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

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► **To cite this version:**

Yahia Chebloune, Guy Trabuchet, Didier Poncet, Michel Cohen-Solal, Claudine Faure, et al.. The Delta-Beta-Crossing-Over Site in the Fusion Gene of the Lepore-Boston Disease Might Be Localized in a Preferential Recombination Region. *Acta Haematologica*, 2004, 69 (5), pp.294-302. 10.1159/000206910 . hal-02965334

HAL Id: hal-02965334

<https://hal.inrae.fr/hal-02965334>

Submitted on 13 Oct 2020

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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.
TI - A new method for detection of small modifications in genomic DNA, applied to the human δ - β globin gene cluster
JO - European Journal of Biochemistry
VL - 142
IS - 3
SN - 0014-2956
UR - <https://doi.org/10.1111/j.1432-1033.1984.tb08310.x>
DO - doi:10.1111/j.1432-1033.1984.tb08310.x
SP - 473
EP - 480
PY - 1984
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 - β globin gene cluster from the total genomic DNA of a β^0 -thalassemic patient. A modification located approximately 530 base pairs upstream from the cap site of the β -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 \rightarrow 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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