عنوان مقاله:

Molecular Identification of G6PD Chatham (1003 G>A) in North-West Iran

محل انتشار:

فصلنامه زیست پزشکی جرجانی, دوره 8, شماره 3 (سال: 1399)

تعداد صفحات اصل مقاله: 9

نویسندگان:

Habib Onsori - *Cellular and Molecular Biology Department, Marand Branch, Islamic Azad University, Marand, Iran*

Davood Poladi - *Department Genetics, Ahar Branch, Islamic Azad University, Ahar, Iran*

Mehdi Valizadeh - *Unit of Genomics Research, Digestive Diseases Research Center, Ardabil University of Medical Sciences, Ardabil, Iran*

Afshin Fathi - *Pediatric Hematology & Oncology Department, Ardabil University of Medical Sciences, Ardabil, Iran*

Mahshid Damandan - *Center for Cell Pathology Research, Department of Life Science, Khazar University, Baku, Azerbaijan/Cellular and Molecular Research Center, School of Medicine, Ardabil University of Medical Sciences, Ardabil, Iran*

Rouhallah Moradpour - *Cellular and Molecular Research Center, School of Medicine, Ardabil University of Medical Sciences, Ardabil, Iran/Center for Cell Pathology Research, Department of Life Science, Khazar University, Baku, Azerbaijan*

خلاصه مقاله:

Background and objective: Glucose 6-phosphate dehydrogenase (G6PD) deficiency is one of the most common human diseases with approximately 400 million people affected worldwide. G6PD Chatham is caused by 1003 G>A mutation leads to a severe enzymatic deficiency. The aim of the present study is to investigate the frequency rate of the Chatham mutations in the population of the North-West of Iran. Material And Method: In this study, by Rapid Genomic DNA Extraction (RGDE) method, from 90 peripheral blood samples of unrelated male and female patients with genetic deficiency of G6PD, DNA was extracted and after digestion by Bstx1 enzymes, in order to search for Chatham mutation, they were analyzed by means of PCR-RFLP and sequencing methods. Result: According to the results, Chatham mutation was observed in 10 samples (11.11%). Conclusion: This study showed that G6PD Chatham (1003 G>A) mutation is the second common mutation, after Mediterranean (563C>T), in the population of .the North-West of Iran. Further studies are recommended to identify the mutation type of other varieties

کلمات کلیدی:

Glucose 6-phosphate dehydrogenase (G6PD), Chatham mutation, PCR-RFLP method, Sequencing