Kuaybe YÜCEBİLGİLİ

THE ALLELE AND HAPLOTYPE ANALYSIS OF 17 Y STR LOCI ON HUMAN POPULATION OF TURKEY

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M.S. Thesis In Biology

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APPROVAL PAGE

I certify that this thesis satisfies all the requirements as a thesis for the degree of Master of Science.

Assist. Prof. Dr. Sevim IŞIK
Head of Department

This is to certify that I have read this thesis and that in my opinion it is fully adequate, in scope and quality, as a thesis for the degree of Master of Science.

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It is approved that this thesis has been written in compliance with the formatting rules laid down by the Graduate Institute of Sciences and Engineering.

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ABSTRACT

The distribution of human genetic diversity has long been a subject of interest. Turkey has a bridge-like location between Europe and Asia. The genetic structure of populations in Turkey is quite complex due to the longstanding human presence. In this study, 116 unrelated male individuals were analyzed for variation at 17 Y-chromosome specific STR loci (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a/b, DYS438, DYS439, DYS456, DYS458, DYS635, Y GATA H4, DYS437 and DYS448) in order to characterize genetic diversity in Turkey and its geographical regions.

All haplotypes were found to be unique. The most common haplotype was observed in 2 (1.72%) individuals when DYS385 locus was excluded. The overall haplotype diversity for 17 Y-STR loci was 1.000 indicating a high potential for differentiating between male individuals in this population. Results of the present Y-STR study reveals high level of genetic diversity in Turkey. A duplication in DYS19 locus is observed. A comparative analysis of the generated data to those previously published data from Romania, Macedonia, Germany, Japan, Poland and Croatia was carried out. The AMOVA test revealed the close relationship of the Black Sea Region of Turkey with Romania (Moldavian) population and significant difference from Polish and Japanese populations. This is the first study that investigates 17 Y-STR loci in population of Turkey.

Keywords: Y-chromosome STRs, Haplotype Analysis, Allele Frequency, Population Genetics, Genetic Diversity, Geographical Regions, Turkey

TÜRKİYE İNSAN POPULASYONUNDA 17 Y-STR LOKUSU İÇİN ALLEL VE HAPLOTİP ANALİZİ

Kuaybe YÜCEBİLGİLİ

Yüksek Lisans Tezi – Biyoloji
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ÖZ

İnsan genetik çeşitliliği her zaman merak konusu olmuştur. Avrupa ve Asya arasındaki köprü konumu, uzun süreli insan varlığı ve sosyal geleneklerin de etkisiyle Türkiye'de yaşayan populasyonun genetik yapısı oldukça komplextir. Bu çalışmada Türkiye geneli ve coğrafi bölgelerdeki genetik çeşitliliği göstermek amacıyla 116 sağlıklı erkek birey 17 Y-STR lokusu için analiz edilmiştir.

17 Y-STR için yapılan analizde herbir bireyin birbirinden farklı haplotip sergilediği görülmüştür. DYS385 lokusu dışlandığında aynı haplotipi sergileyen 2 bireye rastlanmıştır. 17 lokus için haplotip ayırma gücü 1.00 olarak hesaplanmıştır.

Elde edilen Y-STR profilleri diğer populasyonlarla kıyaslanmış MDS grafikleri çizilmiştir. Romanya populasyonuyla genetik bir yakınlık farkedilmiştir. Bu çalışma Türkiye geneli için yapılan ilk 17 Y-STR analiz çalışmasıdır.

Anahtar Kelimeler: Y Kromozomu Kısa Tekrar Dizileri, Haplotip Analizi, Populasyon Genetiği, Genetik Çeşitlilik, Coğrafi Bölgeler, Türkiye

To my mum & dad.

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LIST OF ABBREVIATIONS

ABBREVIATION

AMOVA	: Analysis of Molecular Variance
BP	: Base Pair
CODIS	: Combined DNA Index System
DNA	: Deoxyribonucleic Acid
EDTA	: Ethylen Diamin Tetra Acetic Acid
FBI	: Federal Bureau of Investigations
HV	: Hypervariable Region
IFSG	: International Society of Forensic Genetics
MDS	: Multidimensional Scaling
MSY	: Male-Specific Region
NRY	: Non-Recombining Region of Y Chromosome
PAR	: Pseudoautosomal Regions
PCR	: Polymerase Chain Reaction
RFLP	: Restriction Fragment Length Polymorphism
SINEs	: Short Interspersed Elements
SNP	: Single nucleotide Polymorphisms
SSR	: Simple Sequence Repeats
STR	: Short Tandem Repeat
SWGDAM	: Scientific Working Group on DNA Analysis Methods
UEPs	: Unique Event Polymorphisms
VNTR	: Variable Number Tandem Repeats
YCC	: Y-chromosome Consortium
YHRD	: Y-STR Haplotype Reference Database

CHAPTER I

INTRODUCTION

1.1 POPULATION VARIATION

A very great amount of human DNA (approximately 99.7%) is the same between people and the remaining small part of it differs and thus makes us unique individuals. These variable regions of DNA are useful for human identification. It is possible to locate and characterize the variation at specific sites in the genome. Types of DNA polymorphisms can be classified in two groups; sequence and length polymorphisms. STR markers discussed in this thesis are length polymorphisms (Butler 2005).

A. Sequence polymorphisms (single nucleotide change):

*****GCAT**T**CAAGT**G**ATACA*****

*****GCAT**C**CAAGT**A**ATACA*****

B. Length polymorphisms (number of core repeat unit change):

*****(GATA)(GATA)(GATA)***** 3 repeats

*****(GATA)(GATA)***** 2 repeats

A genotype is used in order to indicate a genetic type or allele state. For example, a genotype of '9, 15' means that the sample contains two alleles, one with 9 and the other with 15 repeat units. Results from multiple samples can be easily compared by this short designation (Butler 2005).

DNA typing is a method in which our genetic material (DNA) is converted into a "barcode" that, ultimately distinguishes each of us from nearly everyone else on earth. In DNA typing multiple markers or loci are examined since it is expensive and time

consuming to evaluate entire DNA sequence of an individual. In order to increase the chance that unrelated individuals have different genotypes, the number of DNA markers should be increased. Namely, DNA the likelihood of having two DNA samples from the same source increases with each marker match. The variability that is observed at these markers is used to compare samples regarding random match probability calculations (Butler 2005).

1.2 MOLECULAR GENETIC MARKERS

1.2.1 Repetitive DNA (STR markers)

Repeats of short DNA sequences are abundant in eukaryotic genomes (Ellegren 2004). These repeated DNA sequences are typically designated in three ways; by the length of the core repeat unit, the number of adjacent repeat units or the full length of the repeat region. As discussed earlier one of the two forms of variation at DNA level was length polymorphisms. Long repeat regions which may contain several hundred to several thousand bases in the core repeat are often referred to as *satellite* DNA because they are mostly found surrounding the chromosomal centromere as in the early experiments involving equilibrium density gradient centrifugation (Britten and Kohne 1968; Primrose 1998).

Variable number tandem repeats (VNTR), “minisatellite” and short tandem repeat (STR), “microsatellite” markers are length polymorphisms, and can vary between individuals based on the number of repeat units within a specific locus. This high degree of variability has led to the widespread use of these sequences as polymorphic markers (Kong, Gudbjartsson et al. 2002; Kopelman, Stone et al. 2009).

VNTRs consist of sets of tandemly repeated base pair sequences that can vary in length from approximately 10–100 bases in length (Tautz 1993; Chambers and MacAvoy 2000). An example of a VNTR is the forensic DNA marker D1S80 (Figure 1.1). The D1S80 marker is a minisatellite with a 16bp repeat unit and contains alleles in the range of 16-41 repeats (Kasai, Nakamura et al. 1990).

Short tandem repeats, also called simple sequence repeats (SSRs), are contiguous repeating units of DNA, where each unit is 2-6 base pairs (bp) in length and were first reported in the late 1980s (Litt and Luty 1989). A schematic of a sample STR is given in Figure 1-2.

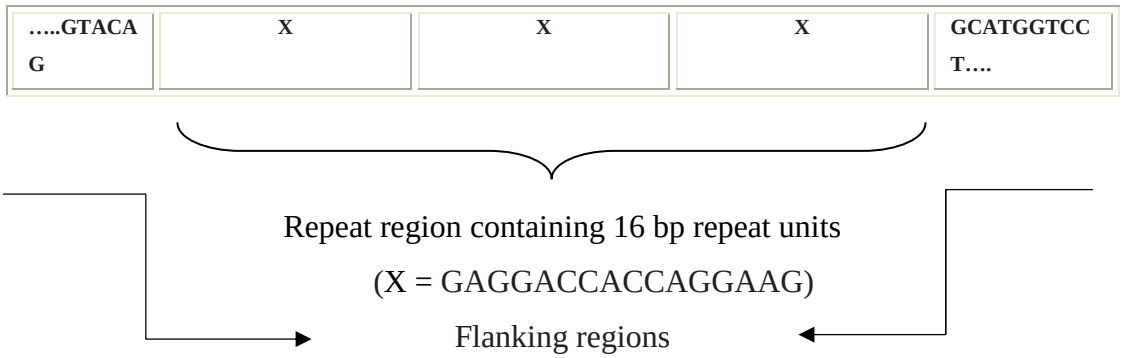


Figure 1.1 Minisatellite Marker (D1S80)

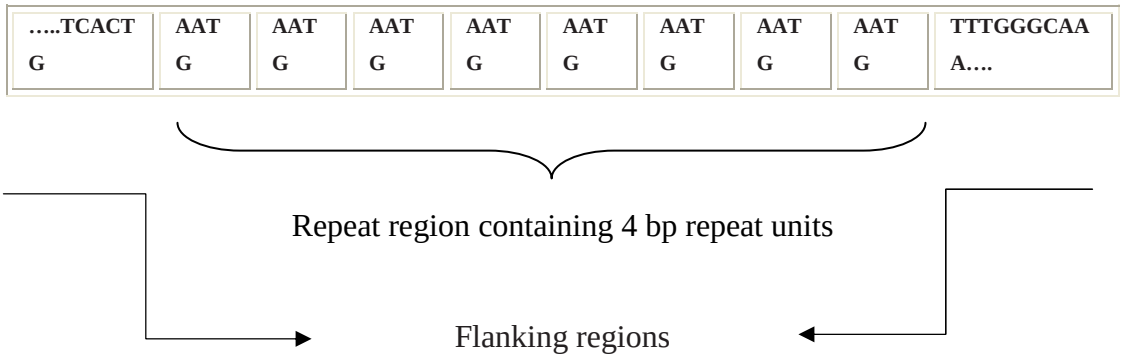


Figure 1.2 STR Marker (TPOX)

Thousands of these polymorphic microsatellites have been identified in human DNA (Ellegren 2004). Microsatellites account for approximately 3% of the total human genome. These markers are scattered throughout the genome and seen approximately in every 10,000 nucleotides (Edwards, Civitello et al. 1991; Collins, Morgan et al. 2003; Subramanian, Mishra et al. 2003).

STRs found within chromosomes 1-22 are termed autosomal DNA markers. These markers are subject to recombination through cell division since half of the genetic material comes from the mother and half comes from the father. An individual

is homozygous if the alleles at a specific location are identical and heterozygous if the alleles are different (Kirby 1990).

1.2.1.1. Isolation and Types of STR Markers

First, flanking regions surrounding the repeat units must be determined in order to study on STR markers. Because when these sequences are known, PCR primers can be designed and the repeat region can properly be amplified. New STR markers are usually characterized in two ways; searching DNA sequence databases such as GenBank for contiguous repeat regions containing more than six repeat units (Weber and May 1989; Collins, Morgan et al. 2003; Subramanian, Mishra et al. 2003) or performing molecular biology isolation methods (Edwards, Civitello et al. 1991; Chambers and MacAvoy 2000).

STR repeat sequences are grouped according to the length of the repeat unit. For example, dinucleotide repeats have two nucleotides in a repeat unit, trinucleotide repeats have three nucleotides and tetranucleotide repeats have four nucleotides, so and so forth. Tetranucleotide repeats have become the most commonly used STR markers for human identification because of their low stutter rate. Stutters are DNA amplification artifacts that result from slipped-strand mispairing in the PCR process affecting the level of polymorphism found at a particular locus. Therefore, it becomes more challenging to interpret DNA profiles, especially in mixtures (Butler 2005).

Another way to categorize STR sequences is based on the repeat pattern. Simple repeats have all repeating units in same length and sequence, compound repeats possess two or more adjacent simple repeats with different repeat motifs, and complex repeats are composed of multiple repeat units of various lengths along with several intervening sequences, such as insertions (Urquhart, Kimpton et al. 1994). The last STR category termed *complex hypervariable repeats* are those with a number of non-consensus alleles that differ in both sequence and size (Butler 2005). Some alleles for a STR locus may contain incomplete repeat units that fall in between alleles with full repeat units. STRs that do not have a whole number of complete repeat units are called microvariants (Puers, Hammond et al. 1993).

1.2.1.2. Advantages of STR Markers

STR markers have become popular DNA repeat markers in a variety of situations such as population studies, forensic identification, gene mapping and disease diagnosis (Hammond, Jin et al. 1994; Fan and Chu 2007).

STRs have large groups of alleles due to their repeat lengths (Lee, Ladd et al. 1994) making them highly variable between populations. They are highly abundant in the human genome with thousands of loci known and classified to date and the possibility of millions throughout the genome (Edwards, Civitello et al. 1991; Collins, Stephens et al. 2003; Ellegren 2004). The most important characteristic of STRs is that they are stable and consistent throughout the body, meaning that the same profile is found in every cell of an individual. Therefore, any kind of biological material can be easily studied and the resulting DNA profile will be the same.

By the way, they are easily amplified by the polymerase chain reaction (PCR) and work with low-quantity DNA templates or degraded DNA samples. STR typing is suitable for automation which involves sensitive fluorescent detection. Thus data collection from these markers becomes faster. When sites on multiple chromosomes are examined, STRs are highly discriminating between unrelated and even closely related individuals. This aspect of STRs makes them excellent markers for human identification. Discrete alleles make results easier to interpret and to compare through the use of computerized DNA databases than RFLP-based systems where similar DNA sizes were grouped together.

Furthermore, short tandem repeat analyses are more popular because of its advantages on VNTR analysis. For example STR allele size is between approximately 100-400 base pairs when compared to VNTR alleles, which has the allele size in 400-1000 base pair range. The small size of STR markers makes them more suitable for studies with degraded samples. VNTR typing is not preferred in lineage studies because of its larger size. This also limits its application in forensic studies. Because of those reasons STRs are preferred over VNTRs (Butler 2005).

1.2.1.3. 13 Core CODIS (Combined DNA Index System) STR Loci

STRs are mostly found in non-coding regions of the genome and dispersed throughout the all chromosomes including X and Y (Edwards, Civitello et al. 1991). Federal Bureau of Investigations (F.B.I.) has selected 13 STR loci as the core set in order to create a common language between worldwide laboratories. DNA profile of a person is derived with the set of alleles for all 13 STR loci.

Table 1.1 Information on the 13 core CODIS STR Markers
(Adapted from Butler 2005 Forensic DNA typing)

Locus	Repeat Motif	GenBank Accession #	Ref Allele
CSF1PO	TAGA	X14720	12
FGA	CTTT	M64982	21
TH01	TCAT	D00269	9
TPOX	GAAT	M68651	11
VWA	[TCTG][TCTA]	M25858	18
D3S1358	[TCTG][TCTA]	NT_005997	18
D5S818	AGAT	G08446	11
D7S820	GATA	G08616	12
D8S1179	[TCTA][TCTG]	G08710	12
D13S317	TATC	G09017	13
D16S539	GATA	G07925	11
D18S51	AGAA	L18333	13
D21S11	Complex [TCTA][TCTG]	AP000433	29

1.2.1.4. STRBASE: A Dynamic Source of Information on STR Markers

As the use of STRs for genetic mapping, forensic identification and population studies new alleles have become widespread new alleles are constantly being discovered, additional STR markers are being developed, and population data increases with each month of published journals. Thus the need of a common dynamic

information source unavoidably arose. Eventually “STRBase” was created in early 1997 by John M. Butler (under the direction of Dennis J. Reeder) and has been available on-line since July 1997 (Butler, Ruitberg et al. 1997).

Presently, over 3400 references may be reached by using the following URL: http://www.cstl.nist.gov/biotech/strbase. The main objective of this web site is to bring together the abundant literature on the subject in a cohesive manner to make future work in this field easier. The database includes the facts and sequence information on STR systems, population data for STR markers, commonly used multiplex STR systems, PCR primers and conditions, various Technologies for STR allele analysis. Addresses for scientists and organizations working in this area have also been included as well as a comprehensive reference listing of material on STRs used for DNA typing purposes. (Butler 2011)

1.2.2 SNP & Other Biallelic Markers

Single nucleotide polymorphisms (SNPs) are another group of genetic markers that have received a lot of attention in recent years due to their abundance throughout the human genome. SNP is a single base polymorphism that occurs between individuals at a single particular position. Single base polymorphisms are abundant in the human genome and can be used for linkage analysis to track genetic diseases (Brookes 1999). It is possible to work with highly degraded biological samples in SNP testing. One of the main advantages of these genetic markers is that they have relatively lower mutation rates than STRs and therefore they are more likely to become ‘fixed’ in a population. This makes SNPs stable genetic markers for lineage-based analyses. Mutation rate for SNPs are one in every 10^8 generations while STR mutation rates are approximately one in a thousand. The presence of rare STR or SNP alleles in particular population groups can be used to estimate the ethnic origin of a sample (Brookes 1999).

SNPs have also some limitations. Most SNPs are biallelic and thus they are less informative for identity testing than STR markers. To obtain the same power of discrimination that exist in STR multiplex systems, a multiplex of 50–100 SNP marker would be needed (Chakraborty, Stivers et al. 1999). Lastly, interpretation of mixtures

involving more than one donor is problematic because of the limited number of alleles per locus.

Today all forensic databases are well-established and based on STR analysis, therefore it is unlikely that SNPs will become the primary forensic markers by replacing STR (Gill, Werrett et al. 2004). However, there is a valuable source of genetic information that can be used in population studies, since approximately 85% of human variation is derived from SNPs (Cooper, Smith et al. 1985; Wang, Fan et al. 1998; Holden 2002).

There is a great number of SNP typing methods are available, each with their own advantages and disadvantages. Some of the primary SNP typing methods in this area are pyrosequencing (Andreasson, Asp et al. 2002), TaqMan (Lareu, Puente et al. 2001), Luminex (Budowle, Planz et al. 2004), and minisequencing or SNaPshot™ (Tully, Sullivan et al. 1996; Sanchez, Borsting et al. 2003). SNP analysis are appropriate for multiplexing, multiple markers can be examined at a time.

There are also other bi-allelic markers used in population variation studies. *Alu* insertion polymorphisms are best studied examples of short, interspersed nuclear elements (SINEs) and found in nearly one million copies per haploid genome (5–10% of the human genome) (Primrose 1998). Alu insertions on the genome are accepted as unique events. *Alu* elements are stable genetic markers and are not subject to loss or rearrangement. Commonly used *Alu* insertion polymorphisms include APO, PV92, TPA25, FXIIIB, D1, ACE, A25, and B65 (Sajantila 1998). *Alu* repeats give information about the geographic/ ethnic origin of the sample being tested (Batzer and Deininger 1991; Bamshad, Wooding et al. 2003). Nevertheless, *Alu* insertions present less variation than multiplex STR profiles. Therefore, they are mostly studied to provide more information of a sample. Insertion-deletions or indels are another form of bi-allelic (or di-allelic) polymorphisms. For instance, STR markers can be thought of as multiallelic indels because the different alleles are typically insertions or deletions of a tandem repeat unit. These markers can be easily typed and may be useful for future genetic studies such as human identification (Butler, 2005).

STRs are more polymorphic but are not as common as SNPs. STR and SNP markers comparison is adapted from Butler 2005 Forensic DNA Typing (Table 1.2).

Table 1.2 Comparison of STR & SNP markers

Characteristics	**Short Tandem Repeats (STRs)**	**Single Nucleotide Polymorphisms (SNPs)**
Occurrence in human genome	~1 in every 15 kb	~1 in every 1 kb
General informativeness	High	Low; only 20–30% as informative as STRs
Marker type	Di-, tri-, tetra-, pentanucleotide repeat markers with many alleles	Mostly bi-allelic markers
Number of alleles per marker	Typically >5	Typically 2
Detection methods	Gel/capillary electrophoresis	Sequence analysis; microchip hybridization
Multiplex capability	> 10 markers with multiple fluorescent dyes	Potential of 1000s on microchip
Major advantage for forensic application	Many alleles enabling higher success rates for detecting and deciphering mixtures	PCR products can be made small potentially enabling higher success rates with degraded DNA samples

1.2.3 Lineage Markers

As described earlier DNA markers found within chromosomes 1-22 are termed autosomal DNA markers and subject to genetic shuffling with each generation because during the process of meiosis half of an individual's genetic information comes from father and half from mother. However, markers on the chromosome X, chromosome Y and mitochondrial DNA represent 'lineage markers'. They are passed down from

generation-to-generation without genetic recombination (except for mutational events) (Butler 2005).

Mitochondrial DNA is being used to trace maternal lineages while Y chromosome markers such as Y-STR and Y-SNPs are being used to trace paternal lineages (Figure 1.3). The genetic information derived from these lineage markers is referred to as a haplotype rather than a genotype since there is only a single allele per individual. Y chromosome markers are linked on the same chromosome, the same as in the mtDNA markers, and are not shuffled with each generation, therefore the statistical calculations for a random match probability does not involve the product rule. As a result, haplotypes obtained from lineage markers alone can never be as effective in differentiating between two individuals as genotypes from autosomal markers that are unlinked and segregate separately through generations. Nevertheless, these markers do have an important role to play in forensic investigations and population studies.

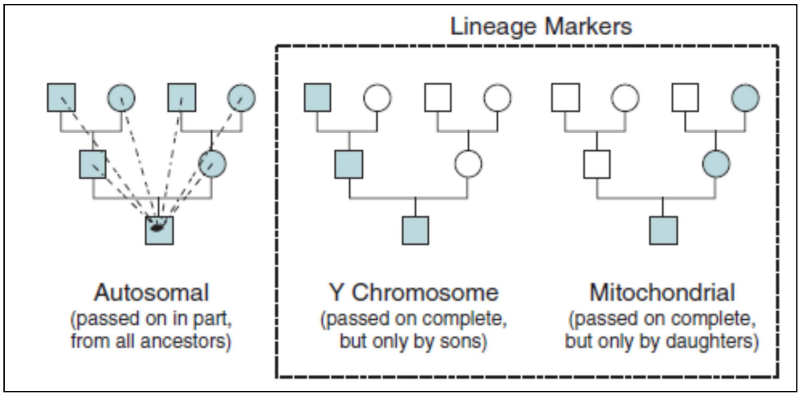


Figure 1.3 Inheritance patterns of autosomal, Y chromosomal and mitochondrial genetic markers

1.2.3.1. Mitochondrial DNA

Analysis of mitochondrial DNA (mtDNA) variation has become a useful tool for human population studies. Mitochondrial DNA (mtDNA) is a circular, double stranded DNA locate inside the mitochondria (Figure 1.4). The number of mitochondrial DNA (mtDNA) molecules varies between cells. However, it is estimated that the average copy number of mtDNA in most cells is about 500 (Satoh and Kuroiwa 1991). Mitochondrial DNA has approximately 16 569 base pairs and possesses 37 ‘genes’ that

code for products used in the oxidative phosphorylation process or cellular energy production.

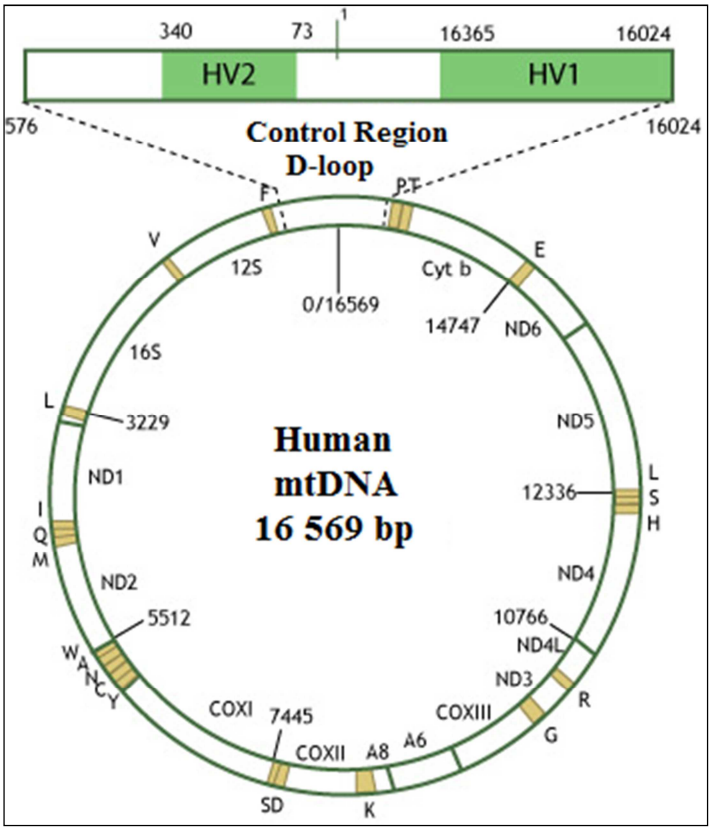


Figure 1.4 Mitochondrial DNA Genome

Because of its characteristics, such as presence of high mutation rate, absence of recombination and its maternal inheritance pattern, mtDNA, especially its first hypervariable region, has been frequently used in genetic genealogical studies.

MtDNA polymorphisms between individuals in the human population are extensively found within the control region (D-loop). Hypervariable region I (HV1) and II (HV2) are amplified with PCR and sequence analysis are done. The DNA sequence between 16024th and 16365th nucleotides in HV1 and 73th and 340 th in HV2 is compared to the reference sequences; Anderson or the revised Cambridge Reference Sequence (Anderson, Bankier et al. 1981). A third hypervariable region (HVIII) is also examined between the 438th –574th nucleotides (Lutz, Wittig et al. 2000; Bini, Ceccardi et al. 2003). Polymorphisms are noted and reported with the altered nucleotides (Butler 2005)

MtDNA is being used for several years in order to try to understand human migration patterns (Relethford 2001; Relethford 2004). Forensic DNA testing, disease diagnostics and anthropological and genealogical research efforts will all continue to be benefited by growth and developments in mitochondrial DNA analysis (Butler 2005).

1.2.3.2. X- STR

Diploid human cells have 22 pairs of homologous chromosomes (autosomes) and 1 pair of sex chromosomes (X and Y chromosomes) which determines if a person is female or male. The length of X chromosome is almost 165 million base pairs making up 5% of the total DNA in females and 2.5% in males since females have two X chromosomes, while males have one X and one Y chromosome.

Typing of STR markers on X chromosome (X-STR typing) as a PCR-based method with lineage entity is today a subject of growing interest for forensic genetics (Toni, Presciuttini et al. 2006). X-STRs have been useful in deficiency paternity testing in which the mother is available for typing. One of the chromosomes on each pair comes from mother and the other comes from father. Both males and females retain one of their mother's X chromosomes, and females retain their second X chromosome from their father. So, female individuals fathered by the same man share their paternal X chromosome. And the other one X chromosome is the same with the mother. Hence in case of deficiency paternity in which the mother is available for typing, the possible X alleles of the putative father can be determined and the paternal profile can be reconstructed. Moreover, there are cases that demonstrate the impact of additional X-STR markers even in identifying case works (special reverse paternity cases) that cannot be solved using autosomal markers (Barbaro and Cormaci 2006). Besides, the use of an X-STR multiplex in conjunction with autosomal STR loci can increase the power of discrimination, than using autosomal STR loci alone (Chen and Pu 2004).

Many researches has been done to utilize specific loci for X-STR. Loci such as GATA172D05, DXS101, HPRTB, STRX1, DXS2390, DXS10011, DXS6789, DXS8377 and DXS9895, DXS6807, DXS8378, DXS9902, DXS7132, ARA, DXS6800, DXS9898, DXS7133, DXS7424, DXS7423, DXS8377 were proven to be at use in X chromosome STR method. Researches on population data on the X chromosome STR

on loci mentioned were also widely reported from all over the globe, confirming the use and benefit that can be taken from them (Edelmann, Deichsel et al. 2002; Chen and Pu 2004).

1.2.3.3. Y Chromosome Polymorphisms

1.2.3.3.1. Structure of the Y Chromosome

The Y-chromosome is one of the two sex determining chromosomes in humans, as in the most of eukaryotic genomes. It is probably the most unusual part of the human genome with a huge non-recombining DNA region and transferred directly from father to son (Bull 1983). Y-DNA analysis may thus be used in genealogy research.

1.2.3.3.2. Sequence of the human Y chromosome

The first complete sequence of a human Y chromosome, from a single male, has been published in 2003 (Skaletsky, Kuroda-Kawaguchi et al. 2003). Although it is stated as 'complete sequence', only a 23 megabase (Mb) euchromatic portion of Y chromosome was reported. However a typical Y chromosome is composed of roughly 60 Mb (Smirnov, Pokrovskaya et al. 2000). The remaining unsequenced ~37 Mb is a heterochromatin region located on the long arm of the Y chromosome (Figure 1.5) that is not transcribed and is composed of highly repetitive sequences, which are impossible to sequence reliably with current technology.

