**Original article:**

**Screening and Molecular Characterization of β-Thalassaemia Mutations in Parents and Siblings of β-Thalassaemia Major Patients**

**Amit Kumar Mishra \*, Archana Tiwari**

School of Biotechnology, UTD, Rajiv Gandhi Proudyogiki Vishwavidyalaya, Airport Road, Bhopal, Madhya Pradesh, India

Corresponding author - Email : btech\_amit@yahoo.com

**Abstract**

**Background:** Hemoglobinopathies are priority genetic diseases for prevention programs. Beta thalassaemia major is one of the single gene blood disorders worldwide. It is also a major health concern in India. Screening of carriers all the way through diverse screening approach is the only way to prevent birth of thalassaemia major child.

**Aims & Objective:** This study was done to screen and molecularly characterize β-thalassaemia mutations in the parents and siblings of thalassaemia major index cases using amplification refractory mutation system polymerase chain reaction for the common Indian mutations and also as an alternative approach to population based screening program for identifying thalassaemia carriers to prevent birth of thalassaemic children in the family members of a thalassaemia index family.

**Material & Methods:** Blood samples were collected from thirty families of thalassaemia index cases. Fifty samples from parents and thirty three siblings of them were given their samples for thalassaemia carrier screening and molecular characterization of five common Indian β-thalassaemia mutations using amplification refractory mutation system polymerase chain reaction.

**Observation & Results:** Seventy five (90%) cases of heterozygous beta thalassaemia were detected in the survey of 83 samples of parents and siblings having beta thalassaemia major children.

**Conclusion:** Screening of siblings of thalassaemia major cases is necessary and facilitate detection of carriers ultimately helps in prevention of birth of Thalassaemic child.

**Keywords:** ARMS PCR; β-thalassaemia mutations; immediate family members; molecular classification