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Ancestry Lineage through Short Tendem Repeats (X-STR)

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Abstract

In the mid 1980s, forensic DNA testing has been introduced It has played a very comprehensive role in providing service to the mankind *via* race finding, population determination, cast analysis, parental lineage, declaring punishment to the culprit and providing relief to the innocent. Remains of the victims of the mass disasters and of the missing people have been recombined for the identification purpose through linking them with the reference samples. The advancement of the new technologies have enhanced the capabilities of the laboratories working in recovering DNA results with more accuracy and improved sensitivity. Forensic laboratories have fully adopted automation for the sample preparation and for data analysis in order to provide efficient results. In forensic DNA analysis Short Tandem Repeat (STR) typing continues to be the most dependable and durable one although other genetic markers are also available and used for specific purposes.

Keywords: SARS-CoV-2 • Hydroxychloroquine • Azithromycin • Chloroquine

Introduction

Short Tandem Repeats (STR) typing of markers that are located on the sex chromosomes have been developed into a method which is in use of many forensic laboratories when providing additional individualizing information while making the use of widely used autosomal STRs over the past two and a half decades [1]. The inheritance patterns of X and Y chromosomes differ both from each other and from the autosomes, markers that are located on the sex chromosomes are durable enough to provide different but complementary information essential in solving forensic caseworks. In the United States especially the markers present on X chromosome are less universally observed Forensic use of Y STR markers is generally accepted and well documented by both the courts of law and the scientific community. However both X and Y marker systems continue to be the topic of pious scientific publication and research. Recent advances providing ever broadening scope in forensic scenarios by offering opportunities to apply these markers [2]. Markers such as Single-Nucleotide Polymorphisms (SNPs) and Insertion/Deletion Markers (INDELs) have been investigated on X and Y chromosomes for the autosomes other than STRs but here in this article the focus is on the forensic development of human specific (no other species) X and Y STR markers specifically. In forensic DNA analysis, the specific part of human sex chromosomes are widely being used specifically in those cases where standard autosomal DNA profiling is found not much informative [3].

A gene fragment of the sex chromosome is applied for the identification of the biological sex of the trace donor of crime scene [4]. Characterization of the haplotypes composed of X or Y chromosomal STRs polymorphism have been done in paternal lineages of unknown trace donors, more suitable when males and females have been associated to the same trace such as in the cases of sexual assault.

Literature Review

DNA testing is always preferred as the most reliable and authentic one whenever there is a need to get essential and accurate information. Advancement in DNA testing has made people enable to know about their genetic ancestry and also get a chance to know that where some of their ancestors actually came [5]. Suppose if I considered myself as an African American, I don't know where my African ancestors actually originate from. I can only point Tennessee as the only geographic location if I have to mention about my ancestral home. This new generation of tests for genetic ancestry has captivated my attention towards itself *via* providing potential knowledge and made me able to know about myself more precisely at genetic level [6].

Both X and Y STRs have played essential role in forensic scenarios although for each marker type the role is different providing varying benefit in addition to autosomal STRs that are already in use.

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The inheritance pattern of both chromosomes is unique and is considered while understanding their application in forensic casework [7]. Other than this with the autosomes where one chromosome of each pair from both the parents is passed to both male and female offspring, in the female offspring X chromosome comes from both the mother and father while in the male offspring X chromosome only comes from the mother. The Y chromosome is present in males only and is entirely passed down from father to its son and a profile known as haplotype is formed from the combination of alleles. Hence, the male offspring have one X chromosome from the mother and one Y chromosome from the father and the female offspring have one X chromosome from the mother and one X chromosome from the father [8-11].

During meiosis the two X chromosomes of female recombine in the same way as in autosomes. Almost the entire chromosome is passed from parent to child unchanged in case of paternally inheritance where recombination only occurs within the two small regions present at the distal ends of the chromosomes known as Pseudo Autosomal Regions (PAR). Additional considerations are needed for linked markers during interpretation of X and Y STR results because currently no STR markers exists within these regions when utilized for forensic purposes [12-16].

In STR typing many commercially multiplexes are available and are in use for the identification of the human remains after mass disasters or in mass graves. In order to get immediate information on the specimen the sample extract is preserve in large multiplex [17-19]. If multiple countries or laboratories are participating in the testing, the decision has to be made as early as possible about which set of markers has to be used. In international collaborations even it is essential that all laboratories must test the same loci. The reagents must be in approach to all the participants and the markers must be well established in the countries and in the forensic community involved [20,21].

