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Y-chromosome microsatellite markers (Y-STRs) provide valuable information in cases of rape and questioned paternity, and they allow for the genetic identification of males. The present study validated a Y-STR 10-plex on the ABI Prism® 3100 Genetic Analyzer for use in forensic and paternity laboratories at Orchid GeneScreen. Following optimization of the polymerase chain reaction, father-son pairs were analyzed to ensure that each pair generated identical haplotypes. The mutation rate varied between 0-0.0238 (+/- 0.046, 95% confidence interval). The present study demonstrated that the 10-plex is sensitive to 0.75 ng and that female samples mixed with male samples did not interfere with Y-STR haplotyping. A population database of 525 males was developed and subsequently analyzed. Three instances of locus multiplication were observed, two at DYS19 and one at DYS435. Overall haplotype diversity was 0.996, suggesting that the 10-plex can efficiently distinguish male Y-STR profiles.

VALIDATION OF A Y-CHROMOSOMAL SHORT TANDEM REPEAT 10-PLEX FOR USE IN FORENSIC AND PATERNITY LABORATORIES

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# VALIDATION OF A Y-CHROMOSOMAL SHORT TANDEM REPEAT 10-PLEX FOR USE IN FORENSIC AND PATERNITY LABORATORIES

INTERNSHIP PRACTICUM REPORT

Presented to the Graduate Council of the

Graduate School of Biomedical Sciences

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For the Degree of

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By

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## LIST OF ABBREVIATIONS

ABI 3100, ABI PRISM® 3100 Genetic Analyzer

AF, alleged father

ATCC, American Type Culture Collection

bp, base pair

BSA, bovine serum albumin

DAB, DNA Advisory Board

DNA, deoxyribonucleic acid

dNTP, deoxynucleotide triphosphate

DTT, dithiothreitol

NIST, National Institute of Standards and Technology

OD, optical density

PCR, polymerase chain reaction

RFU, relative fluorescence unit

SE Hispanic, Southeastern Hispanic

STR, short tandem repeat

UNTHSC, University of North Texas Health Science Center

Y-STR, Y-chromosome short tandem repeat

# CHAPTER 1

## INTRODUCTION

The basis of forensic DNA testing involves interpreting similarities and differences at genetic loci in an effort to determine if two samples may have originated from the same source. In paternity testing, differences at genetic markers between the alleged father and offspring lead to an exclusion of biological paternity (13). Among the genetic markers examined in forensic and paternity testing are microsatellites (short tandem repeats), which are tandemly repeated arrays of 2-7 base pair units of DNA. Their relatively high degree of polymorphism, ubiquitous dispersion in the human genome, and ease of use has resulted in the wide usage of short tandem repeats (STRs) in forensic genetic identification (1). Typically, the DNA markers used in forensics fall outside gene regions and, hence, do not code for proteins. Thus, these regions of the DNA are under decreased selective pressure and consequently exhibit a higher degree of polymorphism among individuals.

In recent years, STRs located on the Y-chromosome (Y-STRs) have attracted the interest of the forensic community. These STRs may be valuable in sexual assault cases, paternity cases (involving male offspring), and genealogical studies (3). Since the Y-chromosome is male-specific, the usage of Y-STRs in forensic evidence will allow for the genetic identification of the male component of a sample. Each individual male has

only one allele per locus, and thus, the genetic information derived from a panel of Y-STR markers is referred to as a haplotype. A haplotype refers to a DNA profile consisting of alleles at linked genes on a given chromosome. With the exception of mutational events, the Y-chromosome is inherited in an unchanged form from father to son(s), and for the most part does not undergo homologous recombination (5, 6). Because the individual Y-chromosome microsatellites are not independently inherited, the results from each locus cannot be combined using the product rule (5). Examining an increased number of Y-STR loci is, therefore, necessary to provide a higher degree of discrimination among males. However, the estimation of the allele and haplotype frequencies is limited by the size of the Y-STR database (9).

Polymerase chain reaction (PCR) allows more than one region of DNA to be concurrently amplified by adding more than one primer set to the reaction (5). The simultaneous amplification of two or more regions of DNA is referred to as multiplexing (5). Development of Y-STR multiplexes is advantageous for several reasons. By establishing multiplex systems, less template DNA and reagents are consumed, decreased labor is required, and the chances of introducing contamination and error are reduced (12). Currently, ReliaGene Technologies, Inc., of New Orleans, LA, is the only forensics laboratory that offers a commercially available Y-STR multiplex – their Y-Plex™6 Amplification Kit analyzes the following six loci: DYS393, DYS19, DYS389, DYS390, DYS391, and DYS385 (8). In contrast, the NIST (National Institute of Standards and Technology) Y-STR 10-plex examines ten loci on the Y-chromosome: DYS436,

DYS439, DYS435, DYS19[†], Y-GATA-A7.1[†], Y-GATA-H4, DYS391, DYS392, DYS438, and DYS437. Thus, the NIST Y-STR 10-plex provides an increased power of discrimination among male lineages. The NIST Y-STR 10-plex uses forward and reverse primer sets that reduce the overall lengths of the PCR amplicons. The smaller PCR amplicons in the NIST Y-STR 10-plex are more advantageous in the analysis of old or degraded samples frequently encountered in forensic casework.

Due to their increased power of discrimination, the more highly polymorphic genetic loci are attractive for forensic purposes. These polymorphic loci are continually evolving through a mutational process, and, thus, understanding the mutational processes and mutation rates for Y-STR systems is critical in correctly interpreting genetic profiles (13). Although understanding the mutation rate of Y-STRs is of less significance for a typical forensic comparison, it is of great importance in kinship studies and body identification. Slipped-strand mispairing appears to constitute the primary mechanism of mutation (10). During this mutational process, alleles of new lengths are produced, in which a gain of repeats occurs more frequently than a loss of repeats (10). In most cases of slipped-strand mispairing, the new allele length is only one repeat larger or smaller than the parent allele. Other mutational mechanisms include either a loss of a larger number of repeat units or a gain of a complete multi-repeat block of DNA, which includes flanking sequences (10). A mutational event involving insertion polymorphisms that results in additional allele formation has also been reported on various occasions (10,

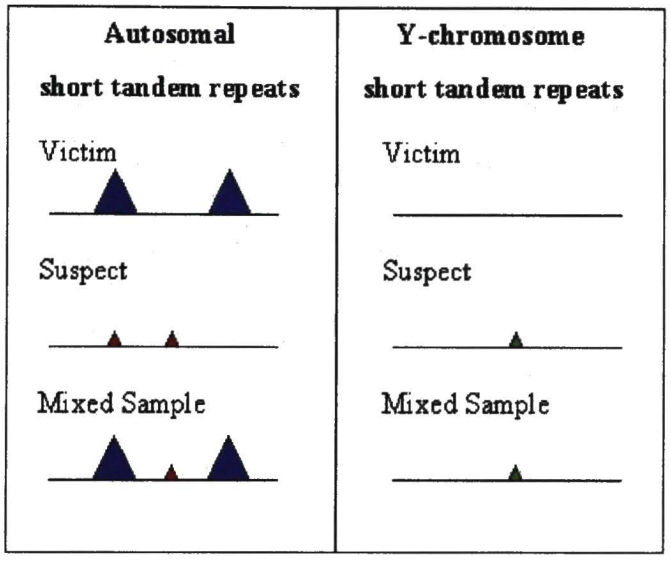
[†] In accordance with the International Society of Forensic Genetics guidelines, DYS19 and Y-GATA-A7.1 have been assigned "D" segment numbers DYS394 and DYS460, respectively (10, 12). However, the names DYS19 and A7.1 will be used throughout the remainder of this document.

