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D28G mutation in congenital glucose-galactose malabsorption

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Abstract

Background: Congenital glucose-galactose malabsorption is a rare autosomal recessive disorder of the intestinal transport of glucose and galactose, leading to watery diarrhea, dehydration, failure to thrive, and early death. Methods: In this study, we analyzed D28G mutation in 16 family members of a patient with typical presentation of congenital glucose-galactose malabsorption with polymerase chain reaction-Restriction Fragment Length Polymorphism method. Results: Nine members of this family were heterozygous for D28G mutation. Conclusion: To the best of our knowledge this is the first report of D28G mutation in Iran. Moreover, this simple typical PCR-Restriction Fragment Length Polymorphism method, allows immediate identification of D28G mutation.

Reaxys Database Information

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Author keywords

Congenital glucose-galactose malabsorption (CGGM); D28G; PCR-RFLP

Indexed Keywords

EMTREE drug terms: aspartic acid; galactose; glucose; glycine; sodium glucose cotransporter 1

EMTREE medical terms: article; clinical article; congenital glucose galactose malabsorption; diarrhea; diet therapy; early diagnosis; family; female; follow up; gene mutation; heterozygote; human; Iran; malabsorption; mutational analysis; pedigree analysis; polymerase chain reaction; restriction fragment length polymorphism

MeSH: Aspartic Acid; Female; Galactose; Glucose; Glycine; Humans; Infant, Newborn; Malabsorption Syndromes; Male; Mutation; Pedigree; Polymerase Chain Reaction; Polymorphism, Restriction Fragment Length; Sodium-Glucose Transporter 1
Medline is the source for the MeSH terms of this document.

Chemicals and CAS Registry Numbers: aspartic acid, 56-84-8, 6899-03-2; galactose, 26566-61-0, 50855-33-9, 59-23-4; glucose, 50-99-7, 84778-64-3; glycine, 56-40-6, 6000-43-7, 6000-44-8;Aspartic Acid, 56-84-8; Galactose, 26566-61-0; Glucose, 50-99-7; Glycine, 56-40-6; Sodium-Glucose Transporter 1

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