A Case Study of Misinterpretation of Results in Forensic DNA Cases Due to Expression of Y- Chromosome in Females

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Abstract : The gender of an individual in forensic DNA analysis is normally accessed by using the STR multiplexes with the incorporated gender based marker amelogenin or in other words by presence or absence of Y-Chromosome, but it may not be true in all the cases. We hereby report an interesting case of a phenotypic female carrying a male karyotype (46XY). In the alleged murder case, the deceased female with XY genotype was noticed. The expression of 18 Y-linked genes was studied to measure the extent of expression. Expression at 4 loci was observed that might have caused the misinterpretation in forensic casework. This clinical situation of the deceased in this case was diagnosed as testicular feminization syndrome, which characterize a female phenotype with a male karyotype (46, XY). Most of these cases have SRY (testis determining factor). The genetic explanation of this phenomenon is not very clear. Here, we are discussing the impact of such situations of genetic discrepancy in forensic interpretation of results. In the presented murder case of a phenotypic female, sexual assault was also suspected. For confirmation vaginal swabs and micro slides were also sent to us for DNA examination. After DNA analysis using STR markers, Y-chromosome was detected in the samples which supporting the suspicion of sexual assault before murder. When the reference blood sample of the deceased was analyzed, it was found to be case of testicular feminization syndrome. Interesting inferences were made from the results obtained.

Keywords: DNA profiling, forensic case study, Y chromosome, females

Conference Title: ICFS 2017: International Conference on Forensic Sciences

Conference Location : Paris, France **Conference Dates :** August 28-29, 2017