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Dried Blood Spot Samples for Pompe, Fabry, Gaucher and Mucopolysacharidosis (Mps): Our First Year Experience

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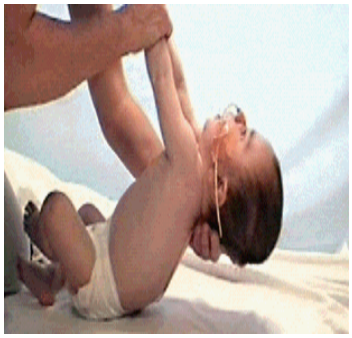
Abstract

Pompe, Gaucher, Fabry and MPS are lysosomal disorders. The new era of enzyme replace therapy (ERT) changed the prognosis of these diseases. Is important to diagnose as soon as possible to reduce the risk of a fatal evolution. We used dried blood spot samples in patients with 2 criteria of one of these diseases during a period of one year. We screened 18 cases (Pompe: 13, MPS 1: 3, Gaucher: 2, Fabry: 0). In our first year experience we confirmed a case of MPS 1 and a Pompe disease.

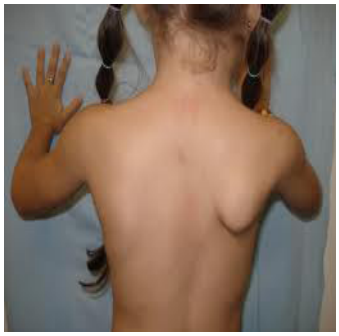
Keywords: *dried blood spot, lysosomal storage disease*

Introduction

Pompe, Gaucher, Fabry and MPS are a group of lysosomal disorders.(1) (Figs.1,2,3,4) The first three diseases and some of the MPS types have enzyme replacement therapy (ERT). We can use dried blood test for screening. The specificity and sensitivity of these tests are respectively 100% and 99.3%.

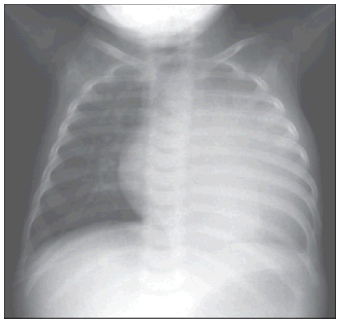


Hypotonia



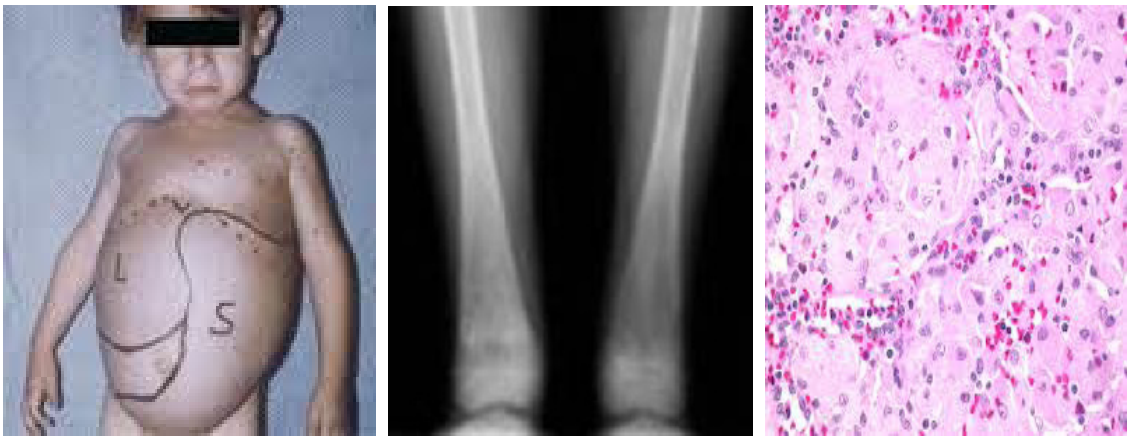
Scapula alatta

Gowers sign



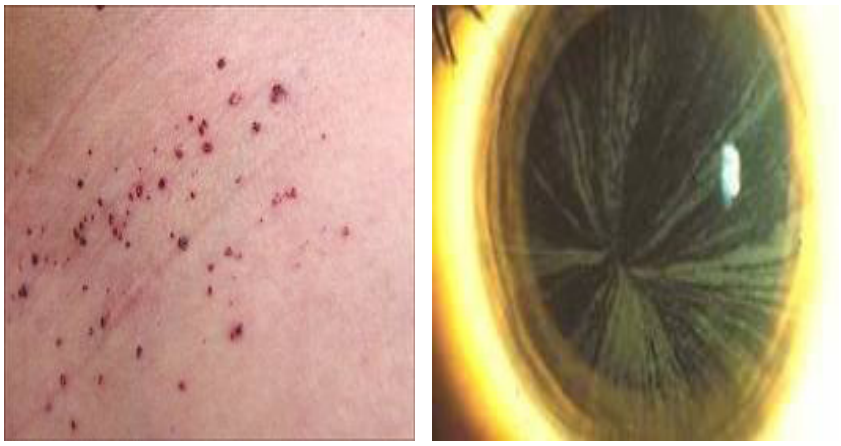
Cardiomegaly

Figure 1. *POMPE*



Hepatosplenomegaly Osteopenia Gaucher cells

Figure 2. *GAUCHER*



Angiokeratoma Cornea verticilata

Figure 3. *FABRY*

Cornea clouding Carpal tunnel syndrome Multiple dysostosis

Figure 4. *MPS*

Purpose

Use dried blood card for enzymatic study during one year (2017) in those cases that showed clinical signs and symptoms of Gaucher , Fabry, Pompe and MPS. (2,3)

