عنوان مقاله:

Whole Exome Sequencing Reveals a Hemizygous Mutation in RPS6KA3 as a Cause of Coffin-Lowry Syndrome

محل انتشار:

نوزدهمین کنگره پژوهشی سالانه دانشجویان علوم پزشکی کشور (سال: 1397)

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خلاصه مقاله:

Background and Objective: Coffin-Lowry syndrome (CLS) is an X-linked disorder in which hemizygous males are more severely affected than females. It has an estimated prevalence of 1 in 50000 to 1 in 100000 male infants. It characterized by mental retardation, short stature, head and facial abnormalities, skeletal anomalies and developmental delays. The signs and symptoms vary in different people. Patient Report: This case study describes a 14-year-old Iranian male patient who was referred to our genetic counseling center for his dysmorphic features and mental retardation. He was second child of non-consanguineous and healthy parents. He had mild intellectual disability and seizure. Facial features included prominent forehead, widely spaced and downward-slanting eyes, broad and short nose, flat nasal bridge, wide mouth with full lips and hypodontia. By whole exome sequencing, we identified a hemizygous missense variant in exon 22 (c.2185C> T; p.Arg729Trp) of RPS6KA3 gene in the patient. Conclusion: We described the clinical and molecular feature of a male Iranian child with CLS due to a de novo missense mutation (c.2185C> T; p.Arg729Trp) in exon 22 of RPS6KA3 gene, displaying a mild mental retardation, seizure and typical facial features.

کلمات کلیدی:

Coffin-Lowry Syndrome, RPS6KA3, RSK2, X-linked Mental Retardation, Whole Exome Sequencing