P-225 - MOLECULAR CARACTERIZATION OF RARE HEMOGLOBINES FOUND BY NEWBORN SCREENING

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INTRODUCTION: In 2013, Uruguay started a nonselective pilot program for newborn screening for hemoglobinopathies. Since then, many variant trait hemoglobin have been found by CE-HPLC. Many clinical complications have been reported due to the presence of rare hemoglobins, even thought if it were found as a trait. This fact gives the importance of their characterization. In consequence, Newborn screening Laboratory in Uruguay is evaluating the incorporation of molecular studies for α and β gen as part of the diagnosis. Objetive Present the results obtained from samples analyzed as a pilot program for molecular studies of rare hemoglobin.

MATERIALS AND METHODS: Molecular studies were performed on seven patients detected by Newborn Screening. All of them presented a HPLC profile of carriers of a rare hemoglobin. Genomic DNA was extracted from the same samples used to the screening studies, obtained from heel prick on filter paper Whatman 903. All exons from β, α1 and α2 and its corresponding intronic sequences were amplified by PCR (polymerase chain reaction), purified and analyzed in an automatic sequencer ABI 310 from Applied Biosystems. Every mutation profile was compared with the databases Ensembl and HGMD. RESULTS: We found five mutations on β gene of hemoglobin chain, one in α1 and one in α2. In gene β we found three different mutations, 20 A>T (Glu6Val) present in three cases, 19 G>A (Glu6Lys) and 23 A>G (Glu7Gly). Moreover, 137A>G (His45Arg) was present in α1 exon 2, and 134C>G (Pro44Arg) in α2 exon 2. All mutations were detected in heterozygosis as newborn screening suggested. These hemoglobins were found in databases like Hb S, Hb C and G-San José, Hb Fort de France, Hb Kawachi respectively. CONCLUSION: Rare hemoglobins are frequent in Uruguayan population, so molecular studies are very important and helpful to characterize these cases. However, to complete the molecular profile is necessary to incorporate thalassemia studies.