

Methylation Profiling and Validation of Candidate Tissue-Specific Differentially Methylated Regions for Identification of Human Blood, Saliva, Semen and Vaginal Fluid and Its Application in Forensics

Authors : Meenu Joshi, Natalie Naidoo, Farzeen Kader

Abstract : Identification of body fluids is an essential step in forensic investigation to aid in crime reconstruction. Tissue-specific differentially methylated regions (tDMRs) of the human genome can be targeted to be used as biomarkers to differentiate between body fluids. The present study was undertaken to establish the methylation status of potential tDMRs in blood, semen, saliva, and vaginal fluid by using methylation-specific PCR (MSP) and bisulfite sequencing (BS). The methylation statuses of 3 potential tDMRS in genes ZNF282, PTPRS, and HPCAL1 were analysed in 10 samples of each body fluid. With MSP analysis, the ZNF282, and PTPRS1 tDMR displayed semen-specific hypomethylation while HPCAL1 tDMR showed saliva-specific hypomethylation. With quantitative analysis by BS, the ZNF282 tDMR showed statistically significant difference in overall methylation between semen and all other body fluids as well as at individual CpG sites ($p < 0.05$). To evaluate the effect of environmental conditions on the stability of methylation profiles of the ZNF282 tDMR, five samples of each body fluid were subjected to five different forensic simulated conditions (dry at room temperature, wet in an exsiccator, outside on the ground, sprayed with alcohol, and sprayed with bleach) for 50 days. Vaginal fluid showed highest DNA recovery under all conditions while semen had least DNA quantity. Under outside on the ground condition, all body fluids except semen showed a decrease in methylation level; however, a significant decrease in methylation level was observed for saliva. A statistical significant difference was observed for saliva and semen ($p < 0.05$) for outside on the ground condition. No differences in methylation level were observed for the ZNF282 tDMR under all conditions for vaginal fluid samples. Thus, in the present study ZNF282 tDMR has been identified as a novel and stable semen-specific hypomethylation marker.

Keywords : body fluids, bisulphite sequencing, forensics, tDMRs, MSP

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