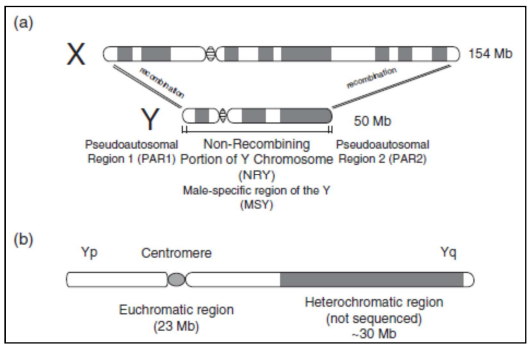


Figure 1.5 (a) Schematic representation of X and Y chromosomes (b) Y chromosome showing both euchromatic and heterochromatic regions

The human Y-chromosome is unable to recombine with the X-chromosome (Figure 1.5), except for small pieces of pseudoautosomal regions (PAR1 and PAR2) at the two tips (Graves, Wakefield et al. 1998). The bulk of the Y-chromosome which does not recombine is called non-recombining region of the Y-chromosome "NRY" or male-specific region "MSY" (Skaletsky, Kuroda-Kawaguchi et al. 2003). Figure 1.6 gives detailed information about this region.

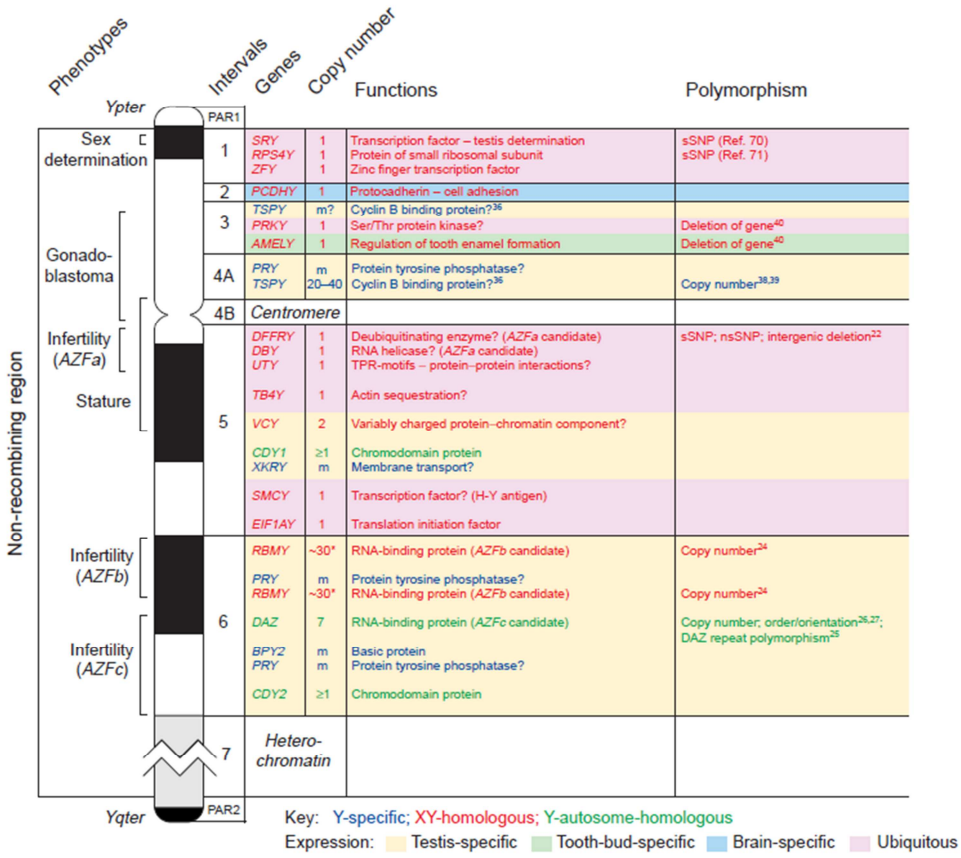


Figure 1.6 Genes and Phenotypes on the non-recombining region of Y chromosome (Jobling and Tyler-Smith 2000)

1.2.3.3.3. Applications of Y Chromosome Analysis

The field of Y-chromosome research has grown rapidly in recent years and Y-chromosome markers have become a widely accepted tool for forensic investigations and population studies. These markers have specific properties useful for many different areas such as human identification applications including forensic casework of sexual assault evidence (Betz, Bassler et al. 2001; Corach, Filgueira Risso et al. 2001), paternity tests (Santos, Epplen et al. 1993; Rolf, Keil et al. 2001), missing persons investigations (Dettlaff-Kakol and Pawlowski 2002), human migration patterns

addressing historical (Foster, Jobling et al. 1998), particularly in the SNP arena (Mitchell and Hammer 1996; Zerjal, Wells et al. 2002; Kayser, Brauer et al. 2003) and genealogical studies (Sykes and Irven 2000; Jobling 2001). The advantages of each application area are given in Table 1.3.

Table 1.3 Application areas of Y Chromosome testing

Use	Advantage
Forensic casework on sexual assault evidence	Male-specific amplification (can avoid differential extraction to separate sperm and epithelial cells)
Paternity testing	Male children can be tied to fathers in motherless paternity cases
Missing persons investigations	Patrilineal male relatives may be used for reference samples
Human migration and evolutionary studies	Lack of recombination enables comparison of male individuals separated by large periods of time
Historical and genealogical research	Surnames usually retained by males; can make links where paper trail is limited

1.2.3.3.4. Categories of Y Chromosome Genetic Markers

Y chromosome specific markers fall into two broad categories: bi-allelic and multi-allelic loci. Bi-allelic markers represent unique events including single nucleotide polymorphisms (Y-SNPs) and an *Alu* insertion, which are sometimes referred as unique event polymorphisms (UEPs) since they have low mutation rates (~10^{-8} per generation) (de Knijff 2000). Multiallelic markers such as multiple microsatellites (average mutation rate m~0.2% per generation) and two minisatellites (m~6–11% per generation) can be used to differentiate Y chromosome due to their higher mutation rates (Kayser, Roewer et al. 2000; Dupuy, Stenersen et al. 2004). Literally, any combination of polymorphic markers along a non-recombining molecule constitutes a haplotype. Combinations of the biallelic markers define stable lineages of Y chromosomes referred to as haplogroups.

1.2.3.3.5. Y-SNPs

Y chromosome-specific single nucleotide polymorphisms (SNPs) are useful for identification of stable paternal lineages because of their low rate mutations and they play an important role in human migration studies because they enable evaluation of major differences between population groups. Because of a lack of recombination and a low mutation rate, these SNPs have been quite informative for kinship analyses, especially in cases where the reference and the evidence samples are separated by several generations. In addition, lineage SNPs are being used for missing person cases or mass disaster identifications in the forensic area.(Jobling and Tyler-Smith 2000)

Compared to another class of markers on Y chromosome (Y-STRs), SNPs have a much lower rate of mutation. SNPs are bi-allelic markers and provide less information per marker when compared to other class of Y chromosome markers, Y-STRs that have much more alleles (or allelic combinations in the case of multi-copy Y-STRs).

In the beginning of Y-SNP studies there has been developed different naming systems for haplogroup designation by the research groups around the world. Examination of different markers with various naming strategies results confusion in interpreting the relationships between markers and populations. In February 2002 a common, unified nomenclature was created by the Y-chromosome Consortium (YCC) and published in Genome Research (YCC 2002).

There are several technologies employed in Y-SNP marker analysis. Real-time PCR method is good at examination of markers one at a time, while others enable multiplex analysis (Lareu, Puente et al. 2001). The most comprehensive approach is the time-of-flight mass spectrometry by the analysis of 118 Y SNPs in 20 multiplexes (Paracchini, Arredi et al. 2002). Moreover SNaPshot assay and Luminex assays are being used for less number of SNP analysis (Jobling and Tyler-Smith 2000)

1.2.3.3.6. Y-STRs

Y-STRs are Short Tandem Repeats found on the male-specific region of Y chromosome. The Y-STRs are polymorphic among unrelated males and are inherited through the paternal line without any change (except mutations) through generations.

Y-chromosome STRs are demonstrating value in tracing paternal lineages. Other applications for Y-STRs include the forensic identification of male DNA from sexual assault cases, missing person's investigations, historical and genealogical studies. Before 2002, only 30 Y-STR loci were described , however today more than 200 Y-STR loci are deposited in the Genome Database (NCBI 2011). Human Genome Project and improved bioinformatics tools result rapid growth in discovery of new Y-STRs (Ayub, Mohyuddin et al. 2000).

In 1997, a core set of Y-STR markers or "minimal haplotype" loci that includes DYS19, DYS389I/II, DYS390, DYS391, DYS392, DYS393, and the highly polymorphic multi-copy locus DYS385 a/b was selected by the European forensic community (Kayser, Caglia et al. 1997; Pascali, Dobosz et al. 1999; Schneider, d'Aloja et al. 1999). The basis for the online Y-STR Haplotype Reference Database (http://www.yhrd.org) was formed by a multicenter study that 48 different subpopulation groups were studied with the minimal haplotype set (de Knijff, Kayser et al. 1997). In early 2003, the U.S. Scientific Working Group on DNA Analysis Methods (SWGDAM) added two more markers (DYS438 and DYS439) to the minimal haplotype set (Figure 1.7).

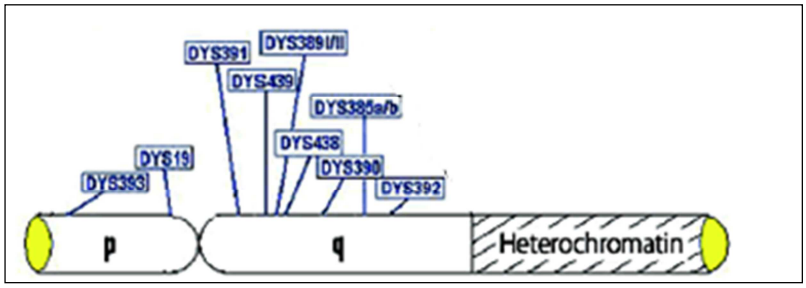


Figure 1.7 Core Y-STR loci recommended by SWGDAM

Chromosomal locations of the markers were determined by performing a BLAT search along the Y-chromosome using the reference sequences (Kent 2002). Relative

positions of the commonly used markers are shown in Figure 1.8. The minimal haplotype loci, which have been used extensively in population studies, are shown under the line, while all of the additional markers above. Only two of the minimal haplotype loci, DYS393 and DYS19 occur along the short arm (p).

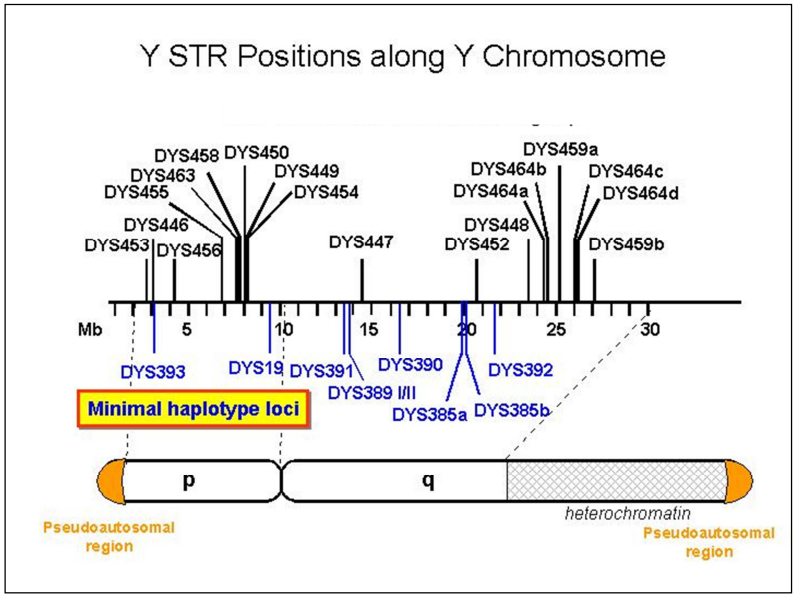


Figure 1.8 Commonly used Y-STR markers

Relative positions of some multicopy Y-STRs are shown in more detail in Figure 1.9. For example, the Y-STR locus DYS385 is present in two regions along the long arm of the Y chromosome 40 000 bp apart from each other and generate two different alleles when amplified with a single set of primers. The two alleles are typically labeled ‘a’ and ‘b’ with the ‘a’ represents smaller sized allele. If both ‘a’ and ‘b’ alleles are the same size (i.e., contain the same number of repeats) only a single peak would appear in an electropherogram (Figure 1.9 a). This duplicated locus is referred to as DYS385 a/b since there are two alleles present. In such a case where the resulting peak is usually twice as high during electrophoretic analysis compared to situations where alleles “a” and “b” are not equal in size and therefore they can be individually resolved.

Two PCR products can also be generated at another locus, DYS389 with a single set of primers. DYS389 has two repeat regions flanked on one side by a similar sequence. The forward PCR primers bind adjacent to both repeats generating amplicons

that differ in size by ~120 bp (Figure 1.9 b). DYS389I PCR product becomes a subset of the DYS389II amplicon so some analyses subtract DYS389II–DYS389I to get DYS389II (Redd, Agellon et al. 2002).

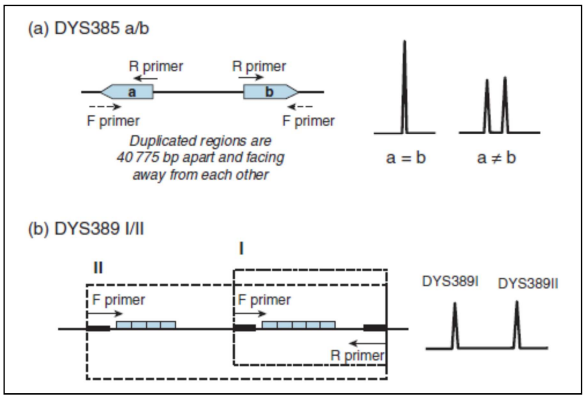


Figure 1.9 (a) DYS385a/b (b) DYS389I/II

There are commercially available kits to perform Y-STR analysis. The first kit was Y-PLEX™ 6 from ReliaGene Technologies (New Orleans, LA), which co-amplifies DYS19, DYS389II, DYS390, DYS391, DYS393, and DYS385 a/b. Table 1.4 lists some of commercially available kits with their release dates and the loci that they amplify. Today, AmpF*l*STR® Y-filer™ PCR Amplification Kit is the most commonly used kit with 17 Y specific STR markers. Minimal haplotype loci, SWDGAM loci and Y-filer™ kit loci are shown on Table 1.5.

Table 1.4 Some of commercially available Y-STR kits

Kit Name	Release Date	Dye Color	Loci Amplified
Y-PLEX™ 6 (ReliaGene Technologies)	January 2001	B Y	DYS393, DYS19, DYS389II, DYS390, DYS391, DYS385 a/b
Y-PLEX™ 5 (ReliaGene Technologies)	July 2002	B G Y	DYS389I, DYS389II, DYS439, DYS438, DYS392
Y-PLEX™ 12 (ReliaGene Technologies)	September 2003	B G Y	DYS392, DYS390, DYS385 a/b, DYS393, DYS389I, DYS391, DYS389II, Amelogenin, DYS19, DYS439, DYS438
PowerPlex® Y (Promega Corporation)	October 2003	B G Y	DYS391, DYS389I, DYS439, DYS389II, DYS438, DYS437, DYS19, DYS392,DYS393, DYS390, DYS385 a/b
Yfiler™ (Applied Biosystems)	Fall 2004	B G Y R	DYS456, DYS389I, DYS390, DYS389II, DYS458, DYS19, DYS385 a/b, DYS393, DYS391, DYS439, C4, DYS392, H4, DYS437, DYS438, DYS448

Table 1.5 Minimal, SWGDAM extended and Y-Filer Extended haplotypes

	Y-STR Markers	**Repeat Motif**	**Allele Range**	**Length (bp)**
Minimal Haplotypes	DYS19	TAGA	10–19	176-211
	DYS385a/b	GAAA	7-25	242–318
	DYS389 I/II	TCTG TCTA	10-15 24-34	142–164 253–293
	DYS390	TCTA TCTG	18-27	192–227
	DYS391	TCTA	7-13	150-176
	DYS392	TAT	7-18	291–326
	DYS393	AGAT	8-16	130-131
SWGDAM extended loci	DYS438	TTTTC	8-13	223-248
	DYS439	AGAT	8-15	197–225
Y-Filer extended loci	DYS437	TCTA	13-17	182–198
	DYS448	AGAGAT	17-24	280-324
	DYS456	AGAT	13-18	104-123
	DYS458	GAAA	14-20	130-155
	Y-GATA-H4	TAGA	8-13	122-142
	DYS635 (Y-GATA-C4)	TSTA compound	20-26	246-270

1.2.3.3.7. Y-STR Haplotype Databases

Several hundred publications now exists describing population data with Y-chromosome markers. This much data result in a need of a common source of information, internet accessible database. The largest and most widely used Y-STR database has been available online since 2000. The information in this database comes from 89 collaborating institutions located in 36 different countries (Roewer 2003).
After 2004, more than 24 000 samples from greater than 224 populations around the world can be searched via the internet at the following web sites: http://www.ystr.org or http://www.yhrd.org. It is important to keep in mind that the samples entered into this database have been typed only at the minimal haplotype loci (DYS19, DYS389I/II, DYS390, DYS391, DYS392, DYS393, and DYS385 a/b). At present, these samples do not include the additional SWGDAM recommended loci DY438 and DYS439 that are now available in commercial Y-STR kits. Other internet-accessible Y-STR databases are also available now, as http://www.ybase.org, http://www.ysearch.org, http://www.promega.com (Butler 2005).

The development of these databases is important not only for haplotype frequency estimation and subsequent match probability calculations in forensic studies but also for comparative population analysis.

1.3 LITERATURE SURVEY FOR TURKEY

During the literature survey, it is found that there are a few notable studies related to Turkish population and most of them focused on Central Anatolia. Besides there are two articles including Turkish male populations living in Germany and Bulgaria. Henke studied on Indian, German and Turkish populations and determined a high degree of diversity in Turks (Henke, Henke et al. 2001). Zaharova also revealed high power of discrimination between Bulgarian and Turkish male groups (99%) (Zaharova, Andonova et al. 2001).

In 2003, a research group from Max Planck Institute for Evolutionary Anthropology typed minimal haplotype loci in populations from the Caucasus, Iran and Turkey (Nasidze, Schadlich et al. 2003). However only 39 samples were from Turkey

and collected from Ankara. This study represents a regional population, not Turkey at all.

In the same year, Council of Forensic Medicine, under the Ministry of Justice, announced Turkish population data. In this study 200 male individuals who were involved in legal proceedings concerning paternity, was genotyped for 6 Y-STR markers (DYS19, DYS389II, DYS390, DYS391, DYS393 and DYS 385) (Asicioglu, Akyuz et al. 2003). However, the study did not include DYS392 and DYS389I loci of the core minimal haplotype loci recommended by YCC.

A comprehensive study in 2004 was conducted by a collaborative group from Stanford University (USA), Stanford Genome Technology Center (USA), Istanbul University (Turkey), University of Pavia (Italy) and Tartu University (Estonia). 553 male individuals were analyzed at DYS19, DYS388, DYS390, DYS391, DYS392, DYS393, DYS389I, DYS389II, DYS439, DYS461 and 89 biallelic polymorphisms in order to reveal Y-Chromosome haplotype strata in Anatolia (Cinnioglu, King et al. 2004). Nevertheless a multi-copy polymorphic marker DYS385a/b was not included in this study which probably would increase the diversity of the population.

Y-STR haplotypes in Central Anatolia region of Turkey was determined by the Criminal Department of Gendarmerie General Command in 2004. 113 unrelated male donors from Central Anatolia were typed at the 11 Y-STR Loci (DYS19, DYS385a/b, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS438 and DYS439) (Cakir, Celebioglu et al. 2004). The study was in concordance with the previous studies showing high degree of diversity in Turkey.

In the same year, another study on Central Anatolian population was conducted by Rustamova and his colleagues (Rustamov, Gumus et al. 2004). 59 male donors were typed at 8 Y-STR loci (DYS19, DYS388, DYS389I/II, DYS 390, DYS 391, DYS 392 and DYS393). In further years Rustamova extended the scope of his study by adding 4 more Y-STR loci (DYS 385a/b and YCAIIa/b) and analyzed 250 unrelated male donors from Turkey. The allelic and haplotypic frequencies were reported in his PhD thesis (PhD thesis Rustamova 2006). This is the first study that all minimal haplotype loci were analyzed in Turkey.

In 2006, Y-STR haplotypes in populations from the Eastern Mediterranean region of Turkey (104 Turkish and Arabic-speaking Eti Turks from Adana, 111 Roma and 110 Turks from Kahramanmaraş) were presented, regarding 10 Y-STR loci namely DXYS156-Y, DYS19, DYS385, DYS 389I/II, DYS 390, DYS 391, DYS 392 and DYS393 (Donbak, Bajanowski et al. 2006).

Last year, 140 male donors living in four rural Central Anatolian villages were analyzed for variation at 17 Y-chromosome STR and 15 autosomal STR loci. The results of the study indicated the fact that distribution of paternal genetic diversity in this region has been influenced by the local social traditions in rural Anatolia (Alakoc, Gokcumen et al.).

1.4 THE OBJECTIVE OF THE STUDY

As seen in the literature survey there are only two studies representing Turkey as a whole (Cinnioglu, King et al. 2004; Rustamov 2006). In these studies number of Y-STR markers is 8 and 12 respectively. The aim of the present study is to demonstrate the high degree of genetic diversity in Turkey therefore number of markers were increased to 17 (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a/b, DYS438, DYS439, DYS437, DYS448, DYS458, DYS456, DYS635, and Y-GATA-H4) in approximately 120 unrelated male individuals. Moreover to represent all regions of Turkey properly, the samples were collected according to population ratio of each region (Figure 2.1).

Furthermore, there is an absence of national Y-STR database in Turkey. A national database would help studies on anthropology, forensic genetics and population. The data produced in this research would also contribute such database building projects.

The objectives of this study can be summarized as follows:

- to calculate allele and haplotype frequencies for each region and Turkey as a whole
- to determine the locus and haplotype diversities of the population in Turkey

- to reveal the intra-population genetic relationship
- to contribute international databases
- to compare Turkey population with worldwide populations according to Y-STR profiles

CHAPTER II

MATERIALS & METHODS

2.1 SAMPLE COLLECTION

Samples for this study were collected from 120 healthy male individuals from different geographical regions of Turkey. The regions of sampling for all of the individuals analyzed presented in Appendix A. Prior to sample collection, a *Request for Review by the Ethic Committee of Fatih University Medical School, Ankara for the Protection of Human Subjects* was submitted and approved for the project. Special care was taken to avoid sampling from related individuals. Blood samples were collected with informed consent from individuals of known ethnic origin and family history. A copy of the questionnaire with *The Informed Consent to Participate in a Research Study* is provided in (Appendix B). The populations included in this study are the Marmara (*n*=23), Black Sea (n=25), Aegean (n=13), Central Anatolia (n=19), Mediterranean (n=13), Eastern Anatolia (n=14), Southeastern Anatolia (n=9), and other countries (n=4) (Figure 2.1). This research was supported by Fatih University Scientific Research Projects Fund (BAP), under the project number P50091001.

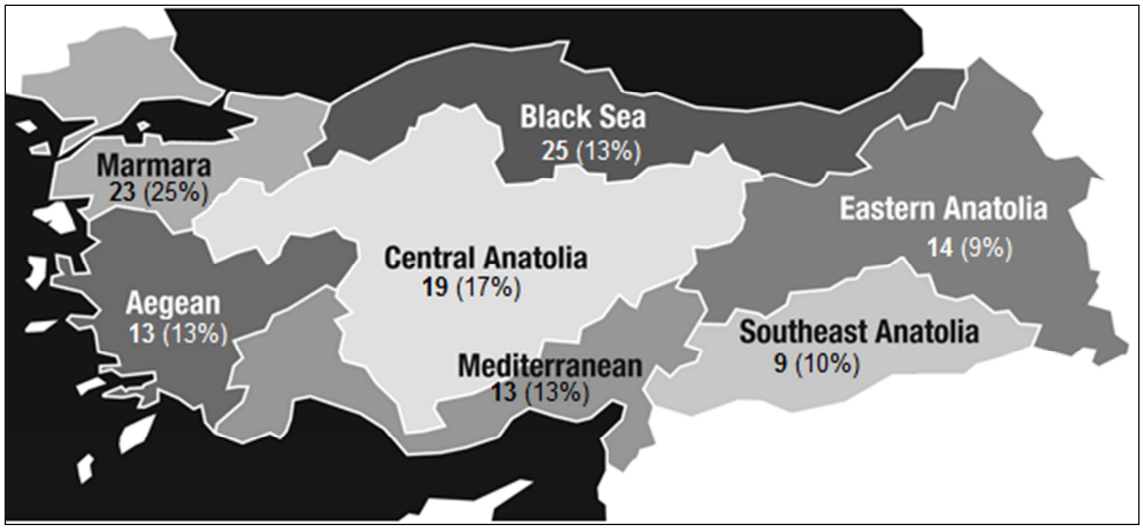
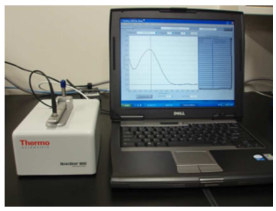


Figure 2.1 Regional Sampling (The numbers represent the sample size for each region and the percentages represent the population ratio of indicated region to the total population of Turkey)

Extract and Quantify DNA



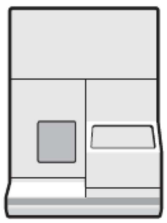
Gentra® Puregene® Blood Kit

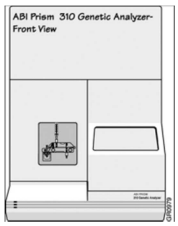
PCR Amplify DNA

AmpF STR® YFiler™ PCR Amplification kit

Techne® TC-412 Thermal Cycler

Perform Electrophoresis





ABI PRISM® 310 Analyzer

Analyze Data

GeneMapper ID Software v.4.0

Figure 2.2 Workflow of the study

2.2 GENERAL INFORMATION OF 17 Y-STR LOCI

Table 2.1 Y-STR Markers in AmpF*l*STR® Y-filer™ PCR Amplification Kit (Applied-Biosystems 2006)

Y-STR Markers	Repeat Motif	Alleles in Allelic Ladder	Length (bp)	GenBank Accession	Reference Allele
DYS19	TAGA	10–19	176-211	AC017019	15
DYS385a/b	GAAA	7-25	242–318	AC022486 (r&c)	11
DYS389 I/II	TCTG TCTA	10-15 24-34	142–164 253–293	AC004617 (r&c)	12, 29
DYS390	TCTA TCTG	18-27	192–227	AC011289	24
DYS391	TCTA	7-13	150-176	AC011302	11
DYS392	TAT	7-18	291–326	AC011745 (r&c)	13
DYS393	AGAT	8-16	130-131	AC006152	12
DYS437	TCTA	13-17	182–198	AC002992	16
DYS438	TTTTC	8-13	223-248	AC002531	10
DYS439	AGAT	8-15	197–225	AC002992	13
DYS448	AGAGAT	17-24	280-324	AC025227	22
DYS456	AGAT	13-18	104-123	AC010106	15
DYS458	GAAA	14-20	130-155	AC010902	16
Y-GATA-H4	TAGA	8-13	122-142	AC011751 (r&c)	12
DYS635	TSTA	20-26	246-270	AC004772 (r&c)	23

2.3 EXPERIMENTAL PROCESSES

2.3.1 DNA isolation and quantitation

The whole blood was obtained by venipuncture and collected into EDTA vacutainer tubes. DNA was isolated from blood using Gentra® Puregene® Blood Kit according to the manufacturer's protocol (Qiagen, Hilden, Germany). Extracted samples were stored at -20° C. The extracted DNA quantity was measured using a Nanodrop® ND-100 spectrophotometer (Nanodrop Technologies, Wilmington, DE, USA).

2.3.2 Amplification of DNA with AmpFlSTR® Y-filer™ PCR Kit

The polymerase chain reaction (PCR) is a technique used for amplification from a small amount of template DNA to generate thousands to millions of copies of a specific DNA sequence. In this study, amplification of the 17 Y-STR loci was performed with the AmpF*l*STR® Y-filer™ PCR Amplification Kit according to manufacturer's instructions (Applied-Biosystems 2006).

This particular kit uses primers that are labeled with fluorescent dyes for detection purposes during capillary electrophoresis. The following table shows the loci that are amplified by the Yfiler kit and the corresponding dyes used. The AmpFlSTR Yfiler Kit Allelic Ladder is used to genotype the analyzed samples. The alleles contained in the allelic ladder are also listed in the Table 2.2.

