عنوان مقاله:

Genetic diagnosis of Congenital hyperinsulinism in a deceased affected child: a case report

محل انتشار:

اولین همایش علوم پایه دربیماریهای ارثی کودکان (سال: 1398)

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خلاصه مقاله:

Introduction: Congenital hyperinsulinism is a condition that causes individuals to have abnormally high levels of insulin. People with this condition have frequent episodes of low blood sugar (hypoglycemia). In infants and young children, these episodes are characterized by a lack of energy (lethargy), irritability, or difficulty feeding. Repeated episodes of low blood sugar increase the risk for serious complications such as breathing difficulties, seizures, intellectual disability, vision loss, brain damage, and coma. individualsMost often, the diffuse form of congenital hyperinsulinism is inherited in an autosomal recessive pattern. Mutations in at least nine genes have been found to cause congenital hyperinsulinism. Mutations in the ABCC8 gene are the most common known cause of the disorder. An asymothomatic couple with the history of having a daughter who was passed away by 2 month year, with possible diagnosis of permanent neonatal hyperinsulinemic hypoglycemia were referred to genetic-counseling clinic. They are second cousins. They wanted to know the disease cause, recurrence risk and prevention possibility in their future pregnancies.Materials & Methods: Whole Exome Sequencing test was performed on the blood sample of the died baby. Sanger sequencing of the identified variant was done. We analyzed co-segregation of the variant in the family.Results: Analysis of exome data showed the proband has homozygous variant c.2754delG in exon 23 of the ABCC8 gene, which is classified as pathogenic variant by prediction tools. It was confirmed in patient and her parents by by sanger sequencing.Conclusion & Discussion: The severity of congenital hyperinsulinism varies widely among affected individuals, even among members of the same family. About 60 percent of infants with this condition experience a hypoglycemic episode within the first month of life. Other affected children develop hypoglycemia by early childhood. Mutations in at least twelve genes have been found to cause congenital hyperinsulinism. Mutations in the ABCC8 gene are the most common known cause of the disorder.The presence of blood sample from the proband made it possible to identify the mutation. Knowing the mutation in this family can prevent future affected pregnancies. In addition, there are other people at risk in this pedigree that detection of the status of being carrier can be and prevention or early treatment ccan be done. This case report emphasizes genetic mutation testing should become ... standard of care for infants with HI and has proven to be useful in projecting prognosis and fa

کلمات کلیدی:

Congenital hyperinsulinism, genetic counseling, Whole Exome Sequencing

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