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Editorial

Screening for Pompe, Fabry, Gaucher and Mucopolysaccharidosis Type 1 (MPS 1): A Clinical and Laboratory Overview

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Received: November 14, 2016; **Published:** November 15, 2016

Fabry, Gaucher, Pompe and Mucopolysaccharidosis type 1 (MPS1) are a group of lysosomal disorders (LSD) with enzyme replacement therapy. We can use Genzyme screening card in filter paper as soon as we have clinical suspicious (Figure 1). In all situations, it is important to perform the biochemical and molecular diagnosis for genetic counseling and prenatal diagnosis (Table 1).

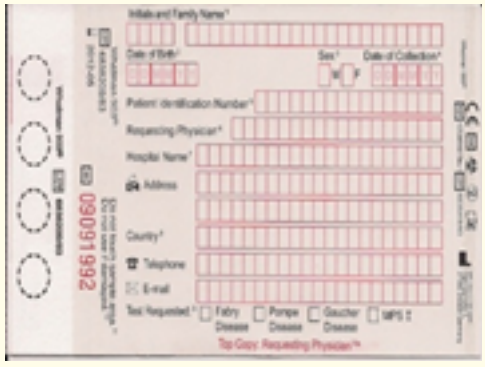

Figure 1: *Genzyme screening card.*

Disease	Enzyme	Gene
Fabry	alfa-galactosidase or alfa-GAL	GLA
Gaucher	B-glucosidase acid (glicocerebrosidase)	GBA
Pompe	alfa-glucosidase acid	GAA
MPS1	α-1-iduronidase	IDUA

Table 1: *Enzyme and molecular screening for Fabry, Gaucher, Pompe and MPS 1.*

Fabry disease is an inherited X-linked autosomal recessive disease caused by an inability to produce an enzyme called alpha-galactosidase or alpha-GAL.

Without this enzyme, a kind of fat called globotriaosylceramide or GL-3, which should be removed from the body, remains in the cells. The result is the accumulation of this material in the blood vessels, leading to malfunctioning of the kidneys, heart and brain. The symptoms of Fabry's disease appear between 6 and 9 years of age. Pain is considered the first and most common of all symptoms. Most people with the disease have two types of pain: acroparasthesia and "Fabry crises". In acroparasthesia, pain affects the hands and feet. It is described as burning pain and may be intermittent or daily. In "Fabry crises", the episodes are intense, with burning pain, initially in the hands and feet and radiating to other parts of the body. They can last from a few minutes to a few days. Other symptoms: hypohidrosis/

anhydrosis, frequent fevers, overheating with physical exercise and intolerance to hot weather, angiokeratoma (Figure 2) - characteristic purple-red rash, is the most visible sign of Fabry's disease are found from the navel to the knees and, in some cases, only on the elbows or knees. Cornea verticillate is similar to the rays of a bicycle wheel (does not affect the vision) and ophthalmoscopy of slit lamp confirm the diagnosis. Other symptoms included, epigastric pain, diarrhea and nausea after meal, renal insufficiency due to excess proteinuria, cardiomyopathy, arrhythmias, heart failure, dizziness, headache, stroke and depression.

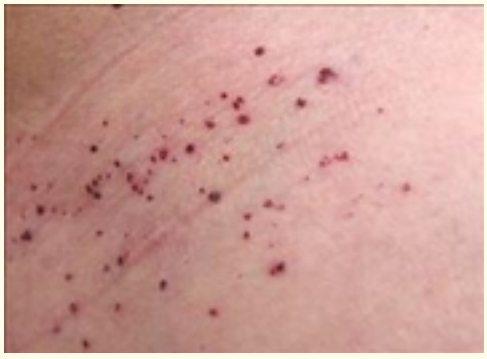

Figure 2: *Angiokeratoma.*

Gaucher, arises from enzymatic deficiency of glucocerebrosidase. This enzyme is not digested within the lysosome, progressively accumulating in the macrophages. These macrophages are increasing in size, being called "Gaucher cells". These cells accumulate mainly in the liver, spleen, marrow, and may cause spontaneous bone fractures. There are 3 types of Gaucher disease: Type 1, is the most frequent, is non-neuropathic or chronic non-neuropathic adult form. Patients may be asymptomatic or present with various forms of hepatosplenomegaly, hematological alterations or bone atrophy. Type 2: acute neuropathy or neuropathic infant form, hepatosplenomegaly, severe hematological changes and death, especially in the first two years of life.

Type 3: neuropathic subacute or juvenile neuropathic form with hepatosplenomegaly, anemia, thrombocytopenia, osteopenia, slowly progressive neuropathy symptoms from childhood and death between 20 and 40 years (Figure 3).

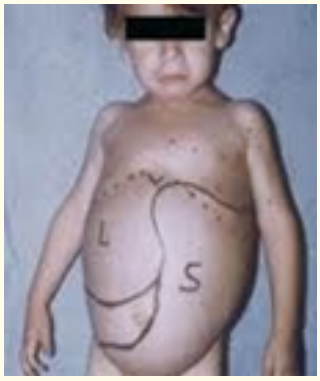

Figure 3: *Hepatosplenomegaly.*

Pompe, is caused by decreased activity of alpha-glucosidase-acid. There are two forms of clinical presentation. The infantile form, revealed progressive muscle weakness, hypotonia, motor delay, macroglossia, areflexia, progressive respiratory weakness, respiratory insufficiency, frequent respiratory infections, cardiorespiratory insufficiency, cardiomegaly, difficulty swallowing, sucking and/or feeding,

delayed psychomotor development and hepatomegaly. The adult form, showed progressive muscle weakness, unsteady gait-toes, difficulty climbing stairs, scapula allata and Gowers sign (Figure 4 and 5).

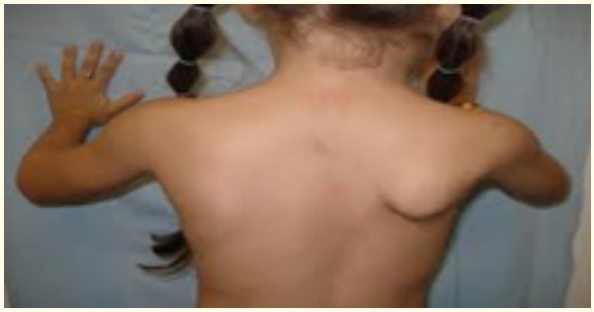

Figure 4: *Scapula allata.*

Figure 5: *Gowers sign.*

MPS 1, showed coarse face, macroglossia, cornea opacity, umbilical hernia, multiple dysostosis, hepatosplenomegaly, psychomotor regression, claw hands and carpal tunnel syndrome (Figure 6).

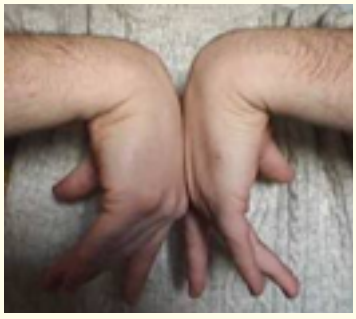

Figure 6: *Carpal tunnel syndrome.*

Fabry, Gaucher, Pompe and MPS 1, are disorders that can be under diagnosed because of the multisystem involvement of many organs. The Genzyme card is very useful for the diagnosis, because is very easy to perform with only some drops of blood and in few days, we have the result of the screening test. After confirmation, we can start ERT in these diseases.

Volume 2 Issue 6 November 2016