P-087 - ENZYMATIC ALPHA-GALACTOSIDASE A (GLA) AND BETA-GLUCURONIDASE (GUSB) RELATION USED TO SUPPORT DIAGNOSTIC IDENTIFICATION IN WOMEN WITH FABRY DISEASE.

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Fabry disease is an sphingolipid metabolism disorder affecting enzymatic activity of GLA. The disorder causes a multisystemic affection with X-linked chromosome (xq22.1) heredity. With an estimate range of 1: 40.000 to 1: 120.000 in men and 1: 117.000 in women both with Gaucher disease are considered the most prevalent sphingolipidoses worldwide. Classic male diagnostic approach has been done demonstrating low enzymatic activity levels of GLA on DBS screenings, followed by confirmation of the screened positive results in leucocytes. Carrier women diagnostic has been more challenging due to the random inactivation of X chromosome causing variable enzymatic activities that can be comparable to normal values until 40% of cases. In this work we present a preliminary approach for a possible identification of carrier women with Fabry disease by means of an analysis of the relation between GLA and beta-glucuronidase performed on DBS and leucocytes. MATERIALS AND METHODS: We quantified the activity of GLA and GUSB by final point fluorometry. DBS and leucocyte samples were obtained from 25 male and 25 female normal controls (ages between 15 and 40 years) to establish normal range. Similar samples were obtained from 14 Fabry confirmed males, 2 obliged carrier females (mothers of affected patients) and 14 patients (both female and male) with clinical suspicion of Fabry disease. RESULTS: Reference ranges obtained for GLA/GUSB relation on DBS: 0.04-0.21 and on leucocytes: 0.1-0.4. Affected males: 0.01-0.08 on DBS and 0.01-0.032 on leucocytes. Obliged carrier females (DBS): 0.034 y 0.038. Females with clinical suspicion presented values on DBS <0.038 and on leucocytes: 0.16. Obliged carrier values were under lower normal controls limit. Is possible that this relation could be a useful tool to better conduct the analysis for female carriers of Fabry disease. More studies are needed to establish this possibility.