13). Locus multiplication (such as allele duplication or triplication) has been demonstrated at several loci, including DYS19, DYS385, and DYS390 (10, 13).

The mutation rate of STR loci depends on the molecular structure of the genetic marker (13). Although the mutation rate of autosomal STRs is known to increase with longer repetitive arrays (tetranucleotide repeats compared to trinucleotide repeats, for example), no such correlation exists with Y-chromosomal STRs (13). However, there does appear to be a direct correlation between the Y-STR mutational rate and the number of repeating DNA units. The likelihood of a mutational event increases when the number of homogeneous repeats accumulates to $\geq$10 repeats (13). Kayser and Sajantila estimated the mutation rate of Y-chromosome microsatellites at up to eight mutations per one thousand father-son pairs (13). The number of mutations is dependent on the Y-STR locus, with an average of three per thousand father-son pairs (13).

The analysis of sexual assault evidence encompasses one of the major areas in which Y-STRs can provide useful information. In 1995, males constituted an estimated 99% of the sexual assault offenders (4), and, therefore, the NIST Y-STR 10-plex could frequently be used to generate a DNA profile from the suspect(s) in a rape case. The traditional method of extracting male DNA from sexual assault evidence involves performing a two-step differential extraction (2). This extraction procedure takes advantage of the fact that the male sperm cells are impervious to the detergents used to lyse epithelial cells. In the differential extraction process, the epithelial cells are first lysed, and then the sperm cells are isolated via centrifugation. The sperm cells can then be treated with a detergent containing dithiothreitol (DTT), which functions to break the

disulfide linkages found on the sperm heads, thus releasing the male DNA. However, because the amount of female DNA in sexual assault evidence is often overwhelming, it tends to mask the DNA profile of the male suspect (see below, left panel). Hence, using the male-specific Y-STRs is advantageous because it allow the analyst to examine the male component of the sexual assault evidence without interference from female DNA (see below, right panel).




Y-STR typing may also be applied to paternity or genealogical investigations involving male children in order to exclude or establish paternity or a paternal lineage. In deficiency paternity cases (those in which the father is unavailable for testing), one can generate a Y-STR profile from a male consanguineous relative (a grandfather or brother, for example) of the alleged father (7). Similarly, because of the direct transmission of the Y-chromosome from father to son(s) without modification (barring any mutations), blood relatives from the same paternal lineage possess the same Y-STR haplotype, thus making it possible to trace patrilineages back several generations (7).

Y-STR testing may also prove advantageous in cases in which there is an apparent male amelogenin deletion or mutation (9). If a deletion or mutation exists, a sample that originated from a male may appear female on an electropherogram since only the X-related amelogenin peak will be seen. In this situation, Y-STRs can help confirm the presence or absence of a Y-chromosome.

In conclusion, Y-STRs can provide valuable information for forensic and paternity laboratories. However, because the Y-chromosome is inherited without modification or recombination, there is decreased discrimination among males. This necessitates the examination of an increased number of Y-STR markers in order to more clearly distinguish among paternal lineages. The forensic community should encourage the research and discovery of additional Y-STRs.

# CHAPTER 2

## NATIONAL INSTITUTE OF STANDARDS AND TECHNOLOGY

## Y-STR 10-PLEX OVERVIEW

The United States National Institute of Standards and Technology (NIST) has developed a Y-chromosome short tandem repeat (Y-STR) 10-plex that analyzes the following loci: DYS436, DYS439, DYS435, DYS19, Y-GATA-A7.1, Y-GATA-H4, DYS391, DYS392, DYS438, and DYS437. The 10 loci examined include 7 tetranucleotides, 2 trinucleotides, and 1 pentanucleotide (Table 1). NIST has designed primer sets (Table 2) that produce polymerase chain reaction (PCR) amplicons that are $\leq$ 200 base pairs (bp) in length (14). The decreased amplicon lengths increase the likelihood of obtaining a complete Y-STR haplotype from degraded samples commonly encountered in forensic casework (14). The primers were tagged with either FAM™, TET, or HEX™ fluorescent dyes, and a ladder labeled with TAMRA™ served as an internal size standard (14). The PCR thermocycling parameters established at NIST are (14):

- 95° C for 10 minutes
- 25 cycles of 94° C for 1 minute, 55° C for 1 minute, and 72° C for 1 minute
- 60° C for 45 minutes

The PCR amplification was optimized for a 20 μL reaction volume, which included 2 units AmpliTaq Gold®, 2 mM $MgCl_2$, 5-20 pmol of each primer, and 5-25 ng template DNA (14). When initially developed, the 10-plex system was designed for usage on the ABI PRISM® 310 Genetic Analyzer.

The aim of the current study was to begin validating the NIST Y-STR 10-plex on the ABI PRISM® 3100 Genetic Analyzer for use at Orchid GeneScreen Dallas. The primers were resynthesized using FAM™, VIC™, and NED™ labels, and GeneScan™-500 LIZ™ served as the internal size standard. The steps involved in the validation were:

1. Verify the conditions of the PCR multiplex reaction.
2. Validate the PCR multiplex conditions with ~40 father-son pairs to ensure the Y-STR 10-plex is producing the expected results.
3. Develop a database of Y-STR haplotypes using DNA samples from Caucasian, African American, and Southeastern Hispanic males.
4. Begin validating the NIST Y-STR 10-plex for forensic casework.

# CHAPTER 3

# MATERIALS AND METHODS

## *3.1 DNA Samples*

The control DNA samples used include the male DNA extract ATCC 45514 (American Type Culture Collection, Manassas, VA) and female extracts K562 (Applied Biosystems, Foster City, CA) and 9947A (Promega, Madison, WI). "BS #1" and "JR #1" DNA refer to DNA extracts obtained from male volunteers.

The paternity laboratory at Orchid GeneScreen Dallas provided male samples from previously characterized parentage analyses for the analysis of father-son pairs and development of a database. The population samples were obtained from unrelated males of Caucasian, African American, and Hispanic origin. DNA was extracted from buccal swabs using the FasTract procedure, a proprietary method of Orchid GeneScreen Dallas.

## *3.2 Polymerase Chain Reaction (PCR)*

Ten Y-chromosome microsatellites were amplified in one PCR multiplex. The multiplex examined the following loci: DYS436, DYS439, DYS435, DYS19, Y-GATA-A7.1, Y-GATA-H4, DYS391, DYS392, DYS438, and DYS437.

The samples were amplified on the Perkin Elmer GeneAmp® PCR System 2400, PE Applied Biosystems 9700, or Applied Biosystems 2700 thermocycler (Applied Biosystems, Foster City, CA). The PCR thermocycling parameters were as follows:

- 95° C for 10 minutes
- 25 cycles of 94° C for 1 minute, 60° C for 1 minute, and 72° C for 1 minute
- 60° C extension for 50 minutes

The PCR was performed in a total reaction volume of 20 μL. The PCR reaction mix contained 2.0 mM $MgCl_2$, 1x GeneAmp® PCR Gold buffer (10x buffer contains 500 mM KCl and 100 mM Tris-HCl, pH 8.0), and 2.5 units AmpliTaq Gold® DNA polymerase (Applied Biosystems, Foster City, CA). The master mix also contained 250 μM of each GeneAmp® dNTP (Applied Biosystems, Foster City, CA), 22% weight per volume stock solution of bovine serum albumin, and primers ranging in concentration from 0.2-1.6 μM. The primers, synthesized by Applied Biosystems, were tagged with one of three dyes: FAM™, VIC™, or NED™. GeneScan™-500 LIZ™ (Applied Biosystems, Foster City, CA) served as the internal size standard. The amount of DNA added to each reaction was 2.5 ng unless otherwise stated, or 2.5 μL in the case of the father-son pairs and database samples.

