A novel frameshift mutation causing β-thalassaemia in Azerbaijan


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The human hereditary syndrome of β-thalassaemia is caused by a number of mutations located in β-globin gene mostly between its TATA-box and second intron |1|. Some β-thalassaemia-inducing mutations found in various regions of the world were identified also in Azerbaijanian population |2, 3|, main areal of this disease in the USSR. Now we have determined the molecular nature of a novel mutation causing β°-thalassaemia.

A segment of β-globin gene (329 bp) comprising end of first intron (42 bp), second exon (223 bp) and beginning of second intron (64 bp) in DNA from leukocytes of peripheral blood of an Azerbaijanian patient with β°-thalassaemia was amplified by means of PCR |4| with synthetic primers 5'-dCACTGACTCTCTCTGCC-TAT-3' and 5'-dTATGACATGAACTTAACCAT-3' (one of them 5'-32p-labelled) and thermostable DNA polymerase from *Thermus thermophilus* |5|. The sequencing of both strands by Maxam-Gilbert method led to the identification of a previously unknown mutation, viz., G deletion in β2/83 codons AAGGGC; the frameshift, resulting in the formation of a nonsense codon TGA 13 nucleotides downstream, prevents synthesis of nature β-globin. Further studies will allow to see, which mutations dominate in that region.

REFERENCES