B176 A Molecular Assessment of DNA Methylation Profiling For Body Fluid Identification in a Forensic Application

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After attending this presentation, attendees will better understand the use of epigenetics and methylated DNA in forensic practice as a tool for the identification of different biological fluids (blood, saliva, semen, and menstrual blood).

This presentation will provide navigation through the roles of epigenetics and DNA methylation in forensic applications and illuminate their uses in the identification of different bio-fluids. This presentation will impact the forensic science community by offering new epigenetic markers that are proven to be of value in this area and will help forensic science practitioners use a simple tool for the identification of blood, saliva, semen, and menstrual blood in different age and gender groups.

Background: Since 1985, when DNA analysis was applied to solving forensic problems, its foremost applications have included criminal investigation, personal identification, and paternity testing. DNA methylation is emerging as an attractive marker in forensic genetics that can provide investigative leads to help solve crimes. Natural roles of DNA methylation in mammalian systems include imprinting, X chromosome inactivation, heterochromatin maintenance, developmental controls, and tissue-specific expression controls.

DNA methylation plays a critical role in normal cellular processes and serves as a mechanism that turns off gene expression. Aberration in DNA methylation has long been linked to diseases such as cancer and is exploited as diagnostic biomarkers, but in forensic analysis, the use of DNA methylation as a tool is still in its infancy; however, there are many advantages to using DNA methylation as a forensic tool, with one of the most important being the stability of the marker, unlike protein or RNA markers that are quickly degraded, thus allowing for quantitative analysis of older samples.

Forensic applications of DNA methylation include: (1) the identification of body fluids; (2) differentiating Monozygotic (MZ) twins; (3) DNA methylation in age estimation; (4) the determination of paternal origin of allele; (5) the determination of cause and circumstances of death; (6) sex determination; and, (7) the authentication of DNA samples.

Goal: This study will explore the different applications of DNA methylation in forensic science with reference to the results of a study on Egyptian DNA methylation profiles of four markers (LINE-1, MT2A, MGMT, and FGF7) in blood, saliva, semen, and menstrual blood in attempt to assess the potential of those markers for identification.

Methodology: For this purpose, 52 samples from female participants and a similar number of samples from male participants were gathered. To explore age variation, each sample was divided into two groups of 26 each, one group more than 30 years of age and the other less than 30 years of age. Each sample was subjected to the following procedures: DNA extraction and amplification of the collected samples, then a bisulfite treatment of the amplified DNA samples. The bisulfate-modified DNA was used as a template for fluorescence quantitative Polymerase Chain Reaction (qPCR) assessment.

Results and Conclusion: The MGMT locus exhibited a differential methylation pattern in blood compared to semen, saliva, and menstrual blood. Therefore, the MGMT marker exhibited significant differences in its methylation patterns for the identification of blood when compared to the other fluids. MT2A was presumed to show a differential methylation pattern in saliva as it displays hyper-methylation state, but hyper-methylation is also seen in semen. No significant difference was seen between menstrual blood and other body fluids. These factors render MT2A not useful in differentiating between body fluids. The FGF7 marker displayed a differential methylation pattern in semen. Methylation values were greater in semen relative to blood, saliva, and menstrual blood. The methylation profile of LINE-1 successfully differentiated saliva from the other three examined biofluids. This marker displayed hyper-methylation in all saliva samples and hypo-methylation in all blood, semen, and menstrual blood samples. Regarding the use of these markers in differentiating between males and females, as well as between the different age groups (less than and more than 30 years of age), highly statistically significant differences were obtained.

DNA Methylation, Biological Fluid, Identification