

STR GENOTYPING USING ION TORRENT PGM AND STR 24-PLEX SYSTEM: PERFORMANCE AND DATA INTERPRETATION

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Massively parallel sequencing (MPS) platforms allow the simultaneous analysis of thousands to millions of DNA fragments, generating large amounts of data in a relatively short time frame compared with traditional sequencing methods. The aim of this study is to examine parameters for data interpretation generated for STR genotyping with a prototype 24-plex panel using Ion Torrent PGM. One male DNA reference sample was amplified with a 24-plex STR panel (kindly provided by Thermo Fisher Scientific) and sequenced in Ion Torrent PGMTM. It was considered the numbers of alleles discriminated and the number of background reads from the number of total reads. Also parameters such as the percentage of stutter peaks, heterozygote peak high ratio (PHR) and sequence variants were evaluated. The full DNA profiles, with DNA input as low as 0.037 ng, corresponded to previous capillary electrophoresis (CE) results. Deep sequencing reads of 10,000 allowed the observation of intra-allelic variation on 5 loci. This study shows that MPS applied to STR genotyping improves accuracy and mixture interpretation.