Review Article

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Y-Chromosome in Medical and Forensic Genetics: A Systematic Review

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Abstract

Y chromosome plays key role in medical and forensic application especially paternal inheritance. Beyond the sex determination, several Y chromosome linked genetic disorders can be diagnosed. 5% of the Y chromosome comprises of pseudoautosomal regions (PAR1 and PAR2) on the Yp arm and Yq arm respectively. Remaining 95% of the Y chromosome does not recombine during meiosis. The non-recombining region of the Y chromosome is designated as "NRY" (non-recombining region). NRY region includes the SRY gene or Male Specific Y (MSY) chromosome. In view of the fact that the Y chromosome is traversed to next generation, it is frequently used in genealogy to track the male ancestry. Both medical and forensic aspects of Y chromosome have been reviewed systematically according to the advancements of technology in the molecular genetics field.

Keywords: Y-STR, Medical aspect, Forensic aspect, Chromosome, Database

Introduction:

Beyond determining sex, the Y chromosome accomplishes remarkable role in solving the crime cases. 5% of the Y chromosome comprises of pseudoautosomal regions (PAR1 and PAR2) on the Yp arm and Yq arm respectively. Remaining 95% of the Y chromosome does not recombine during meiosis (NRY). Because of the non recombining regions of y chromosomes, it has a single copy of genetic material any mutation in this region may cause malfunctioning of spermatogenesis in males. In view of the fact that the Y chromosome is traversed to next generation, it is frequently used in genealogy to track the male ancestry. Each of these sequences is necessary for the spermatogenesis process (Waters et al., 2007).

Medical Genetics Aspect:

It is well established that 5% of human DNA is said to be expressive while 95% is known to be Junk DNA. Some regions of the non-coding DNA may help in defining the chromosome structure and translation location. In the field of medical genetics, expressive segments of Y-chromosome contain 50-60 genes that give directions for creating proteins through translation. However with the newest Malaysian study it is found that the Y chromosome contains 70-200 genes[1]. Out of these, 16 genes are responsible for cell maintenance, 9 genes are concerned with sperm cell production, and if some units in this area are lacking or inoperative, azospermic or physiological state may occur. The SRY gene sequence is accountable for the maleness. This SRY gene regulates the expression of Sox9

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segment of autosomes. If the SRY sequence is mutated, the embryo can develop female genitals in XY genotype individuals. Mutations in Sox9 or other related genes can result in sex reversal or hermaphroditism. The SRY sequence has a simple structure, with only one deoxyribonucleic acid and no introns[2].

Adenine and Thymidine rich two segments are localised in SRY gene which are inverted repeat sequences, implying that this gene originated from the retro position of another gene's transcript to the entire human evolution. SRY is known to regulate the expression of sex-determining genes, amend chromatin architecture, and commence prime cascades that determine the destiny of sertoli cell lineages in the growing testis (Waters et al., 2007). The ampliconic sequences are peculiar, having high density of genes and the most repetitive segments of the Y chromosome. They are built up entirely of large spells of duplicated sequence that are frequently arranged in palindromic pattern to contain multicopy gene pairs with testis-specific coding sequences (Hughes & Rozen, 2012).

Pseudoautosomal regions of Y-chromosome which are localised at telomeres show resemblance with its counterpart X-chromosome. Owing to this crossing over takes place only at these pseudoautosomal regions. Rest of the 95 percent of Chromosome Y is known as ChrY, the male-specific region (MSY), formerly called as the NRY (non-recombining region). Y to Y and X to Y gene conversion and de novo mutations are responsible for the new variant of MSY over the years. One study found that Y-to-Y gene conversion happened at a pace of 600 nucleotides per ChrY in a newborn male during recent human evolution (Parker & Erzurumluoglu, 2020). It was supposed that some genes of Y chromosome are lost and they are displayed to another chromosome.

Ballotter compared the Y chromosomes of eight mammals, including humans, chimps, Rhesus macaque, mice, rats, bulls, and opossums, and discovered that 18 different genes were to be present between these species[4]. The distinction within the organic phenomenon of gene expression may explain the variation in disease risks or symptoms. Recent studies on huge population with detailed health data indicative that mosaic loss of the Y chromosome (mLoy) in circumferential white blood cells have been associated to a range of disorders, including cancer, cardiovascular disease, and Alzheimer's disease (Williams Sarah, 2014).

