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Parental investigation in a child with abnormal karyotype

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Abstract. We present a parental investigation where the father and the mother are dubious, and the alleged child had a previous karyotype with a partial deletion at 13 chromosome (46XX del13). STR typing at D13S317 locus revealed homozygosity alleles in the three individuals involved in this study. The homozygosity of the alleged child at this locus was assumed as the result of a null allele, due to the partial deletion at 13 chromosome. This results showed a paternal inheritance of the chromosomal anomaly. In cases like this, the parental investigation with highly discriminating STR markers could be an additional contribution to the genetic diagnosis as well as an accurate genetic advice to the couple.

Keywords: Paternity testing; Chromosomal anomaly; STRs

1. Introduction

There are several situations in which the assessment of paternity may vary depending on whether a one-banded pattern or only one pick on the electropherogram is assumed as an homozygous allele. Another possibility is to consider that it could represent a gene with two different alleles, one of which is invisible, overlapped or has run off.

‘‘Silent allele’’, of course, includes the possibility of an invisible band and the occurrence of ‘‘primer drop out’’ during the microsatellite amplification by PCR.

In this study, we present a parental investigation where the father and the mother are dubious and the alleged child has a partial deletion of chromosome 13 (46XX del13) in a previous karyotype.

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2. Materials and methods

Genomic DNA was isolated from blood samples with salting out Miller's method. The samples were amplified at the loci FGA, VWA, D3S1358, D5S818, D7S820, D8S1179, D13S317, D18S51 and D21S11 using the AmpFlSTR™ Profiler Plus Kit (PE Biosystems, Foster City, CA) and at the loci CF1PO, TPOX, TH01, D3S1358, D7S820 and D16S539 using the AmpFlSTR Cofiler™ kit (PE Biosystems). The same loci and the D2S1338 and D19S433 were amplified with the AmpFlSTR™ Identifiler Kit (PE Biosystems). Samples were analyzed using the ABI PRISM™ 310 Genetic Analyzer (PE Biosystems), according to the manufacturer's recommendations using as separation medium Performance Optimized Polymer (POP) 4™ (PE Biosystems). The data was acquired by 1.0.2 software ABI PRISM™ 310 Collection and analyzed by GeneScan® Analysis software 3.1 and Genotyper® 2.5 according to the manufacturer's recommendations [1–4].

3. Results

STR-typing at D13S317 locus (Fig. 1) revealed the following results (Table 1).

4. Discussion

The homozygosity of the alleged child at D13S317 locus was assumed as the result of a null allele due to a partial deletion at 13 chromosome. Thus, we considered that allele 8 at D13S317 locus was from maternal origin and that the partial deletion at 13 chromosome (13q 22–31) was by paternal inheritance.

This study revealed a non-exclusion of the maternal–paternal relationship for the other investigated markers.

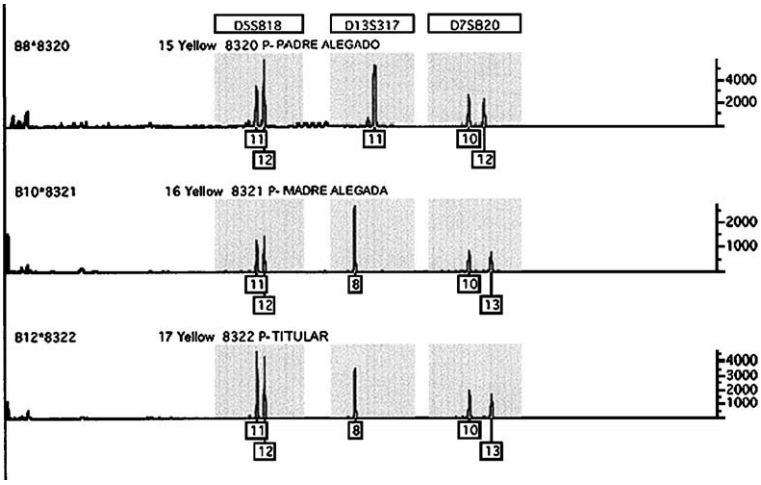

Fig. 1. STR-typing at D13S317 locus.

Table 1
STR-typing at D13S317 locus

Locus		Alleles		
		Mother	Child	Alleged father
1	D8S1179	15–12	13–12	16–13
2	D21S11	31–30	31.2–30	31.2–29
3	D7S820	13–10	13–10	12–10
4	CSF1PO	12–10	12–10	12–10
5	D3S1358	15–15	17–15	17–17
6	TH01	7–6	8–6	8–6
7	D13S317	8–8	8–8	11–11
8	D16S539	12–12	12–12	12–11
9	vWA	18–18	19–18	19–18
10	TPOX	12–11	11–10	11–10
11	D18S51	16–12	14–12	17–14
12	D5S818	12–11	12–11	12–11
13	FGA	25–21	25–24	24–24

In agreement with statistics–mathematics analysis of the obtained results (using Ch. Brenner formulas) [5], the parental probability was 999.999% (without D13S317 marker) and considering a null allele was 99.995%.

5. Conclusion

In cases like the previously described, the parental investigation with highly discriminating STR markers could be an additional contribution to genetic diagnosis as well as an accurate genetic advice to the couple.

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