There are wide range implications of ancestry inference through X-STRs in forensic as reliable genetic source like achievement of identification of skeletal remains at historical and grave sites, perceived ethnicity of deceased individual when information is limited or conflicting, donors ancestry confirmation, refining STR based familial research strategies and for enhancing the sensitivity of forensic studies [22].

Discussion

X and Y STRs provide required genetic information to answers for questions regarding paternity cases, lineage finding, and mass disasters identification. Sex chromosomes related STRs are more preferable as compared to autosomal STRs as are helpful to provide more information about maternal and paternal lineage, migration patterns and inter-relationships [23,24].

Inheritance of X STRs is simpler and are used commonly for paternity cases as compared to autosomal STRs. Whereas, Y STRs are helpful to find the extinct of common ancestry, paternal lineage, race determination and much more. Analysis of autosomal STRs will provide statistical strength to X and Y STRs results, but hereditary inconsistencies complicate the interpretation of autosomal STRs. Similarly, in recent forensic study of paternity case found that alleged father is close relative of girl's true father, universally X and Y STRs are more informative for forensic analysis then autosomal STRs. Alleged criminal, paternity and race determination cases provides detail about more informative side of X and Y STRs. X and Y STRs are inherited in predictable way that's why they outcome the informative side of autosomal STRs [25-29]. Besides routine paternity and lineage investigation, STRs analysis is much more helpful in mass disasters where we don't have any other tool for forensic analysis of victims of different races and origin. STR analysis is cost efficient way to explore more discrimination among mass disasters victim as well as finding of reference family and race [30,31].

So how all of this is done? Multiplex X and Y STRs kits are available commercially that utilized small amplicons for forensic study. Hundreds of X and Y STRs on chromosomes are discovered so far, Y STRs comparison study is based on 9 core markers including DYS19, DYS 385 a/b, DYS390-93, DYS389i, DYS389ii, this heptad is collectively called "minimal haplotype". These are generally used for forensic and genetic analysis including population studies, lineage, paternity and disasters. Commercially available and most commonly used Y STRs analysis kits include "PowerPlex Y" "ampF/STR" "Yfiler Plus" Varied multiplex analysis are available non-commercially with similar and supplementary loci as target for study [32-35].

As compared to Y STRs, X STRs are more diverse, non-standardized globally and are differentially used by different communities [35]. For compiling more relevant information using X STRs, we have to move towards more standardized approach for whole communities, races and continents. Many companies are manufacturing commercially available X STRs kits which include "Qiagen X-12" "Mentype Argus X-UL "Argus X-8 (8-multiplex)". The current variant is X-12 which includes 12 multiplex for X STRs analysis as more and more laboratories are demanding X-STRs as we are moving forward. Increase in literature on X and Y STRs provide the potential for comparison analysis using the data published by different laboratories. Extensive literature was search and selective X-STRs are studied to find the lineage and comparative analysis of major communities and races of the different regions of the globe [34]. Randomly 20 X-STRs markers namely, HumARA, DXS6803, GATA165B12, DXS981, DXS6800, GATA31E08, DXS9898, DXS6807, DXS6809, GATA172D05, DXS7133, DXS8378, DXS7424, DXS7132, DXS8377, DXS6789, DXS7423, HPRTB, DXS101are analyzed and compared from forensic studies conducted in Pakistan and related countries. Following data is generated, that shows similarity and differences among different races in X-STR markers identification studies among masses of different countries.

Conclusion

Forensic analysis at molecular level offers the promise to uncover the underlying important information. Enormous progress was made in last 20 years due to advancement in DNA typing technologies like SNPs, STRs, INDELs, etc. Nowadays Short Tendem Repeat (STR) are prominently used for different aspects of forensic analysis. Though, autosomal STRs have their importance as workhorse in forensic analysis, X-STRs are becoming efficient forensic analysis tools aspect due to more accuracy and enhanced sensitivity. Many different X Chromosomes STR markers are discovered so far and are playing essential role in providing information about origin of individual or remain where other sources of information are absent. Many articles are thoroughly reviewed to extract information on X-STRs, 20 X-STR markers are selected, studied and analyzed in different populations to find their role in realizing the origin. The generated data shows that different populations have some similarity in X-STR markers, showing less and more common descendants among groups studied and geographical linkage between these distinct but related populations.

X-STRs are proving their prominence to the forensic world due to their role in achievement of identification of skeletal remains at historical and grave sites, perceived ethnicity of deceased individual when information is limited or conflicting, donors ancestry confirmation, refining STR based familial research strategies and for enhancing the sensitivity of forensic studies. Further epithet of X-STR is required to enlighten the future of forensic analysis.

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