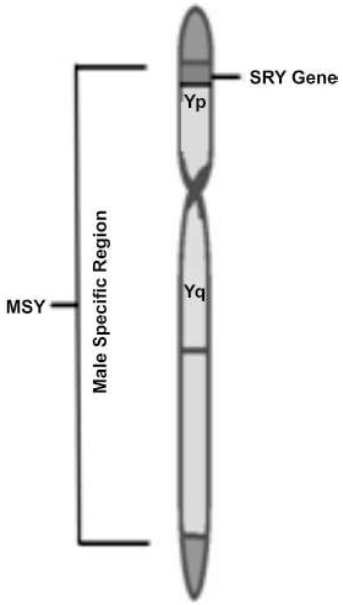

Fig. 1z: Y-chromosome

The inherited genetic content on the Y chromosome, which intended to be the compact and most common gene on the chromosome in mammals, as well as humans, has decreased as a result of evolutionary events (Hughes & Rozen, 2012). According to Cumminy's (year) research, the only approach that can be used to study the link between the fertility of male and Y chromosome is genealogy. Limited recombination during meiosis with homologous mate and expression of male features are the two main characteristics of the Y chromosome. Y chromosomes of rhesus macaque and Chimpanzee have been widely sequenced[5].

In culture that practices patrilineality, surnames can be frequently a rough companion to paternity, though anthropologists are careful to distinguish between biological and juristic fatherhood. Hawks and Wolpoff studied additional criteria for the evolutionary factors of Y chromosomes and it seems to be paradoxically shallow and inconsistent with the degree of inheritable and geographic variation which is seen in the somatic genes of humans (Hawks & Wolpoff, 2003).One more demanding aspect of establishment of Y chromosome haplogroups from the primitive DNA data is that it can discriminate the mutation form introduced by impairment in case of T to C and A to G[7].

The study found variations in some loci of the Y chromosome in mice and human showed the expected range that is based on the mutation rate and the size of the population (Michael W. Nachman). This majority portion of the X chromosome is

because the SNP's and non-combining region of Y chromosome in this segment determines **paternal ancestry.**

Comparing the outcomes determined from distinctive inheritance patterns of autosomes, X, Y chromosome and mt-DNA provides the chance to inspect sex-specific spread and amalgam procedures in the forthcoming years when larger sample sizes are taken from the same topographical area[9].

Forensic Aspect

Non-recombining regions of Y chromosomes are the solid wellspring source of help in forensic DNA

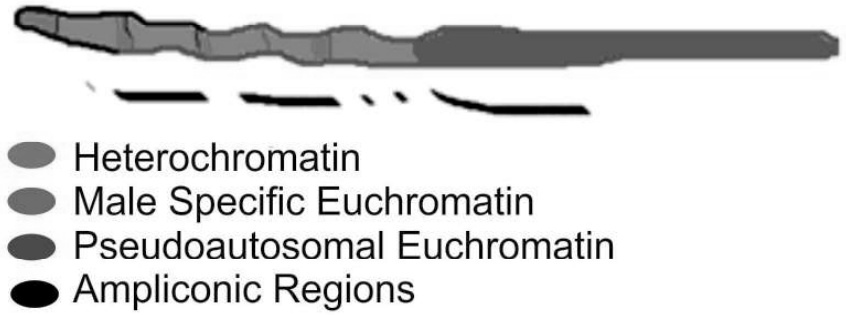

Fig. 2: Regions of Y-chromosome

analysis. These junk DNA segment (enhancer) contains most of the information that can turn on or activate its target genes[10]. Y-STR and Y-SNP are most broadly utilized markers in the forensic analysis of sexual assault due to their diversity[11]. Globally forensic laboratories are performingY-STR analysis for their regular case work and study of population genetics[12-15]. Applications of Y-STR haplotype in crime scene investigations are exclusion of male suspect in crime, presence of multiple male contributors, determination of paternal lineage of male suspects, providing leads in identification of unknown suspects etc.

NRY region can produce the alleles which can straightforwardly assist in filtering out the male DNA in a mixed female DNA[16]. Y-STRs are comprised of 2-6 nucleotide repeats. About 95% of sequence arrangements of this chromosome are supposed to be the male specific region (MSY) or non-recombining region (NRY). Y chromosomes also consist of the pseudoautosomal regions (PARs) which are situated at the far end of both p and q arms. The cytogenetic localization of Y-STR markers were reported at Yp11.2, which reveal the Y-chromosome, band 1, sub-band 2, small arm and region 1.