Table 2.2 AmpFlSTR® Yfiler™ Kit Loci and Dyes

Y-STR Markers	Alleles included in Y-Filer Kit Allelic Ladder	Dye Label
DYS456	13-18	6-FAM™
DYS389 I	10-15	
DYS389 II	24-34	
DYS390	18-27	
DYS385a/b	7-25	VIC®
DYS19	10–19	
DYS458	14-20	
DYS393	8-16	NED™
DYS635	20-26	
DYS391	7-13	
DYS392	7-18	
DYS439	8-15	
DYS437	13-17	PET®
DYS438	8-13	
DYS448	17-24	
Y-GATA-H4	8-13	

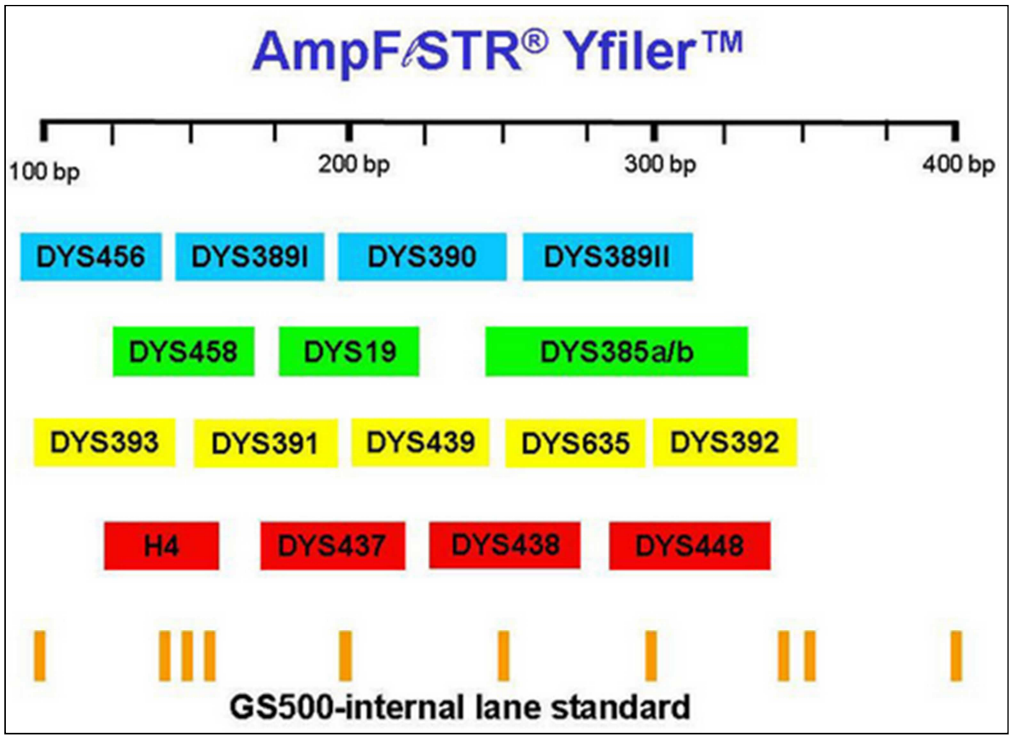


Figure 2.3 Schematic of Y-Filer Kit Loci
(The schematic diagram illustrates the fluorescent dye label color and relative PCR product size ranges for Y-Filer Y-STR Loci)

For a proper PCR set-up, at the beginning necessary dilutions were made to the DNA samples to obtain an optimal concentration ranging from 0.5 ng/µL to 1.5ng/µL. Once proper dilutions have been made, a master mix that will be added to all the reactions is prepared as in the following table:

Table 2.3 Master mix for PCR

Component	Volume Per Reaction (µL)
AmpF*l*STR® PCR reaction mix	9,2
AmpF*l*STR® Primer set	5,0
Amplitaq Gold® DNA polymerase	0,8

The master mix contains AmpF*l*STR® PCR reaction mix, AmpF*l*STR® Primer set, Amplitaq Gold® DNA polymerase. 10µl of samples are loaded to the appropriate wells. Then 15µl of master mix was added onto each sample well, giving a total of 25µl reaction volume as in the Manufacturer's instructions (Applied-Biosystems 2006). PCR amplification was performed on the *TC-412 Techne*® Thermal Cycler according to the

conditions given in the below table and the products were stored at an appropriate temperature (Table 2.4).

Table 2.4 PCR conditions

Initial Incubation Step	**Cycles (30 cycles)**			**Final Extension**	**Final Hold**
	Denature	**Anneal**	**Extend**		
HOLD	CYCLE			HOLD	HOLD
95°C 15 min	94°C 1 min	61°C 1 min	72°C 1 min	60°C 80 min	4°C ∞

Table 2.5 PCR product storage temperatures

Storage time	**Storage temperature**
Less than 2 weeks	2 to 8 °C
More than 2 weeks	–15 to –25 °C.

2.3.3 Capillary Electrophoresis

2.3.3.1. Analysis of AmpFlSTR® Y-filer™ PCR Products

Capillary electrophoresis is an effective tool for the separation of DNA fragments by size in order to be analyzed and is used in the medical and scientific communities. The PCR products generated from the AmpF*l*STR® Y-filer™ PCR amplification kit were analyzed with the ABI Prism® 310 Genetic Analyzer (Applied Biosystems, Foster City, CA). The analysis software program used for this project is Applied Biosystems GeneMapper® Software, which has precise base sizing capabilities and designates appropriate allele calls. In this project, the fragments were separated in POP-4™ polymer.

To set up for capillary electrophoresis, a master mix is prepared containing HiDi Formamide and GeneScan™ 500 LIZ according to Applied Biosystem Corporation's (2003) instructions (Applied-Biosystems 2006). 1µl of amplified STR product and

allelic ladder are added to the appropriate wells. 11.5 µl of master mix is added to all the reactions, giving a total 12.5µl reaction volume as shown in the below table:

Table 2.6 Master mix for Capillary Electrophoresis

Set up for CE	**ABI Prism® 310**
HiDi Formamide	11 µl
LIZ 500	0,5 µl
Sample DNA or Allelic Ladder	1 µl
TOTAL	12,5 µl

Samples were then denatured at 95°C for 3 minutes and immediately chilled at -20°C for 3 minutes. The following conditions were used with the 310 Genetic Analyzer Data Collection Software 3.1.0 (Applied Biosystems).

Table 2.7 Capillary Electrophoresis Conditions

Condition	**Setting**
Dye Set	DS-22 Matrix Standards
Filter Set	G5v2
Size Standard	GeneScan™ 500 LIZ® Size Standard
Run Module	GS STR POP4 (1ml) G5v2.md5
Polymer	310 POP-4™ Polymer (25 mL)
Capillary	310™ Capillary Array 36 cm, 47 cm x 50 µm ID
Running Buffer	10× Genetic Analyzer Buffer with EDTA (25 mL)
Temperature	60°C
Injection voltage	3.0 kV
Injection time	5 seconds
Run voltage	15 kV
Run time	~34 min 30 sec

After data collection, samples were analyzed with GeneMapper® ID version 4.0 for Y-STR peak designation, sizing, and allele identification through size comparisons with allelic ladders provided by the amplification kit. Many samples were analyzed

more than once on the ABI 310™ in order to ensure clear, clean DNA profiles and to verify results.

2.3.3.2. Allelic Ladder

In order to determine the polymorphisms of the seventeen Y-specific STR loci, an allelic ladder which is constructed by Applied Biosystems was used. Allelic ladders provide a consistent currency that aids in quality assurance of results as well as compatibility of results going into DNA databases. The allele nomenclature is based on the number of repetitive sequences according to the recommendations of International Society of Forensic Genetics, ISFG.

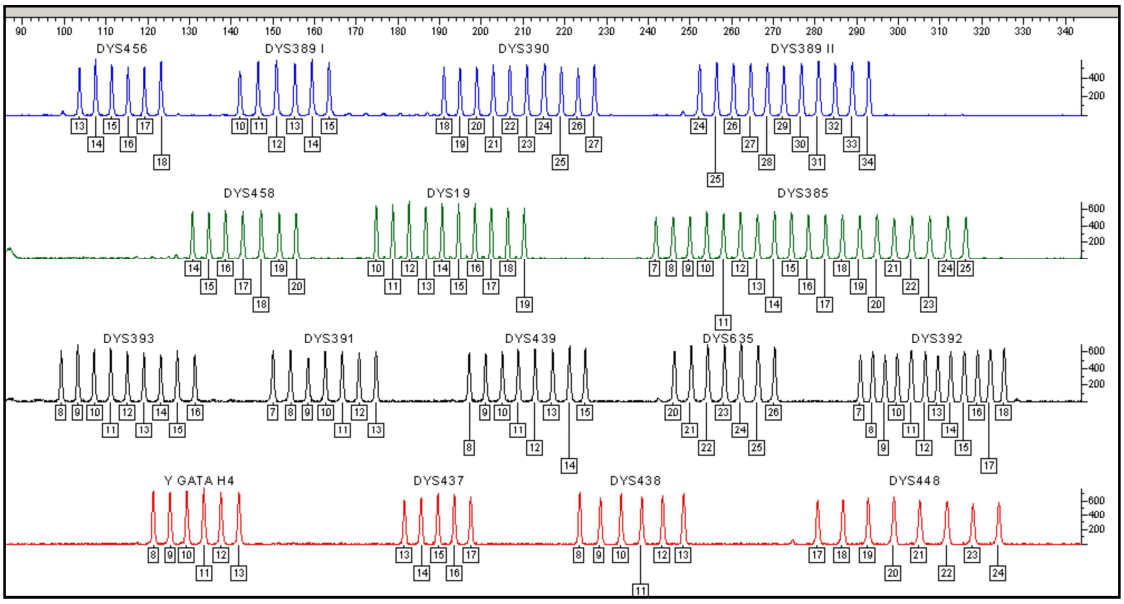


Figure 2.4 Allelic Ladder in AmpF*l*STR® Y-filer™ PCR Amplification Kit

2.3.4 Statistical Analysis

Once allele designations were made for all Y chromosomal STR samples, the frequencies for each locus were determined. In the case of the multi-copy locus DYS385a/b, the allele frequency was evaluated as a combination of two alleles as necessary (Mulero, Chang et al. 2006). All samples were examined to calculate the frequency of occurrence of each specific haplotype profiles. The number of occurrences was divided by the total number of profiles (n=120) to obtain haplotype frequencies. A table of Y-STR haplotype frequencies was also generated. Haplotype frequency, gene

diversity and all other statistic calculations were calculated with the software program Arlequin Version 3.5.1.2 (Excoffier and Lischer 2010).

Population pairwise genetic distances (F_{st}) and P values were calculated using analysis of molecular variance (AMOVA), with 10000 permutations, using an on-line tool of the YHRD. Significance was set at $P < 0.05$. Using this tool multidimensional scaling (MDS) plot was also obtained.

CHAPTER III

RESULTS

In the study 17 Y-STR loci allele frequencies were calculated for Turkey and its geographical regions. The results are listed in Table 3.17 and 3.18. Haplotype frequencies for Turkey are listed in Table 3.19 and Table 3.20 (DYS385 excluded). Haplotype sharing among samples are summarized in Table 3.22. Genetic diversities for each locus are presented in Table 3.23 (Turkey) and Table 3.24 (Regions). Allele duplication is reported in DYS19 locus. Inter population comparison based on pairwise Fst values and P values is done and MDS Plot is drawn.

F st distances between population samples were used for multidimensional scaling analysis (MDS). This technique is used to express the dissimilarity between populations in a multdimensional graph. The distances between points represent the observed genetic differences between populations. “Stress” is defined as the goodness of fit between the distances in the graphic configuration and the original genetic distances. A value of 0 means a perfect fit, a value of 1 means a total mismatch.(Varzari 2006)

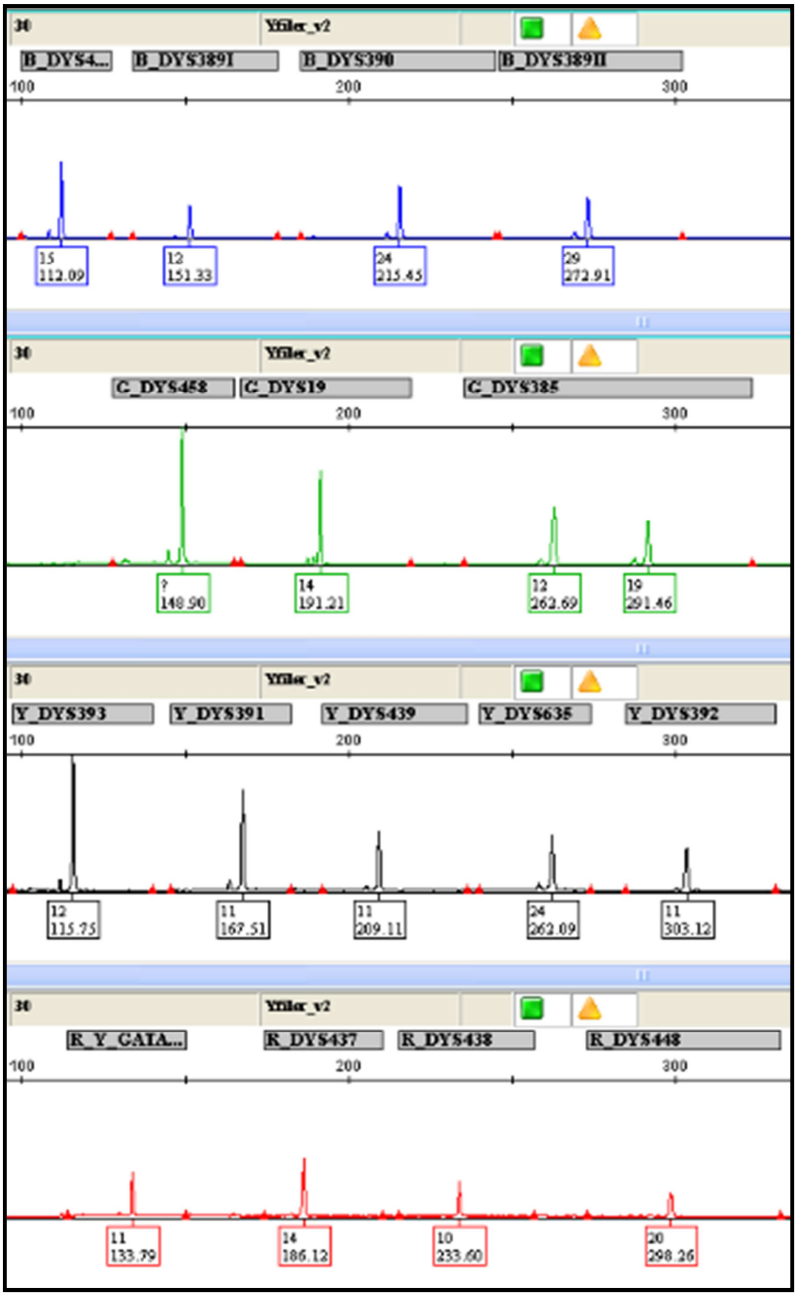


Figure 3.1 A screen shot of an electropherogram representing sample 30

3.1 ALLELE FREQUENCY

Allele frequencies of Turkey and its geographical regions are given in this part.

3.1.1 TURKEY

First, allele frequencies for each locus in population of Turkey are shown in graphs and tables.

3.1.1.1. DYS19

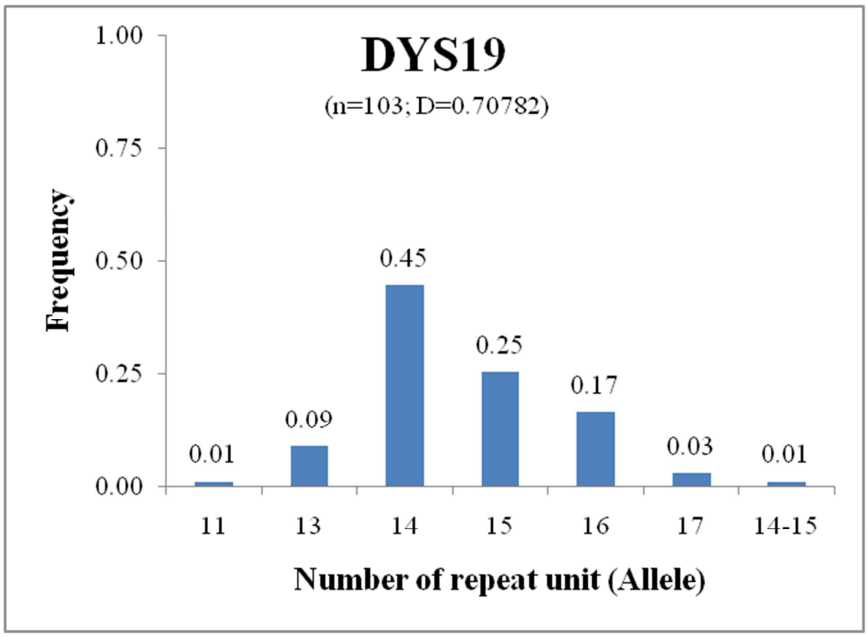


Figure 3.2 Allelic frequencies of DYS19 for Turkey

Table 3.1 Allelic frequencies of DYS19 for Turkey

	Allele	Freq.	Std. Dev.
DYS19	11	0.01	0.0097
	13	0.09	0.0280
	14	0.45	0.0492
	15	0.25	0.0430
	16	0.17	0.0368
	17	0.03	0.0167
	14-15	0.01	0.0097

3.1.1.2. DYS385 a/b

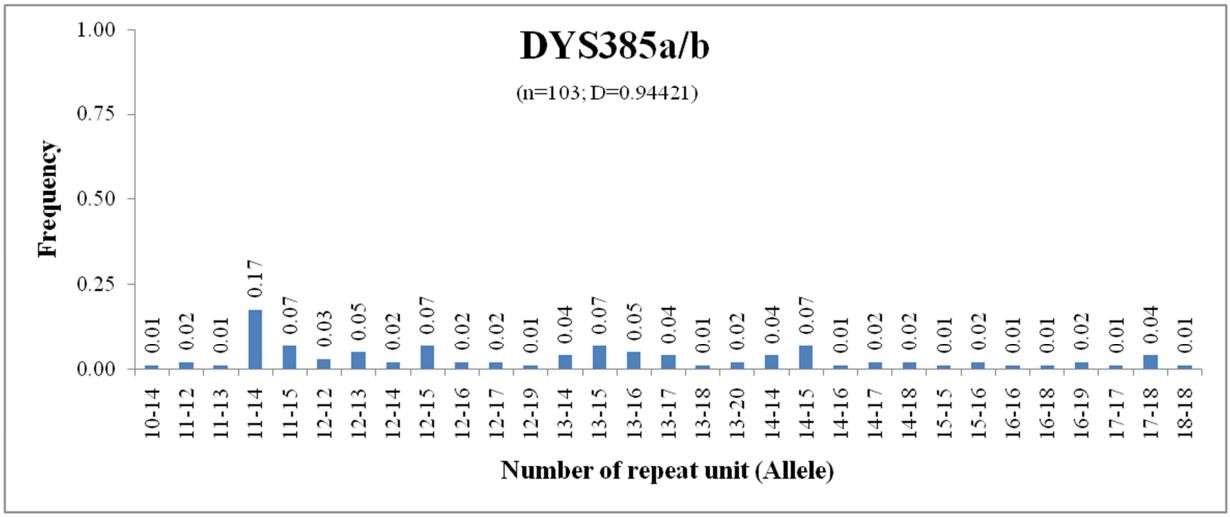


Figure 3.3 Allelic frequencies of DYS385a/b for Turkey

Table 3.2 Allelic frequencies of DYS385a/b for Turkey

	Allele	Freq.	Std. Dev.
DYS385a/b	10-14	0.01	0.0097
	11-12	0.02	0.0137
	11-13	0.01	0.0097
	11-14	0.17	0.0376
	11-15	0.07	0.0249
	12-12	0.03	0.0167
	12-13	0.05	0.0213
	12-14	0.02	0.0137
	12-15	0.07	0.0249
	12-16	0.02	0.0137
	12-17	0.02	0.0137
	12-19	0.01	0.0097
	13-14	0.04	0.0191
	13-15	0.07	0.0249
	13-16	0.05	0.0213
	13-17	0.04	0.0191
	13-18	0.01	0.0097
	13-20	0.02	0.0137
	14-14	0.04	0.0191
	14-15	0.07	0.0249
	14-16	0.01	0.0097
	14-17	0.02	0.0137
	14-18	0.02	0.0137
	15-15	0.01	0.0097
	15-16	0.02	0.0137
	16-16	0.01	0.0097
	16-18	0.01	0.0097
	16-19	0.02	0.0137
	17-17	0.01	0.0097
	17-18	0.04	0.0191
	18-18	0.01	0.0097

3.1.1.3. DYS389 I

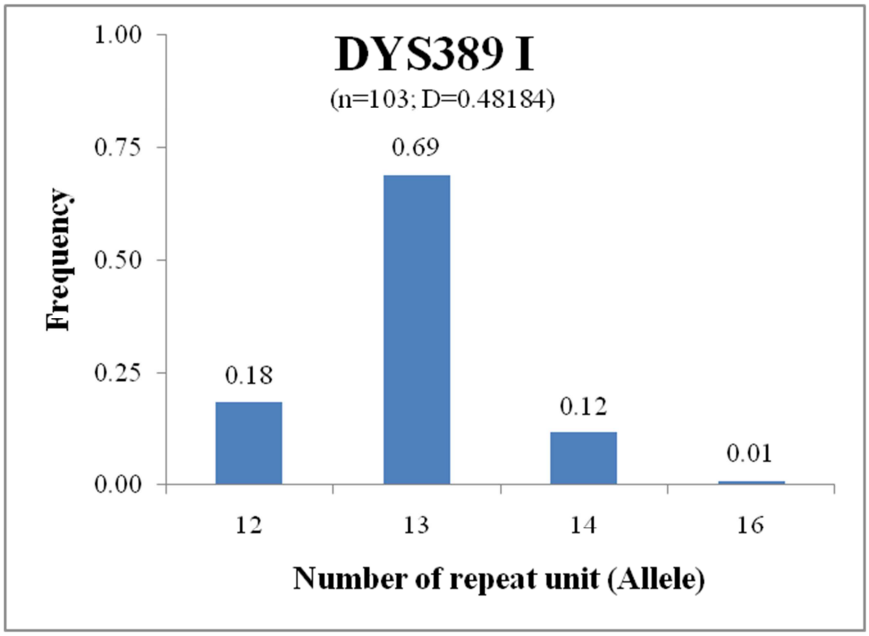


Figure 3.4 Allelic frequencies of DYS389 I for Turkey

Table 3.3 Allelic frequencies of DYS389 I for Turkey

	Allele	Freq.	Std. Dev.
DYS389 I	12	0.18	0.0384
	13	0.69	0.0458
	14	0.12	0.0318
	16	0.01	0.0097

3.1.1.4. DYS389 II

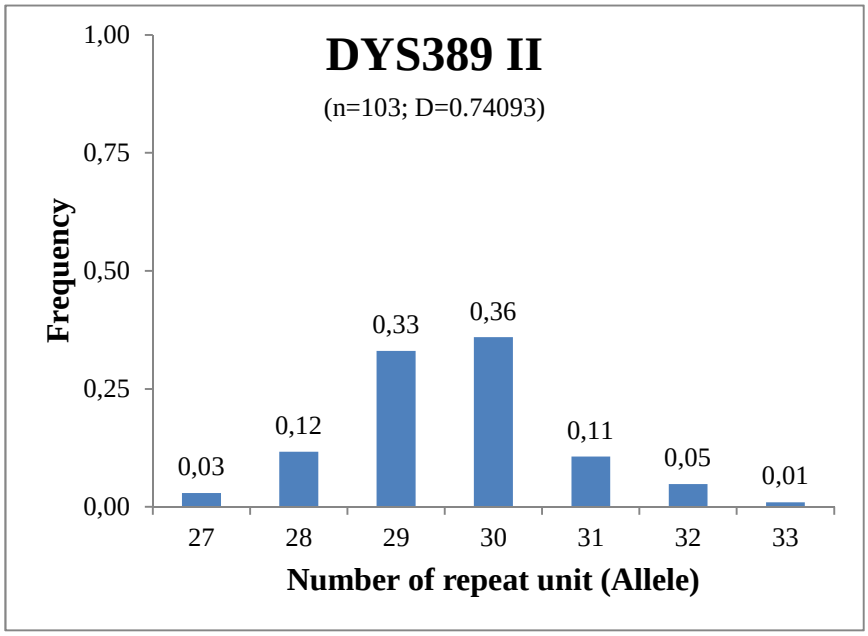


Figure 3.5 Allelic frequencies of DYS389 II for Turkey

Table 3.4 Allelic frequencies of DYS389 II for Turkey

	Allele	Freq.	Std. Dev.
DYS389 II	27	0.03	0.0167
	28	0.12	0.0318
	29	0.33	0.0466
	30	0.36	0.0475
	31	0.11	0.0306
	32	0.05	0.0213
	33	0.01	0.0097

3.1.1.5. DYS390

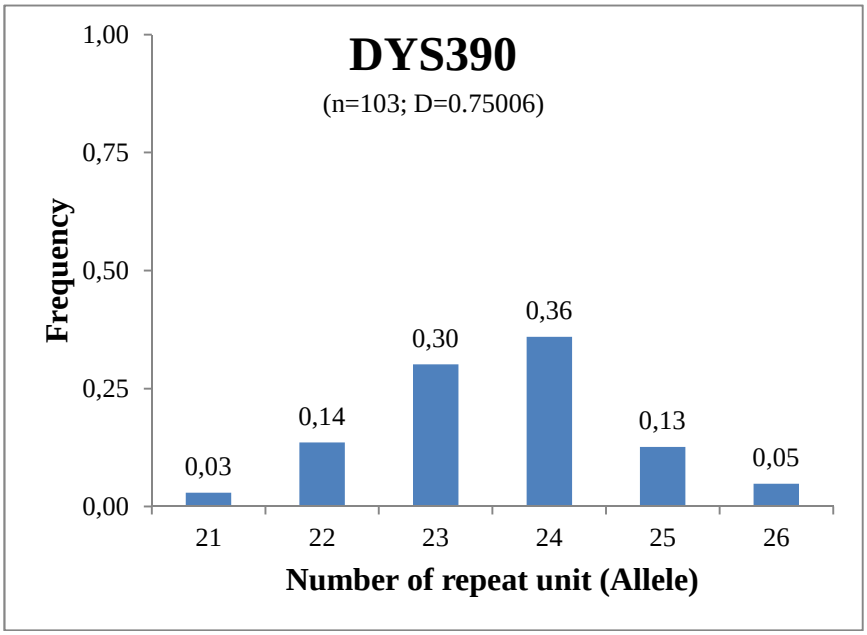


Figure 3.6 Allelic frequencies of DYS390 for Turkey

Table 3.5 Allelic frequencies of DYS390 for Turkey

	Allele	Freq.	Std. Dev.
DYS390	21	0.03	0.0167
	22	0.14	0.0339
	23	0.30	0.0454
	24	0.36	0.0475
	25	0.13	0.0329
	26	0.05	0.0213

3.1.1.6. DYS391

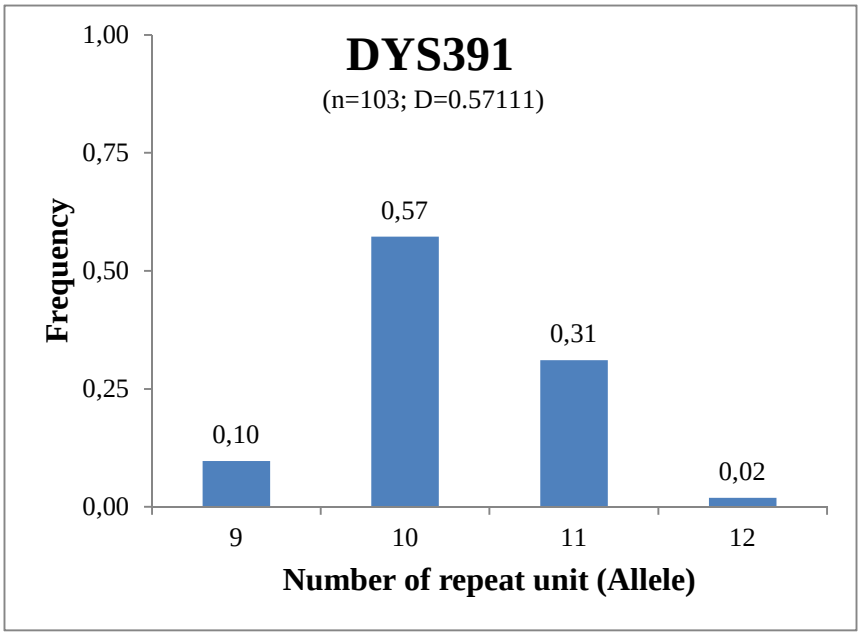


Figure 3.7 Allelic frequencies of DYS391 for Turkey

Table 3.6 Allelic frequencies of DYS391 for Turkey

	Allele	Freq.	Std. Dev.
DYS391	9	0.10	0.0293
	10	0.57	0.049
	11	0.31	0.0458
	12	0.02	0.0137

