عنوان مقاله:

A case of Frank-Ter Haar syndrome with a c.127C> T (p.Arg43Trp) mutation in SH3PXD2B gene

محل انتشار:

دومین کنگره بین المللی و دهمین همایش ملی نوروژنتیک ایران (سال: 1396)

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خلاصه مقاله:

Introduction: Frank-Ter Haar syndrome, is a rare sever progressive disease with a wide range of multisystemicdisorders affecting the skin, bone, joints and heart. FTHS patients usually expire in infancy or in early childhoodbecause of the cardiovascular anomalies and respiratory infections. Homozygous loss-of-function mutations inSH3PXD2B gene on 5q35.1 locus has been considered as one the underlying causes of FTHS.Patient and Method: We studied a 5-year-old affected boy born of healthy consanguineous parents. The patienthad a healthy sister and 2 affected siblings representing similar clinical symptoms including coarse face,prominent eyes, megalocornea, hypertelorism, congenital glaucoma, saddle nose, broad mouth, gingivalhypertrophy, brachydactyly, camptodactyly, flexion deformity of fingers, sever mitral valve collapse,thoracolumbar kyphosis, lordosis and Thick skin. His 2 affected siblings have died at the age of 4 month and 17years respectively due to respiratory infections.Exon 2, 5 and 10 of SH3PXD2B gene were analyzed by PCR-sequencing as hot spot mutations have beenreported in these exons previously.Result: Mutation analysis revealed one homozygous c.127C> T (p.Arg43Trp) mutation in exon2 of SH3PXD2Bgene which is a known pathogenic mutations.Conclusion: Our results confirm that diagnosis of FTHS requires analysis of SH3PXD2B gene for which Sangersequencing is still the most cost-effective method. If a negative result is obtained and the clinical evidence isstrong, whole exome sequencing might be a better approach to take next.

کلمات کلیدی:

Frank-Ter Haar syndrome, SH3PXD2B, PCR sequencing, FTH