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The Molecular Basis of Alpha-Thalassaemias in India: A Review

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KEYWORDS α-Thalassaemias. Haemoglobinopathies. Variation, Ethnic Groups

ABSTRACT Thalassaemias and haemoglobinopathies are the most common genetic disease and impose a major burden on the population of India due to its high degree of morbidity and moderate to severe anaemia among different segments of the society. Amongst the population of 1000 million at the new millennium (2000) forty-five million carriers and fifteen thousand infants with major haemoglobinopathies have been reported in India. A highly heterogenous distribution of a-thalassaemia mutations have been reported in different parts of India. Migration, and gene flow of the mutant alleles of α- thalassaemias by social, political and commercial reasons from different populations of the world are possibly responsible for these heterogenous nature of these mutations. Our study on α-thalassaemias with an special emphasis on the detection of –3.7 and –4.2 deletions showed remarkably high incidence of this disease among the tribal population in Eastern India. We have also reported high prevalence of this disease among nontribal Bengalees in Kolkata and adjacent areas. This article provides a glimpse on the epidemiology, occurrence and different kinds of α- thalassaemia mutations in different parts of India.