International Congress Series 1239 (2003) 569–571

STR typing in a pair of chimeric twins

E.M. Dauber*, I. Faé, S. Stadlbacher, B. Glock, D.W.M. Schwartz, W.R. Mayr

Division of Blood Group Serology, Medical School, University of Vienna, Vienna, Austria

Abstract

In a follow up study, DNA typing of short tandem repeat polymorphisms, HLA and ABO antigen typing was carried out on a pair of chimeric twins, which had already been investigated by serological and cytogenetical methods two decades ago. The aim of the study was to find out whether or not DNA typing of short tandem repeat polymorphisms is an appropriate method to characterize twin chimeras.

Keywords: Short tandem repeat; Twin chimera; Chimerism; DNA typing

1. Introduction

In man, congenital chimerism is due to the coexistence of two genetically different cell lines either in the whole body or limited to the haemopoietic tissue. Blood stem cells can migrate through anastomoses of intrauterine blood vessels during pregnancy of dizygotic twins and produce blood cells in the other embryo for a lifetime. Therefore, these individuals are called ‘‘twin chimeras’’, which differ from ‘‘dispermic chimeras’’, who derive from the fertilization of one or two maternal nuclei by two sperms and their growth into one body.

A pair of chimeric twins, Franz and Johann F., are well known to our laboratory and were extensively investigated with serological, biochemical and cytogenetical methods [1]: two different red and white cell populations had been found in their blood. The proportions in the peripheral blood of Franz and Johann F. had been 19% to 81% and 91% to 9% erythrocytes of group A to O and 1% to 99% and 42% to 58%, respectively, of the

* Corresponding author. AKH-Universitätskliniken, Klinische Abteilung für Blutgruppenserologie, Währinger Gürtel 18-20, A-1090 Wien, Austria. Tel.: +43-1-40400-5320; fax: +43-1-40400-5321.
E-mail address: eva.maria.dauber@univie.ac.at (E.M. Dauber).

0531-5131/03 © 2003 Elsevier Science B.V. All rights reserved.
PII: S0531-5131(02)00218-2

lymphocyte populations I to II, which had been differentiated by studying the polymorphic variants observed in autosomes. Interestingly, both twins had the same lymphocytes of type II as the major population, which should be kept in mind for the following considerations. Further investigations of three dispermic chimeras proved the benefit of mini- and microsatellite typing in these fascinating cases. Therefore, we decided to perform STR typing and DNA typing of red and white cell antigens on these twin brothers to find out whether DNA typing of short tandem repeat polymorphisms is an appropriate method to characterize twin chimeras. We expected to see a mixture of the genetic patterns of the two brothers in the blood but not in other tissues.

2. Materials and methods

Blood samples, buccal swabs, eyebrows and fingernails were taken separately from the twins. DNA was extracted using the Chelex method [2] or the QIAamp® DNA Mini Kit (Qiagen) and examined with the AmpFlSTR SGM Plus™ Kit (Applied Biosystems) and our standard method for SE33 and D12S391 on an ABI Prism 310 capillary electrophoresis instrument (Applied Biosystems) or an A.L.F. DNA Sequencer (Amersham Pharmacia) [3,4]. DNA extraction and PCR for STR typing on the twin's samples were carried out separately.

3. Results and discussion

In ABO-SSP, the same genotypes were found as the twins were supposed to have after the first serological investigations [1]. Three HLA alleles have been found in their blood and at some loci also in their buccal cells. STR typing revealed the coexistence of three different alleles, which showed imbalanced signal intensities in capillary electrophoresis at 6 out of 12 loci in the blood samples. At three loci, two alleles with significant peak height imbalance ($<70\%$) have been found. All the other loci showed two well-balanced peaks. We did not find loci with four alleles. In the samples taken from inside their cheeks, a nearly ''normal'' DNA profile was found in Franz F. except for an additional allele at the D18S51 locus. All other additional alleles were in stutter position to the major alleles and their signals below the threshold for stutter peaks. In the buccal swab of Johann F., we found the same alleles as in his blood, but with increased peak height imbalance. All but one additional allele were clearly identified as they were not in a stutter position to his main alleles. The DNA profiles of Franz F. in his eyebrows and fingernails looked completely normal. Even in the fingernails of his twin brother Johann F., we found the same additional alleles as in his blood and buccal swabs, but with even lower proportions and very low peak heights. Only his eyebrows were free of additional alleles: he was found to be heterozygous at all but three loci. A mixture of the genetic patterns of both twins was found in the blood samples, which was to be expected, but also in the buccal swabs of the twins. This could be due to leucocyte contamination in the oral cavity. Johann F. was found to have about 60% of the leucocyte type II population in his blood, which can be supposed to be the true genetic line of his twin brother Franz F., as STR alleles of his

eyebrows and fingernails and the major alleles of his blood were identical. Leucocyte contamination of the fingernails of Johann F., which were partially covered with detritus, could also account for the additional alleles found in the fingernails of Johann F. All alleles found in the blood of each brother were also found in the blood of his twin brother. This agrees with the definition of twin chimeras, whereas dispermic chimeras are proven, when two populations of cells are found that originate from tissue other than haematopoietic tissue [5]. As the DNA extraction methods used could not differentiate between cells from haematopoietic and nonhaematopoietic tissue in the samples investigated and almost all tissues potentially contain blood cells, DNA-typing results of chimeras must be interpreted with caution.

References

[1] V. Pausch, I. Bleier, E. Dub, M. Kirnbauer, M. Weirather, G. Würger, W.R. Mayr, A further case of chimeric twins: genetic markers of the blood, Vox Sang. 36 (1979) 85–92.
[2] P.S. Walsh, D.A. Metzger, R. Higuchi, Chelex® 100 as a medium for simple extraction of DNA for PCR-based typing for forensic material, BioTechniques 10 (4) (1991) 506–513.
[3] S. Stadlbacher, E.M. Dauber, B. Glock, W.R. Mayr, SE33 (HumACTBP2): native gel electrophoresis versus denaturing capillary electrophoresis, and population data, Int. Congr. Ser. 1239 (2003) 699–701 (this volume).
[4] B. Glock, E.M. Dauber, D.W.M. Schwartz, W.R. Mayr, Additional variability at the D12S391 locus in an Austrian population sample: sequencing data and allele distribution, Forensic Sci. Int. 90 (1997) 197–203.
[5] P. Tippett, Blood group chimeras, Vox Sang. 44 (1983) 333–359.