3.1.1.7. DYS392

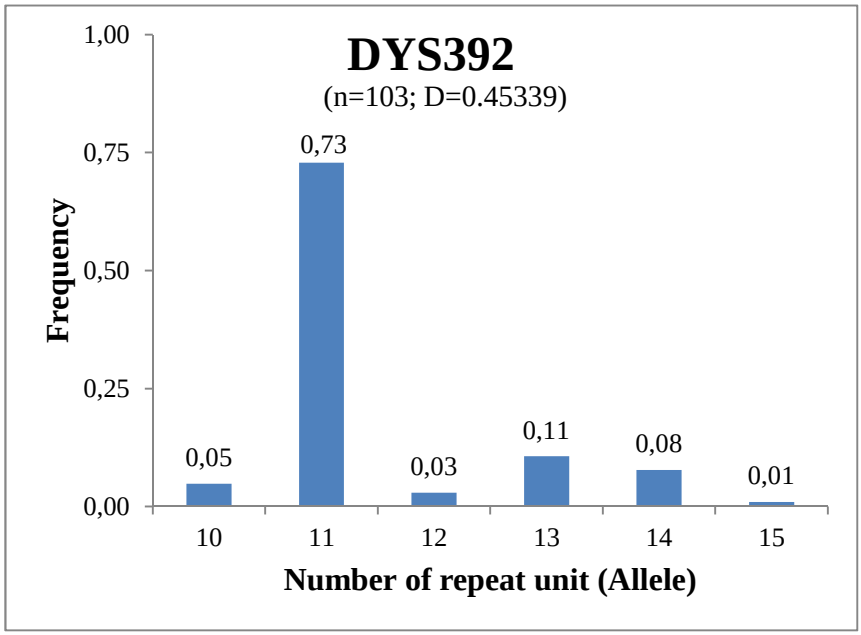


Figure 3.8 Allelic frequencies of DYS392 for Turkey

Table 3.7 Allelic frequencies of DYS392 for Turkey

	Allele	Freq.	Std. Dev.
DYS392	10	0.05	0.0213
	11	0.73	0.0441
	12	0.03	0.0167
	13	0.11	0.0306
	14	0.08	0.0265
	15	0.01	0.0097

3.1.1.8. DYS393

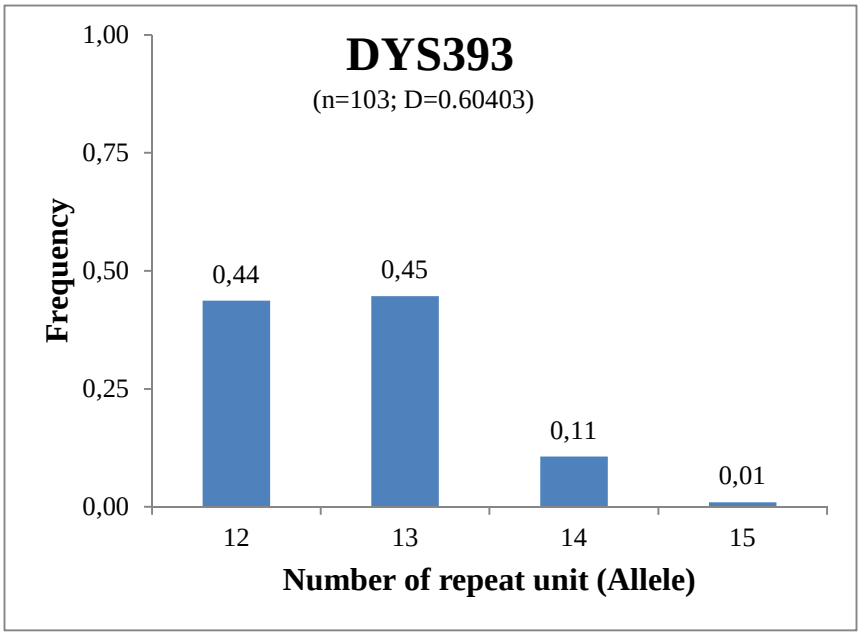


Figure 3.9 Allelic frequencies of DYS393 for Turkey

Table 3.8 Allelic frequencies of DYS393 for Turkey

	Allele	Freq.	Std. Dev.
DYS393	12	0.44	0.0491
	13	0.45	0.0492
	14	0.11	0.0306
	15	0.01	0.0097

3.1.1.9. DYS437

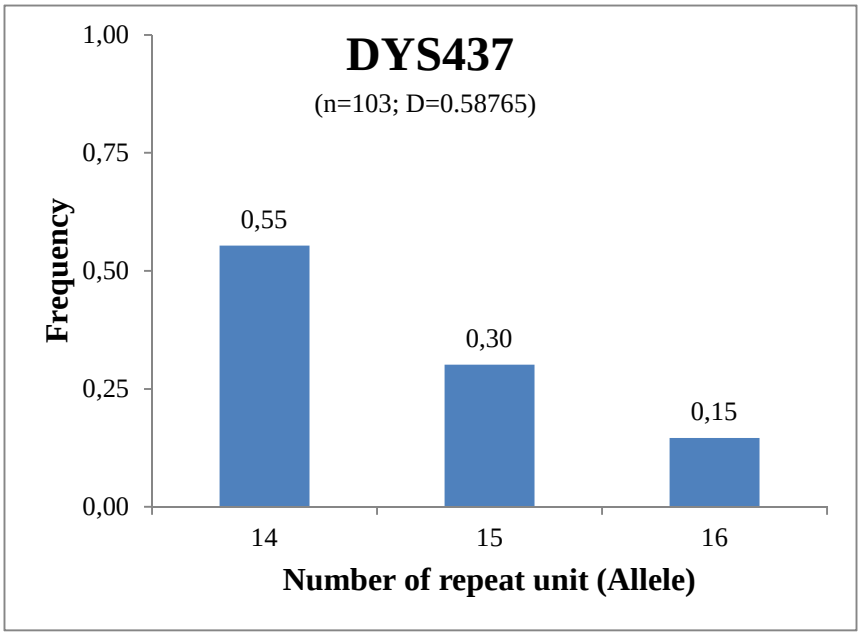


Figure 3.10 Allelic frequencies of DYS437 for Turkey

Table 3.9 Allelic frequencies of DYS437 for Turkey

	Allele	Freq.	Std. Dev.
DYS437	14	0.55	0.0492
	15	0.30	0.0454
	16	0.15	0.0349

3.1.1.10.DYS438

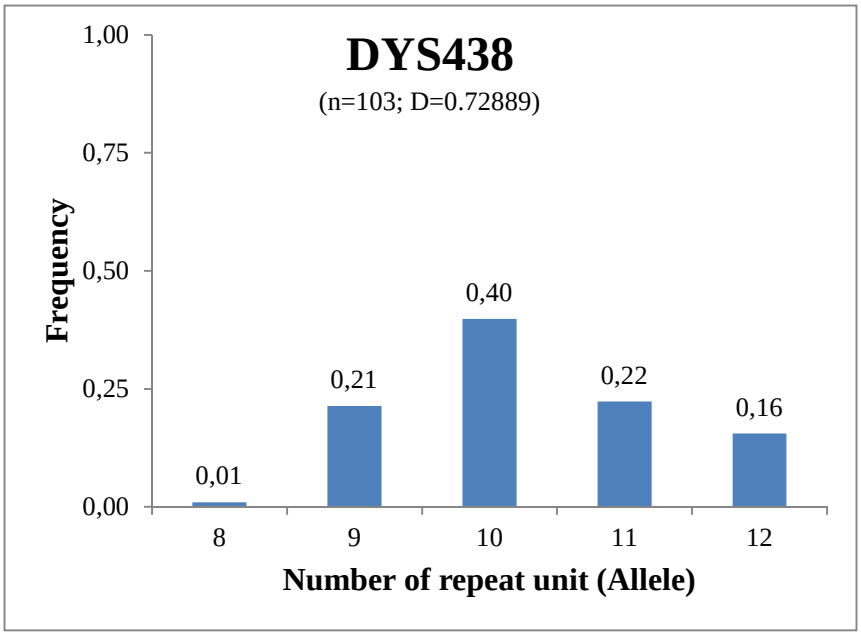


Figure 3.11 Allelic frequencies of DYS438 for Turkey

Table 3.10 Allelic frequencies of DYS438 for Turkey

	Allele	Freq.	Std. Dev.
DYS438	8	0.01	0.0097
	9	0.21	0.0406
	10	0.40	0.0485
	11	0.22	0.0412
	12	0.16	0.0359

3.1.1.11.DYS439

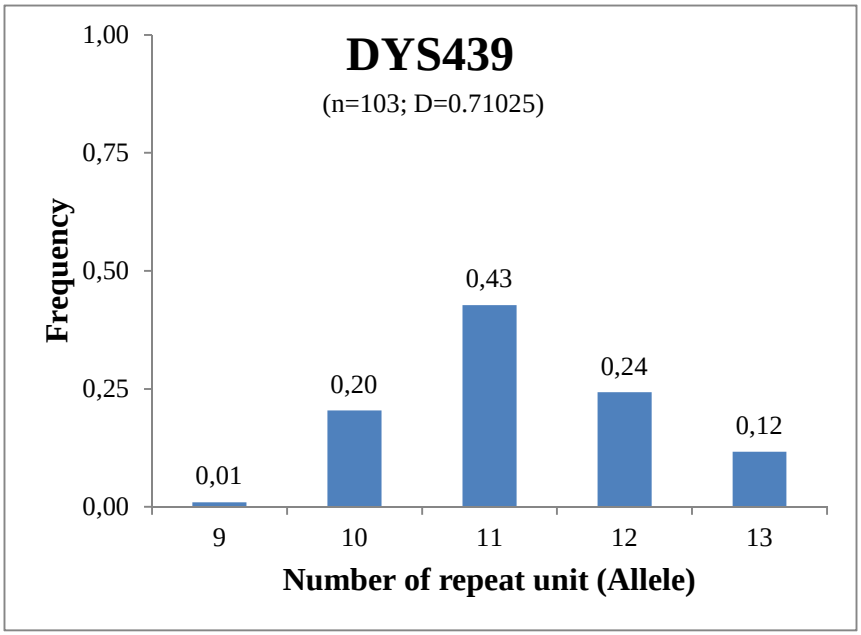


Figure 3.12 Allelic frequencies of DYS439 for Turkey

Table 3.11 Allelic frequencies of DYS439 for Turkey

	Allele	Freq.	Std. Dev.
DYS439	9	0.01	0.0097
	10	0.20	0.0399
	11	0.43	0.049
	12	0.24	0.0425
	13	0.12	0.0318

3.1.1.12.DYS448

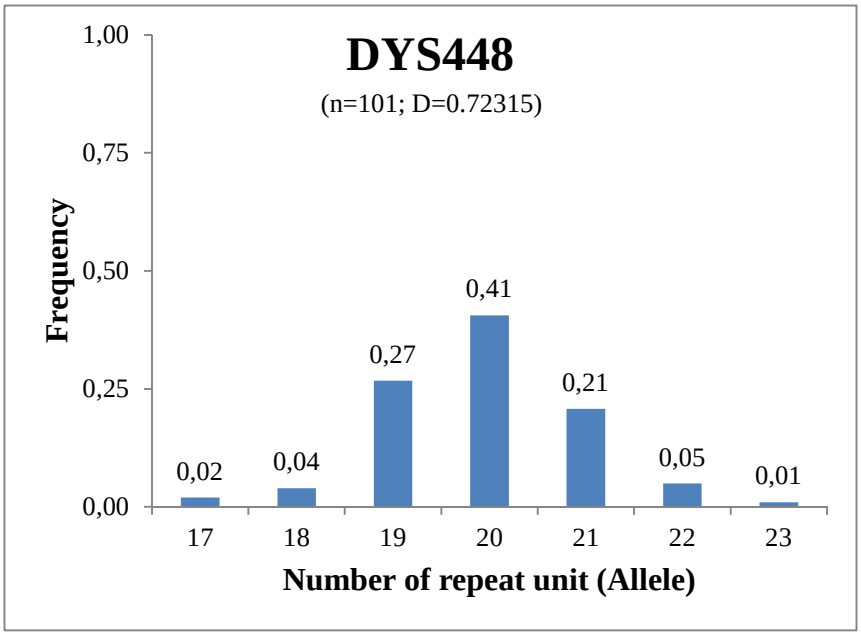


Figure 3.13 Allelic frequencies of DYS448 for Turkey

Table 3.12 Allelic frequencies of DYS448 for Turkey

	Allele	Freq.	Std. Dev.
DYS448	17	0.02	0.0137
	18	0.04	0.0191
	19	0.27	0.0435
	20	0.41	0.0485
	21	0.21	0.0399
	22	0.05	0.0213
	23	0.01	0.0097

3.1.1.13.DYS456

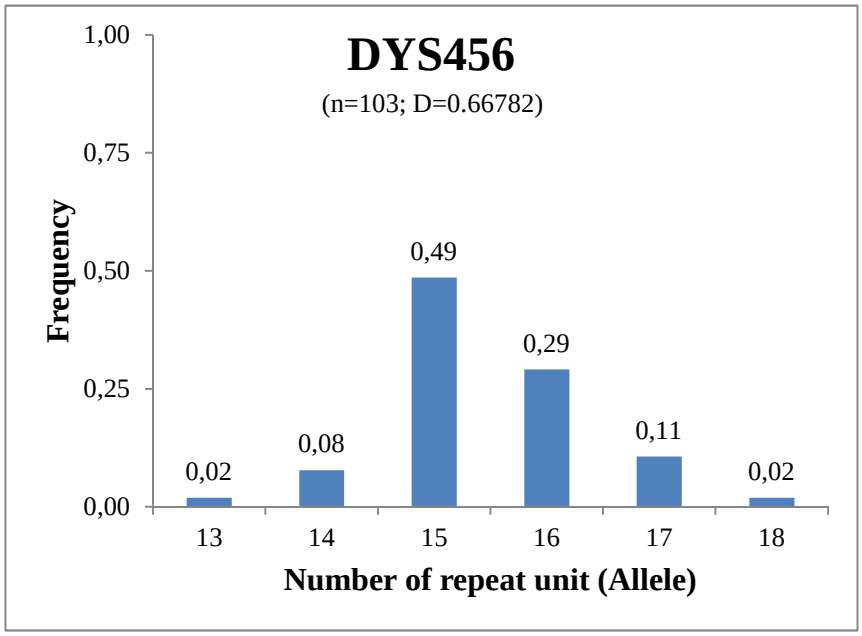


Figure 3.14 Allelic frequencies of DYS456 for Turkey

Table 3.13 Allelic frequencies of DYS456 for Turkey

	Allele	Freq.	Std. Dev.
DYS456	13	0.02	0.0137
	14	0.08	0.0265
	15	0.49	0.0495
	16	0.29	0.045
	17	0.11	0.0306
	18	0.02	0.0137

3.1.1.14.DYS458

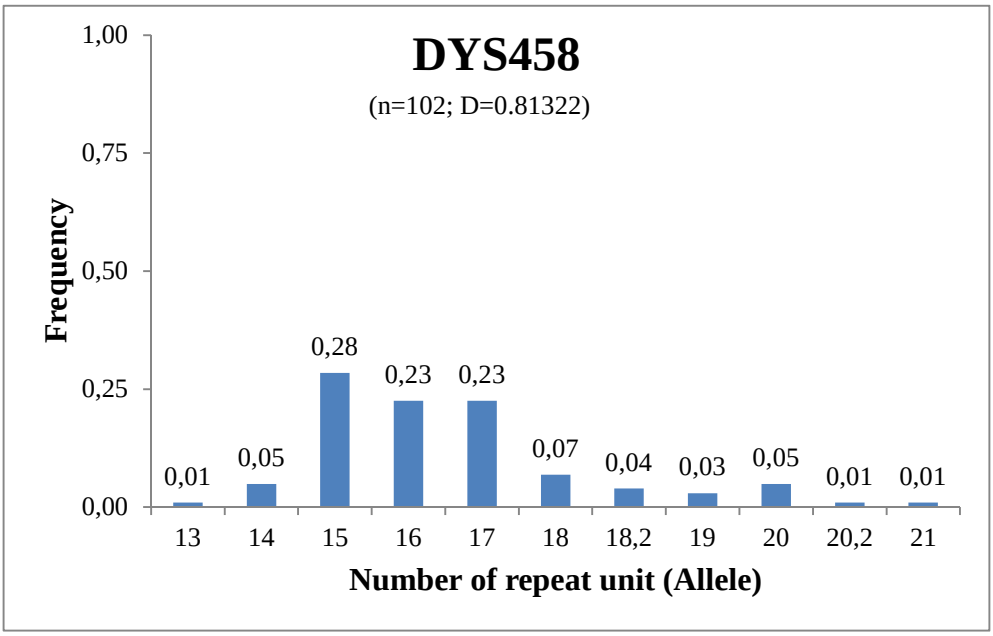


Figure 3.15 Allelic frequencies of DYS458 for Turkey

Table 3.14 Allelic frequencies of DYS458 for Turkey

	Allele	Freq.	Std. Dev.
DYS 458	13	0.01	0.0097
	14	0.05	0.0213
	15	0.28	0.0445
	16	0.23	0.0412
	17	0.23	0.0412
	18	0.07	0.0249
	18.2	0.04	0.0191
	19	0.03	0.0167
	20	0.05	0.0213
	20.2	0.01	0.0097
	21	0.01	0.0097

3.1.1.15.DYS635

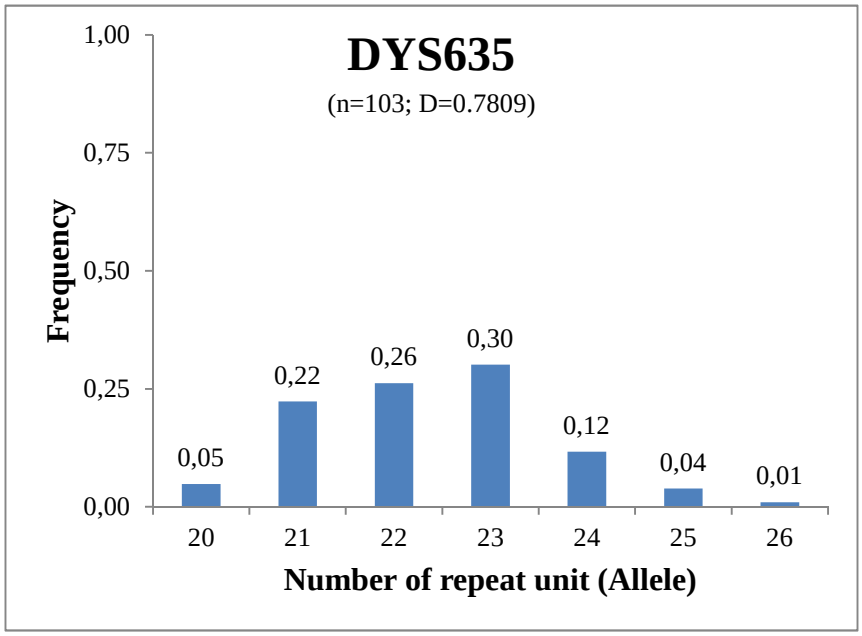


Figure 3.16 Allelic frequencies of DYS635 for Turkey

Table 3.15 Allelic frequencies of DYS635 for Turkey

	Allele	Freq.	Std. Dev.
DYS 635	20	0.05	0.0213
	21	0.22	0.0412
	22	0.26	0.0435
	23	0.30	0.0454
	24	0.12	0.0318
	25	0.04	0.0191
	26	0.01	0.0097

3.1.1.16. Y GATA H4

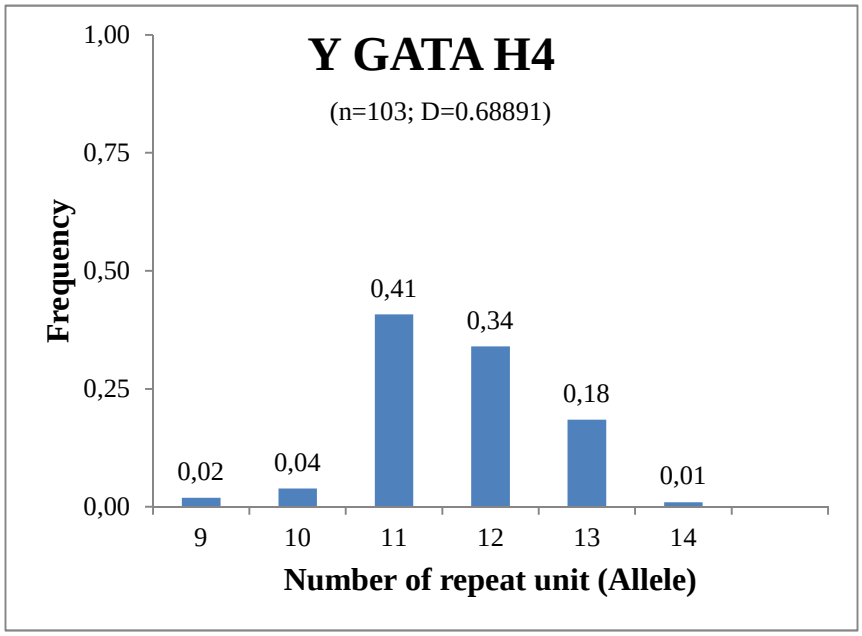


Figure 3.17 Allelic frequencies of Y GATA H4 for Turkey

Table 3.16 Allelic frequencies of Y GATA H4 for Turkey

	Allele	Freq.	Std. Dev.
Y GATA H4	9	0.02	0.0137
	10	0.04	0.0191
	11	0.41	0.0487
	12	0.34	0.0469
	13	0.18	0.0384
	14	0.01	0.0097

Table 3.17 Allele frequencies for 17 Y-STR Loci in Turkey

Allele	DYS 19	DYS 389 I	DYS 389 II	DYS 390	DYS 391	DYS 392	DYS 393	DYS 437	DYS 438	DYS 439	DYS 448	DYS 456	DYS 458	DYS 635	Y GATA H4	Allelic Class	DYS385 a/b
8									0.0097							**10-14**	0.0097
9					0.0971				0.2136	0.0097					0.0194	**11-12**	0.0194
10					0.5728	0.0485			0.3981	0.2039					0.0388	**11-13**	0.0097
11	0.0097				0.3107	0.7282			0.2233	0.4272					0.4078	**11-14**	0.1748
12		0.1845			0.0194	0.0291	0.4369		0.1553	0.2427					0.3398	**11-15**	0.0680
13	0.0874	0.6893				0.1068	0.4466			0.1165		0.0194	0.0098		0.1845	**12-12**	0.0291
14	0.4466	0.1165				0.0777	0.1068	0.5534				0.0777	0.0490		0.0097	**12-13**	0.0485
14-15	0.0097															**12-14**	0.0194
15	0.2524					0.0097	0.0097	0.3010				0.4854	0.2844			**12-15**	0.0680
16	0.1650	0.0097						0.1456				0.2913	0.2255			**12-16**	0.0194
17	0.0291										0.0198	0.1068	0.2255			**12-17**	0.0194
18											0.0396	0.0194	0.0687			**12-19**	0.0097
18.2													0.0392			**13-14**	0.0388
19											0.2673		0.0294			**13-15**	0.0680
20											0.4060		0.0490	0.0485		**13-16**	0.0485
20.2													0.0098			**13-17**	0.0388
21				0.0291							0.2079		0.0098	0.2233		**13-18**	0.0097
22				0.1359							0.0495			0.2621		**13-20**	0.0194
23				0.3010							0.0099			0.3010		**14-14**	0.0388
24				0.3592										0.1165		**14-15**	0.0680
25				0.1262										0.0388		**14-16**	0.0097
26				0.0485										0.0097		**14-17**	0.0194
27			0.0291													**14-18**	0.0194
28			0.1165													**15-15**	0.0097
29			0.3301													**15-16**	0.0194
30			0.3592													**16-16**	0.0097
31			0.1068													**16-18**	0.0097
32			0.0485													**16-19**	0.0194
33			0.0097													**17-17**	0.0097
																17-18	0.0388
																18-18	0.0097
GD:	0.7078	0.4818	0.7409	0.7501	0.5711	0.4534	0.6040	0.5877	0.7289	0.7103	0.7232	0.6678	0.8132	0.7809	0.6889		0.9442

3.1.2 REGIONAL

Distribution of allele frequencies in geographical regions of Turkey is given in Table 3.18.

Table 3.18 Allele frequencies of 17 Y-STR Loci in geographical regions of Turkey

Locus	Allele	Mediterranean	Eastern	Aegean	Southeastern	Central Anatolia	Black Sea	Marmara
DYS19	11	0.0769						
	13	0.0769		0.1538	0.2222	0.0526	0.1600	0.1304
	14	0.3846	0.5714	0.3077	0.2222	0.4737	0.5600	0.4348
	15	0.3077	0.2143	0.2308	0.4444	0.2632	0.2000	0.1304
	16	0.1538	0.1429	0.3077	0.1111	0.2105	0.0800	0.1739
	17							0.1304
	14-15		0.0714					
DYS385a/b	10-14		0.0714					
	11-12	0.0769			0.1111			
	11-13	0.0769					0.0400	
	11-14	0.0769	0.1429	0.2308		0.2105	0.1600	0.2174
	11-15		0.1429	0.0769		0.0526		0.1304
	12-12	0.0769				0.0526		0.0435
	12-13				0.1111	0.0526	0.0800	0.0435
	12-14					0.0526		0.0435
	12-15	0.1538	0.0714			0.0526	0.1200	
	12-16		0.0714			0.0526	0.0400	
	12-17	0.0769			0.1111			
	12-19				0.1111			
	13-14		0.0714			0.0526	0.0800	0.0435
	13-15	0.1538		0.0769		0.1053	0.0800	0.0435
	13-16		0.2143			0.1053	0.0400	
	13-17	0.0769	0.0714			0.0526	0.0400	0.0435
	13-18	0.0769						
	13-19		0.0714					
	13-20			0.1538				
	14-14			0.0769	0.1111	0.0526		0.0435
	14-15		0.0714	0.1538	0.1111		0.0400	0.0870
	14-16							0.0435
	14-17				0.1111			0.0435
	14-18			0.0769			0.0400	0.0435

Locus	Allele	Mediterranean	Eastern	Aegean	Southeastern	Central Anatolia	Black Sea	Marmara
	15-15					0.0526		
	15-16			0.0769			0.0400	
	15-17						0.0400	
	16-16				0.1111		0.0800	
	16-18						0.0400	
	16-19				0.1111			0.0435
	17-17					0.0526	0.0400	
	17-18	0.0769		0.0769				0.0870
	18-18	0.0769						
DYS389 I	12	0.1538	0.2143	0.1538	0.2222	0.0526	0.1600	0.2609
	13	0.6154	0.7143	0.6923	0.6667	0.7368	0.6800	0.6522
	14	0.2308	0.0714	0.1538	0.1111	0.1579	0.1600	0.0870
	16					0.0526		
DYS389 II	27		0.1429					0.0435
	28	0.1538		0.0769		0.1053	0.0800	0.2174
	29	0.2308	0.5000	0.2308	0.4444	0.1579	0.4400	0.3043
	30	0.4615	0.2143	0.4615	0.3333	0.4737	0.1600	0.3478
	31	0.1538	0.0714	0.0769	0.2222	0.2105	0.1600	0.0435
	32		0.0714	0.0769		0.0526	0.1200	0.0435
	33			0.0769			0.0400	
DYS390	21			0.0769		0.1053		
	22		0.2143	0.1538	0.2222	0.1579	0.1200	0.1739
	23	0.5385	0.3571	0.2308	0.1111	0.2105	0.4400	0.2174
	24	0.3077	0.3571	0.3077	0.4444	0.3684	0.3600	0.3478
	25	0.1538		0.2308	0.2222	0.1579	0.0400	0.1304
	26		0.0714				0.0400	0.1304
DYS391	9	0.1538	0.0714	0.1538		0.1579	0.0800	0.0870
	10	0.6923	0.5714	0.3846	0.6667	0.4737	0.6400	0.6522
	11	0.1538	0.3571	0.3846	0.3333	0.3684	0.2800	0.2174
	12			0.0769				0.0435
DYS392	10			0.0769	0.1111	0.0526	0.0400	0.0435
	11	0.7692	0.7143	0.8462	0.7778	0.7368	0.6800	0.6957
	12	0.0769	0.0714					0.0435
	13	0.0769	0.0714		0.1111	0.1053	0.1200	0.1304
	14		0.0714	0.0769		0.1053	0.1200	0.0870
	15	0.0769	0.0714				0.0400	
DYS393	10.3		0.0714					
	12	0.5385	0.6429	0.3077	0.3333	0.4211	0.5200	0.3043
	13	0.3846	0.2143	0.5385	0.5556	0.4211	0.4000	0.5652

Locus	Allele	Mediterranean	Eastern	Aegean	Southeastern	Central Anatolia	Black Sea	Marmara
	14	0.0769	0.0714	0.0769	0.1111	0.1579	0.0800	0.1304
	15			0.0769				
DYS437	14	0.8462	0.3571	0.6154	0.7778	0.4211	0.5600	0.5652
	15	0.1538	0.4286	0.3077	0.1111	0.4211	0.3600	0.2174
	16		0.2143	0.0769	0.1111	0.1579	0.0800	0.2174
DYS438	7						0.0400	
	8						0.0400	0.0435
	9	0.3077	0.2857	0.0769	0.2222	0.2105	0.3200	0.0870
	10	0.2308	0.3571	0.4615	0.5556	0.4737	0.3200	0.4783
	11	0.3077	0.1429	0.3846	0.2222	0.1579	0.1600	0.2174
	12	0.1538	0.2143	0.0769		0.1579	0.1200	0.1739
DYS439	9							0.0435
	10	0.1538	0.1429	0.3846	0.1111	0.2105	0.1600	0.2174
	11	0.4615	0.5000	0.4615	0.5556	0.4211	0.3200	0.4348
	12	0.3077	0.2143	0.1538	0.2222	0.2105	0.2400	0.2609
	13	0.0769	0.1429		0.1111	0.1579	0.2800	0.0435
DYS448	17				0.1111			0.0435
	18	0.0833					0.0833	0.0435
	19	0.1667	0.4286	0.2308	0.1111	0.3158	0.3750	0.2174
	20	0.3333	0.4286	0.5385	0.4444	0.2105	0.2917	0.5652
	21	0.4167	0.1429	0.0769	0.3333	0.3684	0.2083	0.0870
	22			0.0769		0.1053	0.0417	0.0435
	23			0.0769				
DYS456	12						0.0400	
	13		0.1429					
	14	0.0769				0.0526	0.1600	0.1304
	15	0.5385	0.4286	0.4615	0.5556	0.6316	0.4400	0.4783
	16	0.3077	0.4286	0.3846	0.3333	0.2632	0.2000	0.2174
	17			0.1538	0.1111	0.0526	0.1600	0.1304
	18	0.0769						0.0435
DYS458	13	0.0833						
	14	0.1667			0.2222	0.0526	0.0800	
	15	0.1667	0.1429	0.3077		0.3684	0.4000	0.2609
	16	0.1667	0.1429	0.0769	0.3333	0.1579	0.1600	0.4348
	17	0.1667	0.4286	0.3077	0.1111	0.2105	0.2400	0.2174
	17.2		0.0714					
	18		0.1429	0.1538	0.2222	0.0526	0.0400	

Locus	Allele	Mediterranean	Eastern	Aegean	Southeastern	Central Anatolia	Black Sea	Marmara
	18.2	0.0833		0.0769	0.1111			0.0435
	19	0.0833		0.0769		0.0526		
	20					0.1053	0.0800	0.0435
	20.2	0.0833						
	21		0.0714					
DYS635	20		0.1429	0.1538	0.1111	0.0526		0.0435
	21	0.3077	0.2143	0.3077		0.2632	0.1600	0.2174
	22	0.3077	0.2857	0.1538	0.4444	0.1579	0.4000	0.2174
	23	0.3077	0.2143	0.3077	0.1111	0.4211	0.3200	0.3043
	24		0.1429	0.0769	0.1111	0.0526	0.0800	0.2174
	25	0.0769			0.1111	0.0526	0.0400	
	26				0.1111			
Y GATA H4	9	0.0769					0.0400	
	10		0.2143			0.1053	0.0800	
	11	0.2308	0.4286	0.6154	0.3333	0.3684	0.4000	0.4783
	12	0.5385	0.0714	0.1538	0.5556	0.3158	0.3200	0.3913
	13	0.1538	0.2857	0.2308	0.1111	0.1579	0.1600	0.1304
	14					0.0526		

The allelic distribution of 17 Y-STR loci are also shown graphically.

