B41 SNPs by MALDI-TOF MS: A New Tool for Highly Degraded DNA Samples?

Elizabet Petkovski, Bertrand Ludes, MD, PhD, Christine Keyser- Tracqui, Sylvain Amory, and Remi Hienne, PhD, Codgène, 11, rue Humann, Strasbourgh, 67000, France*

After attending this presentation, attendees will have information regarding the Matrix Assisted Laser Desorption Ionization Time-of-Flight Mass Spectrometry (MALDI-TOF MS) method of DNA typing.

This presentation will impact the forensic community and/or humanity by demonstrating to the forensic community the advantages but also the limits of a relatively innovative technique in the field of the DNA typing.

DNA profiling technology has revolutionized the analysis of crime scene evidence, allowing the establishment of genotypes even from minute amounts of biological material. Nowadays, forensic experts are more and more often confronted with the task to obtain results from highly degraded DNA samples, which is difficult with the commonly used STR markers. In this context, efforts have been made in the development of new method- ologies implying the SNPs. Today, several technologies are used to perform SNP genotyping among which the Matrix Assisted Laser Desorption Ionization Time-of-Flight Mass Spectrometry (MALDI-TOF MS).

In order to test the potential of this method in the study of highly degraded DNA, 50 autosomal SNPs were selected on the basis of conser- vation, localisation, lack of phenotypic expression, and allelic frequencies in different populations. A single nucleotide sequence difference between the amelogenin genes carried by the sexual chromosomes has also been selected for sex determination. These 51 SNPs were analysed and vali- dated on a French representative population comprising 21 inclusionary, 3 exclusionary paternity cases and a total of 81 unrelated individuals. This approach to SNP typing is a multiplex PCR based amplification followed by simultaneous detection by primer extension (PEX). Product analysis is accomplished by Matrix Assisted Laser Desorption Ionization Time-of- Flight Mass Spectrometry (MALDI-TOF MS).

The selected SNPs showing independent inheritance and giving clear results in paternity and identification testing (and being therefore suitable as markers in the field of forensic genetics), the usefulness of this approach was investigated on highly degraded samples, the real target of these devel- opments. The results obtained by these assays revealed some difficulties, notably in the reproducibility of the results obtained due to steps which are not entirely controllable. Thus, the aim of this work was to determine whether these obstacles were due to the multiplexing approach or to the degraded nature of the target DNA.

The presentation lists the problems encountered with the development of this new SNP typing method and tries to answer the following question: to what extend is the applied technology a step forward in the analysis of highly degraded samples?

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 ** Presenting Author*