Identification of three novel variants in the UGT1A1 gene as a cause of Crigler-Najjar syndrome type 1

Mohammad Javad Ghorbani[1] and Seyed Mohsen Dehghani[1]

[1]Shiraz University of Medical Sciences

June 11, 2022

Abstract

Crigler-Najjar syndrome type I (CN-I) is a rare inherited disorder with a frequency of one per million at birth. Patients with CN-I have severe hyperbilirubinemia and usually die due to kernicterus. CN-I occurs due to variants within the UGT1A1 gene. The present study aimed to identify genetic defects underlying CN-I.

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`Ghorbani-postdoc.docx` available at https://authorea.com/users/488462/articles/572621-identification-of-three-novel-variants-in-the-ugt1a1-gene-as-a-cause-of-crigler-najjar-syndrome-type-1