ARE TWO EXCLUSIONS ENOUGH TO EXCLUDE IN PATERNITY TESTING? ANALYSIS OF 17 STR, D1S80 AND HLA-A, B, DRB, AND DQB1 IN TWO CASES WITH ONLY TWO EXCLUSIONS

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It is widely accepted that two or more STR exclusions are enough to exclude in a paternity testing dispute. Routinely we analyze 12 Short Tandem repeat loci (STR) in our paternity testing program. When only one or two exclusions are found, we increase the number of STR analyzed to 17 plus D1S80 in order to find additional exclusions (Power of exclusion 99.9999%). Occasionally, the highly discriminating HLA system by molecular typing is also performed if required.

We have come across two unrelated cases were after the analysis of 17 STR, D1S80 and molecular HLA-A, HLA-B, HLA-DRB and HLA-DQB1 loci only two STR exclusions were found in each case. In case #1, CSF1PO alleles were AF 12/11, M 12/12, C 13/12 and D18S849 were AF 17/15, M 19/16 and C 19/16. In case #2, D12S1090 alleles were AF 30/12, M 19/9 and C 29/9 and FGA alleles AF 25/19, M 27/19 and C 28/19. The exclusions were of the first order in both cases, the mother and the alleged father were not related in each case, and there was no additional hypothesis as whether the alleged father could be related to another man--the true father.

STR mutations are produced by slippage of the DNA polymerase during replication of DNA and these mutations occur in a step-by-step fashion (gain or loss of only one repeat unit). Based on the mutation rates calculated for tetranucleotide STR loci, a mutation could be found in one out of 1,000 meiotic events (using very conservative estimates), therefore, the probability of finding two paternal mutations are 1:1 x 10^6 meiotic events, an event unlikely to occur. On the other hand, there are no reports showing paternal and maternal mutations in a paternity testing case.

We postulate as a hypothesis that there are two paternal mutations in case #1 and the existence of both a paternal and a maternal mutation in case #2.

The discussion of cases like the present ones will be of great importance within the scientific community. Should the guidelines for exclusions be modified? Will three or more exclusions be considered instead of two for an exclusion? At the present moment, for cases with only two exclusions we will exclude the paternity if both exclusions are of first order and if the differences are based on more than one repeat unit between the AF alleles and the alleles present in the child.