

Y- STRs and AZF microdeletions in clinical context samples

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Forensic geneticists use several Y-STR PCR amplification kits, which are useful in forensic identification, ancestry studies and genealogies reconstruction. The Y-chromosome regions with forensic interest include the set of 16 loci defined in the Y-Chromosome Haplotype Reference Database. Nevertheless Y chromosome is connected to male infertility and microdeletions in it are the most common cause of genetic origin male infertility. The study of Y chromosome AZF (Azoospermic Factor) region is one of the strategies to diagnose it. This study aimed to characterize the AZF region microdeletions by Y-STRs and STS studies and its possible impact in forensic casework.

Selected samples from fifty-two patients studied for male infertility, collected under informed consent, were characterized at molecular level with specific STS for the presence/absence of the three AZF regions: AZFa - DFFRY3, DBY; AZFb - sY1227, sY1224, sY134, sY119, sY134, RBMY1, sY143; AZFc - sY1192, sY254, RRM3, sY1291, sY283, sY1201. AmpF ℓ STR ® Yfiler ® PCR Amplification ® kit (AB) was used to obtain a 16 Y-STR profile.

All the 52 samples were concordant in forensic and molecular studies, although with different scenarios: a) a normal Y-STR profile and no deletion with STS in the AZF region were revealed in about 32% of the studied samples; b) the majority of samples, about 51%, revealed also a normal Y-STR profile, but with a complete or partial deletion(1 or 2 STS) in the AZFc region; c) deletion in DYS385, DYS392, DYS448 and in AZFb+AZFc were detected in 3 samples. Four samples have the following different scenarios: d) deletion in DYS385, DYS392, DYS448 and in AZFb; e) deletion in DYS390, DYS391, DYS392, DYS385, DYS438, DYS439, DYS458, DYS635 and in AZFa; f) deletion in DYS448, in STS sY1197 and in AZFc; g) deletion in DYS390, DYS392, DYS385, DYS448, GATAH4 and in AZFb+AZFc region. All Y-STRs studied for forensic casework are localized, essentially, in the AZFa or AZFb regions, although DYS448 is located in the distal AZFb region and DYS390 and GATAH4 are localized between AZFa and AZFb regions. Deletion in the AZFc region does not affect results in forensics.

The knowledge of Y-chromosome microdeletions is important in Forensic Genetics as this can be encountered in current casework without possibility to perform clinical studies. So, it is crucial to know how to interpret the results obtained in Y-STR microdeletions samples, according to Y-chromosome structural alterations. As Y-STRs used in Forensic Genetic Laboratories are located in the AZF region, associated to male infertility, this can raise some ethical problems in Forensics.

Key-words: Y-STRs; Y-Filer; STS; AZF regions; microdeletions

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