P-042 - SCREENING OF MUTATION C.1360 C> T IN THE HGSNAT GENE IN TUNJA, BOYACÁ - COLOMBIA.

Bernal IT ¹, Velasco HM ¹, Ahumada MC ², Sánchez Y ³, Cruz LV ², Usaquén W ¹

(1) Institute of Genetics, National University of Colombia. (2) San Rafael Hospital of Tunja. (3) Pedagogical and Technological University of Colombia. Bogotá-Colombia ibernalb@unal.edu.co.

INTRODUCTION: Mucopolysaccharidoses (MPS) are an heterogeneous group of genetic diseases that belong to lysosomal storage disorders. These diseases are characterized for multisystemic affectation associated with progressive motor disability, variable cognitive deficit and decrease in life expectancy. In Colombia, a population cluster was found in Boyacá (Villa de Runta) for MPS IIIC, located with in 5 minutes from the department capital (Tunja) and has an estimated incidence of 1: 200 live births, where the main variant found was g.1360C > T in HGSNAT gene. Given the high incidence of this subtype of MPS in Boyacá, it is considered essential to study the population frequency of the mutation c.1360C> T in HGSNAT gene in Tunja city. OBJECTIVES: To analyze the pathogenic variant c.1360 C> T in HGSNAT gene in 400 individuals of Tunja population, from neonatal TSH dried blood spots, to calculate the frequency of heterozygotes in Tunja. MATERIALS AND METHODS: Cross-sectional study in neonatal screening samples, after signing informed consent. The detection of the different genotypes in HGSNAT gene is carried out by High Resolution Melting - PCR (HRM-PCR); confirming carriers and affected by sequencing. RESULTS: 108 samples analyzed allowed the detection of 2 patients with melting curves similar to that observed in the genotype of the heterozygous control, and 3 patients with melting curves similar to that observed in the genotype of the homozygous mutant control. 103 samples have a melting curve pattern similar to the wild genotype. DISCUSSION: This cost-effective technique has allowed the rapid detection of non-wild type genotypes in a population at high risk for autosomal recessive diseases. These studies are valuable tools to evaluate the need to create specific programs for the study of carriers in inbreeding regions like Boyacá.