عنوان مقاله:

Genetic Association of UGT۱A۱ Promoter Variants (c.-۳۲۷۹T>G and c.-۳۱۵۶G>A) with Neonatal Hyperbili-rubinemia in an Iranian Population

محل انتشار:

مجله علمی ناباروری ایران, دوره 12, شماره 2 (سال: 1400)

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خلاصه مقاله:

Background: Several studies have reported that two promoter variants (c.-۳۲۷۹T>G and c.-۳۱۵۶G>A) in UDP-glucuronosyltransferase (UGT۱A۱) gene may contribute to neonatal hyperbilirubinemia. However, these variants have not been investigated in Iranian neonates. This cross-sectional study aimed to determine if the UGT۱A۱ promoter variants are significant risk factors associated with neonatal hyperbilirubinemia. Methods: A total of ۱۷۸ unrelated neonates, including newborns with neonatal jaundice (n=۹۵) and healthy controls (n=۸۳), were included in this study. Each individual was genotyped by the PCR-RFLP and COP-PCR at nucleotides -۳۲۷۹ and -۳۱۵۶, respectively, using fresh blood DNA. Logistic regression analyses were performed to assess the association of UGT۱A۱ promoter variants with the presence of significant hyperbilirubinemia. Anthropometric indices and clinical variables were also compared between the different genotype groups. Results: Allele and genotype analysis of the c.-۳۲۷۹T>G and c.-۳۱۵۶G>A variants showed no significant association with the risk of neonatal hyperbilirubinemia neither in the crude nor after adjustment for gestational age, gender, and birth weight in different genetic models (P>۰.۰۵). However, in haplotype-association analysis, only one haplotype (A-T) was found to be associated with the risk of neonatal hyperbilirubinemia (OR=۰.۱۹, ۹۵% CI; [۰.۱۸–۰.۲۰], P=۰.۰۰۱). Conclusion: This study failed to demonstrate that c.-۳۲۷۹T>G and c.-۳۱۵۶G>A variants alone might contribute to the risk of neonatal hyperbilirubinemia in Iranian neonates. However, the A-T haplotype may play a significant role in increasing the risk of hyperbilirubinemia.

کلمات کلیدی:

Hyperbilirubinemia, Kernicterus, Polymorphism, UGT۱A۱