International Congress Series 1261 (2004) 502–504

www.ics-elsevier.com

Maternity testing in a chimerical child

M.B. Rodríguez Cardozo, M.V. Cólica, M.A. Abovich, A. Szöcs, A.M. Di Lonardo*

Immunologia, Banco Nacional de Datos Genéticos, Hospital Dr. C. G. Durand, Juan B. Ambrosetti, 743 Planta Baja 1405 Buenos Aires, Argentina

Received 5 September 2003; received in revised form 12 September 2003; accepted 17 September 2003

Abstract. Human congenital chimerism is due to the coexistence of two genetically different cell lines either in the whole body or limited to the blood. We present a case of maternity in which the alleged child has ambiguous genitalia, and chimerism was suspected. The maternity was dubious and the father was not available for the study. STR typing of the child revealed the presence of X an Y chromosomes at Amelogenin locus and a double maternal and paternal alleles contribution in certain autosomical loci. We could not exclude maternal relationship between the alleged mother and the child. Molecular analysis performed with highly discriminating STR systems allowed us to clarify the origin of a chimerical individual. © 2003 Elsevier B.V. All rights reserved.

Keywords: Maternity testing; STRs; Chimerism; Typing

1. Introduction

Human congenital chimerism is due to the coexistence of two genetically different cell lines either in the whole body or limited to the blood. These cells are from different genetic patterns originated from two or more zygotes. Within permanent chimerical individuals "blood chimeras" or "twin chimeras", which result from in-uterus junctions of twins and "whole body chimeras" where the coexistence of the different cell lines is not restricted to hematological cells but spread over various tissues, can be distinguished [1–3].

Whole body chimerism (tetragametic or dispermic chimerism) is characterized by double paternal contribution of markers. In very rare cases, double maternal contribution has also been detected [2].

We present a case of maternity in which the alleged child had ambiguous genitalia, and chimerism was suspected. The maternity was dubious and the father was not available for the study.

* Corresponding author. Tel.: +5411-4982-1716; fax: +5411-4982-0625.
E-mail address: bndg@infovia.com.ar (A.M. Di Lonardo).

doi:10.1016/S0531-5131(03)01624-8

Table 1
STR typing for mother and child

Locus	Child	Alleged mother
D8S1179	14/13/12/11	11/12
D21S11	31/30/29	31/30
D7S820	12/11/9	11/9
CSF1P0	10/10	10/10
D3S1358	18/15	18/15
TH01	6/6	6/6
D13S317	12/11/9	9/9
D16S539	11/10/9	11/9
D2S1338	19/19	21/19
D19S433	15/12	15/12
vWA	17/16/15	16/15
TPOX	12/11	12/8
D18S51	16/13/12	18/13
D5S818	13/11/10	11/10
FGA	25/23/21/19	23/21
Amelogenin	X/Y	X/X

2. Materials and methods

Genomic DNA was isolated from blood samples with salting out Miller's method. The samples were amplified with the AmpFlSTR™ Identifiler Kit (PE Biosystems, Foster City, CA). 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 [4,5].

3. Results

STR typing for mother and child are given in the Table 1.

4. Discussion

STR typing revealed the presence of two maternal and two paternal alleles at two loci in blood of the proposita. In another locus (D13S317), two paternal alleles were present, while the other 11 systems investigated revealed double maternal contribution.

Amelogenin showed the presence of a Y chromosome in misbalance with the X chromosome.

5. Conclusion

We could not exclude maternal relationship between the alleged mother and the child. We confirmed the double maternal contribution, and the possibility that the chimerism of our proposita was the result of the double parental alleles contribution [6,7].

Molecular analysis performed with highly discriminating STR systems allowed us to clarify the origin of a chimerical individual.

References

[1] Yu. Neng, et al., N. Engl. J. Med. 346 (20) (2002) 1546–1552.
[2] J.C. Giltay, et al., Am. J. Hum. Genet. 62 (1998) 937–940.
[3] A.J. Green, et al., J. Med. Genet. 31 (10) (1994 Oct.) 816–817.
[4] K. Lazaruk, et al., Electrophoresis 19 (1998) 86–93.
[5] O. Henegariu, et al., BioTechniques 23 (3) (1997) 504–511.
[6] A. Milde, et al., Int. J. Leg. Med. 112 (5) (1999) 333–335.
[7] L.M. Repas-Humpe, et al., Ann. Hematol. 78 (9) (1999 Sept.) 431–434.