A case report of a- family with a rare mutation in the Amelogenin gene: Comparison of two different approaches to resolve the issue of correct sex typing.

F.H. Alshamali[1], A.I.Alkhayat[1], B. Budowle[2] and N.D.Watson[3].

1.Crime Laboratory, Dubai Police GHQ., Dubai, United Arab Emirates.
2.Laboratory Division, FBI, Quantico, VA, USA
3.Forensic Science Unit, Strathclyde University, UK.

Y Chromosome STRs provide useful information in paternity investigation cases, such as deficiency cases involving male offspring and paternal lineage identification involving immigration cases.

This presentation describes an immigration case of a family comprised of a mother, a father and 5 male offspring. Blood samples were collected on FTA cards. One 1.2 mm washed FTA® disc punch was added to a reaction tube containing 15 µl of master mix and 10 µl of purified water following the manufacturer's recommendations (AmpFlSTR® Identifiler amplification kit, Applied Biosystems, Foster City, CA, USA). Electrophoretic separation was conducted using ABI 310 genetic analyzer. Data were analyzed by GeneScan version 3.7.1 and Genotyper 3.7.2 and compared with manufacturer supplied allelic ladders.

The biological relationship of the mother, father, and 5 male children was verified by autosomal STR typing. The father and all 5 male children showed a dropout of the 112 bp amelogenin Y allele. All were phenotypically normal males.

Two different approaches were followed to investigate the apparent amelogenin Y allele dropout. The first method was Y chromosome STR typing using the PowerPlex® Y System (Promega Corporation, Madison, WI)) which allows co-amplification and three-color detection of twelve loci: DYS19, DYS385a/b, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439. Results showed that the father and the 5 children all carry a complete Y-STR haplotype and except for a variance at one locus for one of the children's, they all had the same haplotype. The second method was using the Quantifiler™ Y Human Male DNA Quantification Kit (Applied Biosystems, Foster City, CA, USA) with the ABI Prism 7000 and SDS software (Applied Biosystems, Foster City, CA, USA), which is a real-time PCR assay. All males tested positive. Results will be shown in detail.

In conclusion, reliable DNA-based sex identification is a major concern for forensic investigation and paternity assessment. This case demonstrate a simple, rapid and cost-effective PCR-based methods for evaluating cases where amelogenin results may be questionable.