### *3.3 Capillary Electrophoresis (CE)*

PCR amplification products were subjected to capillary electrophoresis on the ABI PRISM® 3100 Genetic Analyzer (Applied Biosystems, Foster City, CA). Sample

preparation was as follows: 9.3 μL Hi-Di™ formamide (Applied Biosystems, Foster City, CA), 0.2 μL GeneScan™-500 LIZ™ Size Standard (Applied Biosystems, Foster City, CA), and 0.5 μL PCR product per sample. The samples were denatured for 5 minutes at 95° C and subsequently snap-cooled on ice for 5 minutes. They were run on the ABI PRISM® 3100 Genetic Analyzer using dye set G5 and the "GeneScan36_POP4 Default Module". Following capillary electrophoresis, the samples were analyzed using GeneScan® Analysis (minimum of 100 RFU [relative fluorescence units] peak heights were called) and Genotyper® version 2.5.2 software (Applied Biosystems, Foster City, CA).

### *3.4 Statistical Analysis*

All alleles were reported in base pair sizes and were rounded to a whole number for purposes of analysis. Allele frequencies were calculated for each locus using the formula $p_i$= # *occurrences/n*, where $p_i$ is the allelic frequency and *n* represents the sample size. Gene diversity, *D*, of each locus was computed using the formula $D=(n/n-1)(1-p_i^2)$ (12). Haplotype diversity (*HD*) was calculated with the same equation using haplotype frequencies rather than allele frequencies. For the Y-chromosome, haplotype diversity identically corresponds with the power of discrimination (12). The number of haplotype occurrences and their relative frequencies were calculated using Arlequin software (20). Similarly, the tables of significant linkage disequilibrium (significance level of 0.05) were generated by Arlequin using exact tests. The mutation rate was calculated using the formula # *of mutations/total number of father-son pairs*. The 95% confidence intervals

were calculated using the equation $p+/-1.96(pq/n)^{1/2}$, where $p$ is calculated as $x/n$ ($n$ represents the sample size and $x$ represents the number of observations) and $q$ is calculated as $1-p$.

### *3.5 Trademarks*

ABI PRISM, GeneScan, and Genotyper are registered trademarks, and FAM, HEX, LIZ, NED, VIC, and Hi-Di are trademarks of Applera Corporation or its subsidiaries in the United States and certain other countries. AmpliTaq and GeneAmp are both registered trademarks of Roche Molecular Systems, Inc.

# CHAPTER 4

## INTERNSHIP PRACTICUM DAILY LOG

**February 22, 2002**

- 10:00 AM Meeting with Dr. Rick Staub, Judy Floyd, and Joe Warren

  The goal of the internship project is to validate a 10-plex for Y-STRs (short tandem repeats)

  The steps involved include:

  1. Verify the amplification conditions [balance the reaction using known, quantitated DNA]
     - Tentative deadline = 3-8-02
  2. Validate the PCR (polymerase chain reaction) amplification reaction set-up with 40 father:son pairs
     - Tentative deadline = 3-29-02
  3. Database samples
     - Obtain Y-STR profiles from ~200 males each of Caucasians, African Americans, and Hispanics
     - The haplotype results from the fathers in step 2 can be included.
     - Tentative deadline = 4-30-02
  4. Validation for forensic casework (DAB validation study)

- Perform Y-STR analysis on mixed samples (male:male and female:male mixtures), and on samples isolated from different sources [ex: blood, buccal, stains, etc]. Sensitivity and male specificity assays will also be performed.
- Tentative deadline = 5-10-02

Future goals for the Y-STR project include:

* Develop or find an allelic ladder or standard male reference to compare other haplotypes to.
    - Check on the internet for Leiden University regarding YSTR ladders. In order for the ladder to be of use, the loci and primers must be identical to those that GeneScreen utilizes.
    - Talk with John Butler at NIST to see if he has developed an allelic ladder from him, even if it isn't a complete ladder.
    - Tentative deadline = 3-22-02
* Modify the Genotyper template with the help of Joe Warren at GeneScreen.
    - Check the STRBase for downloads of Genotyper macros
    - Tentative deadline = 3-8-02

- In this study, the 2400 thermocyclers will primarily be used for amplifications.

- This project is a continuation of the work done by Joseph E. Warren, Ph.D. Dr. Warren was working on the ABI 377 using primers tagged with the following dyes: FAM, HEX, TET, and TAMRA (internal size standard)

**February 23, 2002**

- An internet search for Leiden University (a link from STRBase) and allelic ladders was performed.

  Alleles in the Leiden University ladder include:

  DYS19,DYS388, DYS389, DYS390, DYS393, DYS391, DYS392

  I did not find any Genotyper macros for download on STRBase

**February 25, 2002**

- Experiment #1: SENSITIVITY TEST USING 9947A
  - Purpose: Determine the sensitivity of the PCR conditions established by Dr. John Butler by adding varying amounts (0-1.0 ng) of control DNA (ABI 9947A, 0.1 ng/μL stock) to the amplification reaction.
  - Quantities of 9947A tested: 0 ng, 0.1 ng, 0.25 ng, 0.5 ng, 0.75 ng, 1 ng
  - For each amplification reaction, I added 10 μL of 9947A DNA of varying concentrations. I made a 100 μL stock of each:

    - 0 ng/μL → 0 μL DNA + 100 μL water
    - 0.01 ng/μL → 10 μL DNA + 90 μL water
    - 0.025 ng/μL → 25 μL DNA + 75 μL water

- 0.05 ng/μL → 50 μL DNA + 50 μL water

- 0.075 ng/μL → 75 μL DNA + 25 μL water

- 0.1 ng/μL → 100 μL DNA + 0 μL water

Example calculation: to make 0.075 ng/μL DNA → $V_1C_1 = V_2C_2$

$(V_1)(0.1\ ng/\mu L) = (100\ \mu L)(0.075\ ng/\mu L) = 75\ \mu L\ DNA$

- Amplifications:
  - Tubes #1, 2 = 0 ng DNA
  - Tubes #3, 4 = 0.1 ng DNA
  - Tubes #5, 6 = 0.25 ng DNA
  - Tubes #7, 8 = 0.5 ng DNA
  - Tubes #9, 10 = 0.75 ng DNA
  - Tubes #11, 12 = 1.0 ng DNA

- A 50 μM stock of Y392 primers was made.

**February 26, 2002**

- After conversations with Dr. Rick Staub and Joe Warren, the decision was made to do the Y-STR project on the Applied Biosystems (ABI) 3100 rather than the ABI 377.

**February 28, 2002**

- ABI does not support dye set C (HEX, FAM, TET, and TAMRA) on the ABI 3100, and therefore does not want to provide GeneScreen with the matrix

standards. I will try to get the matrix standards through the University of North Texas Health Science Center (UNTHSC).

