



Presence of two mutations between father/child in two cases of paternity testing

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The presence of mutations and specially step mutations (loss or gain of a tandem repeat) is well known and relatively frequent. We report here two cases of paternity testing presenting two different mutations between father and child.

In the first case, two mutations were observed at the VWA locus: the mother had alleles 16–17, the child had 15–16 and the alleged father had 17, and at the D5S818 locus: mother had 11, child had 11–12 and alleged father had 11–13.

The second case also showed two inconsistencies with the following phenotypes for the VWA locus: mother had 14, child had 18 and alleged father had 14–19 and the D5S818 locus: mother had 11–12, child had 11–13 and alleged father had 12.

The two cases were examined with more than 15 PCR-based loci and four RFLP Southern blot probes and the probability of paternity for both was >99.999% (including the mutations in the calculation).

It is interesting to notice that, in both cases, the mutations were present at the same two loci. Furthermore in the first case a double-step mutation, which is quite rare, was observed at the VWA locus between father and child. In the second case, a single-step mutation between father and child and an inconsistency between mother and child were observed.

In spite of the fact that the mutation rates for the different loci used in the forensic analysis are reported in the literature, the presence of a double mutation in a paternity testing is not common. The double mutation (father/child) rate observed in the two laboratories concerned shows a value of about 0.7% of all paternal meiosis ($N=294$). Therefore, when working with STRs, we believe that a “three exclusion rule” should be used when forming a judgement of non-paternity.

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