

**On-line Table: Clinical and instrumental features of the 3 patients**

	Patient No.		
	1	2	3
Sex	Male	Male	Female
Parental consanguinity	No	No	Yes (second-grade cousins)
Unaffected siblings	No	No	No
Affected siblings	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 (yr)	14	8	11
Psychomotor development	Delayed: walked unaided at 6 yr of age; no acquisition of expressive language	Delayed: walked unaided at 3 yr of age; no acquisition of expressive language	Delayed
IQ (WISC III-R)	Moderate intellectual disability (PIQ = 46); only performance skills could be evaluated	Moderate intellectual disability (PIQ = 41); only performance skills could be evaluated	Severe intellectual disability
Behavior, social skills	Good social skills	Good social skills	Poor social skills
Learning	Able to read and write words in uppercase letters printed	No acquisition	No acquisition
Language	Absent expressive language; learning sign language; oromotor dyspraxia	Absent expressive language; learning sign language; oromotor dyspraxia	Poor expressive language and social skills
Neurologic findings	Diffuse hypotonia, hand dysmetria, and gait ataxia	Diffuse hypotonia, hand dysmetria, and gait ataxia	Diffuse hypotonia and clumsiness
Neuro-ophthalmologic 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 congenital amaurosis)
Electroretinogram	Amplitude reduction (early stage)	Normal	Response was absent
Brain stem auditory evoked potentials	Normal	Normal	Normal
EEG	Normal	Normal	Not available
Dimorphisms and extrabrain malformations	No	No	Facial dysmorphism
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

**Note:**—PIQ indicates Performance Intelligence Quotient; WISC III-R, Wechsler Intelligence Scale for Children-Revised, Third Edition; EEG, electroencephalography.