- 12:30 PM Samples #1-12 from the sensitivity test using ABI 9947A DNA (amplified on 2-25-02) were loaded onto the ABI 377.
    - Sample sheet name: "CJ Expt #1 2-27-02"
    - Run name: "CJ Expt #1 2-27-02"
- To prepare samples for loading onto the ABI 377:
    1. Make a solution of FLS (formamide loading solution)

        5 μL HiDi formamide + 1 μL loading buffer per sample
    2. Mix together 5 μL FLS + 1 μL TAMRA per sample
    3. Mix together 3 μL amplified sample and 3 μL FLS/TAMRA solution
    4. Load 0.7 μL of prepared sample into the loading tray, skipping wells between each sample.
    5. The gel runs for 2.5 hours using filter set C
- Results of Experiment #1: No bands were detected during the GeneScan analysis of the gel. This is because ABI 9947A is *female*, rather than male DNA.
- Because the TAMRA looked very faint on the gel image, from now on, I will use 2 μL of TAMRA per sample if running samples on the ABI 377.

**March 1, 2002**

- I looked up information on standard reference male DNA

- The positive control used by ReliaGene Technologies, Inc in their Y-Plex™6 Amplification Kit is ATCC #45514.
- John Butler's presentation (link on STRBase) mentioned SRM 2395 as a Y-chromosome standard reference. This is going to be a new release in 2002.

**March 4, 2002**

- I looked into the usage of dye set C matrix standards for the ABI 3100. A representative from ABI said that catalog part number #401546 (fluorescent amedite standards) should work on the ABI 3100, but ABI technical support says they will not work on the 3100, but will work on the 310. I will try running the matrix standards on the ABI 3100 to determine if the matrix can be made successfully.

**March 5, 2002**

- Experiment #2: SENSITIVITY TEST USING MALE DNA (JR#1, 5 ng/μL stock):
  - Purpose: Determine the sensitivity of the PCR conditions established by Dr. John Butler by adding varying amounts (0-1.0 ng) of male DNA (JR #1) to the amplification reaction.
  - Made 50 μL of 1 ng/μL DNA from the 5 ng/μL stock JR #1 DNA (10 μL of 5 ng/μL + 40 μL water)
  - Made 250 μL of 0.1 ng/μL DNA from the 1 ng/μL
  - Using JR's 0.1 ng/μL DNA, I made 50 μL solutions of 0.01 ng/μL, 0.025 ng/μL, 0.05 ng/μL, and 0.075 ng/μL DNA

Example calculation: to make 0.075 ng/μL DNA → $V_1C_1 = V_2C_2$

$(V_1)(0.1\ ng/\mu L) = (50\ \mu L)(0.075\ ng/\mu L) = 37.5\ \mu L$ DNA

- The samples were amplified in duplicates in orange tubes (labeled as: #1-12, 3-5-02 CJ) using 10 μL each DNA
- Amplifications:
  - Tubes #1, 2 = 0 ng DNA
  - Tubes #3, 4 = 0.1 ng DNA
  - Tubes #5, 6 = 0.25 ng DNA
  - Tubes #7, 8 = 0.5 ng DNA
  - Tubes #9, 10 = 0.75 ng DNA
  - Tubes #11, 12 = 1.0 ng DNA

- I ran the fluorescent amedite standards (ABI #401546) on the ABI 3100. The spectral calibration failed. Some of the peaks were overlapping, and peak heights were ~8000-9000 RFUs (relative fluorescent units), with the exception of the TAMRA. It is possible that the HEX, TET, and 6-FAM need to be diluted in order for the matrix to be made successfully.

**March 7, 2002**

- I reran the fluorescent amedite standards (ABI #401546) on the ABI 3100, but diluted the dyes to decrease the intensity of the RFU values.
  - Preparation of the matrix standards: 197 μL formamide, 1.25 μL TAMRA, 0.625 μL each of TET, HEX, and 6-FAM.

- The spectral calibration failed, and the ABI technical support was not able to offer any suggestions as to how to improve the matrix calibration.

- Experiment #2 samples #1-12 (amplified on 3-5-02) were given to the paternity lab for them to run on the Spectrumedix.

**March 11, 2002**

- The Y-STR primers were reordered with new dyes (the same dyes as used with the Identifiler kit) that will work on the ABI 3100. The new dye labels include FAM, VIC, NED, and LIZ as the internal size standard.

**March 12, 2002**

- I looked at the results from my Experiment #2 samples (#1-12) that were run on the Spectrumedix. The TAMRA peaks were clearly visible, but I was unable to detect any additional peaks.

**March 14, 2002**

- I attended the ABI 3100 training at the UNTHSC.

**March 15, 2002**

- I attended the ABI 3100 training at the UNTHSC.

**March 18, 2002**

- Standard male reference DNA (ATCC 45514) was ordered through Suzanne Constance at Orchid. This is the same control DNA used by ReliaGene.

**March 20, 2002**

- The new primers arrived and I diluted them to ~200 μM.

  (Example calculation: 72,000 pmol = 72,000 μM/μL; 100 μM = 72,000/720 μL water; 200 μM = 72,000/360 μL water)

- I read the OD values of all primers (newly labeled primers as well as the original primers) at an absorbance of 260 nm. The OD values were all within an acceptable range with the exception of the A7.1 forward primer. It is possible that the low OD values for the A7.1 forward primer is caused by degradation of the DNA. Therefore, the A7.1 forward primer was reordered from Applied Biosystems.

- Based on the OD readings, I diluted the new primer sets (except A7.1) and made a 50 μM stock solution of each.

**March 22, 2002**

- The ordering process through ATCC is much more extensive than originally thought and requires going through a lengthy application process. Therefore, at this time the ATCC #45514 was not ordered. The UNTHSC has a ReliaGene Y-Plex™6 Amplification Kit containing the positive control, ATCC #45514. I will use this

positive control at GeneScreen for the Y-STR project until additional DNA can be ordered.

**March 25, 2002**

- Experiment #3: SIMPLEXING WITH Y-STR PRIMERS
    - Purpose: To amplify 1 ng of positive control DNA (ATCC #45514) using the individual primer sets. This will ensure that the primers function properly by themselves before multiplexing them.
    - Amplifications: Each reaction contained 1 ng of standard reference male DNA (ATCC #45514)
        - Tubes #3-1, 3-11 = Y435
        - Tubes #3-2, 3-12 = Y19
        - Tubes #3-3, 3-13, 3-20 = Y437
        - Tubes #3-4, 3-14, 3-21 = Y392
        - Tubes #3-5, 3-15, 3-22 = H4
        - Tubes #3-6, 3-16, 3-23 = Y438
        - Tubes #3-7, 3-17, 3-24 = Y439
        - Tubes #3-8, 3-18 = Y436
        - Tubes #3-9, 3-19 = Y391
        - Tube #3-10 = negative control (blank)

- Each sample, with the exception of the negative control, was amplified with the same primer concentration as would be used if the system was being multiplexed. The negative control contained all primers (except A7.1).

**March 26, 2002**

- The A7.1 forward primer arrived from ABI. I diluted the primer to ~200 μM (calculation: 127,000 pmol/635 μL water = 200 μM). After taking the OD reading of the primer (A-260 = 0.240), a 50 μM stock of the A7.1 primer set was made.
- Experiment #4: SIMPLEXING WITH A7.1 PRIMERS AND AMPLIFICATION OF PATERNITY SAMPLE #185989
    - Purpose: To amplify 1 ng of positive control DNA (ATCC #45514) using the A7.1 primer set. This will ensure that the primers function properly by themselves before multiplexing them.

