DEALING WITH APPARENT MUTATIONS IN RELATIONSHIP ANALYSIS – DEVELOPMENT OF A MATHEMATICAL MODEL AND ITS INCORPORATION INTO THE FSS DNA LINEAGE SOFTWARE

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In the context of relationship analysis there may be a mutation or recombination event during replication at the meiosis stage i.e. at the point at which genetic material is transmitted from parents to child. This may be perceived as an apparent inconsistency in the Mendelian pattern of inheritance.

Such apparent mutation events have an impact upon the calculated Likelihood ratio (or Paternity Index), potentially providing support for an alternate hypothesis. When conducting relationship analysis by utilising autosomal STR loci it is incorrect for a scientist to ignore a locus (or loci) in which such genetic inconsistencies appear. By analysing many thousands of parent/child transmissions it is clear that the apparent mutation rates vary by locus and depend on whether the allele was maternally or paternally inherited.

As a consequence FSS has created a 'mutation' model which invokes stepwise changes to the repeat unit and takes into consideration the identity of the locus, the position in the allele range of the specific allele involved (terminal or mid-range), the number of step changes involved and whether the apparent inconsistency arose in the maternal or paternal lineage.

This model has been incorporated into FSS DNA Lineage™; a relationship analysis application created by FSS. This software application allows a user to conduct simple (or complex) pedigree analyses by drawing the pedigrees associated with each of the hypotheses under test.

User-definable STR marker systems and allele frequency databases can be added to the FSS DNA Lineage application. In this way the user defines the parameters used to calculate probabilities and likelihood ratios from the hypotheses under test. The analysis parameters can be chosen to reflect laboratory-specific requirements to take account of rare alleles (minimum default frequencies or size bias corrections), mutations and Theta values (coefficient of inbreeding of a population).

FSS DNA Lineage also creates a technical audit file which contains the case details, process settings, allele frequencies, probabilities for each hypothesis by locus, likelihood ratio by locus and overall likelihood ratio. The application also has the functionality to produce user-defined statements or reports.