Activity of Alpha-Glucosidase in Dried Blood Spots and Fibroblasts From Healthy Controls and Pompe Disease Patients

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Pompe’s Disease (PD) is a lysosomal storage disorders (LSDs) autosomal recessive, rare and progressive. In PD, the enzyme acid alpha-D-glucosidase (GAA) is deficient leading to an accumulation of glycogen in the lysosomes of cells. Early diagnosis of PD is an essential step of the efficiency of the treatment, since the enzyme replacement therapy for PD is available. For this, has been developed screening methods which measure the activity of lysosomal enzymes is performed directly on dried blood spots (DBS). However, the confirmatory diagnosis for PD is performed in fibroblasts because they do not present a neutral alpha-glucosidase isoenzyme. In this work, the aim was to compare the activity of GAA in samples of DBS and fibroblasts from healthy controls and PD patients. We used samples from volunteer health donors and PD patients. The activities in DBS and fibroblasts were performed according to Castilhos et al. (2011) and Droadhead & Butterworth (1977) and compared used Student’s t test. The mean activity of the enzyme in samples from healthy controls was 10.06 ± 5.83 nmol/20h/mL (DBS) and 155.40 ± 92.29 nmol/h/mg prot (fibroblasts) whereas for the PD patients, the mean activities were 0.09 ± 0.15 nmol/20h/mL (DBS) and 1.45 ± 1.61 nmol/h/mg prot (fibroblasts), demonstrating a significant difference between the groups (p < 0.05). Thus, our data shows that the groups can be clearly differentiated and the DBS is a great material for screening tests of this disease.

Word Keys: Dried Blood Spots, Lysosomal Disease, Pompe Disease

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