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BETA THALASSAEMIA MUTATION IN POPULATION OF BHOPAL, CENTRAL INDIA

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ABSTRACT

Thalassaemia is a major haemoglobinopathy of wide occurrence in the Indian sub-continent. It is distributed to different degrees in different sub-populations. The treatment of this disorder is quite expensive and counseling seems to be the only way for controlling it. Genetic analysis for Beta-thalassaemia blood disorder is detected by ARMS(Amplification Refractory Mutation System) tecnique. A total number of 43 cases obtain from OPD of Gandhi Medical College & Delta Pathology Bhopal were tested for this disorder. Out of seven common Beta thal mutation (D.J. Weatherall & J.B. Clegg), IVS1 nt 5 (G→C), IVS1nt 1(G→T), 619 bp deletion, Cap+1(A→C), -88 C→T,Fr 8-9(+G), Fr 41-42(-4bp) 39.52%, 16.27%, 18.59%, 6.97%, 0%, 0%, 0% respectively found in population of Bhopal. Early detection of thalassaemia is therefore important not only from treatment point of view, but also for the prevention by genetic counseling.

Key words: Beta-thalassaemia, Central India, Mutation, PCR, Genetic Counseling and Prenatal Diagosis.

Thalassaemia is one of the major hereditary disorder involving haemoglobin in the human blood, which account for high mortality in childhood. It is estimated that these are over 25 million carrier of Beta-thalassaemia genes in India with mean prevalence of 3.3% (Verma *et al*., 1992) and 6000 to 8000 chidren are born every year with thalassaemia major (Varawalla *et al*., 1991). Only 10 to 15% of these children receive optimal treatment (Choudhry *et al*., 1991). The cost of treatment for one thalassaemic child amount to Rs.90,000 to 1,00,000 annually at around three year of age, which increase as the child grows. The only cure available today is bone marrow transplantation which is not affordable to almost all of our patients. It is necessary to identify individuals with beta thalassaemia trait (BTT) for several reasons: to avoid unnecessary iron medication for mild hypochromic anaemia caused by BTT misdiagnosed as iron deficiency anemia, to avoid marriages of two individuals with BTT to prevent the possibility of progeny are found to be BTT carriers, to advise them of the risk of having children and to opt for parental diagnosis. As majority of individuals with BTT are asymptomatic, a population survey is the best way to identify them.

Methodology

Mutation present in the population of Bhopal, Central India and in carrier parents seeking parental diagnosis were detected by PCR-based technique of ARMS (Amplification Refractory Mutation System). Take 6 ml whole blood for molecular study. DNA extraction by standard method.

Observation

Mutation for beta thalassaemia found in population of Bhopal are Del 619 bp, IVS1 nt 5 (G→S), IVS1 nt 1 (G→T), Cap + 1 (A→C).

Four different beta thal. mutation have been found in population of Central India are IVS1 nt (G→C) IVS1 nt 1 (G→T). Cap + 1 (A→C), Del 619 bp. In Sindhi Colony community Del 619 bp, Jain community IVS1 nt 5 (G→C) and Muslim Community have Cap +1 (A→C) mutation in Bhopal (Table -1). Population of this region is conscious and willing to accept parental diagnosis as a mean of control of thalassaemia.

Table - 1: Beta thalassaemia mutation in population of Bhopal

Type of mutation	Beta thal major (%)	Beta thal minor (%)	Beta thal intermedia (%)
IVS1 nt 5 (G→C)	11.62	27.90	Nil
IVS1 nt 1 (G→T)	4.65	11.62	Nil
619 bp deletion	2.32	16.27	Nil
Cap + (A→C)	4.65	2.32	Nil
-88 C → T	Nil	Nil	Nil
IVS1 nt 5 (G→C) & Cap+1	Nil	4.65	2.32
IVS1 nt 1 (G→T) & Cap+1	2.32	2.32	Nil
IVS1 nt 5 (G→C) & -88	2.32	Nil	Nil
IVS1 nt 1 (G→T) & 619 bp del.	2.32	Nil	Nil
IVS1 nt 5 (G→C), IVS nt (G→T) & Cap+1	Nil	2.32	Nil
Total (No. of cases)	30.23	67.44	2.32

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