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Copy Number Variation in Forensic Science

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Legal importance of DNA evidence for forensic cases is much vital, so there is constant and consistent research going on to make this type of evidence more compelling [1]. Investigators are searching for the pattern of deleted and duplicated regions of large chunks of DNA sequences consisted of ten thousand to five million letters, which is known as Copy Number Variation (CNV) [2]. Such mutation has often been overlooked in previous surveys of mutations that cause genetic diseases. It's not yet completely understood to researchers that what proportion of a genetic disease is caused by CNV. A number of advanced technologies allow us to scan the entire human genome for CNV in a single experiment. Researchers are characterizing functionally-relevant CNVs at the highest possible resolution, incorporating these variants within association studies for complex diseases, and developing a public resource to facilitate integration of CNV within medical genetic studies [3].

Compared to Short Tandem Repeats (STRs), Single Nucleotide Polymorphisms (SNPs) and Variable Number of Tandem Repeats (VNTRs), CNVs are larger in size and can often involve complex repetitive DNA sequences. Due to its large size, many researchers have proposed it for designing personalized medicines [4]. They are reported to be associated with different functions. Some CNVs could be employed to add favoritism power in forensics. Forensic DNA typing often requires the use of techniques that allow the detection of genetic variations among humans, usually short repeats [5]. VNTRs polymorphism was used till few years ago which was then replaced by STR by the new century scientists. CNVs are supposed to be major determinants of human traits, and they may become useful in forensic science, particularly in the determination of the population substructures [6].

Observation of anomalous STR haplotypes in forensic cases and population studies may lead to discovery of new CNVs [7-9]. While studying haplotypes, when a single-copy STR is absent not due to primer-site mutation, it is indication of a deletion. Alternatively, if there is a double peak, it may signal the presence of a duplication variant, although care should be taken to distinguish this from mosaicism, particularly in cell-line DNA [10,11]. STR based duplication can be detected only when there is a sizeable difference between the two alleles but Quantitative methods are used when the two alleles are identical in length. When more than one copy of STRs are present in reference sequence, other CNVs can be observed. For instance, there may be three or four alleles observed for the DYS385, normally a bi-locus allele [9]. The physical extent of CNVs can give the indication of putative deletion or duplication involving two or more STRs [12].

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In conclusion: CNVs may be used for personal identification and population based studies. It is suggested that high throughput sequencing of CNV regions be done using next generation sequencing technology. In the future, forensic scientists and bioinformaticians may work together for finding possible ways to learn more about the variation of gene dosage and its influence on personal identification in forensic sciences. Further research may improve the utilization and benefits of Copy Number Variation in forensic investigations.

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