The Delta-Beta-Crossing-Over Site in the Fusion Gene of the Lepore-Boston Disease Might Be Localized in a Preferential Recombination Region

Yahia Cheboune, Guy Trabuchet, Didier Poncet, Michel Cohen-Solal, Claudine Faure, Geard Verdier, Victor Nigon

TY - JOUR
AU - CHEBLOUNE, Yahia
AU - TRABUCHET, Guy
AU - PONCET, Didier
AU - COHEN-SOLAL, Michel
AU - FAURE, Claudine
AU - VERDIER, Géard
AU - NIGON, Victor M.
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AB - Cloned DNA fragments were subcloned in filamentous coliphages fd 103 or M 13; the recombinant single-stranded DNAs were then used to form hybrids with genomic DNA as well as with complementary recombinant single-stranded DNA. Hybrids were submitted to S1-nuclease treatment alone or in combination with restriction enzyme digestions. This method was used to analyze the δ-? globin gene cluster from the total genomic DNA of a ?°-thalassemic patient. A modification located approximately 530 base pairs upstream from the cap site of the ?-globin gene was detected in only one thalassemic chromosome of this patient. Sequence analysis have shown that the patient was homozygous for a single nucleoside change (dC?dT) which remains undetected by our hybridization method, leading to a codon 39 nonsense mutation; they have demonstrated too that he was heterozygous for the modification mentioned and detected by S1-nuclease, which corresponds to an additional sequence d(T-A-T-A) in a 52 alternating purine-pyrimidine run, leading to a complex change from d[(A-T)7(T)7] to d[(A-T)11(T)3].
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