Int J Hum Genet, 1(2): 97-100 (2001)
PRINT: ISSN 0972-3757 ONLINE: ISSN 2456-6330
DOI: 10.31901/24566330.2001/01.02.04

Structural Hemoglobin Variants: Mutation, Hematology and Its Application in Prenatal Diagnosis

Sarita Agarwal, M. Pradhan, U.R. Gupta, R.S. Yadav and S.S.Agarwal

Department of Genetics, Sanjay Gandhi Post Graduate Institute of Medical Sciences, Lucknow 226 014, Uttar Pradesh, India

KEY WORDS Hemoglobin variants; prenatal diagnosis; mutation; polymerase chain reaction; thalassemia; hematology.

ABSTRACT Referred cases of Genetics OPD, SGPGI between 1988-2000 were screened for cause of anemia. Out of which one hundred and ninety eight individuals were found as abnormal hemoglobin variants and associated with ß-thalassemia mutations viz: EßT, SßT, DßT. These phenotypes were of severe type. Hematology and mutation analysis were performed in these subjects by ARMS-PCR technique. Due to high prevalence of IVS I-5 (G-C) mutation, most of the structural variants (69%) were found associated with this mutation. Severity of the thalassemia syndromes (EßT, SßT, DßT) emphasized the need of establishment of prenatal diagnosis for common structural hemoglobin variants along with beta thalassemia mutations.

Home Back