International Congress Series 1261 (2004) 485–487

www.ics-elsevier.com

Forensic DNA analysis of a foetal histological section and chromosome sample from chorionic villi (CV) in Down syndrome

H. Pamjav[a,*], B. Egyed[b], S. Füredi[b], J. Woller[b], Zs. Pádár[b], E. Susa[a], B. Baraczka[a]

[a] *Institute of Forensic Medicine, Ministry of Justice, DNA Laboratory, 1363 Budapest, Pf. 31, Hungary*
[b] *Institute of Forensic Sciences, Hungarian National Police HQ, Budapest, Hungary*

Abstract. According to the opinion of a plaintiff (Mrs. B) and a co-plaintiff (Mrs. A), two samples had been exchanged during cytogenetic investigation of foetal chorionic villi (CV), because the child of Mrs. A was born with Down syndrome in spite of the negative CV result (46, XY). The CV investigation of Mrs. B had given a positive result (47, XY, +21), so the pregnancy was terminated, but histological sections of her abortus showed no pathological changes. DNA profiles were successfully obtained from the participants and both of the slides. Two complicating factors were observed during the investigation. Between mother (Mrs. A) and the Down syndrome child, a one-step germ line mutation was found together with a three-allele D3S1358 pattern in the same child's profile. The genetic investigation and its statistical interpretation strongly support the hypothesis that the putative exchange of samples did not occur.

Keywords: Three-banded pattern; Paternity testing; Down syndrome; STR; Germline mutation

1. Case report

A civil liability claim was initiated against one of the Budapest hospitals in 1992. According to the opinion of the plaintiff (Mrs. B) and the co-plaintiff (Mrs. A), two samples had been exchanged during cytogenetic investigation of foetal chorionic villi (CV), because the child of Mrs. A was born with Down syndrome in spite of the negative CV result (46, XY). The CV investigation of Mrs. B had given a positive result (47, XY, +21) and so the pregnancy was terminated, but histological sections of her abortus showed no pathological changes.

* Corresponding author. Tel.: +36-1-457-0183; fax: +36-1-201-6166.
E-mail address: phorolma@hotmail.com (H. Pamjav).

0531-5131/
doi:10.1016/j.ics.2003.11.011

2. Materials and methods

Forensic DNA analysis was carried out on the Giemsa-stained CV slide (chromosome sample) from Mrs. A, the histological slide stained with hematoxylin–eosin from the abortus of Mrs. B and on blood samples from Mrs. A, her husband Mr. A, their child with Down syndrome and Mrs. B.

The histological slide and the chromosome sample were placed in xylene and acetone, respectively, until the cover glasses could be easily removed. The slides were then placed in ethanol solutions of decreasing concentration, and finally in distilled water. The tissue and mitotic chromosomes were removed with cotton swabs. DNA was isolated by phenol–chloroform extraction and Microcon YM-100 (Millipore, USA) purification. DNA was extracted from blood samples using the QIAmp DNA Blood Mini Kit (Qiagen, Germany). STR loci were amplified with the ProfilerPlus AmpF1STR kit (Applied Biosystems). All samples were analysed on an ABI Prism 310 Genetic Analyzer (Applied Biosystems).

3. Results and discussion

Profiles were successfully obtained from the participants and off the slides originated from Mrs. A and Mrs. B (Table 1). The child of Mrs. A had three different alleles at the locus D21S11, consistent with Down syndrome. The profiles indicated that Mr. and Mrs. A were the biological parents of the child (PP>99.99%), although there were two complicating factors. First, one allele of the child (D8S1179) did not match either parent but could have arisen as a one-step mutation within the mother. Second, a three-allele pattern was found at the locus D3S1358 from the child's blood, hair and buccal samples, indicating that a second mutation had also occurred (Table 1). The DNA profile from the CV slide matched that of the child's, except that only two alleles at D3S1358 were present, suggesting that the third allele (allele 19) had arisen as a one-step somatic mutation in embryoblast cells during embryonic life (Table 1). The histological slide from the abortus also showed three alleles at D21S11, consistent with the original cytogenetic diagnosis of Down syndrome. The DNA profiles justified that the histological slide had been prepared from the abortus of Mrs. B. The genetic investigation and its statistical interpretation [1,2] therefore strongly support the hypothesis that the putative exchange of samples did not occur.

Table 1
Genotypes

Locus	Father "A"	Mother "A"	Child	CV Sample	Mother "B"	Abortus Sample
D3S1358	17-18	15-16	16-18-19	16-18	14-15	14-17
VWA	16-17	17-19	16-19	16-19	15-19	15-18
FGA	21-23	19-21	21-21	21-21	-	-
D8S1179	13-14	13-16	13-17	13-17	13-14	13-14
D21S11	30-32.2	30.2-31.2	30.2-31.2-32.2	30.2-31.2-32.2	28-29	28-29-30
D5S818	10-10	11-12	10-12	10-12	12-13	13-13
D13S317	9-12	12-12	12-12	12-12	-	-

Acknowledgements

Authors express their gratitude to Dr. Chris Tyler Smith and Dr. Charles Brenner for providing essential help with consultation on the subject.

References

[1] B. Egyed, S. Füredi, M. Angyal, L. Boutrand, A. Vandenberghe, J. Woller, Z. Pádár, Analysis of eight STR loci in two Hungarian populations, Int. J. Leg. Med. 113 (2000) 272–275.
[2] C. Brenner, Paternity calculation with 3-banded pattern, Available at: http://www.dna-view.com/3band.htm, 1999.