Alpha-galactosidase A: Comparison Between the Activity in Dried Blood Samples on Filter Paper, Leukocytes and Plasma

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The enzyme alpha-galactosidase A (GLA) is a lysosomal hydrolase that degrades glycosphingolipids. A deficiency of this enzyme and the symptoms resulting from the accumulation of its substrate characterize Fabry disease (FD), an X linked recessive genetic disease. Considering the difficulty around the establishment of a conclusive and accurate biochemical diagnosis of FD, the objective of this study was to compare the measurement of GLA activity using dried blood samples on filter paper (DBS), plasma and leukocytes of FD patients and healthy individuals. We used 9ml blood with heparin for obtaining DBS, leukocytes and plasma samples. GLA activity was determined by fluorometric assays using the different sample materials. For DBS average activity for the healthy controls was 6.02 ± 2.84 nmol/h/mL for patients with DF was 1.25 ± 0.80 nmol/h/mL. In plasma of healthy controls was 6.78 ± 1.71 nmol/h/mL and for patients was 1.30 ± 1.79 nmol/h/mL, while activities for leukocytes were respectively 29.13 ± 11.12 nmol/h/mg prot and 5.13 ± 3.39 nmol/h/mg prot. The correlation between DBS and plasma was significant, r=0.52 and p<0.005, as well as between plasma and leukocytes (r=0.72 and p<0.0001). There was no significant correlation between the results in leukocyte and DBS (p>0.05). There is a correlation between the GLA activity obtained for most of the different samples, leading to good agreement between the different techniques. Activity in DBS is used for screening and the results demonstrate the need to improve this technique for diagnostic purposes.

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