P-049 - BETA-GLUCORONIDASE QUANTIFICATION IN DBS

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INTRODUCTION: Beta-glucuronidase (GUSB), is a lysosomal enzyme in charge of hydrolyzing the glycosidic bond, releasing glucuronic acid present in the structure of complex carbohydrates. Specifically the enzyme participates in the glycosaminoglycans degradation. GUSB deficiency results in the presentation of a skeletal dysplasia known as Mucopolysaccharidosis type VII (MPS VII) or Sly syndrome. AIM: This study shows the implementation of a fluorometric method of quantification of GUSB in DBS and the construction of reference values for our population. MATERIALS AND METHODS: DBS samples were obtained from healthy subjects of different ages and both sexes, prior signed informed consent. For the quantification of GUSB a modification of Civallero 2006 was used. We establish reproducibility, stability and sensibility of the technique using normal and affected individuals. RESULTS: Once the enzyme test was standardized by modifying the Civarello method, an intraassay and interoperative variation coefficient of 5% was achieved, the samples stability was tested for up to 210 days at 4 °c or room temperature; the reference values range for our population were established using of 79 normal samples (50.8-173.7 nmol/h/ml). CONCLUSIONS: The study allowed verifying that the enzyme beta-glucuronidase is highly stable in dry blood samples over filter paper. The samples can be stored at 4 °C or at room temperature for up to 8 months. In addition, the reference values for our population were established providing the medical community with an additional tool for preliminary evaluation in the diagnostic process of patients with suspected mucopolysaccharidosis type VII.