# Homozygous Mutation on the β-Globin Polyadenylation Signal in a Tunisian Patient with β-Thalassemia Intermedia and Coinheritance of Gilbert's Syndrome

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**Abstract:**

We report here the clinical, hematological and molecular data in a 50-year-old patient with β-thalassemia intermedia (β-TI) caused by a homozygous β+ mutation on the β-globin gene polyadenylation (polyA) signal (AATAAA>AAAAAA). β Haplotype analysis was accomplished by polymerase chain reaction-restriction fragment length polymorphism (PCR-RFLP). Haplotype and framework analysis showed that this mutation is associated with the [βββββ+++] β haplotype and framework 1 (CCGCT) (FW1). This mutation was previously reported in the heterozygous state in association with the codon 9 (+TA) mutation in a β-TI patient originating from Tunisia. To the best of our knowledge, this is the first report describing this mutation in the homozygous state. The case reported here, coinherited Gilbert's syndrome, which is characterized by hyperbilirubinemia. This conclusion was reached by the investigation of the promoter region [A(TA)nTAA] motif of the UGT1A1 gene, showing the (TA)6/(TA)7 genotype.

**Biography:**

Nawel Trabelsi is a Laboratory of Molecular and Cellular Hematology, Pasteur Institute of Tunis-Tunis EI Manar Unviversity, Tunis, Tunisia. Faculté des Sciences Mathématiques, Physiques et Naturelles de Tunis.

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