The fundamental characteristics of genetic markers are referenced and documented in the guidelines of Scientific Working Group on DNA Analysis Methods (SWGDAM). A little number of Y-STR loci was typed for forensic investigations until the mid-1990s. DYS19 was originally known as Y-27H39, and was discovered in 1992 by Lutz Roewer as the first polymorphic Y-STR marker[17]. Lida described five STRs loci DYS442,DYS445, DYS443, DYS441 and DYS444[18].

Additionally, 14 novel Y-STR loci DYS45 6,DYS459,DYS453,DYS458,DYS450,DYS447, DYS452,DYS448, DYS449, DYS454, DYS455, DYS446,DYS463, and DYS464 were recognized[19].

Y-PlexR was the pioneer kit for Y-STR typing developed by Reliagene Technology. Scientists founded the Forensic Y user group and demonstrated the functionality of thirteen Y-STR markers for forensic inference[20,21]. Initially 15 STR markers were constructed and discovered, out of which 7 core loci were used for forensic DNA examination purposes with two dyes and six markers (DYS393, DYS389II,DYS385a/b, DYS391, DYS19 and DYS390) in 2001[22,23].

DYS385a/b,DYS391, DYS390, DYS389I/II, DYS392, DYS19 and DYS393 were taken out by the Forensic Y User group or European Forensic Community in 1997 as the core group of minimum haplotypes. Expanded haplotype was produced by combining the YCAIIa/b marker. In 2000, Ayub

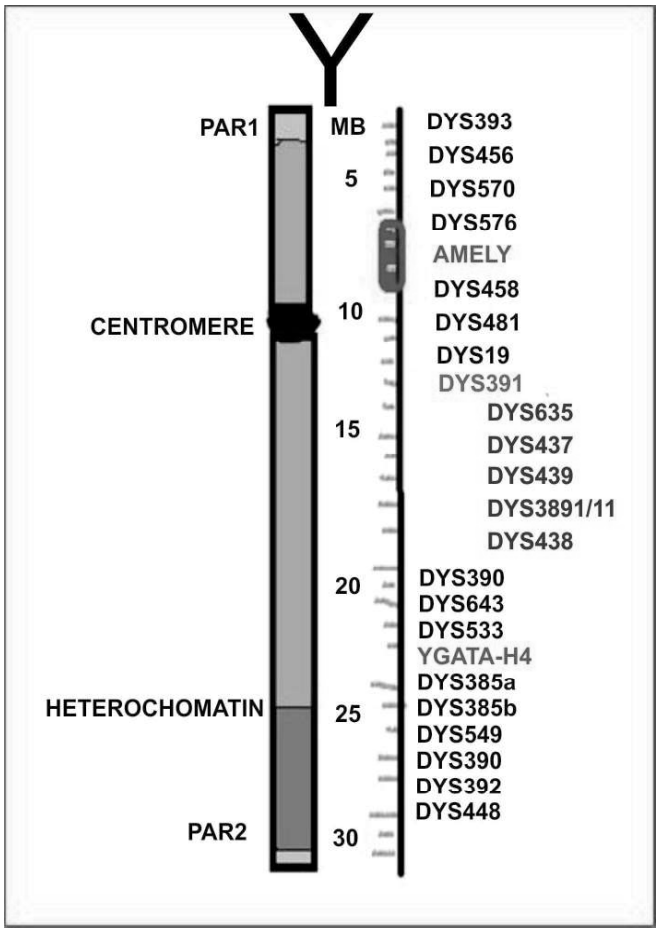

Fig. 3: Defined locus of Y-chromosome

identified 18 unique STRs, but only three of them were acknowledged by forensic experts: DYS438, DYS439 and DYS437and the YCAIIa / b extended haplotype markers were replaced with the DYS439 and DYS438 markers[24].

The next year, the same business produced Y-Plex5®, which uses three dyes to create five distinct markers (DYS389I, DYS389II, DYS392, DYS438 and DYS439). The same company developed the Y-Plex12® kit in 2003, which includes SWGDAM proposed Y-STR markers and amelogenin. In the year (2003), Promega Corporation launched a new Y-Plex12® kit having all Y-Plex12® markers excluding amelogenin and DYS437 as well as DYS439. Applied Biosystems launched a Y filer® kit with 17 Y-STR markers in same year[25]. Promega produced the PowerPlex® Y23System kit in 2013, which contains 23 markers, while Applied Biosystems published the Yfiler® plus kit in the same year, which accommodate 27 Y-markers. After that, China introduced a set of in-house kits having gradually more Y-STR markers, varying from 20 to 50. These kits are also essential in population genetics and forensic inference[15].

