عنوان مقاله:

Mutational analysis of DMD gene reveals two novel small deletions in patients bearing no large deletions or duplications

محل انتشار:

اولین کنگره بین المللی و سیزدهمین کنگره ژنتیک ایران (سال: 1393)

تعداد صفحات اصل مقاله: 1

نویسندگان:

Zohreh Fattahi - *Genetics Research Centre, University of Social Welfare and Rehabilitation Sciences, Tehran, Iran. .Kariminejad - Najmabadi Pathology & Genetics Center, Tehran, Iran*

Gholamerza Zamani - *Department of Pediatric Neurology, Children's Medical Center, Tehran University of Medical .Sciences, Tehran, Iran*

Mahsa Fadaee - *Genetics Research Centre, University of Social Welfare and Rehabilitation Sciences, Tehran, Iran. .Kariminejad - Najmabadi Pathology & Genetics Center, Tehran, Iran*

.Mohammad Reza Akbari - *Women's College Research Institute, University of Toronto, Toronto, ON, Canada*

خلاصه مقاله:

Dystrinopathies are inherited muscular dystrophies with mutations in DMD gene which contains 79 exons and is the largest human gene. DMD encodes Dystrophin protein and is the only gene responsible for the spectrum of dystrinopathies. Genotype analysis has indicated that deletions of one or more exon account for ~65% of all cases, while 5-10% are in-frame or out-frame duplications and the remaining 30% of affected individuals may have point mutations, small deletions orinsertions within the gene. In the present study, two patients diagnosed with Duchenne muscular dystrophy (DMD) are investigated. Multiple ligationdependent probe amplification (MLPA) analysis which detects up to 98% of all deletions and duplications in DMD gene didnot reveal any large rearrangement in these patients. Further investigation with application of two different techniques led toidentification of two novel small deletions in DMD gene. In the first patient, all coding as well as flanking intronic regions of the dystrophin gene were PCR-amplified followed by sequencing and a hemizygous novel deletion; c.650-16_653del20 (p.D217V fs11X) was detected. Next, whole exomesequencing was applied to investigate the causative variant in the second patient that revealed another novel hemizygousdeletion defined as c.8297delT (p.Leu2766Arg fsX17). The identified variant was confirmed by Sanger sequencing in the proband as well as other family members. This result extends the mutational spectrum of the disease by introducing two novel mutations in DMD gene and is another indication that the advent of recent technologies such as whole exome sequencing has made the diagnostic approaches much easier

کلمات کلیدی:

Duchenne muscular dystrophy, MLPA, whole exome sequencing