

Original Paper

Molecular evaluation of hemoglobin D mutations in Mazandaran province, Iran

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Abstract

Background and Objective: Hemoglobinopathies are among the most prevalent genetic disorders worldwide, and occur as a result of mutations in the gene involved in synthesizing hemoglobin chains. By now more than 1000 defects in hemoglobin chains are discovered. Hemoglobin D (Hb D) is one of these disorders, identified by a single nucleotide mutation on codon 121 of beta globin chain. This study was carried out to evaluate Hb D mutations through molecular methods in Mazandaran province of Iran.

Materials and Methods: This descriptive laboratory study was done on 70 patients with an electrophoresis band in hemoglobin-S zone in Mazandaran province of Iran during 2010-11. Capillary zone electrophoresis was done to find out Hb D in 51 patients. Subsequently, PCR-RFLP was performed to evaluate the samples at molecular level.

Results: Molecular investigation revealed all cases are carriers of hemoglobin D-Punjab. Two patients were shown to be homozygote carriers of the abnormal gene.

Conclusion: This study showed all Hb D affected patients were carriers of Hb D Punjab.

Keywords: Hemoglobin D Punjab, PCR-RFLP, Hemoglobinopathy, Genetic mutation

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Received 11 March 2012

Revised 4 August 2012

Accepted 8 August 2012