Other kits include the SureID®PathFinder Plus kit, GFS Y24 STR, YfilerTM Platinum system, AGCU Y24,Y Direct ID System, DNATyperTMY24 amplification kit, Goldeneye® 26Y system, Microreader TM29, Sure ID® Path Finder, Goldeneye® DNA ID 27Y kit,AGCU Y24 Plus PCR, Goldeneye®Y-PLUS kit, GoldeneyeTM 20Y amplification kit, and DNA Typer™ Y29 PCR amplification kits which were utilized by the scientific community for the study of Y-STR in Chinese populations. For all forms of forensic investigations, 475 Y-STR loci have been recognized so far. The PowerPlex® Y23 system kit was used to identify 1950 haplotypes in India, while the total number of haplotypes found in China is 28502. Millions of STR profiles of Y-STR markers can be found in massive YHRD databases[23].

Till date 475 Y-STR loci have been acknowledged for diverse forensic study. In India, 1950 haplotypes were discovered utilizing the PowerPlex® Y23 system kit, however in China, 28502 haplotypes were revealed. Large YHRD databases now contain millions of STR profiles based on Y-STR markers[26]. These kits are broadly utilized throughout the

world for tackling sexual assault and paternity cases.

According to the current state of database in YHRD, the following kits for Y-STR have been updated (https://yhrd.org/pages/resources/national_databases):-

At present, 25 Y STR loci are incorporated in the Y-FilerTMPlus system for forensic DNA typing. These are:

Name	Description	Loci
Minimal	YHRD core loci	DYS393, DYS19, DYS390, DYS389I, DYS392, DYS391, DYS398II, DYS385
Power Plex Y	Promega PowerPlex® Y	DYS437, DYS389I, DYS390, DYS438, DYS393, DYS385, DYS391, DYS19, DYS392, DYS389II,
Y-Filer	Applied Biosystems Amp FLSTR® Y-Filer®	DYS19, DYS389I, DYS438, DYS390, DYS391, DYS456, DYS389II, DYS439, DYS393, DYS458 DYS385, DYS448, DYS635, DYS392, YGATAH4, DYS437
Powerplex Y-23	Promega PowerPlex®Y-23	DYS389I, DYS393, DYS439, DYS448, DYS533, DYS19, DYS449, DYS481, DYS437, DYS391, DYS570, DYS458, DYS635, DYS576, DYS389II, DYS390, DYS643, DYS392, DYS385, DYS438, DYS456, YGATAH4
Y-Filer Plus	Applied Biosystems Amp FLSTR® Y-Filer ® Plus	DYS391, DYS627, DYS635, DYS389II, DYS385, DYS460, DYS456, DYS458, YGATAH4, DYS390, DYS576, DYS448, DYS393, DYS533, DYS438, DYS392, DYS481, DYS389I, DYS518, DYS19, DYS439, DYS570, DYF387S1, DYS437, DYS449
Maximal	YHRD max loci	DYS576, DYS549, DYS389I, DYS643, DYS635, DYS627, DYS392, DYS389II, DYS385, DYS391, DYS460, DYS393, DYS19, DYS458, YGATAH4, DYS570, DYS448, DYS456, DYS481, DYS518, DYS390, DYS449, DYS438, DYS437, DYS439, DYS533, DYF387S1

Name	Description	Loci
Minimal	YHRD core loci	DYS19, DYS393, DYS389I, DYS390, DYS391, DYS392, DYS385, DYS398II
Power Plex Y	Promega PowerPlex® Y	DYS389I, DYS437 DYS393, DYS438, DYS391, DYS19, DYS392, , DYS390, DYS385, DYS389II
Y-Filer	Applied Biosystems Amp FLSTR® Y-Filer®	DYS393, DYS456, DYS391, DYS389I, DYS390, DYS389II, DYS439, DYS458 DYS19, DYS392, DYS385, DYS635, YGATAH4,DYS448, DYS437, DYS438
Powerplex Y-23	Promega PowerPlex®Y-23	DYS393, DYS389I, DYS448, DYS439, DYS19, DYS481, DYS449, DYS533, DYS437, DYS570, DYS458, DYS635, DYS391 DYS576, DYS389II, DYS390, DYS392, DYS643, DYS385, DYS456, YGATAH4, DYS438
Y-Filer Plus	Applied Biosystems Amp FLSTR® Y-Filer ® Plus	DYS385, DYS635, DYS391, DYS389II, DYS576, DYS627, DYS19, DYS460, DYS458, DYS481, YGATAH4, DYS456, DYS448, DYS570, DYS390, DYS393, DYS438, DYS392, DYS389I, DYS518, DYS533, DYS439, DYS437, DYS449, DYF387S1,
Maximal	YHRD max loci	DYS549, DYS576, DYS643, DYS389I, DYS627, DYS635, DYS389II, DYS392, DYS391, DYS385, DYS393, DYS460, DYS458, DYS19, YGATAH4, DYS448, DYS570, DYS481 ,DYS456, DYS390, DYS438, DYS518, DYS437, DYS449 ,DYS439, DYF387S1, DYS533

