عنوان مقاله:

A novel mutation in AGL gene causes Glycogen storage disease type III

محل انتشار:

چهارمین کنگره بین المللی و شانزدهمین کنگره ملی ژنتیک (سال: 1399)

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خلاصه مقاله:

Background and Aim: Glycogen storage disease type III (GSDIII) is a rare autosomal recessive disorder caused by deficiency of the glycogen debranching enzyme. (amylo-۱,۶-glucosidase, ۴-alpha-glucanotransferase (AGL)) activity. GSD III, also known as Forbes or Cori's disease, is characterized by the accumulation of abnormal glycogen in the liver and muscles.Methods: We collected ۵-mL of peripheral blood from a patient presenting GSD symptoms. DNA was isolated using salting out method. The coding region and intron–exon boundaries of the AGL were amplified using PCR. PCR purification and bidirectional Sanger sequencing of the PCR products were done.Results: We identified a novel substitution; c.۳۶۸۲C>T, p.Arg۱۲۲۸X. According to the insilico analysis we suggest the variation is probably damaging.Conclusion: A novel mutation in the AGL gene, c.۳۶۸۲C>T in an Iranian affected person was identified. This mutation affects the C-terminal region of the protein. Mutation of this region disrupts glucosidase activity of the .enzyme. It should be considered in genetic assessment of the patients suffering GSD in Southwest of Iran

کلمات کلیدی:

GSDIII, AGL, c.۳۶۸۲C>T, Southwest of Iran

لینک ثابت مقاله در پایگاه سیویلیکا: