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Insights on Short Tandem Repeats

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Description

Short Tandem Repeat (STR) typing methods are widely used today for human identity testing applications such as forensic DNA analysis. Following multiplex PCR amplification, DNA samples containing the length-variant STR alleles are typically separated by capillary electrophoresis and genotyped by comparison to an allelic ladder supplied with a commercial kit. This article provides a brief overview of the technologies and issues involved in STR typing. Short tandem repeats (STRs), also known as microsatellites or simple sequence repeats (SSRs) [1-3] are accordion-like stretches of DNA that contain core repeat units ranging in length from two to seven nucleotides and are randemly repeated from a half dozen to several dozen times. Despite the fact that the human genome contains thousands upon thousands of STR markers, only a small core set of loci has been chosen for use in forensic DNA and human identity testing. Core loci, similar to using a single, common currency in a financial sense, allow equivalent genetic information to be shared and compared. Commercial kits for generating DNA profiles containing these core STR loci are now available.

STR data interpretation typically begins with a comprehensive examination of a DNA profile. Off-scale data and the resulting pull-up peaks may indicate that there was too much DNA template in the multiplex PCR reaction. The absence of signal from larger-sized STR loci indicates the presence of PCR inhibitors or degraded DNA. The presence of more than two alleles at a locus can indicate a potential mixture though it is critical not to focus on a single locus due to the possibility of tri-allelic patterns. The X/Y allele ratio from the amelogenin sex-typing primer pair can also help with DNA mixture interpretation. Inter-locus balance within a dye channel can provide an analyst with information about potential PCR inhibition or DNA degradation. The inter-locus balance of dye channels can provide information to a STR kit developer about dye sensitivity and PCR primer balance. This is a well-known mechanism for the production of large blocks of satellite DNA [4,5] It is linked to the transfer of repeat units between homologous chromosomes. However, because this process involves multiple chromosomes, it has a limited role in STR mutation. Nonetheless, this mechanism may be responsible for STR multistep mutations, as discussed later. This mechanism postulates that A-rich STRs are produced by a 3' extension of retrotranscripts, similar to how mRNA is polyadenylated.

There is evidence of a link between the most common human STRs with A-rich content and transposable elements. A high density of transposable elements, on the other hand, does not always correspond to a high density of STRs. More research is needed to determine whether this is a true mechanism for STR mutation. According to this mechanism, A-rich STRs are produced by a 3' extension of retrotranscripts, similar to how mRNA is polyadenylated. There is evidence that there is a connection between the most common human STRs with A-rich content and transposable elements. However, a high density of transposable elements does not always correspond to a high density of STRs. More research is needed to determine whether this is a true STR mutation mechanism.

Conflict of Interest

None.

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