Molecular Genetics, Genomics, Mechanisms of Diseases

AB095. Prenatal diagnosis of Duchenne muscular dystrophy by combining of multiplex Polymerase Chain Reaction and Multiplex Ligation dependent Probe Amplification

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Background: Duchenne muscular dystrophy (DMD) is the most common allelic X-linked muscular disorder caused by mutation in the dystrophin gene. The dystrophin gene located at Xp21.2, is one of the largest known human genes, covering 2.2 Mb, containing 79 exons and at least eight independent, tissue specific promoters and two poly-A-addition sites. The goal of this study is prenatal diagnosis for pregnancies at high risk of DMD disease.

Methods: Twenty-one pregnant women who had children with DMD, were identified the dystrophin mutations by multiplex Polymerase Chain Reaction (Multiplex PCR) and Multiplex Ligation dependent Probe Amplification (MLPA).

Results: Among 21 fetuses, we found 18 males and 3 females. In 18 male fetuses, 5 cases showed deletion mutation and 13 cases have not deleted in 25 exons by multiplex PCR. In 3 female fetuses, 2 cases were heterozygous of deletion mutation and 1 case has not found deletion and duplication mutation in 79 exons by MLPA.

Conclusions: Combination of multiplex PCR and MLPA could detect deletion and duplication mutations for determination of the carrier and prenatal diagnosis of DMD.

Keywords: Duchenne muscular dystrophy (DMD); multiplex Polymerase Chain Reaction (PCR); Multiplex Ligation dependent Probe Amplification (MLPA)

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