      Paternity sample #185989 will be amplified with all 10 Y-STR primers using 10 μL DNA.
    - Amplifications:
        - Tube #4-1, 4-2 = A7.1 (1ng ATCC #45514)
        - Tube #4-3 = multiplexing of paternity sample #185989AF (10 μL DNA)
        - Tube #4-4 = multiplexing of 1 ng ATCC #45514
        - Tube #4-5 = negative control multiplex

**March 28, 2002**

- Meeting with Dr. Rick Staub and Joe Warren:
  - John Butler is willing to provide GeneScreen with the ABI 3100 base pair typings of ATCC #45514.
  - Toshimichi Yamamoto from the Department of Legal Medicine and Bioethics, Postgraduate School of Medicine, Nagoya University has developed a ladder for the 10-plex of Y-STRs. I will try to contact him to determine what he uses as a positive control and what the typings are for the 10 systems.

**March 29, 2002**

- Samples from Experiments #3 and #4 were loaded onto the ABI 3100. Sample preparation guidelines were as follows:
  - Mix the following for a final volume of 10 μL per sample: 1 μL PCR product, 0.3 μL GeneScan-500 LIZ size standard, and 8.7 μL Hi-Di formamide
  - Denature the samples at 95° C for 5 minutes and then immediately place the samples on ice for 3 minutes.
  - The samples were run using "Dye Set G5" and "GeneScan36_POP4default module".
  - Results from Experiments #3 and #4: The peaks on the simplex reactions seemed very low, possibly because there was not enough DNA in the PCR reactions. The paternity sample (#185989 AF) had numerous peaks, leading

me to believe that the sample is indeed male, yet because of the high baselines, analysis was difficult.

**April 1, 2002**

- Experiment #5: SENSITIVITY TEST (ATCC 45514, BS #1, K562)
    - Purpose: Determine the sensitivity of the PCR conditions established by Dr. John Butler by adding varying amounts (1.0-5.0 ng) of male DNA (ATCC 45514 or BS #1) to the amplification reaction. Female DNA (K562) will be used as a negative control.
    - Amplifications:
        - Tube #5-1 = 1 ng ATCC 45514
        - Tube #5-2 = 2.5 ng ATCC 45514
        - Tube #5-3 = 5 ng ATCC 45514
        - Tube #5-4 = 1 ng BS #1
        - Tube #5-5 = 2.5 ng BS #1
        - Tube #5-6 = 5 ng BS #1
        - Tube #5-7 = blank (10 μL water)
        - Tube #5-8 = 1 ng K562
        - Tube #5-9 = 2.5 ng K562
        - Tube #5-10 = 5 ng K562

**April 2, 2002**

- Samples from Experiment #5, as well as samples #4-3 and #4-4, were loaded onto the ABI 3100. Varying amounts of PCR product, ranging from 0.1 μL to 1.0 μL, were loaded. This was done to see if lowering the amount of PCR product added to each reaction helped to decrease the baseline levels.
    - Sample preparation for loading 1 μL PCR product onto the ABI 3100:

      Per sample = 1 μL PCR product, 0.3 μL GeneScan-500 LIZ size standard, 8.7 μL Hi-Di formamide
    - Sample preparation for loading 0.5 μL PCR product onto the ABI 3100:

      Per sample = 0.5 μL PCR product, 0.3 μL GeneScan-500 LIZ size standard, 9.2 μL Hi-Di formamide
    - Sample preparation for loading 0.1 μL PCR product onto the ABI 3100:

      Per sample = 0.1 μL PCR product, 0.3 μL GeneScan-500 LIZ size standard, 9.6 μL Hi-Di formamide
- Results from Experiment #5: The optimal amount of DNA to use in the PCR reaction appears to be ~2.5 ng. Therefore, unless otherwise noted, I will use 2.5 ng DNA in future experiments. Loading 0.5 μL versus 1 μL PCR product onto the ABI 3100 gives comparable results. Thus, in future experiments, I will use 0.5 μL PCR product. The LIZ peaks are plenty high, and therefore, from now on, I will use 0.2 μL LIZ during sample preparation for the ABI 3100. See electropherograms for more detailed results.

**April 4, 2002**

- Experiment #6: INCORPORATION OF BSA INTO THE PCR REACTION
  - Purpose: To determine if the addition of BSA (bovine serum albumin) to the PCR reaction mix helps to decrease the baseline levels observed on the ABI 3100.
  - Amplifications:
    - Tube #6-1 = 2.5 ng ATCC 45514
    - Tube #6-2 = 2.5 ng BS #1
    - Tube #6-3 = paternity sample #185989 AF (10 μL DNA)
    - Tube #6-4 = 2.5 ng K562
    - Tube #6-5 = blank (10 μL water)

**April 5, 2002**

- A new spectral calibration for dye set G5 was performed on the ABI 3100.
- Samples from Experiment #6, as well as samples #4-3, #5-2, #5-5, and #5-9 were loaded onto the ABI 3100. Sample preparation was as follows:

  0.5 μL PCR product, 0.2 μL GeneScan-500 LIZ size standard, and 9.3 μL Hi-Di formamide per sample
- Results from Experiment #6: The electropherograms looked much cleaner and the baseline levels were improved. Therefore, I will continue to add BSA to the PCR reaction mix. Several extraneous peaks were observed in the blank and female control samples. This is potentially due to a relatively low annealing temperature (55° C).

**April 8, 2002**

- Experiment #7: EFFECT OF ANNEALING TEMPERATURE ON PCR ARTIFACTS
  - Purpose: To determine if raising the annealing temperature (compared to John Butler's annealing temperature of 55° C) helps to decrease the peak heights and/or number of PCR artifacts.
  - Amplifications:
    - Tube #7-1 = BS #1 (2.5 ng), 57° C
    - Tube #7-2 = K562 (2.5 ng), 57° C
    - Tube #7-3 = blank, 57° C
    - Tube #7-4 = BS #1 (2.5 ng), 59° C
    - Tube #7-5 = K562 (2.5 ng), 59° C
    - Tube #7-6 = blank, 59° C
    - Tube #7-7 = BS #1 (2.5 ng), 56° C
    - Tube #7-8 = K562 (2.5 ng), 56° C
    - Tube #7-9 = blank, 56° C
    - Tube #7-10 = BS #1 (2.5 ng), 58° C
    - Tube #7-11 = K562 (2.5 ng), 58° C
    - Tube #7-12 = blank, 58° C
  - Results from Experiment #7: Using an annealing temperature of 59° C appears to work better than 56° C, 57° C, or 58° C, in that the expected Y-

STR peaks are clearly identifiable and the artifact peaks on the electropherograms are minimized.

**April 9, 2002**

- Experiment #8: EFFECT OF MAGNESIUM CONCENTRATION AND ANNEALING TEMPERATURE
  - Purpose: To determine if increasing both the magnesium concentration and annealing temperature helps decrease the observed artifacts.
  - Amplifications:
    - Tube #8-1 = 2.5 ng BS #1, 59° C annealing temperature, 2.5 mM $MgCl_2$
    - Tube #8-2 = 2.5 ng K562, 59° C annealing temperature, 2.5 mM $MgCl_2$
    - Tube #8-3 = blank, 59° C annealing temperature, 2.5 mM $MgCl_2$
    - Tube #8-4 = 2.5 ng BS #1, 60° C annealing temperature, 2.5 mM $MgCl_2$
    - Tube #8-5 = 2.5 ng K562, 60° C annealing temperature, 2.5 mM $MgCl_2$
    - Tube #8-6 = blank, 60° C annealing temperature, 2.5 mM $MgCl_2$

**April 12, 2002**

- Samples from Experiment #8 were loaded onto the ABI 3100.
- Results from Experiment #8: Increasing both the magnesium concentration to 2.5 mM and the annealing temperature got rid of the majority of the nonspecific peaks previously seen. One nonspecific peak of significant height still appears at ~95-96

base pairs with the NED dye. I will further increase both the magnesium concentration and the annealing temperature in attempt to get rid of this peak.

