عنوان مقاله:

Homozygous Deletion of exon ۷ in SMN۱ gene without phenotypic features of spinal muscular atrophy

محل انتشار:

کنفرانس بین المللی ژنتیک و ژنومیکس انسانی (سال: 1400)

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خلاصه مقاله:

Backgrounds: Spinal muscular atrophy (SMA) is an autosomal recessive disorder, resulting insymmetrical progressive weakness of skeletal and respiratory muscles and atrophy. Thecorresponding gene for the disease is the survival motor neuron ۱ (SMN۱) and SMN۲ genes.Homozygous deletion of SMN۱ exons is the most common underlying cause of the disease, andSMN۲ copy numbers modify the disease phenotype. However, homozygous deletion of exon ۷of SMN۱ in a completely asymptomatic individual is an extremely rare finding. The presentreport discusses a case of homozygote deletion of exon ۷ of SMN۱ in a healthy female.Materials and Methods: A healthy couple with a family history of infected family memberswith SMA was referred for genetic counseling. Genomic DNA was extracted from the peripheralblood of the couple and the copy number of exon ۷ of the SMN۱ gene was assessed using realtimepolymerase chain reaction (PCR) and PCR-Restriction fragment length polymorphism(RFLP).Results: Assessment of SMN۱-related ct in the female compared with control samples showedthat the female had a homozygous deletion in the SMN۱ gene. PCR-RFLP and gelelectrophoresis results also confirmed the homozygous deletion of exon ۷ in the female SMN۱gene.Conclusion: According to the results of this study and also other findings in previous studies, thelack of symptoms in the female with biallelic deletion of SMN۱ may be related to the presenceof SMN۲ copies or other modifier genes.

کلمات کلیدی:

spinal muscular atrophy, SMA, SMN۱, homozygous deletion, biallelic deletion

لینک ثابت مقاله در پایگاه سیویلیکا: