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Type 3 Gaucher disease, diagnostic in adulthood

El-Beshlawy reported in the issue of the Journal the largest experience to date of patients with type 3 Gaucher disease (GD3) treated by imiglucerase and enrolled in an International Registry [1]. They confirmed both a very early clinical presentation (median age at diagnosis: 1.7 year) and a good prognosis (probability of surviving for at least 5 years after starting imiglucerase of 92%). However, for unknown reason, they excluded of the study, patients diagnosed after 18. From the French registry experience, we would like to report the rare possibility of diagnostic GD3 in adulthood.

Among 496 patients enrolled in French registry, 18 alive patients had GD3. The median age was 19.6 years (2.16–51.8) age and the median at diagnosis: 1.5 year (0–26.5 years) and as in El-Beshlawy's study. However, 3 patients out of 18 (16%) were diagnosed after 18 and one after 16. Main characteristics of these patients are summarized in Table 1.The reason of the delay diagnosis were: mild neurologic symptoms $n = 2$ (isolated ophtalmoplegia, isolated status seizure), challenging molecular form $n = 1$ and poor access to healthcare system in the country of birth $n = 1$. 50% of our patient diagnosed at adulthood had at least one D409 mutation, contrary to 14.9% in El-Beshlawy cohort, in which L444P mutation represents 77% of cases. Patients with D409 mutation have been previously reported possibly lately diagnosed and could exhibited particular cardiac and corneal involvement [2]. Molecular deficit in Saposine C is a well-known but very rare challenging diagnostic [3]. Our small series corroborate the El-Beshlawy results about the good outcome and earliness of symptoms of GD3. However we would like add that the heterogeneity of GD3 could lead to unusual diagnostic in adulthood (16% of cases) and non-pediatrics physicians may also be aware of this possibility.

Table 1
Main characteristics of patients diagnosed > 16 with GD3 in French Gaucher disease registry.

	Age of 1st symptom (year)	Main symptoms	Age at diagnosis (years)	Main reason of diagnostic delay	Type of mutation
1	7.4	- Frontal/generelalized/myoclonic jerks (11 years) - Loss of intellectual abilities (15 years) - Cerebellar syndrome - Mild splenomegaly	16.4 years	- Stability of epilepsy between 11 and 15 - Late onset of intellectual disability. - Delay for definite diagnostic due to rare mutation	Saposine C
2	18.3	- PAUCI-symptomatic - Slight ophtalmoplegia - Mild Splenomgaly/hepatomegaly - Mild thrombopenia	18.5	- Moderate ophtalmoplegia	L444P/RECTL
3	0,4	- Ophtalmoplegia (falsely linked to strabism with surgical treatment) - Splenomegaly, cytopenia in childhood - Mental retardation	21	- Born in Algeria: poor access to efficient health care system	D409H/D409H
4	11	- Myoclonic seizures - Mental retardation Cerebellar syndrome	26.5	- Stable seizure until adulthood	D409H/L444P

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