DYS389I,YS576,DYS389II,DYS635,DYS46, DYS627,DYS19,DYS458,DYS448,YGATAH4,DYS 456,DYS391,DYS438,DYS390,DYS518,DYS392,D YS437,DYS570,DYS449,DYS385,DYS439,DYS393 ,DYF387S1,DYS533andDYS481[27]. Y-chromosome haplotype is supposed to be a single locus due to absence of recombination[10]. The genes in the NRY sequence are expected to become dedicated for male function or deteriorate through time as a result of the collection of deleterious mutations through mechanisms like Hitchhiking with beneficial mutations[28]. The possibilities of sporadic translocation within the extremely large chromosome segments have been reported. Y-STRs are useful to develop the population data[29]. Analysis of Y-STR indicated a lack of significant geographic structure and shape among African American and Asian American population, minor heterogeneity among European American and Hispanic American population, and extensive scale

subdivision among Native American population[20]. Y chromosome heterogeneity is displayed by some markers between and within the population and supposed to be beneficial to locate human evolutionary process during an ancient time i.e. recently delineating and these population are still linked[30].

Gillian cooper et. al found very remarkable haplotype diversity and authenticated that most of the mutation comprise the loss or gain of a single repeat unit. Further recommended that any given microsatellite haplotype might have been unfolding independently on a pair or more Y chromosome lineages.

Restricted duplication of a new locus, demonstrate the significance of whole distinguishing such duplications linked not simply treating each product as an separate locus[31].

Every submitted population sample is considered to be a collection of various populations sharing a common geographic, genetic, background linguistic or demography which keeps YHRD structurally intact (metapopulation). Due to a significant increase in sample numbers, this trait facilitates statistical evaluation of haplotype matching[32,27].

The YHRD now recognises seven primary meta populations: African, Eurasian, Afroeurasian, Amerindian, East Asian, Eskimo Aleut, Australian Aboriginal, and one intermingle metapopulation (large contribution of various historical populations). They grouped these seven meta populations into twenty subgroups. On the basis of genetic analysis the subgroups Europeans is further divided into Eastern, Western and South-eastern Europeans. For the calculation of genetic distances(Fst-values) a customised AMOVA algorithim (http:// rprojekt.org/amova) was used and to observe if the classification of selected meta population is according to the available haplotype database (Willuweit & Roewer, 2007).

SiiriRootsi et al. reported the different domains of gene flow in the ancient segment of Europe which defines the overarching super-haplogroups of the European ancestors that shares the non-African gene pool. But, Volga region of South France shows a thinly STR variation, suggesting a mutational event in this region[33]

Michael Hammer et al. cited one of the kind rationalizations for the presence of extraordinary excessive range of founding's lineage within the cohanim. Within the cohanim, one is responsible for setting up lineages. One hypothesis is that numerous men were chosen as cohanim early in the priesthood's existence. (M. Hammer& Redd, 2006). Anand et al.studied 310 unrelated individuals of population of Rajasthan, India. Out of 310 haplotype groups, 30 are unique and haplotypes, which concludes a vast discrimination capacity for the studied loci i.e. 0.997[15].

The absence of haplotypic structure linked to the putative ancient haplotype in almost all groups supports a recently shared ancestry and/ or significant male migrations across human evolution. They are located in the centre, and they comprise of people from most of the racial groups. It implies that male migration is a widespread phenomenon for human evolution. Rapid mutation rate at various loci and huge migration of male population causes an insufficient haplotypic composition in the globe[34].