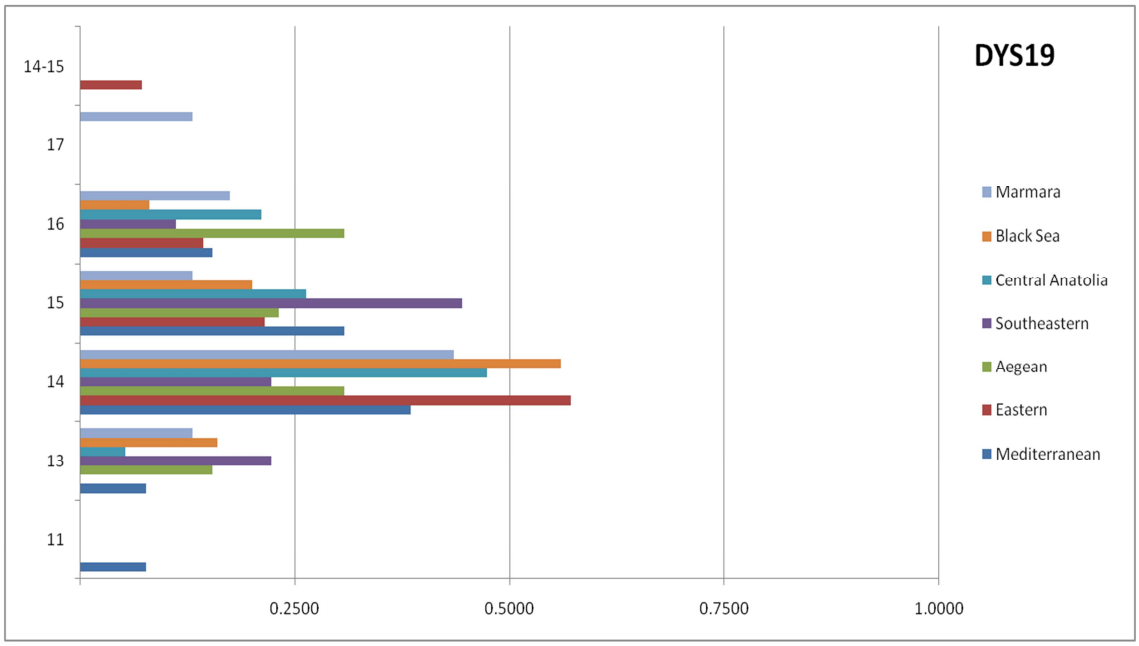


Figure 3.18 Allele frequency distribution of DYS19 in regions of Turkey

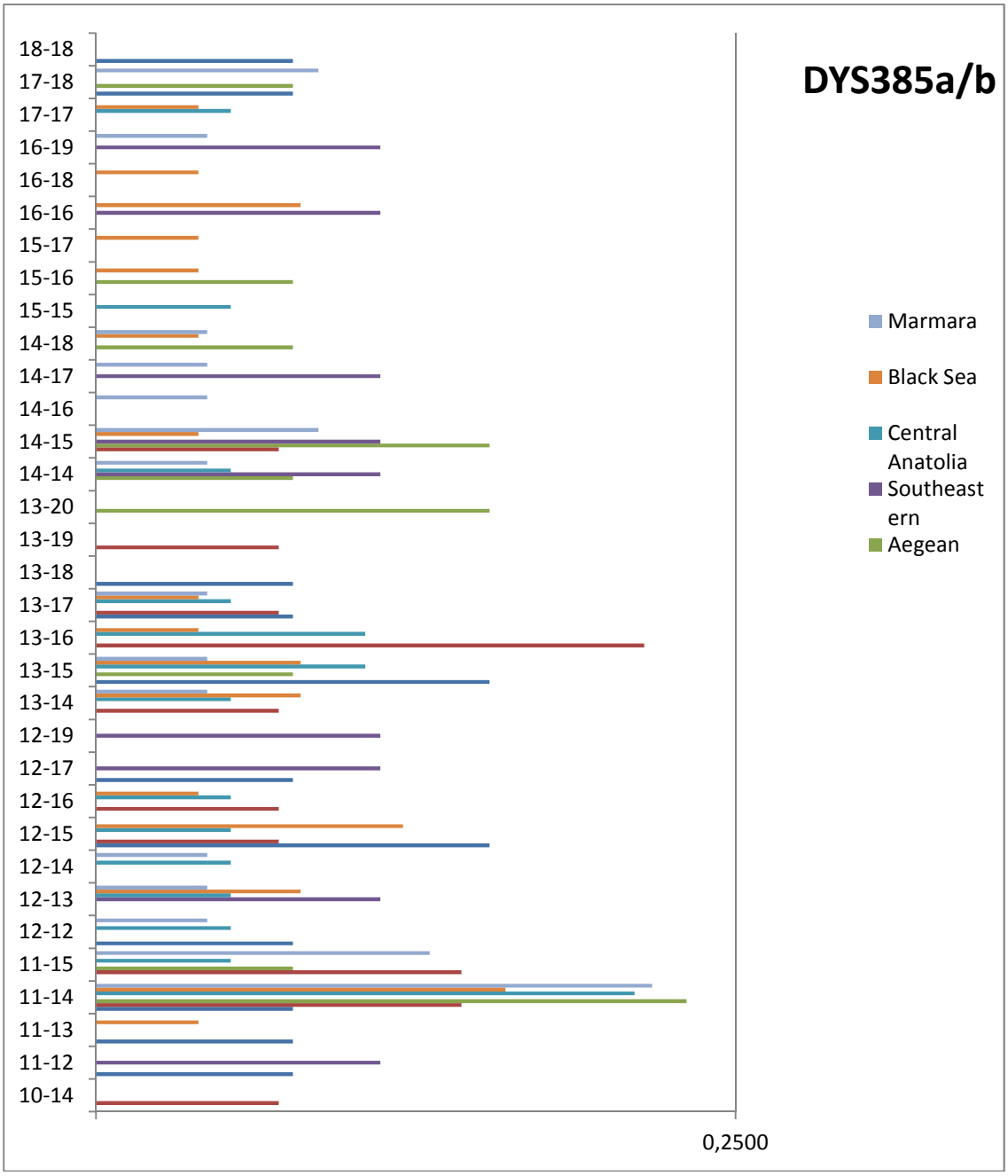


Figure 3.19 Allele frequency distribution of DYS385a/b in regions of Turkey

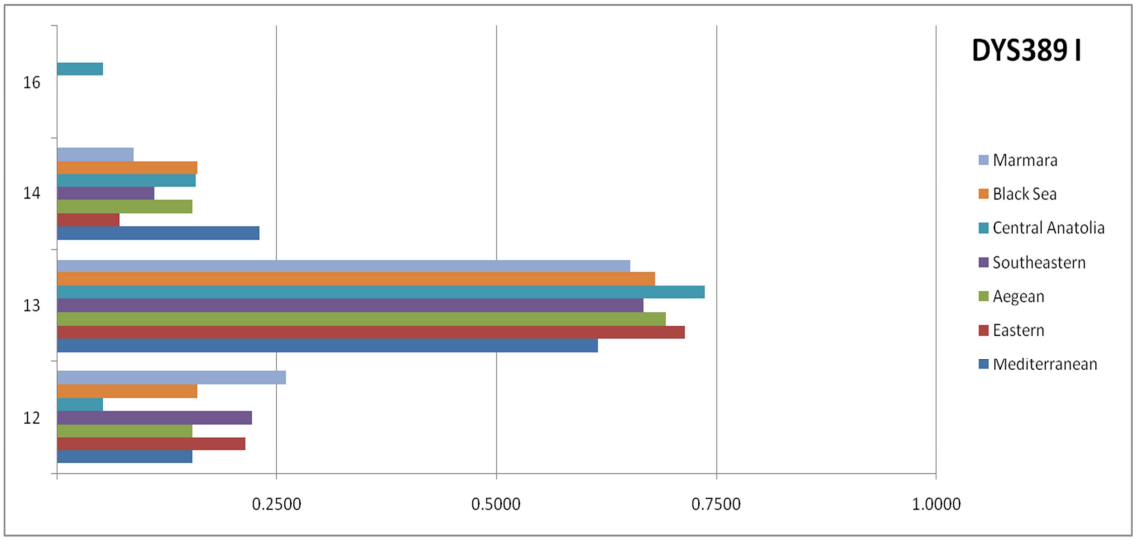


Figure 3.20 Allele frequency distribution of DYS389 I in regions of Turkey

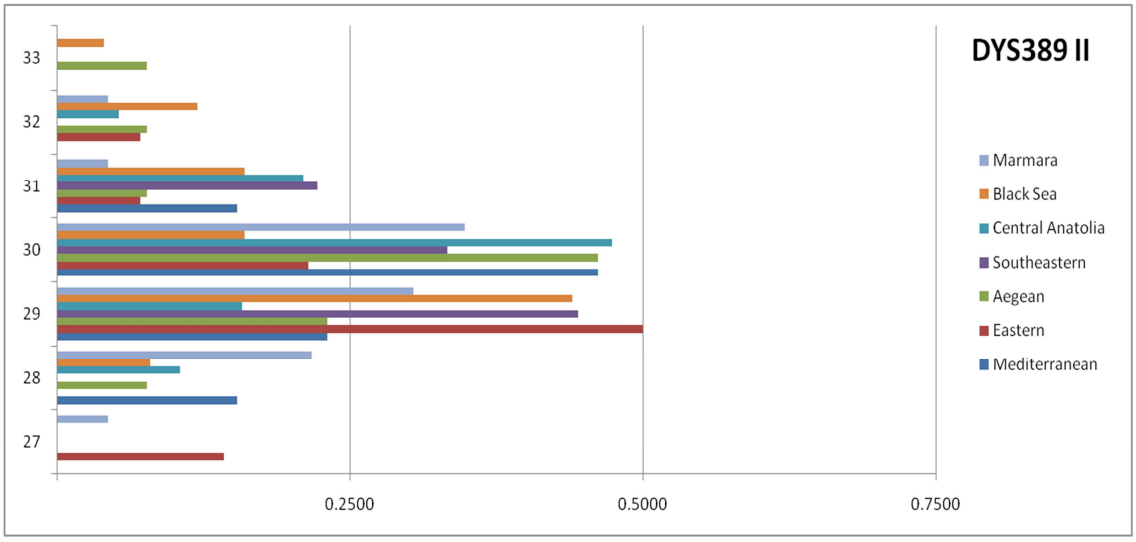


Figure 3.21 Allele frequency distribution of DYS389 II in regions of Turkey

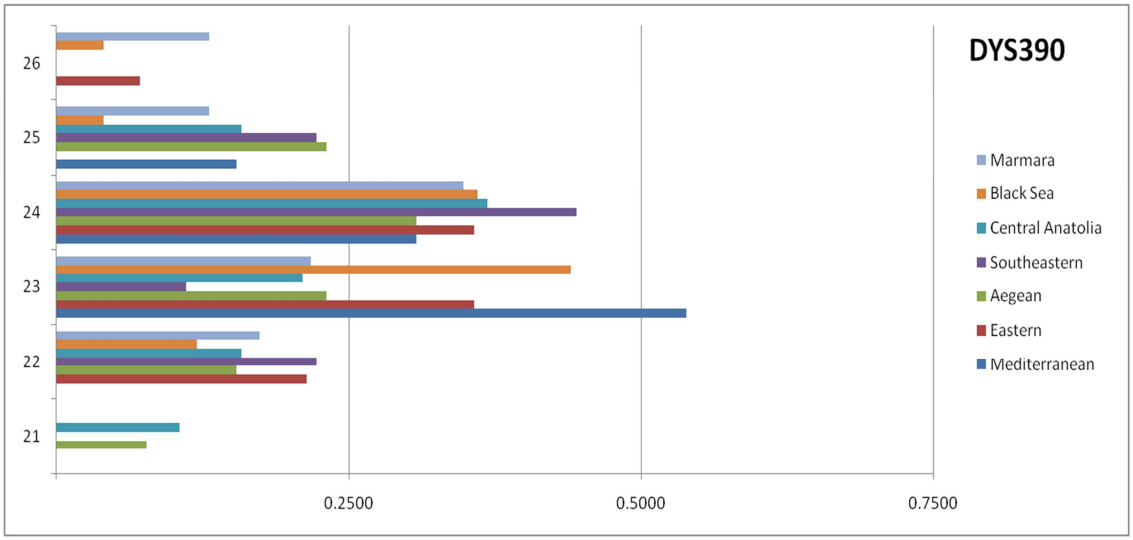


Figure 3.22 Allele frequency distribution of DYS390 in regions of Turkey

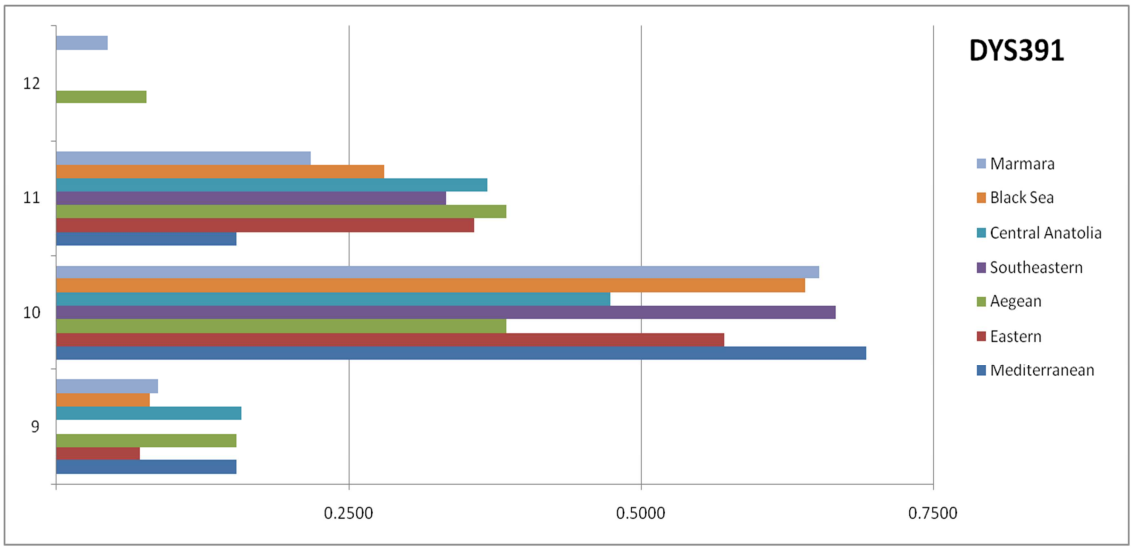


Figure 3.23 Allele frequency distribution of DYS391 in regions of Turkey

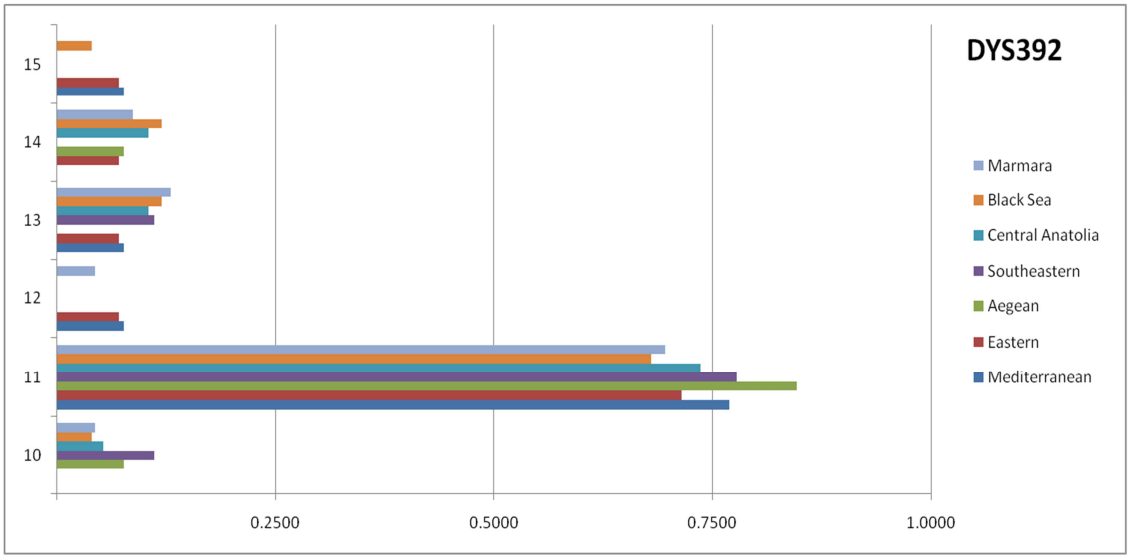


Figure 3.24 Allele frequency distribution of DYS392 in regions of Turkey

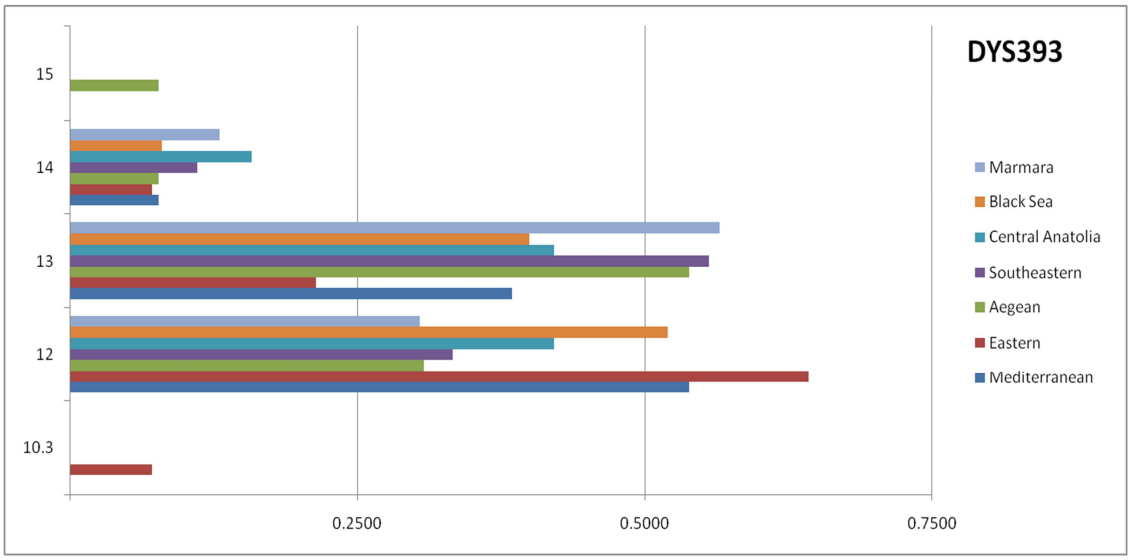


Figure 3.25 Allele frequency distribution of DYS393 in regions of Turkey

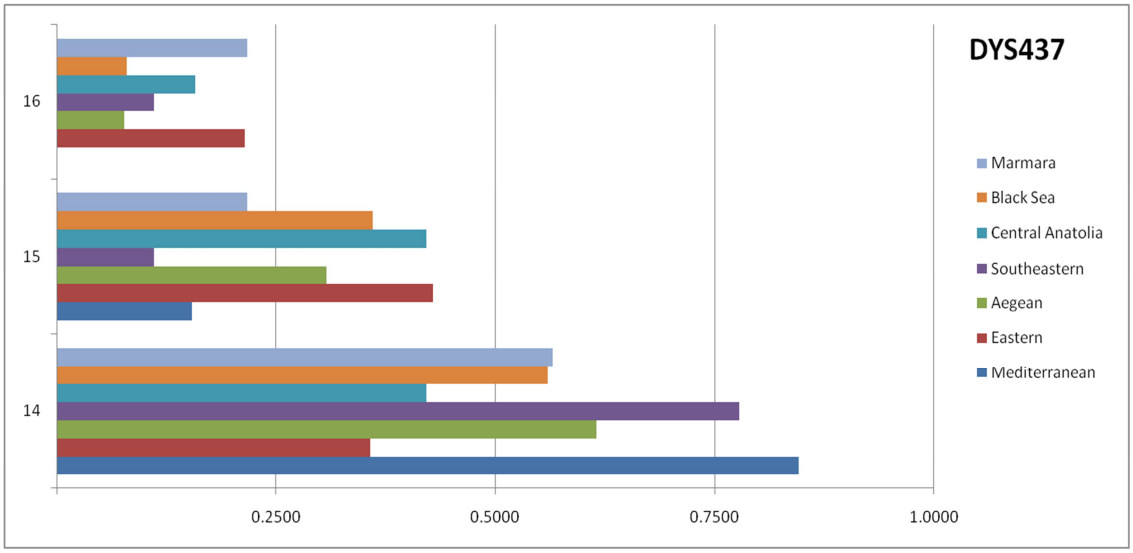


Figure 3.26 Allele frequency distribution of DYS437 in regions of Turkey

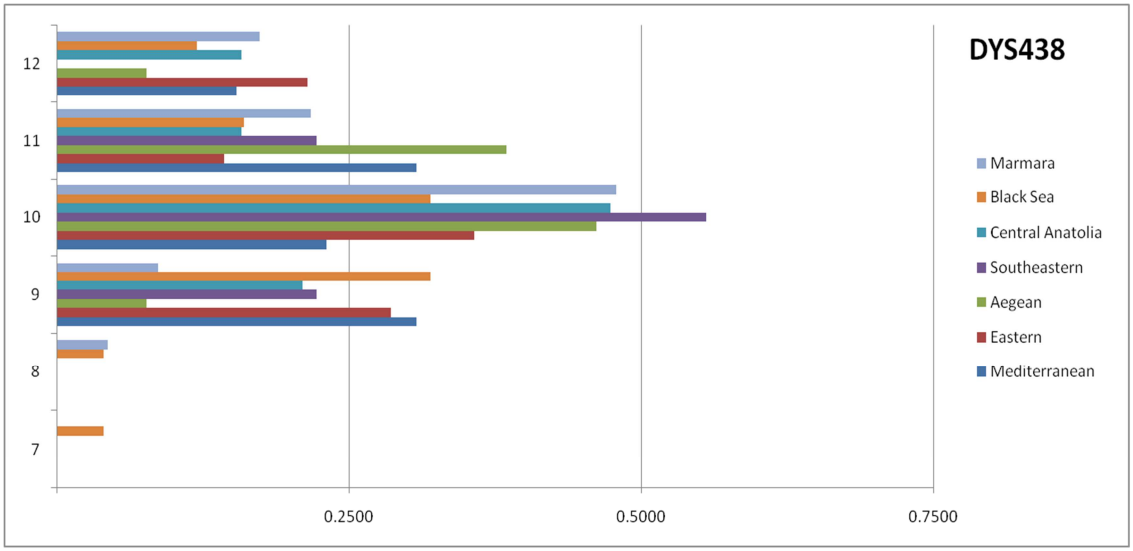


Figure 3.27 Allele frequency distribution of DYS438 in regions of Turkey

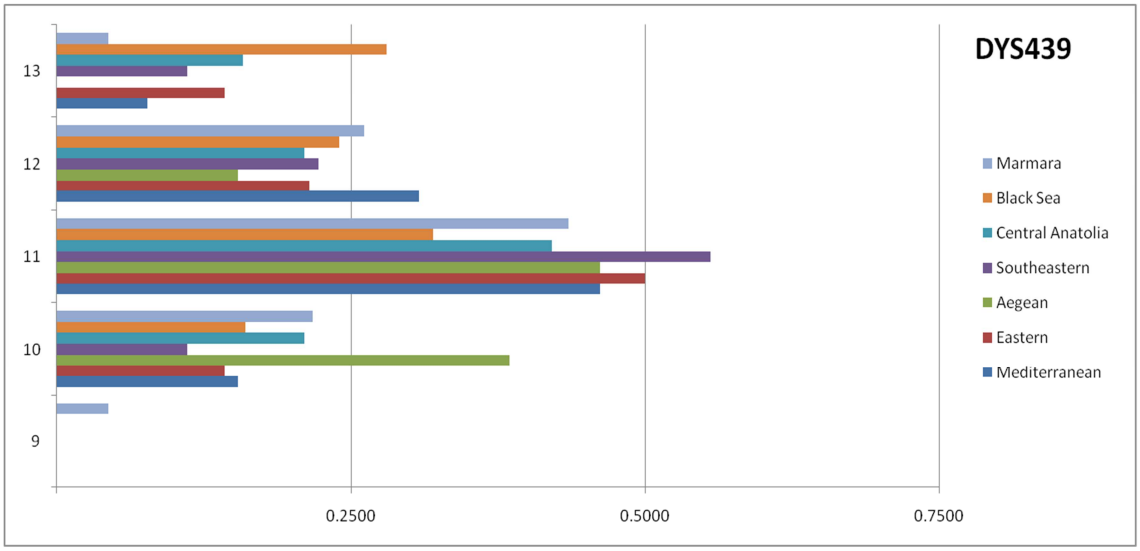


Figure 3.28 Allele frequency distribution of DYS439 in regions of Turkey

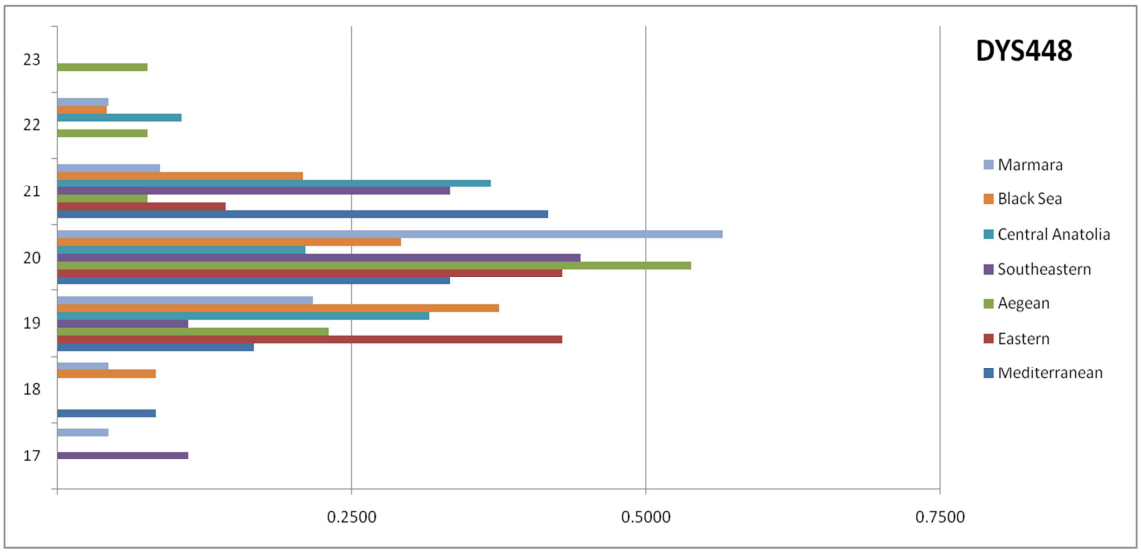


Figure 3.29 Allele frequency distribution of DYS448 in regions of Turkey

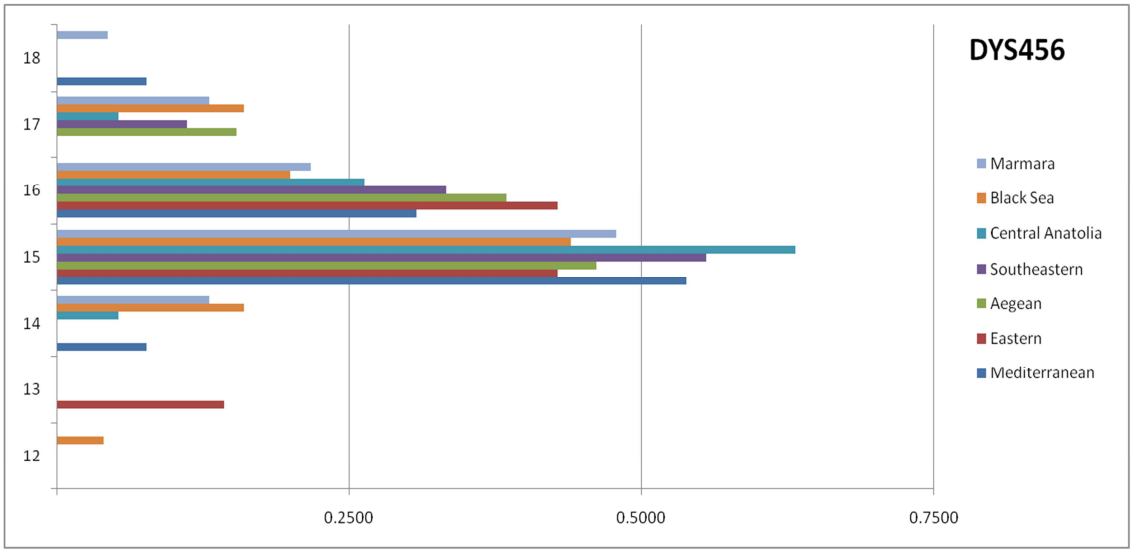


Figure 3.30 Allele frequency distribution of DYS456 in regions of Turkey

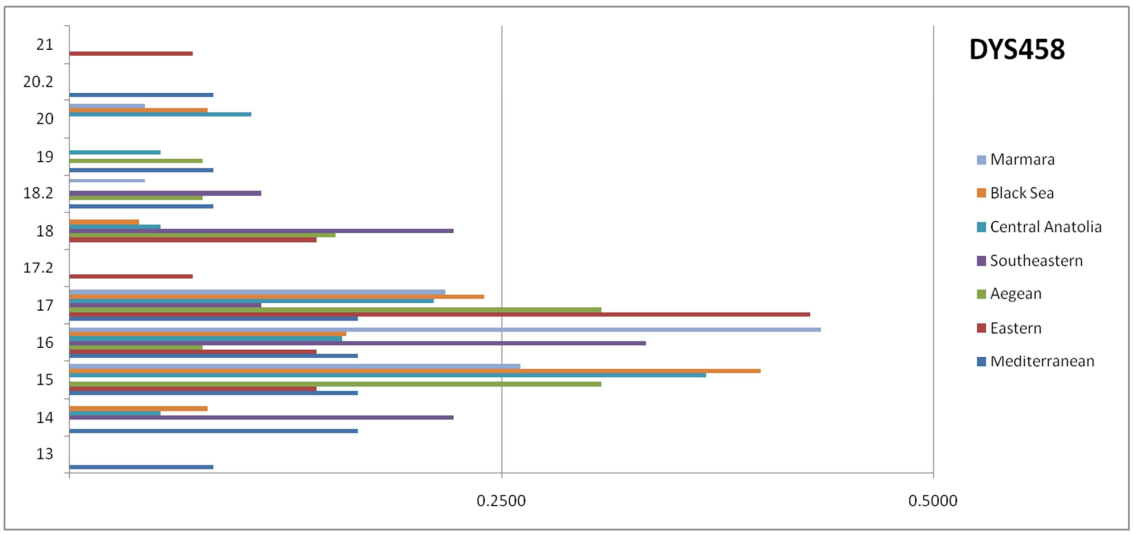


Figure 3.31 Allele frequency distribution of DYS458 in regions of Turkey

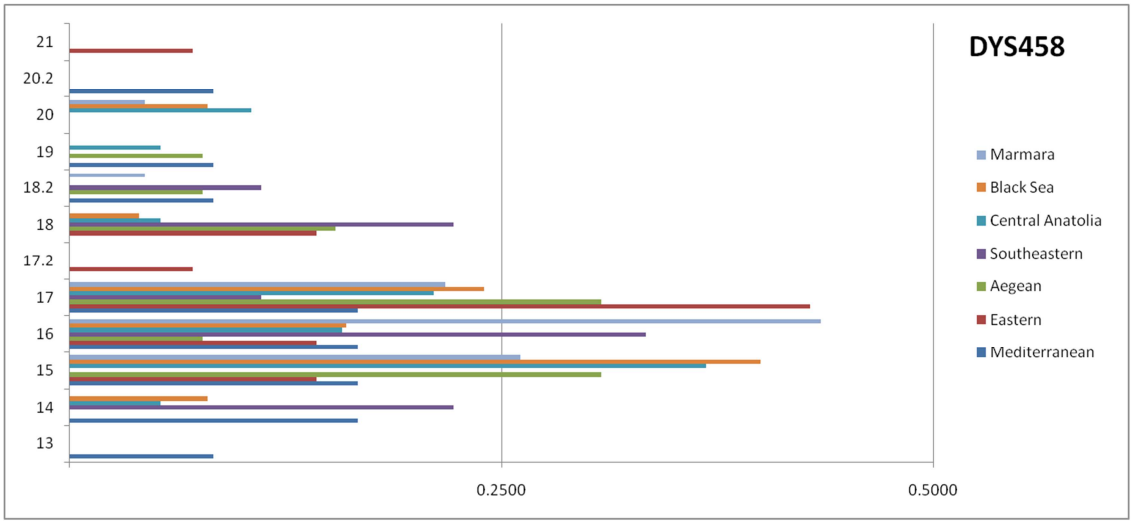


Figure 3.32 Allele frequency distribution of DYS458 in regions of Turkey

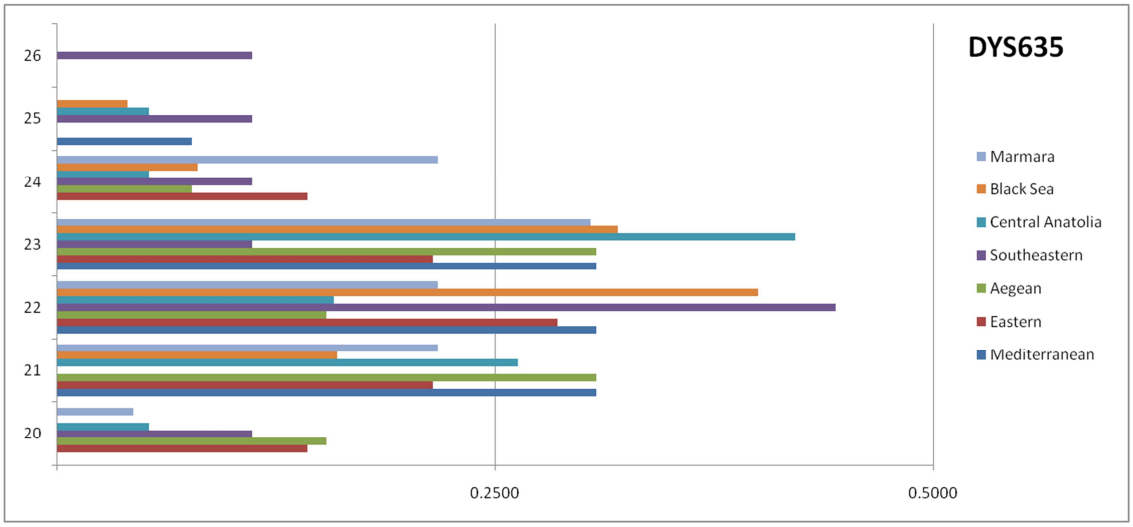


Figure 3.33 Allele frequency distribution of DYS635 in regions of Turkey

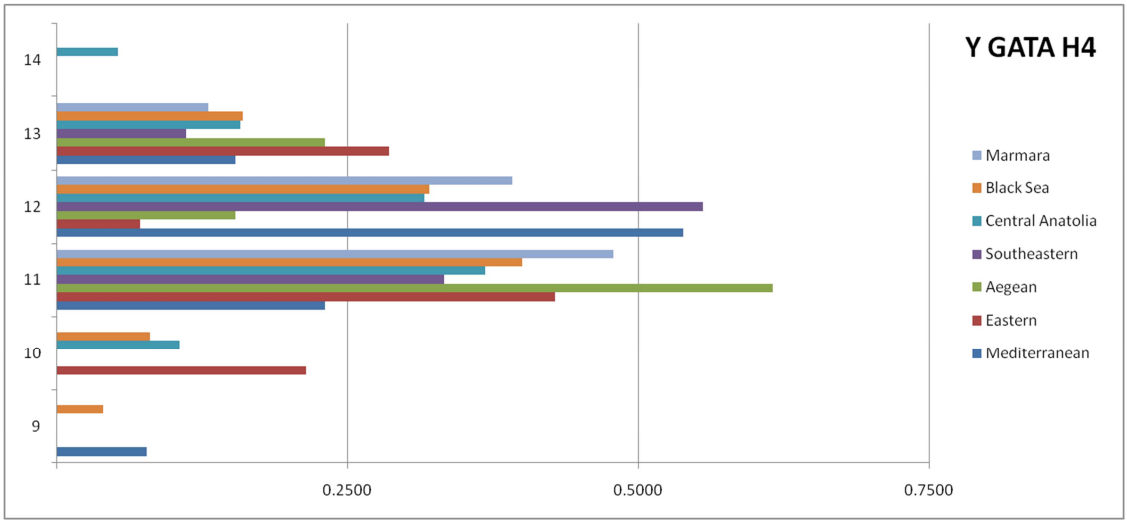


Figure 3.34 Allele frequency distribution of Y GATA H4 in regions of Turkey

3.2 HAPLOTYPE ANALYSIS

Haplotype frequencies for minimal, SWGDAM and Y-Filer extended loci are given in the following tables.

In Table 3.20 same analysis were done without the locus DYS385a/b in order to reveal more similarities between samples for Turkey.

Haplotype sharing among samples are summarized in Table 3.22.

Table 3.19 The 17-loci Y-STR haplotypes in samples from Turkey

	Minimal Haplotype										SWGDAM (extended)				Y-Filer (extended)							
Haplotype	**n**	**Freq**	**DYS 19**	**DYS 385**	**DYS389 I**	**DYS389 II**	**DYS390**	**DYS 391**	**DYS392**	**DYS 393**	**n**	**Freq**	**DYS438**	**DYS 439**	**n**	**Freq**	**DYS437**	**DYS448**	**DYS456**	**DYS 458**	**DYS635**	**Y GATA H4**
TR1	**1**	0.00971	15	12-13	13	29	24	10	13	13	1	0.00971	11	11	1	0.00971	14	17	15	14	26	13
TR2	**1**	0.00971	14	10-14	13	29	24	11	12	12	1	0.00971	12	12	1	0.00971	16	19	16	17	24	13
TR3	**1**	0.00971	16	12-12	14	31	24	9	11	13	1	0.00971	10	11	1	0.00971	14	20	15	19	23	10
TR4	**1**	0.00971	16	11-15	13	30	26	10	11	13	1	0.00971	11	10	1	0.00971	14	20	16	15	23	12
TR5	**1**	0.00971	13	16-16	13	31	24	10	11	13	1	0.00971	10	12	1	0.00971	14	19	16	17	22	12
TR6	**1**	0.00971	15	14-14	13	31	22	10	11	14	1	0.00971	10	11	1	0.00971	15	21	15	18	25	11
TR7	**1**	0.00971	15	14-18	13	29	23	10	11	12	1	0.00971	10	11	1	0.00971	14	19	16	18.2	20	12
TR8	**1**	0.00971	15	13-15	13	30	23	9	11	14	1	0.00971	9	11	1	0.00971	14	21	16	14	22	12
TR9	**1**	0.00971	15	13-16	12	27	24	10	11	12	1	0.00971	9	13	1	0.00971	15	19	13	17	21	11
TR10	**1**	0.00971	14	12-17	14	30	23	10	11	12	1	0.00971	10	11	1	0.00971	14	21	16	20.2	21	12
TR11	**1**	0.00971	14	13-15	13	30	24	10	11	12	1	0.00971	10	11	1	0.00971	15	19	15	20	21	11
TR12	**1**	0.00971	14	17-18	13	30	24	10	11	13	1	0.00971	10	12	1	0.00971	14	20	15	15	21	12
TR13	**1**	0.00971	13	16-19	13	30	24	11	11	13	1	0.00971	10	11	1	0.00971	15	20	17	16	24	12
TR14	**1**	0.00971	16	13-17	13	30	23	10	11	12	1	0.00971	9	11	1	0.00971	15	21	16	15	22	12
TR15	**1**	0.00971	15	12-15	13	30	24	10	11	13	1	0.00971	11	10	1	0.00971	14	21	16	15	23	13
TR16	**1**	0.00971	14	13-16	13	29	24	10	11	12	1	0.00971	9	12	1	0.00971	15	19	16	21	21	11
TR17	**1**	0.00971	15	14-17	12	29	22	10	10	13	1	0.00971	10	12	1	0.00971	16	20	15	18	20	11
TR18	**1**	0.00971	15	12-14	12	29	22	10	11	13	1	0.00971	10	11	1	0.00971	16	22	15	17	22	11
TR19	**1**	0.00971	14	17-17	13	30	25	10	11	14	1	0.00971	10	11	1	0.00971	14	20	15	15	22	10
TR20	**1**	0.00971	15	13-17	13	29	23	9	11	12	1	0.00971	9	11	1	0.00971	14	21	15	14	23	12
TR21	**1**	0.00971	14	13-15	12	29	22	10	11	13	1	0.00971	10	11	1	0.00971	16	20	15	15	21	11

	Minimal Haplotype										SWGDAM (extended)				Y-Filer (extended)							
Haplotype	**n**	**Freq**	**DYS 19**	**DYS 385**	**DYS389 I**	**DYS389 II**	**DYS390**	**DYS 391**	**DYS392**	**DYS 393**	**n**	**Freq**	**DYS438**	**DYS 439**	**n**	**Freq**	**DYS437**	**DYS448**	**DYS456**	**DYS 458**	**DYS635**	**Y GATA H4**
TR22	**1**	0.00971	13	17-18	14	31	24	10	11	13	1	0.00971	10	12	1	0.00971	14	20	17	16	22	12
TR23	**1**	0.00971	15	13-17	13	29	23	10	11	12	1	0.00971	9	9	1	0.00971	15	21	15	15	21	11
TR24	**1**	0.00971	16	11-15	13	30	23	11	11	13	1	0.00971	11	10	1	0.00971	14	20	16	15	24	11
TR25	**1**	0.00971	11	18-18	12	30	23	10	12	12	1	0.00971	12	11	1	0.00971	14	21	15	13	22	12
TR26	**1**	0.00971	14	11-14	13	29	24	11	13	12	1	0.00971	12	12	1	0.00971	15	19	15	16	23	13
TR27	**1**	0.00971	14	12-19	12	29	24	11	11	12	1	0.00971	10	11	1	0.00971	14	20	15	18.2	24	11
TR28	**2**	0.01942	14	12-13	13	29	23	10	14	13	1	0.00971	10	10	1	0.00971	14	18	15	15	25	12
TR72											1	0.00971	10	11	1	0.00971	14	18	15	16	24	12
TR29	**1**	0.00971	14	11-14	13	29	23	11	13	12	1	0.00971	12	13	1	0.00971	15	19	15	16	23	12
TR30	**1**	0.00971	14	12-13	13	30	23	10	14	13	1	0.00971	10	11	1	0.00971	14	18	15	15	24	11
TR31	**1**	0.00971	14	12-16	13	30	24	10	11	12	1	0.00971	10	11	1	0.00971	15	19	15	20	21	11
TR32	**1**	0.00971	15	14-15	13	28	24	10	11	12	1	0.00971	9	11	1	0.00971	14	20	16	17	23	11
TR33	**1**	0.00971	14	13-15	13	30	21	10	11	14	1	0.00971	10	11	1	0.00971	16	22	16	18	21	11
TR34	**1**	0.00971	14-15	12-15	13	30	22	9	11	14	1	0.00971	10	11	1	0.00971	16	20	13	18	21	11
TR35	**1**	0.00971	14	12-15	13	29	24	11	13	12	1	0.00971	12	13	1	0.00971	15	20	17	15	23	13
TR36	**1**	0.00971	14	13-15	13	29	23	10	11	12	1	0.00971	10	12	1	0.00971	14	20	15	18.2	21	11
TR37	**1**	0.00971	16	11-14	13	33	25	12	11	13	1	0.00971	11	10	1	0.00971	15	20	15	15	23	13
TR38	**1**	0.00971	15	13-16	13	28	22	9	10	12	1	0.00971	9	12	1	0.00971	14	21	15	14	21	12
TR39	**1**	0.00971	14	14-14	12	28	23	10	11	13	1	0.00971	10	11	1	0.00971	16	20	14	16	22	11
