

A Parentage Testing Study Revealing a Possible Deletion at the PLG Locus

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INTRODUCTION

A routine paternity study of a black trio revealed reverse homozygosity (single indirect exclusion) between the alleged father and child in the PLG system. Additional testing revealed a second possible exclusion based on a single band in the RFLP system D6S132. Since both apparent exclusions are loci found on the same segment of Chromosome 6 (6q26-q27) it is unlikely that the discrepant results between father and child are evidence of non paternity.

METHODS

Standard methods were used to test for red cell antigens (ABO, RH, MNSs, Kell, Duffy, and Kidd), red cell enzymes (ESDi, ACP, and PGM1i), and serum proteins (Gci, BF, F13A, F13B, and PLG). Eight single locus DNA RFLP systems (D1S47, D2S44, D6S132, D7S104, D7S467, D12S11, D17S79, and D18S27) were tested using Pst1 for restrictions and colorimetric detection with alkaline phosphatase labeled probes. For each DNA probe/enzyme combination, members of the trio were tested in the same batch and each included a mix of the alleged father and child as well as appropriate controls. A multi locus probe, DNF24, was also used to test this trio. Calculations of the paternity index was based on in house frequency tables for the majority of systems.

RESULTS

Initial testing showed only a possible exclusion in PLG (alleged father PLG 2, child PLG 1). Because of a high residual paternity index (26,362 if PLG assigned a value of 1 or 403 if a "PLG null" is assumed with a frequency of 0.0153, [Polesky 1987]) a family study was suggested. Testing of the parents (both PLG 1-2) and sister of the alleged father ruled out the possibility of a PLG null (see figure 1). Additional RFLP testing showed a second discrepancy in expected inheritance at the D6S132 locus using the probe/enzyme combination Sli1090/Pst1. The results (see figure 1) showed the child to have a single band (1.92 Kb) that matched with one of the maternal bands. The alleged father had two bands, 3.47 and 1.89. Although the 1.89 and 1.92 Kb bands are similar in size, the mix of child and alleged father showed three distinct bands (1.89, 1.92, 3.47). The paternity index based on the red cell antigens, red cell enzymes, serum proteins (except PLG), and seven single locus

RFLP systems (D6S132 not included) exceeded 2.7 billion.

DISCUSSION and CONCLUSION

The two loci that potentially exclude this alleged father are both assigned to the q26-q27 region of chromosome 6 [Ziegler, 1991]. Since extensive testing of other genetic loci fail to exclude this man and testing of his family rules out the presence of a "PLG null", we have concluded that he is the father of the child in question. The finding of two apparent discrepancies at the same chromosomal region can best be explained by a deletion involving a single locus or two closely linked loci. This case illustrates the importance of comparing chromosomal locations for classic markers with the assignments of various DNA probes.

REFERENCES

Polesky HF, Souhrada JM (1987) A protocol for reporting single indirect exclusions. *Advances in Forensic Haemogenetics* 2: 600 - 602.

Ziegler A, Field LL, Sakaguchi AY (1991) Report of the committee on the genetic constitution of chromosome 6. *Cytogenet Cell Genet* 58: 295 - 336.

Fig 1. DW Family: PLG and D6S132 Results

