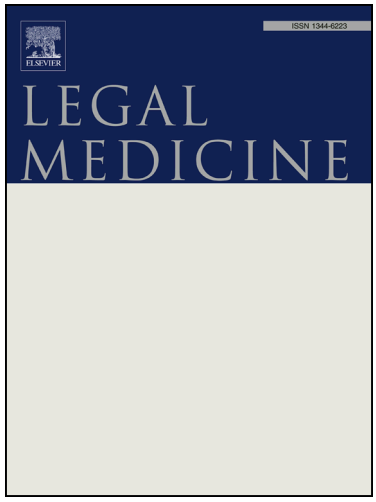

Case Report

Tri-allelic patterns of STRs and partially homologous non-sister chromatid crossover observed in a parentage test

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Tri-allelic patterns of STRs and partially homologous non-sister chromatid crossover observed in a parentage test

Abstract

A maternity testing case is reported, in which the child showed tri-allelic patterns in two STR loci. The genotypes of Penta D of the mother and the child were 9,13 and 9,10,13, respectively. Those of D21S11 were 32.2,35 and 29,35, respectively, but intensity ratio of alleles 29 and 35 of the child was 1:2. These results suggested the copy number variations (CNVs) or trisomy of chromosome 21. By further examination using STR-based chromosome aneuploidy detection kit, three alleles were detected in D21S1411, LFG21 and Penta D, and 2 alleles with intensity ratio of 1:2 were observed in D21S2502, D21S1435, D21S11 and D21S1246. Karyotype and whole-genome SNP array analyses showed that the child had a free trisomy 21. In addition, partially homologous non-sister chromatid crossover occurred at the region 19181770-39499178 on the long arm of chromosome 21.

Keywords STR, parentage test, trisomy 21

1. Introduction

In the parentage test using STR typing, detection of tri-allelic pattern in a locus suggests the copy number variations (CNVs) or trisomy. However, it is difficult to identify whether the pattern was caused by CNVs or trisomy. Trisomy is caused by Robertsonian translocation, reciprocal translocation, inverted duplication of chromosome, recombinant duplication of chromosome resulted by ring chromosome or parental inverted, and partial duplications [1]. CNVs are caused by structural rearrangements of the genome including translocations, duplications, inversion, and deletions [2]. Since both trisomy and CNVs correspond to relatively large regions of the chromosome, in which several short tandem repeat (STR) markers may be located, STR genotypes analyzed for parentage testing may result in tri-allelic patterns or peak height imbalance. Then, further analysis should be performed to confirm or exclude the parentage relationship. Here we report a rare duo maternity testing case showing tri-allelic pattern and two alleles with intensity ratio of 1:2 in the child's DNA profile, while the mother's profile was normal.

2. Materials and methods

2.1. Samples and DNA isolation

Blood samples from the mother and the child (male) was collected with informed consent. This research protocol was given permission by the ethical review committees of Beijing institute of Genomics. DNA was extracted by Chlex-100 resin method [3] for the STR amplifications, according to the manufacture's protocol. The concentration and quantity of the isolated DNA was determined by Qubit® Quantitation System(Invitrogen, CA, USA), according to the manufacturer's specifications.