**April 16, 2002**

- Experiment #9: 60° C AND 61° C ANNEALING TEMPERATURES WITH 2.5mM or 2.75 mM MAGNESIUM
  - Purpose: To determine if increasing the magnesium concentrations and annealing temperature helps to get rid of the nonspecific band observed at ~95-96 base pairs with the NED dye.
  - Amplifications (using 2.5 ng DNA):
    - Tube #9-1 = BS #1, 60° C, 2.5 mM $MgCl_2$
    - Tube #9-2 = K562, 60° C, 2.5 mM $MgCl_2$
    - Tube #9-3 = blank, 60° C, 2.5 mM $MgCl_2$
    - Tube #9-4 = BS #1, 60° C, 2.75 mM $MgCl_2$
    - Tube #9-5 = K562, 60° C, 2.75 mM $MgCl_2$
    - Tube #9-6 = blank, 60° C, 2.75 mM $MgCl_2$
    - Tube #9-7 = BS #1, 61° C, 2.5 mM $MgCl_2$
    - Tube #9-8 = K562, 61° C, 2.5 mM $MgCl_2$
    - Tube #9-9 = blank, 61° C, 2.5 mM $MgCl_2$
    - Tube #9-10 = BS #1, 61° C, 2.75 mM $MgCl_2$
    - Tube #9-11 = K562, 61° C, 2.75 mM $MgCl_2$

- Tube #9-12 = blank, 61° C, 2.75 mM $MgCl_2$

**April 19, 2002**

- Samples from Experiment #9 were run on the ABI 3100.
- Results from Experiment #9: The results with 60° C versus 61° C and 2.5mM $MgCl_2$ versus 2.75 mM $MgCl_2$ seemed relatively comparable. Some artifacts, namely at ~95 base pairs with NED, still remain. I will continue to use a 60° C annealing temperature unless otherwise noted.
- Experiment #10: SENSITIVITY TEST USING 2.75 mM MAGESIUM AND 60° C; TEST PATERNITY SAMPLE #187816 AF.
    - Purpose: To test the sensitivity of the Y-STR 10-plex using varying amounts of DNA (from 0.25-2.5 ng). I will also run paternity sample #187816 to determine if it came from a male.
    - Amplifications:
        - Tube #10-1 = 2.5 ng BS#1 male DNA
        - Tube #10-2 = 2 ng BS#1 male DNA
        - Tube #10-3 = 1.5 ng BS#1 male DNA
        - Tube #10-4 = 1 ng BS#1 male DNA
        - Tube #10-5 = 0.75 ng BS#1 male DNA
        - Tube #10-6 = 0.5 ng BS#1 male DNA
        - Tube #10-7 = 0.25 ng BS#1 male DNA
        - Tube #10-8 = paternity sample #187816 AF (5 µL DNA)

- Tube #10-9 = 2.5 ng K562 female DNA
- Tube #10-10 = blank

**April 22, 2002**

- Samples from Experiment #10 were loaded onto the ABI 3100. For tubes #10-1 to #10-10, 0.5 μL of each sample was loaded, as well as 1 μL of samples #10-4 to #10-9.
- Results from Experiment #10: The ten Y-STR systems were visible down to 0.75 ng male DNA, yet some of the systems had RFU (relative fluorescent units) that were quite low.
- Experiment #11: EFFECT OF MAGNESIUM CONCENTRATION USING AMPLIFIED AND NON-AMPLIFIED SAMPLES
  - Purpose: To determine what effect magnesium concentration has on extraneous bands observed in male, female, and blank samples. In order to determine if these bands are due to a problem in the PCR amplification process, some samples will be amplified whereas others will not be amplified.
  - Amplifications (using 2.5 ng DNA and 60° C annealing temperature):

    Samples #11-1 to #11-12 were amplified. Samples #11-13 to #11-21 were immediately placed at 4° C following PCR set-up.

    - Tube #11-1, #11-13 = BS#1 male DNA, 1.5 mM $MgCl_2$
    - Tube #11-2 = ATCC 45514, 1.5 mM $MgCl_2$
    - Tube #11-3, #11-14 = K562, 1.5 mM $MgCl_2$

- Tube #11-4, #11-15 = blank, 1.5 mM $MgCl_2$
- Tube #11-5, #11-16 = BS#1 male DNA, 2 mM $MgCl_2$
- Tube #11-6 = ATCC 45514, 2 mM $MgCl_2$
- Tube #11-7, #11-17 = K562, 2 mM $MgCl_2$
- Tube #11-8, #11-18 = blank, 2 mM $MgCl_2$
- Tube #11-9, #11-19 = BS#1 male DNA, 2.5 mM $MgCl_2$
- Tube #11-10 = ATCC 45514, 2.5 mM $MgCl_2$
- Tube #11-11, #11-20 = K562, 2.5 mM $MgCl_2$
- Tube #11-12, #11-21 = blank, 2.5 mM $MgCl_2$

**April 23, 2002**

- Samples from Experiment #11 were loaded onto the ABI 3100.
- Results from Experiment #11: The ~95 base pair band seen in the NED loci appeared in both the amplified and non-amplified samples, regardless of what magnesium concentration was used. This suggests that the 95 base pair band is not appearing as a result of the amplification process, but instead may be an impurity in one of the primer sets. I did not observe any significant differences when using 2.0 versus 2.5 mM $MgCl_2$. Therefore, I will use the lower $MgCl_2$ concentration unless otherwise noted.
- After reviewing the results of Experiment #11, I looked back to the results from Experiment #3 in which I performed simplex reactions of the ten Y-STR loci. The electropherograms from simplexes of Y392, Y437, and Y438 loci all showed the 95

base pair peak. This suggests that the 95 base pair peak may be occurring because of something inherent to the NED dye rather than an impurity in one of the primer sets.

- The status of the Y-STR 10-plex research was discussed with representatives from Cellmark Germantown.

**April 24, 2002**

- Experiment #12: SIMPLEX REACTIONS WITH NED LOCI; INCREASING THE Y19 PRIMER CONCENTRATIONS
  - Purpose: The Y392, Y437, and Y438 loci will be amplified individually to determine the origin of the non-specific 95 base pair fragment. Additionally, the Y19 primer concentration will be increased in an effort to increase the peak height.
  - Amplifications (using 2.5 ng DNA, 1.5 mM $MgCl_2$, and 60° C annealing temperatures):

    Samples #12-1 to #12-12 were amplified, but samples #12-13 to #12-24 were not amplified. Samples #12-1 to #12-9, and samples #12-13 to #12-21 were simplex reactions. Samples #12-10 to #12-12 were multiplex reactions using an increased concentration of the Y19 primers (1.2 μM).

    - Tube #12-1, #12-13 = Y392 locus → BS#1 male DNA
    - Tube #12-2, #12-14 = Y392 locus → K562
    - Tube #12-3, #12-15 = Y392 locus → blank
    - Tube #12-4, #12-16 = Y437 locus → BS#1 male DNA

- Tube #12-5, #12-17 = Y437 locus → K562
- Tube #12-6, #12-18 = Y437 locus → blank
- Tube #12-7, #12-19 = Y438 locus → BS#1 male DNA
- Tube #12-8, #12-20 = Y438 locus → K562
- Tube #12-9, #12-21 = Y438 locus → blank
- Tube #12-10 = BS#1 male DNA
- Tube #12-11 = K562
- Tube #12-12 = blank
- Tube #12-22 = Y392 primer (0.95 μL) + water (79.05 μL)
- Tube #12-23 = Y437 primer (0.5 μL) + water (79.5 μL)
- Tube #12-24 = Y438 primer (0.5 μL) + water (79.5 μL)

**April 26, 2002**

- Samples from Experiment #12 were run on the ABI 3100.
- Results from Experiment #12: The ~95 base pair peak was seen in all amplified and non-amplified samples from the Y392 and Y438 loci, and to a lesser degree in the Y437 locus.

