THE EGYPTIAN JOURNAL OF IMMUNOLOGY Vol. 12 (1), 2005
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Molecular basis of beta-thalassemia in Alexandria

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PMID: 16734135

Thalassaemia is a group of inherited haemoglobin disorders characterized by reduced synthesis of one or more of the globin chains leading to imbalanced a /non-a globin synthesis which is the major factor in determining the severity of the disease in the thalassaemia syndromes. In Egypt, beta-thalassemia is the commonest cause of chronic haemolytic anaemia and it represents a major genetic disease and a public health problem. This study included 50 transfusion dependent beta-thalassaemic cases. They were subjected to detailed history taking, physical examination to assess the size of liver and spleen and laboratory investigations including complete haemogram, bone marrow (BM) aspiration, haemoglobin electrophoresis and serum ferritin. Genetic analysis for detection of point mutations was done by PCR amplification refractory mutation system (ARMS) which is a PCR method based on allele specific priming. Using this technology, it was possible to characterize mutations in 73% of beta-thalassaemia cases, while 27% remained uncharacterized. The five common mutations used were: IVS-1-110 (G-->A), IVS-1-6 (T-->C) IVS-I-1 (G-->T), IVS-II-1 (G-->A) and codon 39 (C-->T). The commonest was IVS-I-110 (62%) followed by IVS-1-6 (7%), then IVS-I-1 (4%). On the other hand mutations such as IVS-II-1 and Cd- 39 were not found in any of our patients. No significant difference was found between different genotypes regarding the frequency of blood transfusion needed, degree of anaemia and microcytosis, HbF% or serum ferritin levels. This may be due to the small sample size of some of the genotypes (IVS-I-110/IVS-I-1 & IVS-I-110/IVS-I-6) or due to repeated blood transfusions which mask the patient original CBC and Hb electrophoresis pattern or due to co-inheritance of other genetic modifying factors that alter the typical phenotype.