In the human population, the forensic application of Y-STR results from their excessive polymorphism ranges. They have huge capacity to multiplex many Y-STR in a single PCR reaction and their tiny size in base pairs (100-400bp). The shortage of individual identity of those markers on non-recombining areas is one of Y STR's limitations in forensic and paternity analysis. Due to the difficulty in differentiating paternally related males in a population, Y-STR haplotypes may have lower inclusion probabilities than autosomal STR haplotypes[35].

As a result, population surveys are essential, and quality-controlled population records have been produced by creating the (YHRD) Y-chromosome haplotype reference database, as a global database. YHRD can be used to search for all single alleles and all genotype combinations in a constantly increasing and frequently updated data collection because new data can substitute the previous ones[27].

Due to non- recombination feature of Y-chromosomes, few mutations are being propagated into the next generations[23].Chandler calculated the rates of mutation in the Y-STR gene[36].Most Y-STR mutation rates are similar to autosomal STR mutation rates, ranging from 0.1%-0.4% generational events. Only the loci DYS458 and DYS439 can able to mutate faster than 0.5%. Out of these 27 Y chromosome-STR markers in the Yfiler Plus® PCR amplification kit, seven markers are rapidly mutating viz. DYS449, DYF387S1a/b, DYS576, DYS518, DYS627, and DYS570[14].

Here, are the lists of combined mutation rates per locus markers for Y-Filer Plus Kit.

Locus	Combined Mutation Rates
DYS576	1.2 e – 02(72 in 5941)
DYS389I	2.44 e -03 (44 in 18051)
DYS635	4.24-03 (50-11788)
DYS389II	4.64 e -03(84 in 1804)
DYS627	1.47 e -02 (69 in 4678)
DYS460	4.41 e -03 (19 in 4311)
DYS458	6.60-03 (72-10914)
DYS19	2.12 e -03 (42 in 19807)
YGATAH4	2.51 e -03 (30 in 11970)
DYS448	1.37 e -03 (15 in 10935)
DYS391	2.40 e-03 (46 in 19199)
DYS456	4.30-03 (47 in 10932)
DYS390	2.03 e -03 (38-18685)
DYS438	3.48e-04(5 in 14385)
DYS392	5.23 e -04 (10 in 19127)
DYS518	11.9 e -02 (53 in 4465)
DYS570	9.22 e -03 (52 in 5642)
DYS437	1.32 e -03 (19 in 14360)
DYS385	2.86 e -03 (95 in 33252)
DYS449	1.02 e -02 (46 in 4532)
DYS393	1.22 e -03 (22 in 17977)
DYS439	5.15 e -03 (74 in 14363)
DYS481	4.58 e -03 (25 in 5456)
DYF387S1	7.91 e -03 (51 in 64479)
DYS533	2.94 e -03 (16 in 5449)

Online reference database of Y-STR haplotype

Supplier	Type of the database	Y-STR markers	No.of haplotype	Url
National Institute for forensic Sciences, Orlando, USA	US Y-STR database	17	17,216	Usystrdatabase.org
Institute of legal medicine and forensic Sciences, Berlin, Germany	YHRD	17	72.055	Yhrd.org
Applied Biosystems, inc, Foster city, USA	Y-Filer haplotype database	17	3,561	Appliedbiosystems.com/Yfilerdatabase
Promega Corporation, Madison, USA	Power Plex Y-haplotype database	12	4,004	Promega.com/techserv/tools/pplexy

Some, interbreeding between the colonizing modern humans and the local population would account for the apparent morphological continuity in some regions, particularly Australasia (Archaeology & Petraglia).

Conclusion

Both exon and intron regions of Y-chromosome can be helpful for tracking various factors in medical and forensic field. Gene identification at Y-chromosome is aprominent area of research in medical genetics that can lead to the successful diagnosis of many male related inheritable diseases. In forensic investigations such as identification of mass disaster's victim, paternity testing, human remains burning, sexual assault cases, and population genetics can make Y-chromosome a powerful tool for dealing these areas. Highly mutating regions of Y chromosome can provide a useful hand towards the genealogical studies. Y-chromosome is useful for the study of ancient migration of global population. It can also determine the similarity and comparison of genetic distances. Y-STR markers have been used in many evolutionary categories that solved many historical and geographical puzzles of human population.

Advance studies on Y chromosomes from well-defined lineages could test the feasibility of distinguishing between patrilineality related people of varying degrees utilising highly changing Y-STRs, which would be extremely useful in population and forensic genetics.

Acknowledgement

Conflict of interest

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Authors have no conflict of interest.

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