



E10 Development Process Validation for Kinship Analysis Algorithm

Sharada Vijaychander, MS, Thermo Fisher Scientific, 180-120 Oyster Point Boulevard, South San Francisco, CA 94080*

After attending this presentation, attendees will gain an understanding of the established Life Technologies™ (LT) kinship algorithm analysis program, its features, flexible implementation, and standard calculations for trio analysis, including complex pedigree trees such as incest, motherless/fatherless cases, and inclusion/exclusion of mutations, and rare alleles based on currently used methods. The analysis program has established processes for Short Tandem Repeat (STR) analysis but a feasibility study using Single Nucleotide Polymorphism (SNP) data has also been proven.

This presentation will impact the forensic science community by not only using a standardized approach but will also show the ability to include concepts of both STRs and SNPs for better discrimination power.

Computing likelihood ratio in kinship analysis for autosomal markers is straightforward and well defined. Such calculation provides a value for evidence given the prosecution-versus-the-defense proposition. It is recommended and widely used in forensics, missing persons cases, and paternity cases. The forensics community has validated stand-alone software for calculating Likelihood Ratio (LR) using trios and many biologically related family members. Software such as Familias and MPKin™ FS Edition are used regularly for such calculations.¹⁻³ Because these implementations are stand-alone, transcription errors can occur on transferring data from data collection, table input, and result storage; in addition, it can also be time consuming.

LT has incorporated its version of a kinship algorithm, based on the Error Standard (ES) algorithm, to data collection and storage for ease of use and reliability of results, therefore avoiding human transcription errors.²⁻⁵ This presentation will encompass the steps taken by this study to validate the kinship analysis algorithm, given the available methods, data, and external collaborators. Building on previous literature, this study used the National Institute of Standards and Technology (NIST), the Council on Education for Public Health (CEPH), and real data from collaborators to compare results of the kinship algorithm to those currently used in the paternity and forensics laboratories. This study shows that the standard calculations, including complex pedigree trees, mutations, and rare alleles concur with currently used methods.

Through this work, the LT-kinship algorithm, a more flexible implementation with state-of-the-art models, has been established as accurate. The algorithm with SNP data has been further tested, showing that, for a small number of SNPs, the algorithm produces LR values, which may be an option once expert data and tables become available.

For Research, Forensic, or Paternity Use Only. Not for use in diagnostic procedures.

References:

1. Azevedo, D.A., Souza, G.R.B., Silva, I.H.E.F. & Silva, L.A.F. Genetic kinship analysis: A concordance study between calculations performed with the software Familias and algebraic formulas of the American Association of Blood Banks. *Forensic Sci. Int. Genet. Suppl. Ser. 3*, e186–e187 (2011).
2. Ge, J., Budowle, B. & Chakraborty, R. DNA identification by pedigree likelihood ratio accommodating population substructure and mutations. *Investig. Genet.* 1, 8 (2010).
3. Budowle, B., Monson, K.L. & Chakraborty, R. Estimating minimum allele frequencies for DNA profile frequency estimates for PCR-based loci. *Int. J. Legal Med.* 108, 173–6 (1996).
4. Brenner, C.H. Symbolic kinship program. *Genetics* 145, 535–542 (1997).
5. Minn, A., Deng, J., Ge, J., K, Makesh., Rajagopalan, N. Integrated Forensic DNA Data Analysis and Management – A scalable enterprise solution for Next Generation Sequencing.

Kinship Algorithm, STRs, SNPs