TR40	**1**	0.00971	15	11-14	13	31	24	11	11	13	1	0.00971	11	10	1	0.00971	14	19	17	15	23	14
TR41	**1**	0.00971	14	12-17	13	29	23	10	11	12	1	0.00971	9	11	1	0.00971	14	21	15	14	22	12
TR42	**1**	0.00971	13	11-14	14	31	25	9	13	12	1	0.00971	12	12	1	0.00971	15	21	14	16	23	12
TR43	**1**	0.00971	14	14-15	13	29	23	11	11	12	1	0.00971	9	11	1	0.00971	14	20	16	17	22	10

	Minimal Haplotype										SWGDAM (extended)				Y-Filer (extended)							
Haplotype	**n**	**Freq**	**DYS 19**	**DYS 385**	**DYS389 I**	**DYS389 II**	**DYS390**	**DYS 391**	**DYS392**	**DYS 393**	**n**	**Freq**	**DYS438**	**DYS 439**	**n**	**Freq**	**DYS437**	**DYS448**	**DYS456**	**DYS 458**	**DYS635**	**Y GATA H4**
TR44	**1**	0.00971	14	11-14	13	30	25	11	11	13	1	0.00971	11	10	1	0.00971	14	20	15	15	23	13
TR45	**1**	0.00971	13	17-18	13	30	26	9	11	14	1	0.00971	10	11	1	0.00971	14	20	15	17	22	11
TR46	**1**	0.00971	14	11-15	12	27	24	10	14	12	1	0.00971	11	12	1	0.00971	15	19	15	17	23	13
TR47	**1**	0.00971	16	15-15	13	31	24	11	11	13	1	0.00971	10	12	1	0.00971	15	20	15	17	23	11
TR48	**1**	0.00971	14	11-15	13	28	24	11	14	12	1	0.00971	12	11	1	0.00971	14	19	16	18	23	11
TR49	**1**	0.00971	16	12-12	13	29	26	10	11	13	1	0.00971	10	10	1	0.00971	14	20	15	20	21	11
TR50	**1**	0.00971	16	11-15	13	29	24	10	11	13	1	0.00971	11	11	1	0.00971	14	20	17	16	24	12
TR51	**1**	0.00971	14	12-14	12	29	23	9	11	12	1	0.00971	10	11	1	0.00971	14	19	15	16	22	13
TR52	**1**	0.00971	15	16-18	13	29	24	10	11	12	1	0.00971	9	13	1	0.00971	14	19	14	15	22	12
TR53	**1**	0.00971	16	11-12	13	31	25	11	11	13	1	0.00971	11	10	1	0.00971	14	19	15	16	23	13
TR54	**1**	0.00971	14	13-17	12	28	23	10	11	12	1	0.00971	9	12	1	0.00971	14	21	14	20	21	11
TR55	**1**	0.00971	14	12-13	14	30	23	10	14	13	1	0.00971	10	10	1	0.00971	14	19	15	16	24	12
TR56	**1**	0.00971	14	13-15	13	30	22	10	11	12	1	0.00971	9	13	1	0.00971	15	21	15	15	23	11
TR57	**1**	0.00971	14	12-16	13	32	23	11	11	12	1	0.00971	9	11	1	0.00971	16	22	17	15	22	11
TR58	**2**	0.01942	16	12-15	13	29	24	10	11	13	1	0.00971	11	12	1	0.00971	14	?	17	17	22	11
TR75											1	0.00971	11	11	1	0.00971	14	?	15	17	21	11
TR59	**1**	0.00971	14	13-16	13	29	24	11	11	12	1	0.00971	10	12	1	0.00971	15	19	15	20	21	11
TR60	**1**	0.00971	14	12-15	13	29	23	10	11	12	1	0.00971	9	12	1	0.00971	15	20	16	17	24	9
TR61	**1**	0.00971	13	16-19	13	30	24	10	11	13	1	0.00971	10	11	1	0.00971	14	20	17	16	22	12
TR62	**1**	0.00971	15	14-15	14	30	25	11	11	12	1	0.00971	9	13	1	0.00971	14	21	16	16	22	12
TR63	**1**	0.00971	14	13-16	13	29	23	10	11	12	1	0.00971	9	11	1	0.00971	15	20	15	17	22	11
TR64	**1**	0.00971	14	14-18	13	29	22	11	11	12	1	0.00971	10	12	1	0.00971	14	20	14	18.2	21	11
TR65	**1**	0.00971	13	11-13	13	28	24	10	15	12	1	0.00971	12	12	1	0.00971	14	19	16	17	23	13

	Minimal Haplotype										SWGDAM (extended)				Y-Filer (extended)							
Haplotype	**n**	**Freq**	**DYS 19**	**DYS 385**	**DYS389 I**	**DYS389 II**	**DYS390**	**DYS 391**	**DYS392**	**DYS 393**	**n**	**Freq**	**DYS438**	**DYS 439**	**n**	**Freq**	**DYS437**	**DYS448**	**DYS456**	**DYS 458**	**DYS635**	**Y GATA H4**
TR66	**1**	0.00971	14	13-14	13	30	23	10	11	12	1	0.00971	9	11	1	0.00971	15	21	15	15	22	12
TR67															1	0.00971	14	20	16	15	25	13
TR89	**3**	0.02913	16	11-14	13	30	25	11	11	13	3	0.02913	11	10	1	0.00971	14	20	17	15	24	13
TR100															1	0.00971	14	20	16	15	23	11
TR68	**1**	0.00971	14	11-15	13	29	24	11	13	13	1	0.00971	12	13	1	0.00971	15	19	15	17	23	12
TR69	**1**	0.00971	14	13-14	16	32	23	10	11	12	1	0.00971	9	11	1	0.00971	15	21	15	17	21	11
TR70	**1**	0.00971	15	13-14	12	29	22	10	11	13	1	0.00971	10	11	1	0.00971	16	20	16	18	22	13
TR71	**1**	0.00971	15	11-14	13	30	25	11	11	13	1	0.00971	11	10	1	0.00971	14	20	16	15	23	12
TR73	**1**	0.00971	15	11-14	13	32	26	11	11	13	1	0.00971	11	10	1	0.00971	14	19	17	15	23	13
TR74	**1**	0.00971	14	11-14	13	31	24	11	13	12	1	0.00971	12	13	1	0.00971	15	19	16	15	23	10
TR76	**1**	0.00971	16	11-12	13	30	25	11	11	13	1	0.00971	11	10	1	0.00971	14	20	16	16	23	12
TR77	**1**	0.00971	15	14-14	13	30	21	11	11	14	1	0.00971	10	11	1	0.00971	16	21	15	16	20	12
TR78	**1**	0.00971	14	13-18	12	28	23	10	11	12	1	0.00971	9	11	1	0.00971	15	21	15	19	22	11
TR79	**1**	0.00971	15	15-16	12	29	22	10	10	14	1	0.00971	10	13	1	0.00971	14	21	15	16	22	12
TR80	**1**	0.00971	14	11-14	13	29	25	12	13	13	1	0.00971	12	12	1	0.00971	15	19	15	17	24	12
TR81	**2**	0.01942	14	11-14	13	30	24	11	13	12	1	0.00971	12	12	1	0.00971	14	19	15	16	23	13
TR85											1	0.00971	12	13	1	0.00971	14	19	15	16	23	12
TR82	**1**	0.00971	15	14-15	12	28	23	10	11	13	1	0.00971	10	11	1	0.00971	16	20	14	15	22	11
TR83	**1**	0.00971	15	14-14	13	32	23	10	11	14	1	0.00971	10	11	1	0.00971	15	20	16	18	21	11
TR84	**1**	0.00971	17	11-14	13	32	25	10	11	13	1	0.00971	11	10	1	0.00971	14	20	16	16	23	12
TR86	**1**	0.00971	16	14-15	13	31	24	11	11	13	1	0.00971	10	12	1	0.00971	15	19	15	17	22	11
TR87	**1**	0.00971	14	14-17	14	30	23	10	10	14	1	0.00971	11	10	1	0.00971	15	17	18	17	24	11

	Minimal Haplotype										SWGDAM (extended)				Y-Filer (extended)							
Haplotype	**n**	**Freq**	**DYS 19**	**DYS 385**	**DYS389 I**	**DYS389 II**	**DYS390**	**DYS 391**	**DYS392**	**DYS 393**	**n**	**Freq**	**DYS438**	**DYS 439**	**n**	**Freq**	**DYS437**	**DYS448**	**DYS456**	**DYS 458**	**DYS635**	**Y GATA H4**
TR88	**1**	0.00971	14	13-20	14	30	22	9	11	12	1	0.00971	9	10	1	0.00971	15	20	15	16	23	11
TR90	**1**	0.00971	14	11-15	13	28	24	10	14	12	1	0.00971	12	12	1	0.00971	15	19	15	16	23	12
TR91	**1**	0.00971	14	13-20	13	30	23	10	11	12	1	0.00971	10	12	1	0.00971	14	20	16	17	20	11
TR92	**1**	0.00971	14	11-14	13	28	23	11	14	12	1	0.00971	12	13	1	0.00971	14	19	16	15	23	13
TR93	**1**	0.00971	16	13-15	12	29	21	11	11	15	1	0.00971	10	11	1	0.00971	16	23	16	17	21	11
TR94	**1**	0.00971	14	12-15	14	31	23	10	11	12	1	0.00971	9	12	1	0.00971	15	20	18	16	22	9
TR95	**1**	0.00971	13	17-18	13	30	24	10	11	13	1	0.00971	11	11	1	0.00971	14	21	17	15	22	11
TR96	**1**	0.00971	17	14-16	12	28	22	10	11	13	1	0.00971	10	12	1	0.00971	16	21	16	16	21	12
TR97	**1**	0.00971	13	14-15	14	30	24	9	11	13	1	0.00971	10	10	1	0.00971	14	20	15	19	21	12
TR98	**1**	0.00971	15	15-16	12	29	22	10	10	13	1	0.00971	11	11	1	0.00971	14	22	15	17	21	11
TR99	**1**	0.00971	15	12-12	14	30	24	10	13	13	1	0.00971	11	13	1	0.00971	14	18	14	?	25	12
TR101	**1**	0.00971	15	13-14	12	28	22	10	11	13	1	0.00971	10	11	1	0.00971	16	20	14	15	20	11
TR102	**1**	0.00971	17	14-15	12	27	24	10	12	14	1	0.00971	8	11	1	0.00971	16	22	15	17	22	13
TR103	**1**	0.00971	15	11-14	14	30	26	11	11	13	1	0.00971	12	10	1	0.00971	14	20	16	15	23	13

Table 3.20: The minimal ht + SWGDAM loci Y-STR haplotypes in samples from Turkey [DYS385 excluded]

