

Title:

Genetic Screening of Thalassemia in region of Gujrat.



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Abstract

Thalassemia is an autosomal recessive blood disorder caused by mutations in the globin gene, which result in insufficient hemoglobin production. Like many parts of Punjab, Gujarat has a high prevalence of this disease, largely as a result of consanguineous marriages and low public awareness. Despite breakthroughs in molecular diagnostics, public awareness is still low, and genetic testing is not generally available. The lack of structured screening programs and genetic counseling has contributed to the continuing transmission of thalassemia in the area, resulting in an increasing number of children born with thalassemia major.

The major goal of this study is to determine the causative variation in the HBB gene in thalassemia affected families from Gujarat. Five families affected by thalassemia will be registered. Blood samples will be collected from the diseased and normal individuals with informed consent. DNA will be extracted using standard organic method. Primers will be designed for HBB gene. PCR will be performed, and amplicons will be sequenced. Data will be analyzed for mutational assessment. Families will be informed about the causative mutation and counselling session will be arranged for the betterment of the affected family.