**April 29, 2002**

- Experiment #13: ALLEGED FATHER (AF) SAMPLES USING 2.5 μL DNA; INCREASING Y19 CONCENTRATION (1.2 μM AND 1.6 μM); PATERNITY SAMPLE #188926 AF

- Purpose: 1) To obtain Y-STR haplotypes from AF samples
  2) To determine if increasing the Y19 primer concentration helps to increase peak height.
  3) To determine if paternity sample #188926 AF originated from a male.
- Amplifications:
  - Samples #13-1 to 13-24 are AF samples
  - Sample #13-25 is paternity sample #188926 AF
  - Samples #13-26 to #13-31 test different concentrations of the Y19 primer.

**April 30, 2002**

- Samples from Experiment #13 were loaded onto the ABI 3100.
- Results from Experiment #13: Results from the AF samples were varied. I was able to obtain haplotypes on some samples, but not on others. In many cases, all loci worked well except Y19. However, increasing the Y19 primer concentration did seem to increase the Y19 peak height.

**May 1, 2002**

- Experiment #14: VALIDATION USING FATHER:SON PAIRS, CONTINUED
  - Purpose: To validate the Y-STR 10-plex using father:son pairs. I will reamplify many of the AF samples that didn't work from Experiment #13, as well as numerous son samples.

- Amplifications:
  - Samples #14-1 to #14-20 were son (C) samples
  - Samples #14-21 to #14-32 were AF samples

**May 2, 2002**

- Samples from Experiment #14 were run on the ABI 3100.
- Results from Experiment #14: The haplotype from each son's sample matched the haplotype from the respective AF sample.

**May 3, 2002**

- Experiment #15: FATHER:SON PAIRS AND DATABASING
  - Purpose: To compare haplotypes between fathers and sons and to begin databasing male Caucasian samples.
  - Amplifications (60° C, 2 mM $MgCl_2$):
    - Samples were amplified in a 96-well tray.
    - Wells #A1 through #G2 were father:son pairs using 2.5 µL DNA.
    - Wells #G3 through #H9 were male Caucasian samples using 2.5 µL DNA.
    - Well #H10 contained 2.5 ng BS#1 male DNA
    - Well #H11 contained 2.5 ng K562
    - Well #H12 served as a blank sample.

**May 6, 2002**

- Samples #A1 through #D12 from Experiment #15 were loaded on the ABI 3100.
- Results from Experiment #15: Essentially all of the samples amplified at all 10 loci. Because of the high degree of success, I will continue to use 2.5 μL DNA, 2.0 mM $MgCl_2$, and 60° C annealing temperatures for the remainder of the databasing.

**May 7, 2002**

- Samples #E1 through #H12 from Experiment #15 were loaded on the ABI 3100.
- Results from Experiment #15, continued: Virtually all of the samples amplified at all ten Y-STR loci. The haplotypes of the sons matched the haplotypes from their respective fathers. One mutation was observed at the H4 locus in which the father typed as 139 bp and the son was 135 bp. Some degree of "-A" was observed, and therefore I will increase the extension time in future experiments.

**May 8, 2002**

- I prepared database samples for amplification.

**May 9, 2002**

- I prepared database samples for amplification.

**May 10, 2002**

- I received the following primers from ABI, which had been ordered on May 2, 2002:

Y439 forward, Y19 forward, Y436 reverse, and Y435 reverse

- Each of the new primers was diluted to ~200 μM and made into 50 μM stocks based on OD readings taken at 260 nm.
- Experiment #16: DATABASING
  - Purpose: To database ~200 each of Caucasian, African American, and Hispanic males in order to obtain their Y-STR haplotypes.
  - The database samples had been previously collected from the paternity division of Orchid GeneScreen Dallas. The samples were amplified using 2 mM $MgCl_2$, an annealing temperature of 60° C, and a 55 minute extension time.
  - Amplifications (databasing plate names):
    - Caucasian #1
    - Caucasian #2
    - African American #1
    - African American #2
    - Hispanic #1
    - Miscellaneous databasing
  - The Caucasian #1 plate was amplified using the original PCR reagents from ABI. Accidentally however, 50 mM $MgCl_2$ stock was used instead of 25 mM $MgCl_2$. The remainder of the plates were amplified with new PCR reagents from Invitrogen (dNTPs, 10x PCR buffer, 50 mM $MgCl_2$).

**May 11, 2002**

- Database samples were loaded onto the ABI 3100.
- Database plates Caucasian #1 and Hispanic #1 were loaded on the ABI 3100.
- Database plates African American #1 and #2 were loaded on the ABI 3100.

**May 12, 2002**

- The Miscellaneous databasing plate was loaded on the ABI 3100.
- Results from Experiment #16 (databasing plates Caucasian #1, Hispanic #1, African American #1 and #2, and Miscellaneous): Essentially none of the samples worked, in that I was unable to obtain any Y-STR haplotypes. It is likely that something is wrong with one of the new Invitrogen PCR reagents (possibly the dNTPs). The few samples that did work came from the Caucasian #1 plate in which I used the remainder of the original PCR reagents from ABI. All other plates were amplified using Invitrogen reagents.

**May 13, 2002**

- Because none of the databasing samples worked from Experiment #16, new reagents (10x PCR buffer, 25 mM $MgCl_2$, dNTPs, and AmpliTaq Gold DNA polymerase) were ordered from ABI.

**May 14, 2002**

- Experiment #17: QUALITY ASSURANCE OF THE NEW ABI PCR REAGENTS

- Purpose: I received the PCR reagents from ABI. In order to ensure they were working properly, I will amplify several samples, including positive controls.
- The samples were amplified using 2 mM $MgCl_2$, a 60° C annealing temperature, and a 50 minute extension.
- The samples were subsequently loaded on the ABI 3100.

**May 15, 2002**

- Results from Experiment #17: I was able to obtain Y-STR haplotypes from the samples, and therefore determined that the reaction and PCR reagents are functioning properly.
- Experiment #18: DATABASING
  - Purpose: To obtain Y-STR haplotypes from ~200 Caucasian, African American, and Hispanic males.
  - Caucasian #1 and #2 databasing plates were amplified using 2 mM $MgCl_2$, a 60° C annealing temperature, and a 50 minute extension.

**May 16, 2002**

- The Caucasian #2 databasing plate was loaded on the ABI 3100 and subsequently analyzed.
- African American #1 and #2 databasing plates were amplified using 2 mM $MgCl_2$, a 60° C annealing temperature, and a 50 minute extension.

**May 17, 2002**

- The Caucasian #1 databasing plate was loaded on the ABI 3100 and subsequently analyzed.
- The Hispanic and Miscellaneous databasing plates were amplified using 2 mM $MgCl_2$, a 60° C annealing temperature, and a 50 minute extension.

**May 18, 2002**

- The African American databasing plates #1 and #2 were loaded on the ABI 3100.