	Minimal Haplotype									SWGDAM (extended)				Y-Filer (extended)							
Haplotype	**n**	**Freq**	**DYS 19**	**DYS389 I**	**DYS389 II**	**DYS390**	**DYS 391**	**DYS392**	**DYS 393**	**n**	**Freq**	**DYS438**	**DYS 439**	**n**	**Freq**	**DYS437**	**DYS448**	**DYS456**	**DYS 458**	**DYS635**	**YGATA H4**
TR1	**1**	0.00971	15	13	29	24	10	13	13	1	0.00971	11	11	1	0.00971	14	17	15	14	26	13
TR2	**1**	0.00971	14	13	29	24	11	12	12	1	0.00971	12	12	1	0.00971	16	19	16	17	24	13
TR3	**1**	0.00971	16	14	31	24	9	11	13	1	0.00971	10	11	1	0.00971	14	20	15	19	23	10
TR4	**1**	0.00971	16	13	30	26	10	11	13	1	0.00971	11	10	1	0.00971	14	20	16	15	23	12
TR5	**1**	0.00971	13	13	31	24	10	11	13	1	0.00971	10	12	1	0.00971	14	19	16	17	22	12
TR6	**1**	0.00971	15	13	31	22	10	11	14	1	0.00971	10	11	1	0.00971	15	21	15	18	25	11
TR7	**2**	0.01942	15	13	29	23	10	11	12	1	0.00971	10	11	1	0.00971	14	19	16	18	20	12
TR23										1	0.00971	9	9	1	0.00971	15	21	15	15	21	11
TR8	**1**	0.00971	15	13	30	23	9	11	14	1	0.00971	9	11	1	0.00971	14	21	16	14	22	12
TR9	**1**	0.00971	15	12	27	24	10	11	12	1	0.00971	9	13	1	0.00971	15	19	13	17	21	11
TR10	**1**	0.00971	14	14	30	23	10	11	12	1	0.00971	10	11	1	0.00971	14	21	16	20	21	12
TR11	**2**	0.01942	14	13	30	24	10	11	12	2	0.01942	10	11	2	0.01942	15	19	15	20	21	11
TR31																					
TR12	**1**	0.00971	14	13	30	24	10	11	13	1	0.00971	10	12	1	0.00971	14	20	15	15	21	12
TR13	**1**	0.00971	13	13	30	24	11	11	13	1	0.00971	10	11	1	0.00971	15	20	17	16	24	12
TR14	**1**	0.00971	16	13	30	23	10	11	12	1	0.00971	9	11	1	0.00971	15	21	16	15	22	12
TR15	**1**	0.00971	15	13	30	24	10	11	13	1	0.00971	11	10	1	0.00971	14	21	16	15	23	13
TR16	**1**	0.00971	14	13	29	24	10	11	12	1	0.00971	9	12	1	0.00971	15	19	16	21	21	11
TR17	**2**	0.01942	15	12	29	22	10	10	13	1	0.00971	10	12	1	0.00971	16	20	15	18	20	11
TR98										1	0.00971	11	11	1	0.00971	14	22	15	17	21	11

	Minimal Haplotype									SWGDAM (extended)				Y-Filer (extended)							
Haplotype	**n**	**Freq**	**DYS 19**	**DYS389 I**	**DYS389 II**	**DYS390**	**DYS 391**	**DYS392**	**DYS 393**	**n**	**Freq**	**DYS438**	**DYS 439**	**n**	**Freq**	**DYS437**	**DYS448**	**DYS456**	**DYS 458**	**DYS635**	**YGATA H4**
TR18	**2**	0.01942	15	12	29	22	10	11	13	2	0.01942	10	11	1	0.00971	16	22	15	17	22	11
TR70														1	0.00971	16	20	16	18	22	13
TR19	**1**	0.00971	14	13	30	25	10	11	14	1	0.00971	10	11	1	0.00971	14	20	15	15	22	10
TR20	**1**	0.00971	15	13	29	23	9	11	12	1	0.00971	9	11	1	0.00971	14	21	15	14	23	12
TR21	**1**	0.00971	14	12	29	22	10	11	13	1	0.00971	10	11	1	0.00971	16	20	15	15	21	11
TR22	**1**	0.00971	13	14	31	24	10	11	13	1	0.00971	10	12	1	0.00971	14	20	17	16	22	12
TR24	**1**	0.00971	16	13	30	23	11	11	13	1	0.00971	11	10	1	0.00971	14	20	16	15	24	11
TR25	**1**	0.00971	11	12	30	23	10	12	12	1	0.00971	12	11	1	0.00971	14	21	15	13	22	12
TR26	**2**	0.01942	14	13	29	24	11	13	12	1	0.00971	12	12	1	0.00971	15	19	15	16	23	13
TR35										1	0.00971	12	13	1	0.00971	15	20	17	15	23	13
TR27	**1**	0.00971	14	12	29	24	11	11	12	1	0.00971	10	11	1	0.00971	14	20	15	18	24	11
TR28	**2**	0.01942	14	13	29	23	10	14	13	1	0.00971	10	10	1	0.00971	14	18	15	15	25	12
TR72										1	0.00971	10	11	1	0.00971	14	18	15	16	24	12
TR29	**1**	0.00971	14	13	29	23	11	13	12	1	0.00971	12	13	1	0.00971	15	19	15	16	23	12
TR30	**1**	0.00971	14	13	30	23	10	14	13	1	0.00971	10	11	1	0.00971	14	18	15	15	24	11
TR32	**1**	0.00971	15	13	28	24	10	11	12	1	0.00971	9	11	1	0.00971	14	20	16	17	23	11
TR33	**1**	0.00971	14	13	30	21	10	11	14	1	0.00971	10	11	1	0.00971	16	22	16	18	21	11
TR34	**1**	0.00971	14-15	13	30	22	9	11	14	1	0.00971	10	11	1	0.00971	16	20	13	18	21	11
TR36	**4**	0.03883	14	13	29	23	10	11	12	1	0.00971	10	12	1	0.00971	14	20	15	18	21	11
TR60										1	0.00971	9	12	1	0.00971	15	20	16	17	24	9
TR41										2	0.01942	9	11	1	0.00971	14	21	15	14	22	12
TR63														1	0.00971	15	20	15	17	22	11

	Minimal Haplotype									SWGDAM (extended)				Y-Filer (extended)						
Haplotype	**n**	**Freq**	**DYS 19**	**DYS389 I**	**DYS389 II**	**DYS390**	**DYS 391**	**DYS392**	**DYS 393**	**n**	**Freq**	**DYS438**	**DYS 439**	**n**	**Freq**	**DYS437**	**DYS448**	**DYS456**	**DYS 458**	**DYS635**	**YGATA H4**
TR37	**1**	0.00971	16	13	33	25	12	11	13	1	0.00971	11	10	1	0.00971	15	20	15	15	23	13
TR38	**1**	0.00971	15	13	28	22	9	10	12	1	0.00971	9	12	1	0.00971	14	21	15	14	21	12
TR39	**1**	0.00971	14	12	28	23	10	11	13	1	0.00971	10	11	1	0.00971	16	20	14	16	22	11
TR40	**1**	0.00971	15	13	31	24	11	11	13	1	0.00971	11	10	1	0.00971	14	19	17	15	23	14
TR42	**1**	0.00971	13	14	31	25	9	13	12	1	0.00971	12	12	1	0.00971	15	21	14	16	23	12
TR43	**1**	0.00971	14	13	29	23	11	11	12	1	0.00971	9	11	1	0.00971	14	20	16	17	22	10
TR44	**1**	0.00971	14	13	30	25	11	11	13	1	0.00971	11	10	1	0.00971	14	20	15	15	23	13
TR45	**1**	0.00971	13	13	30	26	9	11	14	1	0.00971	10	11	1	0.00971	14	20	15	17	22	11
TR46	**1**	0.00971	14	12	27	24	10	14	12	1	0.00971	11	12	1	0.00971	15	19	15	17	23	13
TR47	**2**	0.01942	16	13	31	24	11	11	13	2	0.01942	10	12	1	0.00971	15	20	15	17	23	11
TR86														1	0.00971	15	19	15	17	22	11
TR48	**1**	0.00971	14	13	28	24	11	14	12	1	0.00971	12	11	1	0.00971	14	19	16	18	23	11
TR49	**1**	0.00971	16	13	29	26	10	11	13	1	0.00971	10	10	1	0.00971	14	20	15	20	21	11
TR50	**3**	0.02913	16	13	29	24	10	11	13	2	0.01942	11	11	1	0.00971	14	20	17	16	24	12
TR75														1	0.00971	14	?	15	17	21	11
TR58										1	0.00971	11	12	1	0.00971	14	?	17	17	22	11
TR51	**1**	0.00971	14	12	29	23	9	11	12	1	0.00971	10	11	1	0.00971	14	19	15	16	22	13
TR52	**1**	0.00971	15	13	29	24	10	11	12	1	0.00971	9	13	1	0.00971	14	19	14	15	22	12
TR53	**1**	0.00971	16	13	31	25	11	11	13	1	0.00971	11	10	1	0.00971	14	19	15	16	23	13
TR54	**2**	0.01942	14	12	28	23	10	11	12	1	0.00971	9	12	1	0.00971	14	21	14	20	21	11
TR78										1	0.00971	9	11	1	0.00971	15	21	15	19	22	11
TR55	**1**	0.00971	14	14	30	23	10	14	13	1	0.00971	10	10	1	0.00971	14	19	15	16	24	12

	Minimal Haplotype									SWGDAM (extended)				Y-Filer (extended)						
Haplotype	**n**	**Freq**	**DYS 19**	**DYS389 I**	**DYS389 II**	**DYS390**	**DYS 391**	**DYS392**	**DYS 393**	**n**	**Freq**	**DYS438**	**DYS 439**	**n**	**Freq**	**DYS437**	**DYS448**	**DYS456**	**DYS 458**	**DYS635**	**YGATA H4**
TR56	**1**	0.00971	14	13	30	22	10	11	12	1	0.00971	9	13	1	0.00971	15	21	15	15	23	11
TR57	**1**	0.00971	14	13	32	23	11	11	12	1	0.00971	9	11	1	0.00971	16	22	17	15	22	11
TR59	**1**	0.00971	14	13	29	24	11	11	12	1	0.00971	10	12	1	0.00971	15	19	15	20	21	11
TR61	**2**	0.01942	13	13	30	24	10	11	13	1	0.00971	10	11	1	0.00971	14	20	17	16	22	12
TR95										1	0.00971	11	11	1	0.00971	14	21	17	15	22	11
TR62	**1**	0.00971	15	14	30	25	11	11	12	1	0.00971	9	13	1	0.00971	14	21	16	16	22	12
TR64	**1**	0.00971	14	13	29	22	11	11	12	1	0.00971	10	12	1	0.00971	14	20	14	18	21	11
TR65	**1**	0.00971	13	13	28	24	10	15	12	1	0.00971	12	12	1	0.00971	14	19	16	17	23	13
TR66	**2**	0.01942	14	13	30	23	10	11	12	1	0.00971	9	11	1	0.00971	15	21	15	15	22	12
TR91										1	0.00971	10	12	1	0.00971	14	20	16	17	20	11
TR67	**4**	0.03883	16	13	30	25	11	11	13	4	0.03883	11	10	1	0.00971	14	20	16	15	25	13
TR76														1	0.00971	14	20	16	16	23	12
TR89														1	0.00971	14	20	17	15	24	13
TR100														1	0.00971	14	20	16	15	23	11
TR68	**1**	0.00971	14	13	29	24	11	13	13	1	0.00971	12	13	1	0.00971	15	19	15	17	23	12
TR69	**1**	0.00971	14	16	32	23	10	11	12	1	0.00971	9	11	1	0.00971	15	21	15	17	21	11
TR71	**1**	0.00971	15	13	30	25	11	11	13	1	0.00971	11	10	1	0.00971	14	20	16	15	23	12
TR73	**1**	0.00971	15	13	32	26	11	11	13	1	0.00971	11	10	1	0.00971	14	19	17	15	23	13
TR74	**1**	0.00971	14	13	31	24	11	13	12	1	0.00971	12	13	1	0.00971	15	19	16	15	23	10
TR77	**1**	0.00971	15	13	30	21	11	11	14	1	0.00971	10	11	1	0.00971	16	21	15	16	20	12
TR79	**1**	0.00971	15	12	29	22	10	10	14	1	0.00971	10	13	1	0.00971	14	21	15	16	22	12
TR80	**1**	0.00971	14	13	29	25	12	13	13	1	0.00971	12	12	1	0.00971	15	19	15	17	24	12

	Minimal Haplotype									SWGDAM (extended)				Y-Filer (extended)							
Haplotype	**n**	**Freq**	**DYS 19**	**DYS389 I**	**DYS389 II**	**DYS390**	**DYS 391**	**DYS392**	**DYS 393**	**n**	**Freq**	**DYS438**	**DYS 439**	**n**	**Freq**	**DYS437**	**DYS448**	**DYS456**	**DYS 458**	**DYS635**	**YGATA H4**
TR81	**2**	0.01942	14	13	30	24	11	13	12	1	0.00971	12	12	1	0.00971	14	19	15	16	23	13
TR85										1	0.00971	12	13	1	0.00971	14	19	15	16	23	12
TR82	**1**	0.00971	15	12	28	23	10	11	13	1	0.00971	10	11	1	0.00971	16	20	14	15	22	11
TR83	**1**	0.00971	15	13	32	23	10	11	14	1	0.00971	10	11	1	0.00971	15	20	16	18	21	11
TR84	**1**	0.00971	17	13	32	25	10	11	13	1	0.00971	11	10	1	0.00971	14	20	16	16	23	12
TR87	**1**	0.00971	14	14	30	23	10	10	14	1	0.00971	11	10	1	0.00971	15	17	18	17	24	11
TR88	**1**	0.00971	14	14	30	22	9	11	12	1	0.00971	9	10	1	0.00971	15	20	15	16	23	11
TR90	**1**	0.00971	14	13	28	24	10	14	12	1	0.00971	12	12	1	0.00971	15	19	15	16	23	12
TR92	**1**	0.00971	14	13	28	23	11	14	12	1	0.00971	12	13	1	0.00971	14	19	16	15	23	13
TR93	**1**	0.00971	16	12	29	21	11	11	15	1	0.00971	10	11	1	0.00971	16	23	16	17	21	11
TR94	**1**	0.00971	14	14	31	23	10	11	12	1	0.00971	9	12	1	0.00971	15	20	18	16	22	9
TR96	**1**	0.00971	17	12	28	22	10	11	13	1	0.00971	10	12	1	0.00971	16	21	16	16	21	12
TR97	**1**	0.00971	13	14	30	24	9	11	13	1	0.00971	10	10	1	0.00971	14	20	15	19	21	12
TR99	**1**	0.00971	15	14	30	24	10	13	13	1	0.00971	11	13	1	0.00971	14	18	14	?	25	12
TR101	**1**	0.00971	15	12	28	22	10	11	13	1	0.00971	10	11	1	0.00971	16	20	14	15	20	11
TR102	**1**	0.00971	17	12	27	24	10	12	14	1	0.00971	8	11	1	0.00971	16	22	15	17	22	13
TR103	**1**	0.00971	15	14	30	26	11	11	13	1	0.00971	12	10	1	0.00971	14	20	16	15	23	13

Table 3.21 Number of occurrence of the same haplotype for minimal, SWGDAM and Y-Filer Extended Loci

#of shared HT	min HT	SWGDAM	Y-Filer
DYS 385 Excluded	14	6	1
DYS 385 Included	4	1	0

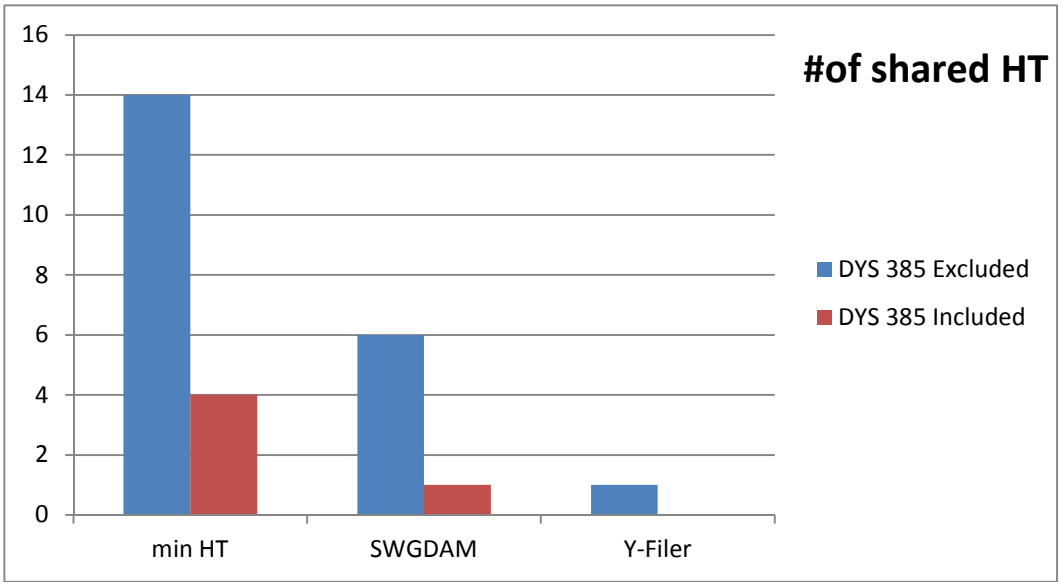


Figure 3.35 Number of occurrence of the same haplotype for minimal, SWGDAM and Y-Filer Extended Loci

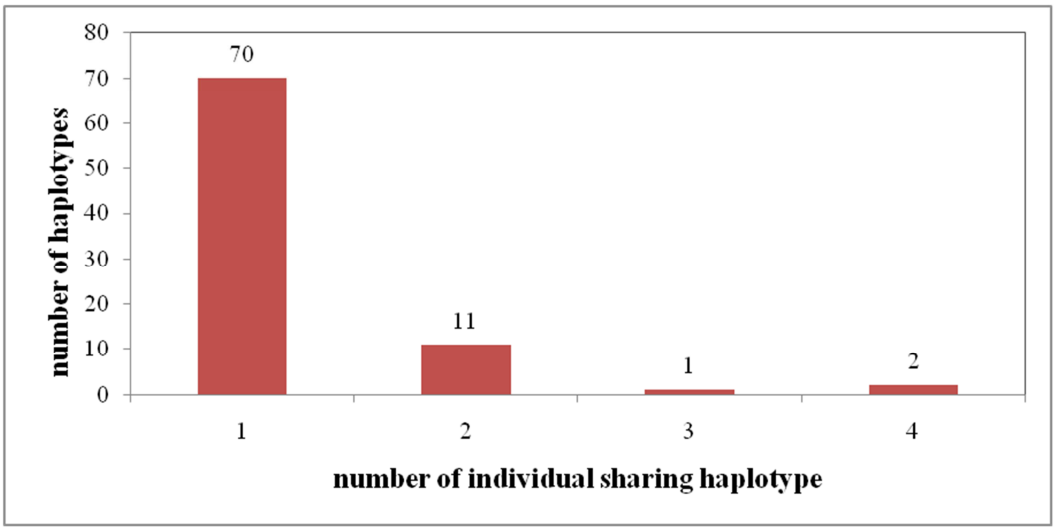


Figure 3.36 Haplotype frequency distribution for DYS385 excluded minHT

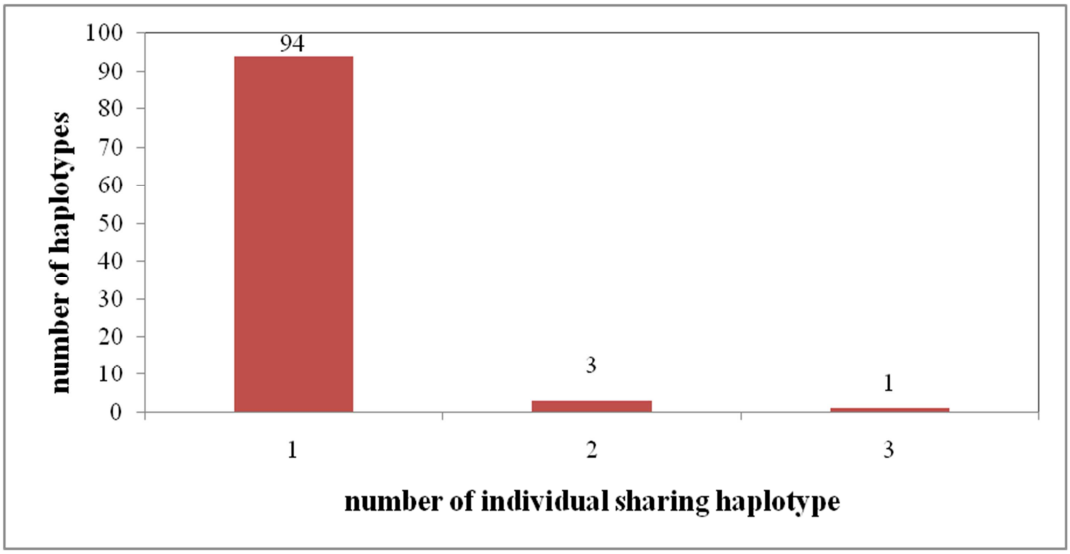


Figure 3.37 Haplotype frequency distribution for DYS385 included minHT

Table 3.22 Haplotype Sharing Summary

Population (n=103)	**DYS385 included**	**DYS385 excluded**
Y-filer 17 Y-STR haplotype		
Number of haplotypes	103	102
# of haplotypes observed once	103	101
# of haplotypes observed twice	0	1
# of haplotypes observed 3 times	0	0
# of haplotypes observed 4 times	0	0
Haplotype diversity	1.0000 +/- 0.0014	1.0000 +/- 0.0014
Extended SWGDAM 11 Y-STR haplotype		
Number of haplotypes	101	95
# of haplotypes observed once	100	89
# of haplotypes observed twice	0	5
# of haplotypes observed 3 times	1	0
# of haplotypes observed 4 times	0	1
Haplotype diversity	1.0000 +/- 0.0014	1.0000 +/- 0.0014
Minimal 9 Y-STR haplotype		
Number of haplotypes	98	84
# of haplotypes observed once	94	70
# of haplotypes observed twice	3	11
# of haplotypes observed 3 times	1	1
# of haplotypes observed 4 times	0	2
Haplotype diversity	1.0000 +/- 0.0014	1.0000 +/- 0.0014

3.3 GENE DIVERSITY

3.3.1 TURKEY

Table 3.23 Gene diversity values for each Y-STR locus

Locus	# of alleles	Gene Diversity
DYS19	7	0.7078
DYS385a/b	31	0.9442
DYS389 I	4	0.4818
DYS389 II	7	0.7409
DYS390	6	0.7501
DYS391	4	0.5711
DYS392	6	0.4534
DYS393	4	0.6040
DYS437	3	0.5877
DYS438	5	0.7289
DYS439	5	0.7103
DYS448	7	0.7232
DYS456	6	0.6678
DYS458	11	0.8132
DYS635	7	0.7809
Y GATA H4	6	0.6889

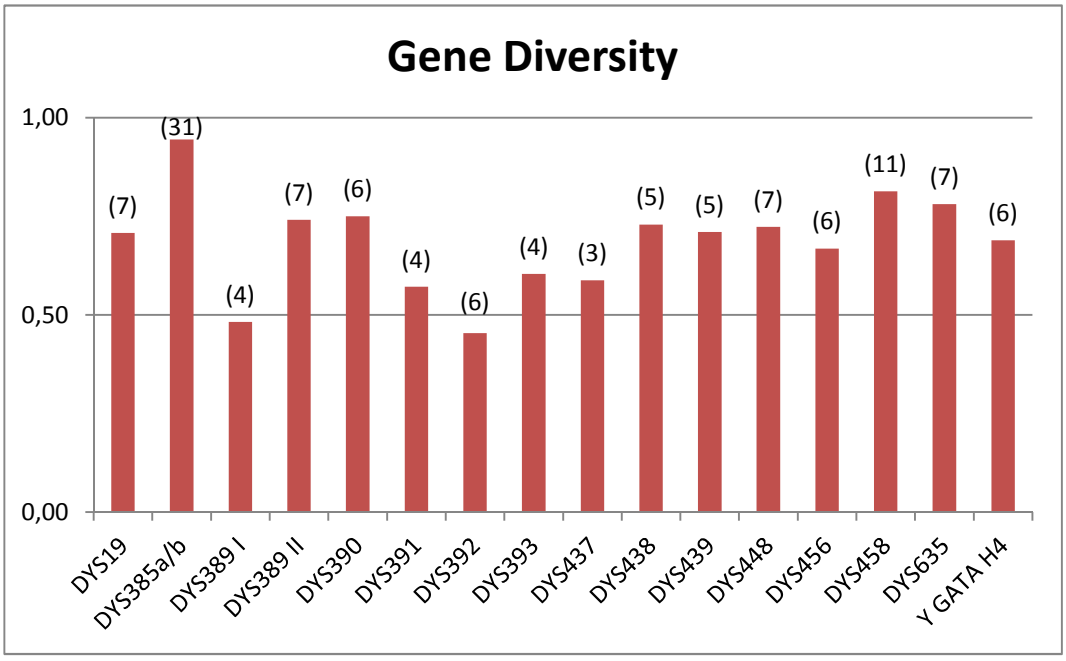


Figure 3.38 Genetic diversity for Y-STR loci

3.3.2 REGIONAL

Table 3.24 Locus diversities for each geographical region of Turkey

	Mediterranean	Eastern	Aegean	Southeastern	Central Anatolia	Black Sea	Marmara
DYS19	0.7821	0.6484	0.7949	0.7778	0.6959	0.6400	0.7629
DYS385a/b	0.9744	0.9451	0.9359	1.0000	0.9532	0.9567	0.9407
DYS389 I	0.5897	0.4725	0.5128	0.5556	0.4503	0.5067	0.5217
DYS389 II	0.7436	0.7253	0.7692	0.7222	0.7310	0.7633	0.7668
DYS390	0.6410	0.7473	0.8333	0.7778	0.8012	0.6867	0.8024
DYS391	0.5128	0.5824	0.7308	0.5000	0.6491	0.5267	0.5415
DYS392	0.4231	0.5055	0.2949	0.4167	0.4561	0.5267	0.5099
DYS393	0.6026	0.5714	0.6539	0.6389	0.6550	0.5867	0.5968
DYS437	0.2821	0.6923	0.5641	0.4167	0.6550	0.5733	0.6127
DYS438	0.7949	0.7802	0.6795	0.6667	0.7193	0.7833	0.7154
DYS439	0.7180	0.7143	0.6667	0.6944	0.7485	0.7667	0.7233
DYS448	0.7424	0.6593	0.6923	0.7500	0.7485	0.7536	0.6482
DYS456	0.6539	0.6593	0.6667	0.6389	0.5556	0.7433	0.7194
DYS458	0.9394	0.8022	0.8333	0.8611	0.8187	0.7733	0.7233
DYS635	0.7692	0.8462	0.8205	0.8333	0.7602	0.7333	0.7984
Y GATA H4	0.6795	0.7363	0.5897	0.6389	0.7661	0.7333	0.6285

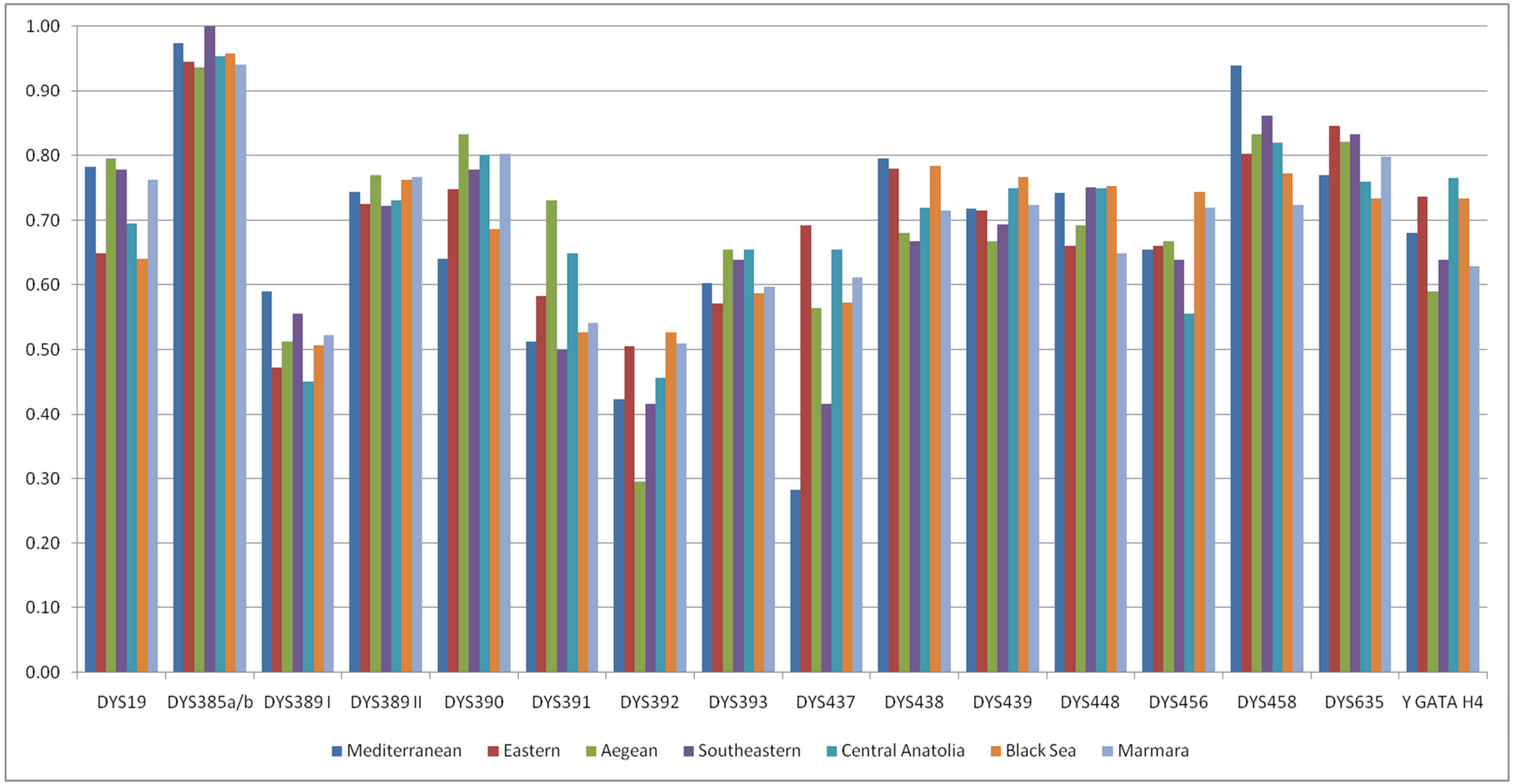


Figure 3.39 Locus diversities of geographical regions

3.4 DYS 19 DUPLICATION

In this study, an individual is found to exhibit biallelic pattern for DYS19 (14 and 15). When the father was typed for the same locus, it has been observed to inherit his haplotype to his son (Figure 3.40).