Methods

All patients with at least 2 criteria of one of these diseases were screened by Genzyme/Sanofi screening card with dried blood spot samples for enzymatic study during the period of 1/1/2017 until 31/12/2017. This card can screen Gaucher, Fabry, Pompe and MPS 1, 2, 3B, 4A, 6 and 7.

All positive tests need to be confirmed by whole blood test and DNA study. The responsible genes for each disease are: GAA (Pompe), GBA (Gaucher), GLA (Fabry), IDUA (MPS 1), IDS (MPS 2), NAGLU (MPS 3 B), GALNS (MPS 4 A), ARSB (MPS 6) and GUSB (MPS 7).

The patients were selected in 2 main hospitals of Macau, one public (CHCSJ) and other private (KW).

After confirmation of the disease, they will start ERT for treatment.

Results

A total of 18 patients were screened. The age distribution was between 1 month to 6 years of age, with the gender relation of 1:1. The 18 cases were distributed as follow: Pompe-13, MPS1-3, Gaucher-2, Fabry-0 (Fig 5). In **Pompe** disease, the main clinic reasons for the screening were: hypotonia, scoliosis, scapula alatta, hypertrophic cardiomyopathy, weak muscle and high creatine kinase. In **Gaucher** disorder, the reasons for the test were spontaneous fractures and pancytopenia. In **MPS 1**, the reasons for the spot test examination were corneal clouding, coarse face and hypotonia . We didn't found any indication for **Fabry** screening. We found one case of MPS 1 and is now under ERT. The other case was a DBS positive for Pompe disease (Fig. 6)

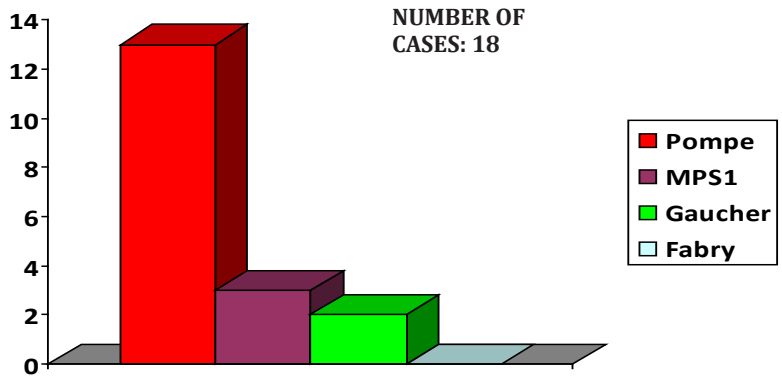


Figure 5- *Number of Cases Screened in Dried Blood Spot*

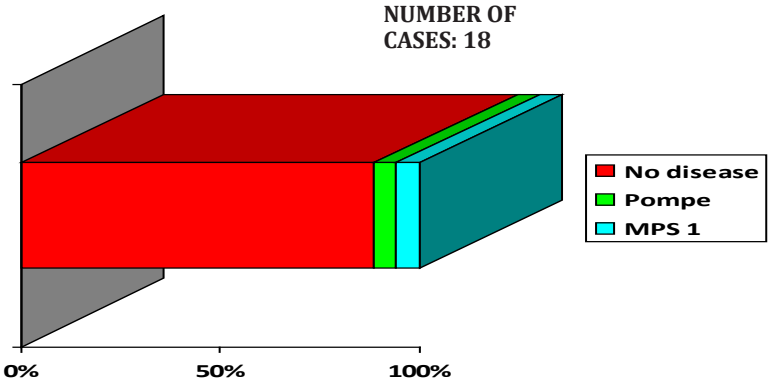


Figure 6 – *Number of Disorders Detected*

Conclusions

Pompe, Fabry, Gaucher and MPS, are disorders that can be under diagnosed because of the multisystem involvement of many organs.

In our first year experience we confirmed a MPS 1 case that is now under treatment and another case of Pompe disease.

Reference

[1] Lysosomal storage diseases Ferreira CR, Gahl WA Transl Sci dis 2017,May 25;2:1-71

[2] Methods for determination of alfa-glycosidase, beta-glycosidase and alfa-galactosidase activities in dried blood spot samples Sozmen EY, SEzer ED Methods Mol. Biol. 2017;1594: 255-264

[3] Inherited metabolic disorders: efficacy of enzyme as says on dried blood spots for the diagnosis of lysosomal storage disorders VermaJ, Thomas DC, Kasper DC, Sharma S, Puri RD, Bijarnia-Mahay S, Mistry PK, Verma IC JIMD Rep 2017;31:15-27.

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