A20 An Interesting Mutation at Locus DYS385 in an Uncle-Nephew Pair in a Fatherless Paternity Case

Massimo Lancia, MD, University of Perugia, Via del Giochetto, snc, Perugia, 06126, ITALY; Eugenia Carnevali, MD, Hospital of Terni, Via T. di Joannuccio, snc, Terni, 05100, ITALY; Alessio Coletti, MD, INPS, Via Canali, 5, Perugia, 06122, ITALY; Simona Severini, PhD, and Susanna Massetti, PhD, University of Perugia, Via del Giochetto, snc, Perugia, 06126, ITALY; Luigi Carlini, MD, Hospital of Terni, Via T. di Joannuccio, snc, Terni, 05100, ITALY; and Marina Dobosz, PhD, and Mauro Bacci, MD, University of Perugia, Via del Giochetto, snc, Perugia, 06126, ITALY*

After attending this presentation, attendees will understand how the Y-chromosomal short tandem repeat polymorphisms (Y-STRs) included in the AmpFlSTR® Yfiler® amplification kit are currently used for forensic and evolutionary applications, therefore a consistent knowledge on mutation properties is necessary for correct data interpretation.

This presentation will impact the forensic science community by presenting the necessity to know mutation rate of Y-STRs according to ISFG guidelines.

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Recently, in a fatherless paternity case, an interesting mutation at locus DYS385 in an uncle/nephew pair was observed. The alleged father was unavailable due to his death several years ago in South America; where he is currently buried. Consequently the paternity test was conducted without the father's DNA profile.

DNA typing was conducted on the reference samples from the son, his mother, and his alleged uncle (father's brother) using AmpFlSTR® Identifiler® amplification kit. The obtained data showed that in this particular fatherless case the analysis of 15 STRs was not sufficient to establish the paternity. The paternity probability value was improved by conducting additional DNA typing using three miniSTRs markers (NC01 systems: D10S1248, D14S1434, D22S1045) and AmpFlSTR® Yfiler®. The combined DNA profiles obtained were statistically analyzed with Probabilistic Expert Systems (PES) FINEX and Familias; a high paternity probability value was of P=0.9993 obtained.

The relationship between the son and his uncle was confirmed by conducting Y STR typing using the AmpFlSTR® Yfiler® amplification kit. However, an incompatibility at locus DYS385 was observed; the uncle's genotype was 15-16 opposing the nephew's genotype of 14-16 at that locus. A second amplification of the son and the alleged uncle samples was conducted using the PowerPlex Y amplification kit (Promega) and the mutation was confirmed.

PCR was carried out using AmpFlSTR® Identifiler®, AmpFlSTR® Yfiler® (Applied Biosystems), PowerPlex Y (Promega) amplification kits, and three miniSTRs markers with a homemade multiplex. All PCR products were detected by capillary electrophoresis in the ABI Prism310 Genetic Analyzer and alleles were typed using Genemapper software. Experiments were performed according to the ISFG guidelines.

Sequencing of the DYS385 locus was performed. DNA was extracted from two blood samples using DNA IQ™ System (Promega) according to the manufacturer's protocol. DYS385 alleles were amplified using available primers on Gene Bank (Accession code: AC022486). The separated alleles were removed by means of surgical scalpel and placed in a spin tube with Chelex (20%) incubated overnight at 56°C. Three cycles of freezing – thawing were performed. The amplification products were recovered, re-amplified using the same conditions, purified with Exosap, sequenced with BigDye Terminator v 1.1 kit (Applied Biosystems), re-purified with Centrisep Columns and finally detected by capillary electrophoresis in the ABI Prism 310 Genetic Analyzer. The sequenced alleles showed a regular repeat structure [GAAA] with 15 repeats in one case and 14 repeats in the other.

Mutation Rate, Y-STR, Probabilistic Expert Systems

 ** Presenting Author*