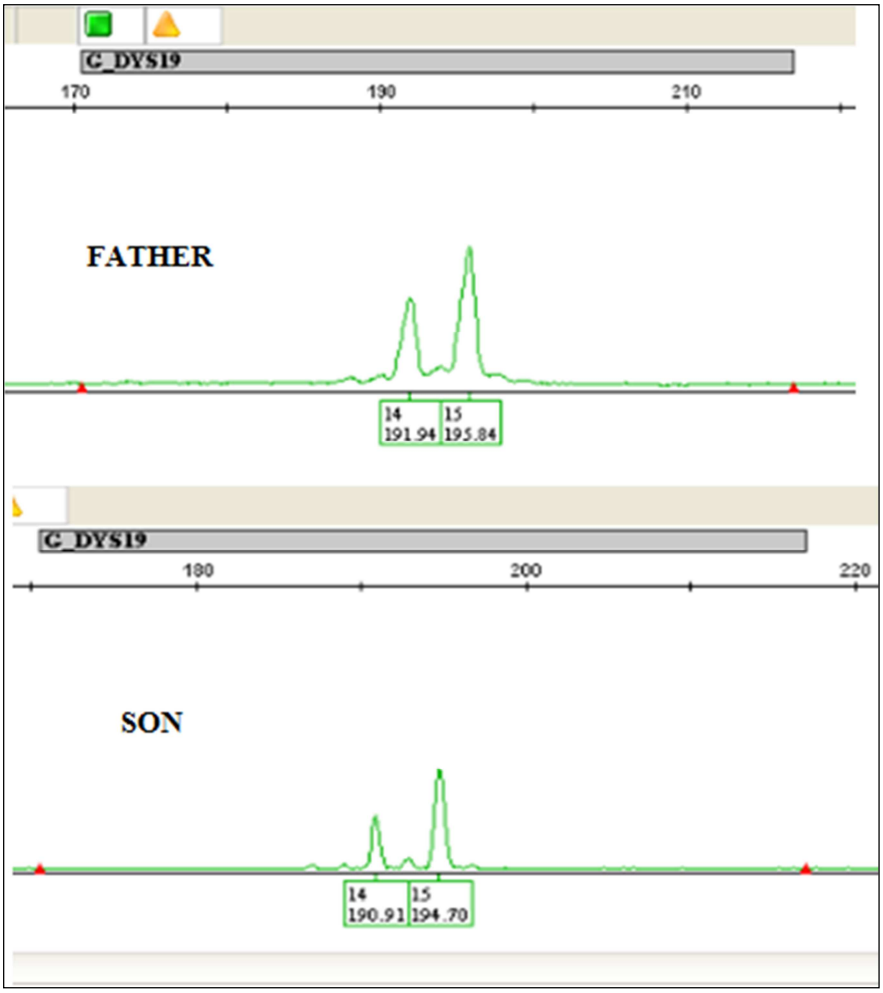


Figure 3.40 Duplication at locus DYS19 (father - son pair)

3.5 A COMPARATIVE INTER-POPULATION ANALYSIS

Using an implementation of Analysis of Molecular Variance (AMOVA) provided at the YHRD website, an MDS analysis based on Fst genetic distances was performed. A good fit between the two dimensional plot and pairwise Fst values was obtained with a low stress value 0.0273.

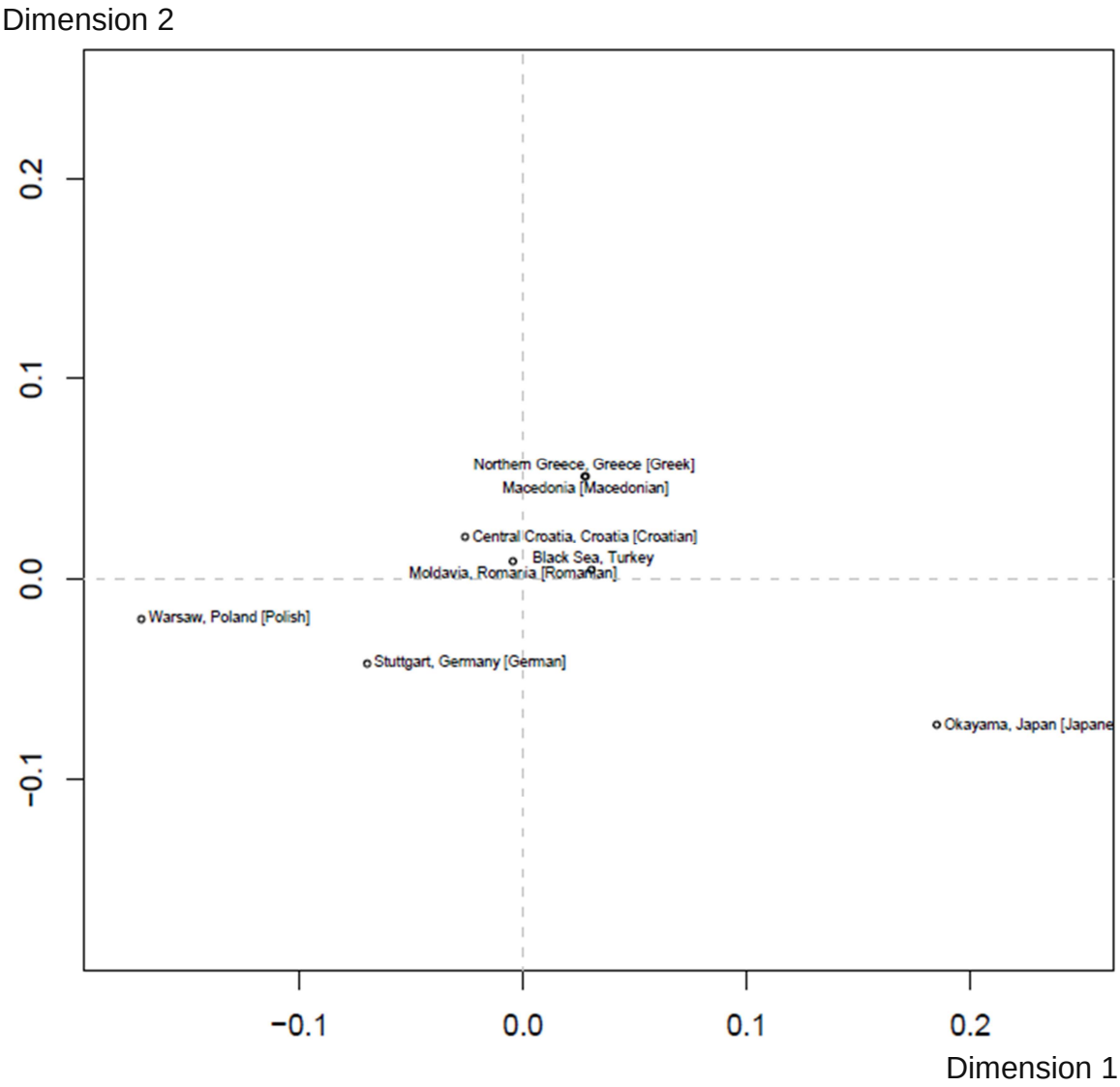


Figure 3.41 Multidimensional scaling plot (MDS)
(MDS plot is based on Fst genetic distances between Turkey Black Sea Region and seven reference populations; stress value = 0.0273)

Table 3.25 AMOVA pairwise distance
(based on Fst values between Black Sea Region of Turkey and eight reference population samples; P values are shown above, Fst values below the diagonal)

Population	Central Croatia, [Croatian]	Macedo-nia [Macedo-nian]	Moldavia, Romania [Roman-ian]	Northern Greece, [Greek]	Okayama, Japan [Japanese]	Stuttgart, Germany [German]	Warsaw, Poland [Polish]	Black Sea, Turkey
Central Croatia, [Croatian]	-	<0.0001	0.0534	<0.0001	<0.0001	<0.0001	<0.0001	0.0022
Macedo-nia [Macedo-nian]	0.0579	-	0.0034	0.3394	<0.0001	<0.0001	<0.0001	0.0209
Moldavia, Romania [Romanian]	0.0170	0.0524	-	0.0046	<0.0001	0.0004	<0.0001	**0.0691**
Northern Greece, [Greek]	0.0541	0.0005	0.0398	-	<0.0001	<0.0001	<0.0001	0.0274
Okayama, Japan [Japanese]	0.2599	0.2194	0.1744	0.2218	-	<0.0001	<0.0001	<0.0001
Stuttgart, Germany [German]	0.0859	0.1787	0.0531	0.1505	0.2865	-	<0.0001	0.0002
Warsaw, Poland [Polish]	0.1161	0.2667	0.1498	0.2315	0.3738	0.1051	-	<0.0001
Black Sea, Turkey	0.0787	0.0460	0.0226	0.0439	0.1403	0.1161	0.2182	-

Comparisons of pairs of population samples yielded significant Fst values for all the population pairs with the exception of the population pair Black Sea Region of Turkey- Moldavia, Romania. Comparative study revealed the close relationship of our population with Moldavia population from Romania (Fst = 0.0226, P = 0.0691) followed by Northern Greece, [Greek] population (Fst = 0439, P = 0.0274). Okayama, Japan [Japanese], Stuttgart, Germany [German], Warsaw, Poland [Polish] populations are significantly different (Fst = 0. 1403, P = <0.0001), (Fst = 0. 1161, P = 0.0002), (Fst = 0.2182, P = <0.0001) respectively. The largest distances occurred between population pairs: Turkey- Poland, Turkey-Japan, Turkey-Germany, Turkey- Central Croatia, Turkey-Macedonia, Turkey- Northern Greece, and Turkey-Moldavia.

CHAPTER IV

DISCUSSION & CONCLUSION

In this study, it is aimed to show high degree genetic diversity in Turkey. Therefore, 116 healthy male donors from Turkey were typed for variation at 17 Y chromosomal STR loci (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS385a/b, DYS438, DYS439, DYS437, DYS448, DYS458, DYS456, DYS635, and Y-GATA-H4). By the way this is the first study that investigates 17 Y-STR loci in population of Turkey.

Allele frequencies for each locus are calculated. The most common alleles are; 14 for DYS19, 11-14 for DYS385a/b, 13 for DYS389 I, 30 for DYS389 II, 24 for DYS390, 10 for DYS391, 11 for DYS392, 11 for DYS393, 14 for DYS437, 10 for DYS438, 11 for DYS439, 19 for DYS448, 15 for DYS456, 16 AND 17 for DYS458, 23 for DYS635, 11 for Y GATA H4 (Table 3.17).

Regional allele frequencies were also calculated and compared to each other. Distinctive results were obtained in some loci. For instance, according to DYS19 analysis allele 11 was observed in only Mediterranean Region while allele 17 was observed in only Marmara Region. So, this may be a distinctive feature for DYS19.

Among 103 male individuals typed for all loci, all haplotypes were found to be unique (GD: 1.00). Henke et al showed a high degree of haplotype diversity in German Turks, with the highest frequency of 2.5%. Asicioglu 2003 also found the two most frequent haplotypes occurring three times with the frequency 1.5%. In this study the most common haplotype for 17 Y-STR loci (in DYS385 excluded case) was observed two times with the frequency of 1.9% which confirms the precious observations.

However, when the number of analyzed loci decreased to SWGDAM or minimal haplotype loci, it is observed that some individuals share the same haplotype. In order to

show the case, “minimal” and “extended” haplotypes were compared and the results are given in Table 3.22. The number of haplotypes was less than the total number of samples, since there were some individuals sharing the same haplotype.

The increasing number of same haplotype combinations, after exclusion of DYS385 locus, confirms this locus as one of the most informative Y-STR loci so far (Aşıcıoğlu 2003).

Moreover, locus diversities were calculated for both Turkey and its regions. When Turkey was analyzed as a whole, DYS385a/b was found to be the most polymorphic locus with 94.43% gene diversity (GD) while DYS392 was the least polymorphic locus (GD= 45,34%). Besides, loci with highest and lowest diversity were determined for each region and listed in Table 3.24.

The compilation of haplotypes shows that the vast majority of haplotypes was observed only once, reflecting the enormous genetic heterogeneity in Turkey. This heterogeneity could be seen more detailed when the regional analysis were taken into account.

Duplicated alleles at Y-STR loci were described earlier (Kayser, Caglia et al. 1997; Kayser, Roewer et al. 2000). In this study an individual with a biallelic pattern for DYS19 was detected (allele duplication:14,15) and in order to be sure, the father was also analyzed for the same locus. The resulting profile was the same with the son, as expected (Figure 3.40). Two similar case, duplicated allele at locus DYS19, were observed in a Turkish male living in Germany and a Turkish male in Central Anatolia Region of Turkey (Henke, Henke et al. 2001; Cakir, Celebioglu et al. 2004). In conclusion, the seventeen Y-STR polymorphisms provide a powerful means for male identification in forensic cases and parentage testing in Turkey.

For having an illustration of the genetic relation a comparative inter-population analysis among Anatolian (Black Sea Region), Moldavia- Romanian (Stanciu, Cutar et al.) , Macedonia (Jakovski, Nikolova et al. ; Spiroski, Arsov et al. 2005), Polish (Ploski, Wozniak et al. 2002), German (Roewer, Krawczak et al. 2001) , Croatian, Greek

(Kovatsi, Saunier et al. 2009) and Japanese populations (Mizuno, Nakahara et al. 2008) is done.

Figure 4.1 Black Sea Map

The analysis of the Y-STR variation pattern indicated that the contemporary northern Turkish population (Black Sea Region) shows the most similarity to Romanian, while the other available Y-STR data suggested significant differences from the other populations. Romanian samples were from Moldavia which includes the Gagauz settlement. The Gagauzes are a small Turkish-speaking ethnic group living mostly in southern Moldova. The Gagauzes speak the Oghuz version of the Turkic languages, which also includes the Azeri, Turkish, and Turkmen languages. The origin of the Gagauzes is unclear. Several hypotheses about the ethnogenesis of the Gagauzes have been proposed (Pokrovskaya 1964; Guboglo 1967). Two of them are more popular and suggest different structure of the Gagauzes being either closer to the Central Asian or Anatolian one. They may be the descendants of the Turkic nomadic tribes from the South Russian steppes (Kumans, Pechenegs, or Torks) as suggested by the Steppe hypothesis, or have a complex Anatolian-steppe origin, as postulated by the Seljuk or Anatolian hypothesis.

The genetic similarity between Moldavia and Black sea region of Turkey discovered in this study provides some support of the hypothesis of the Seljuk origin of

the Gagauzes and Anatolian route of the Gagauz migration instead of northern way from the Eurasian steppes.

This information therefore becomes not only useful for forensic investigation, but also population genetics and history as it can be used to infer some major demographic events related to the population dynamics based on the contemporary genetic data, and even possibly be used to confirm some historic information.

In conclusion, results of the present Y chromosomal STR study reveals high level of genetic diversity in Turkey.

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APPENDIX A

Regions	Sample	DYS 19	DYS 385	DYS389 I	DYS389 II	DYS390	DYS 391	DYS392	DYS 393	DYS437	DYS438	DYS 439	DYS448	DYS456	DYS 458	DYS635	Y GATA H4
S	h1	15	12-13	13	29	24	10	13	13	14	11	11	17	15	14	26	13
E	h2	14	10-14	13	29	24	11	12	12	16	12	12	19	16	17	24	13
C	h3	16	12-12	14	31	24	9	11	13	14	10	11	20	15	19	23	10
Mar	h4	16	11-15	13	30	26	10	11	13	14	11	10	20	16	15	23	12
S	h5	13	16-16	13	31	24	10	11	13	14	10	12	19	16	17	22	12
S	h6	15	14-14	13	31	22	10	11	14	15	10	11	21	15	18	25	11
A	h7	15	14-18	13	29	23	10	11	12	14	10	11	19	16	18	20	12
Med	h8	15	13-15	13	30	23	9	11	14	14	9	11	21	16	14	22	12
E	h9	15	13-16	12	27	24	10	11	12	15	9	13	19	13	17	21	11
Med	h10	14	12-17	14	30	23	10	11	12	14	10	11	21	16	20	21	12
B	h11	14	13-15	13	30	24	10	11	12	15	10	11	19	15	20	21	11
Med	h12	14	17-18	13	30	24	10	11	13	14	10	12	20	15	15	21	12
Mar	h13	13	16-19	13	30	24	11	11	13	15	10	11	20	17	16	24	12
C	h14	16	13-17	13	30	23	10	11	12	15	9	11	21	16	15	22	12
C	h15	15	12-15	13	30	24	10	11	13	14	11	10	21	16	15	23	13
B	h16	13	15-17	14	33	24	11	11	13	14	10	12	19	15	16	21	11
E	h17	14	13-16	13	29	24	10	11	12	15	9	12	19	16	21	21	11
S	h18	15	14-17	12	29	22	10	10	13	16	10	12	20	15	18	20	11
C	h19	15	12-14	12	29	22	10	11	13	16	10	11	22	15	17	22	11
C	h20	14	17-17	13	30	25	10	11	14	14	10	11	20	15	15	22	10
Med	h21	15	13-17	13	29	23	9	11	12	14	9	11	21	15	14	23	12
Mar	h22	14	13-15	12	29	22	10	11	13	16	10	11	20	15	15	21	11
Mar	h23	13	17-18	14	31	24	10	11	13	14	10	12	20	17	16	22	12
Mar	h24	15	13-17	13	29	23	10	11	12	15	9	9	21	15	15	21	11
E	h25	16	11-15	13	30	23	11	11	13	14	11	10	20	16	15	24	11
A	h26	11	18-18	12	30	23	10	12	12	14	12	11	21	15	13	22	12
B	h27	14	11-14	13	29	24	11	13	12	15	12	12	19	15	16	23	13
S	h28	14	12-19	12	29	24	11	11	12	14	10	11	20	15	18	24	11
B	h29	14	12-13	13	29	23	10	14	13	14	10	10	18	15	15	25	12
E	h30	14	11-14	13	29	23	11	13	12	15	12	13	19	15	16	23	12

Regions	Sample	DYS 19	DYS 385	DYS389 I	DYS389 II	DYS390	DYS 391	DYS392	DYS 393	DYS437	DYS438	DYS 439	DYS448	DYS456	DYS 458	DYS635	Y GATA H4
B	h31	14	13-14	13	31	23	9	11	12	15	9	10	20	15	15	22	11
Mar	h32	14	12-13	13	30	23	10	14	13	14	10	11	18	15	15	24	11
C	h33	14	12-16	13	30	24	10	11	12	15	10	11	19	15	20	21	11
Mar	h34	15	14-15	13	28	24	10	11	12	14	9	11	20	16	17	23	11
C	h35	14	13-15	13	30	21	10	11	14	16	10	11	22	16	18	21	11
E	h36	14-15	12-15	13	30	22	9	11	14	16	10	11	20	13	18	21	11
B	h37	14	12-15	13	29	24	11	13	12	15	12	13	20	17	15	23	13
Med	h38	14	13-15	13	29	23	10	11	12	14	10	12	20	15	18	21	11
A	h39	16	11-14	13	33	25	12	11	13	15	11	10	20	15	15	23	13
C	h40	15	13-16	13	28	22	9	10	12	14	9	12	21	15	14	21	12
Mar	h41	14	14-14	12	28	23	10	11	13	16	10	11	20	14	16	22	11
C	h42	15	11-14	13	31	24	11	11	13	14	11	10	19	17	15	23	14
S	h43	14	12-17	13	29	23	10	11	12	14	9	11	21	15	14	22	12
E	h44	14	13-17	13	32	22	10	11	12	14	10	11	21	15	17	20	10
C	h45	13	11-14	14	31	25	9	13	12	15	12	12	21	14	16	23	12
E	h46	14	14-15	13	29	23	11	11	12	14	9	11	20	16	17	22	10
A	h47	14	11-14	13	30	25	11	11	13	14	11	10	20	15	15	23	13
Mar	h48	13	17-18	13	30	26	9	11	14	14	10	11	20	15	17	22	11
E	h49	14	11-15	12	27	24	10	14	12	15	11	12	19	15	17	23	13
C	h50	16	15-15	13	31	24	11	11	13	15	10	12	20	15	17	23	11
K	h51	13	14-18	14	30	22	10	15	13	14	11	13	19	16	18	22	10
K	h52	13	16-16	13	32	24	10	11	13	14	10	13	20	15	17	22	11
A	h53	14	11-15	13	28	24	11	14	12	14	12	11	19	16	18	23	11
Mar	h54	16	12-12	13	29	26	10	11	13	14	10	10	20	15	20	21	11
Mar	h55	16	11-15	13	29	24	10	11	13	14	11	11	20	17	16	24	12
Mar	h56	14	12-14	12	29	23	9	11	12	14	10	11	19	15	16	22	13
K	h57	15	16-18	13	29	24	10	11	12	14	9	13	19	14	15	22	12
Med	h58	16	11-12	13	31	25	11	11	13	14	11	10	19	15	16	23	13
K	h59	14	13-17	12	28	23	10	11	12	14	9	12	21	14	20	21	11
C	h60	14	12-13	14	30	23	10	14	13	14	10	10	19	15	16	24	12
C	h61	14	13-15	13	30	22	10	11	12	15	9	13	21	15	15	23	11
K	h62	14	11-13	14	29	23	11	14	14	14	10	10	19	14	17	23	12
K	h63	14	12-16	13	32	23	11	11	12	16	9	11	22	17	15	22	11
K	h64	16	12-15	13	29	24	10	11	13	14	11	12	?	17	17	22	11
C	h65	14	13-16	13	29	24	11	11	12	15	10	12	19	15	20	21	11
K	h66	14	12-15	13	29	23	10	11	12	15	9	12	20	16	17	24	9
S	h67	13	16-19	13	30	24	10	11	13	14	10	11	20	17	16	22	12

Regions	Sample	DYS 19	DYS 385	DYS389 I	DYS389 II	DYS390	DYS 391	DYS392	DYS 393	DYS437	DYS438	DYS 439	DYS448	DYS456	DYS 458	DYS635	Y GATA H4
S	h68	15	14-15	14	30	25	11	11	12	14	9	13	21	16	16	22	12
E	h69	14	13-16	13	29	23	10	11	12	15	9	11	20	15	17	22	11
Mar	h70	14	14-18	13	29	22	11	11	12	14	10	12	20	14	18	21	11
Med	h71	13	11-13	13	28	24	10	15	12	14	12	12	19	16	17	23	13
K	h72	14	13-14	13	30	23	10	11	12	15	9	11	21	15	15	22	12
K	h73	13	17-17	12	31	24	10	11	13	14	8	13	20	12	17	22	11
K	h74	16	16-16	13	29	23	9	11	12	14	9	12	21	16	14	23	12
C	h75	16	11-14	13	30	25	11	11	13	14	11	10	20	16	15	25	13
C	h76	14	11-15	13	29	24	11	13	13	15	12	13	19	15	17	23	12
E	h77	16	12-16	13	29	24	10	15	10	15	10	11	19	15	16	22	11
C	h78	14	13-14	16	32	23	10	11	12	15	9	11	21	15	17	21	11
K	h79	15	11-14	14	31	25	10	11	13	14	11	11	20	15	17	23	13
E	h80	15	13-14	12	29	22	10	11	13	16	10	11	20	16	18	22	13
Med	h81	15	11-14	13	30	25	11	11	13	14	11	10	20	16	15	23	12
K	h82	14	12-13	13	29	23	10	14	13	14	10	11	18	15	16	24	12
K	h83	15	11-14	13	32	26	11	11	13	14	11	10	19	17	15	23	13
K	h84	14	11-14	13	31	24	11	13	12	15	12	13	19	16	15	23	10
Med	h85	16	12-15	13	29	24	10	11	13	14	11	11	?	15	17	21	11
S	h86	16	11-12	13	30	25	11	11	13	14	11	10	20	16	16	23	12
C	h87	15	14-14	13	30	21	11	11	14	16	10	11	21	15	16	20	12
K	h88	14	13-15	13	29	22	10	11	12	15	9	11	21	15	14	21	11
Med	h89	14	13-18	12	28	23	10	11	12	15	9	11	21	15	19	22	11
K	h90	15	15-16	12	29	22	10	10	14	14	10	13	21	15	16	22	12
Mar	h91	14	11-14	13	29	25	12	13	13	15	12	12	19	15	17	24	12
Mar	h92	14	11-14	13	30	24	11	13	12	14	12	12	19	15	16	23	13
K	h93	15	14-15	12	28	23	10	11	13	16	10	11	20	14	15	22	11
A	h94	15	14-14	13	32	23	10	11	14	15	10	11	20	16	18	21	11
Mar	h95	17	11-14	13	32	25	10	11	13	14	11	10	20	16	16	23	12
Mar	h96	14	11-14	13	30	24	11	13	12	14	12	13	19	15	16	23	12
A	h97	16	14-15	13	31	24	11	11	13	15	10	12	19	15	17	22	11
Mar	h98	14	14-17	14	30	23	10	10	14	15	11	10	17	18	17	24	11
A	h99	14	13-20	14	30	22	9	11	12	15	9	10	20	15	16	23	11
A	h100	16	11-14	13	30	25	11	11	13	14	11	10	20	17	15	24	13
Mar	h101	14	11-15	13	28	24	10	14	12	15	12	12	19	15	16	23	12
E	h102	14	13-19	13	31	23	10	11	12	14	10	11	21	15	17	20	10
A	h103	14	13-20	13	30	23	10	11	12	14	10	12	20	16	17	20	11
C	h104	14	11-14	13	28	23	11	14	12	14	12	13	19	16	15	23	13

Regions	Sample	DYS 19	DYS 385	DYS389 I	DYS389 II	DYS390	DYS 391	DYS392	DYS 393	DYS437	DYS438	DYS 439	DYS448	DYS456	DYS 458	DYS635	Y GATA H4
A	h105	16	13-15	12	29	21	11	11	15	16	10	11	23	16	17	21	11
Med	h106	14	12-15	14	31	23	10	11	12	15	9	12	20	18	16	22	9
A	h107	13	17-18	13	30	24	10	11	13	14	11	11	21	17	15	22	11
Mar	h108	17	14-16	12	28	22	10	11	13	16	10	12	21	16	16	21	12
A	h109	13	14-15	14	30	24	9	11	13	14	10	10	20	15	19	21	12
A	h110	15	15-16	12	29	22	10	10	13	14	11	11	22	15	17	21	11
Med	h111	15	12-12	14	30	24	10	13	13	14	11	13	18	14	?	25	12
Mar	h112	16	11-14	13	30	25	11	11	13	14	11	10	20	16	15	23	11
Mar	h113	15	13-14	12	28	22	10	11	13	16	10	11	20	14	15	20	11
K	h114	14	13-16	13	30	23	10	11	12	15	7	11	19	16	15	23	12
Mar	h115	17	14-15	12	27	24	10	12	14	16	8	11	22	15	17	22	13
E	h116	15	11-14	14	30	26	11	11	13	14	12	10	20	16	15	23	13

The 17-loci Y-STR haplotypes of samples from Turkey

(A: Aegean Region, C: Central Anatolia Region, E: Eastern Anatolia Region, K: Black Sea Region, Mar: Marmara Region, Med: Mediterranean Region, S: Southern Eastern Region)

APPENDIX B

TÜRKİYE POPULASYONUNUN Y-STR ANALIZI

ANKET FORMU

1. Adı Soyadı:

2. Telefon:

3. E-mail:

4. Cinsiyet:

5. Memleket:

6. Doğum yeri:

7. Etnik köken:

8. Doğum yılı:

9. Ailede genetik hastalık var mı?

10. Akraba evliliği durumu?

11. Baba tarafından dedeleri

	Adı	Yaşadığı Yer	Yaklaşık Doğumyılı
Baba			
Dede 1			
Dede 2			
Dede 3			
Dede 4			
Dede 5			

Alınan numunenin sadece bu araştırmada kullanılmasına müsaade ediyorum.

İmza Tarih: