MUTATIONS, VARIATIONS AND MISSING ALLELES IN THE STR SYSTEMS

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The Short Tandem Repeat (STR) loci that we have used consist of repetitive sequence elements of 4 basepairs in length and are a rich source of polymorphism. In the past three years, extensive use of the STR systems has resulted in the observation of many mutations between both mother/child and father/child pairs in paternity cases. A paternity case most commonly involves the testing of a mother, child and alleged father. We have also observed a variety of variant and new alleles. Missed alleles have occurred at both the Amelogenin and D13S317 loci.

Mutations were observed at the CSF1PO, TPOX, vWA, F13AO1, FESFPS, D16S539, D7S820, D13S317 and D5S818 loci. A single exclusionary event was deemed to be a mutation where no other evidence of exclusion could be found in a total of 15 to 28 loci tested. Mutations at the loci between mother/child and father/child pairs have always resulted in either an increase or decrease of one STR repeat unit from the parent to the child.

A number of variants and new alleles have been observed by differences in electrophoretic mobility and size when compared to the allelic ladders on the ABI™373A/377 Sequencers.

We report the failure to amplify and detect fragments at both the D13S317 and Amelogenin loci. Both the Promega PowerPlex™ and PE Applied Biosystems AmpF/STR™ Profiler Plus kits failed to amplify in both instances. This may indicate sequence variation at these primer positions.

The combination of many STR loci has produced confounding variables such as mutations, variations and missed alleles in approximately 1% of our Australian paternity cases. This adds an element of complexity to analysis and therefore many tests including VNTRs, DQA1, Y-chromosome loci and 17 STR loci are available for use in the laboratory.