**May 19, 2002**

- The Hispanic and Miscellaneous databasing plates were loaded on the ABI 3100.
- Experiment #19: FORENSIC VALIDATION
  - Purpose: To determine the specificity and sensitivity of the Y-STR 10-plex, as well as determine the ability of the Y-STR 10-plex to distinguish mixtures.
  - Amplifications:
    - 25 female samples will be tested to ensure that the Y-STR 10-plex is specific to males.
    - A sensitivity test will be performed in which varying amounts of male DNA (BS#1), ranging from 0.25 ng to 2.5 ng, will be amplified.
    - Both female:male (9947A:ATCC 45514) and male:male (BS#1:ATCC 45514) mixtures will be amplified in the following ratios:
      100:0, 90:10, 75:25, 50:50, 25:75, 10:90, 0:100

- The Foresic Validation samples were loaded on the ABI 3100 following amplification.

**May 20, 2002**

- Results of Experiment #18 (Databasing): I was able to successfully obtain Y-STR haplotypes from 206 African Americans, 224 Caucasians, and 95 Hispanics. On sample #185410 AF (African American), I saw 2 peaks at Y435. On samples #185790 AF (Caucasian) and #186507 AF (Caucasian), I observed 2 peaks at Y19. Are these possibly gene duplications?
- Results of Experiment #19: A complete Y-STR haplotype was observed down to 0.75 ng male DNA. At 0.5 ng, two of the larger loci dropped out. No peaks were observed at 0.25 ng. The female samples did not produce any peaks, thereby suggesting that the 10-plex is specific to males. In the female:male mixture studies, the male YSTR profile was complete at a 50:50 ratio. At 75:25 however, some Y-STR loci began to drop out. In the male:male mixture studies, 2 Y-STR profiles could be distinguished at a 75:25 ratio.

**May 21, 2002**

- Experiment #20: SENSITIVITY TEST AND MIXTURES USING 1.5x BUFFER; TESTING PLATINUM TAQ/TAQ GOLD AND INVITROGEN dNTPs; MISCELLANEOUS SAMPLES

- Purpose: 1) To determine if increasing the amount of buffer to 1.5x will help to increase the sensitivity of the Y-STR 10-plex.
  2) To determine if using ¼ the volume of the Invitrogen dNTPs will produce the same results as using the full volume of ABI dNTPs.
  3) To determine if the double peaks seen at the Y435 and Y19 loci in several of the AF samples is passed on to their sons.
- The samples were amplified using a 60° C annealing temperature, 2 mM $MgCl_2$, and a 50 minute extension time.

**May 22, 2002**

- Samples from Experiment #20 were loaded on the ABI 3100.
- Results from Experiment #20: Increasing the concentration of buffer did not appear to substantially increase the sensitivity of the Y-STR 10-plex. Using ¼ the volume of Invitrogen dNTPs produced the same haplotype results as using the full amount of ABI dNTPs. This suggests that the amount of Invitrogen dNTPs previously used in the databasing experiments quenched the reaction. No amplification was observed in the children's samples. It is possible that the DNA was too concentrated, and therefore I will use a decreased amount of DNA in a future experiment in order to determine if the double peak phenomenon is passed on from father to son. Platinum Taq produced the same results as Taq Gold, and could therefore be used as a less expensive alternative for DNA polymerase.

# CHAPTER 5

# RESULTS

## *5.1 Verification of the Polymerase Chain Reaction (PCR) Amplification Conditions*

In order to perform Y-chromosome short tandem repeat (Y-STR) testing on the ABI Prism® 3100 Genetic Analyzer (ABI 3100), the NIST (National Institute of Standards and Technology) 10-plex primer sets had to be resynthesized with fluorescent tags that were compatible with the ABI 3100. The fluorescent dyes used in the 10-plex consisted of FAM™, VIC™, NED™, and LIZ™. The loci DYS436, DYS439, DYS435, and DYS19 were labeled with FAM™; A7.1, H4, and DYS391 with VIC™; and DYS392, DYS438, and DYS437 with NED™. A LIZ™-labeled 500 base pair ladder served as the internal size standard.

To determine the optimal amount of DNA needed for the Y-STR 10-plex, varying amounts of male DNA (ATCC 45514 or BS #1), ranging from 1.0-5.0 ng, were subjected to the PCR amplification conditions established by NIST. The resulting electropherograms suggested that the optimal amount of DNA to incorporate in the PCR reaction is ~2.5 ng. However, the baseline levels were very high, thus making analysis difficult (Figure 1). Addition of bovine serum albumin (BSA) to the PCR reaction substantially decreased the baseline levels (Figure 2), yet several non-specific peaks were

still observed, possibly due to the relatively low annealing temperature (55° C) utilized by NIST.

The effect of annealing temperature on the number and peak heights of PCR artifacts was examined by varying the annealing temperature from 56° C to 60° C. As shown in Figure 3, when utilizing an annealing temperature of 60° C, the ten Y-STR peaks remained clearly identifiable. Although the increased annealing temperature helped to minimize the PCR artifacts, a few non-specific peaks still remained. The non-specific peaks observed in Genotyper® analysis were ~123 base pairs (bp) and ~213 bp in the FAM™-labeled loci, ~116 bp in the VIC™-labeled loci, and ~95 bp in the NED™-labeled loci.

In an effort to get rid of the non-specific peaks, samples were amplified using a 60° C annealing temperature and varying concentrations of magnesium chloride, ranging from 1.5-2.5 mM. Increasing the magnesium concentration to 2.5 mM did not have a substantial effect on the disappearance of non-specific peaks, and thus a magnesium concentration of 2.0 mM was used in the development of the database. During Genotyper® analysis, split peaks (minus A) were observed, thereby prompting an increase in the extension time to 50 minutes, as compared to the 45 minute extension time established by NIST.

### *5.2 Validation of the NIST Y-STR 10-Plex Using Father-Son Pairs*

Because the Y-chromosome is transmitted from father to son(s) in an unmodified form, male relatives possess the same Y-STR haplotype, barring any mutational events.

Therefore, to ensure that the Y-STR 10-plex was working properly, the Y-chromosome haplotypes of 42 father-son pairs were analyzed to confirm that each pair generated identical haplotypes.

Caucasian, African American, and Southeastern (SE) Hispanic father-son pairs were obtained from the paternity division of Orchid GeneScreen Dallas. Following capillary electrophoresis, the samples were analyzed with GeneScan® and Genotyper® software. Each father-son pair exhibited the same Y-STR profile, with one exception. One mutation was observed at the Y-GATA-H4 locus, in which the father carried a 139 bp allele and the son carried a 135 bp allele. Based on the 42 father-son pairs analyzed, the mutation rate for the H4 locus was computed to be 0.0238 +/– 0.046 (95% confidence interval).

### *5.3 Development of a Y-STR Database*

Due to the mode of inheritance of the Y-chromosome, it is reasonable to expect that certain Y-STR alleles and haplotypes will be distributed in a population-specific manner. Thus, development of a Y-chromosome haplotype database is essential in correctly understanding the frequency of occurrence of a particular haplotype in a population. For the development of such a database, unrelated Caucasian, African American, and SE Hispanic male samples were obtained from the paternity division of Orchid GeneScreen Dallas.

Allele frequencies for each locus were calculated by the counting method (Table 3 A-D). As shown in the cumulative allele frequency table (Table 3-D), overall gene

diversity (*D*) ranged from 0.071 (DYS436) to 0.699 (DYS19). Through analysis with Arlequin software, 365 different haplotypes were detected among the three populations ($n = 525$) (Table 4). The most frequent haplotype across all populations was found in 20 individuals (3.8%) and consisted of: DYS436 (85) - DYS439 (124) - DYS435 (146) - DYS19 (189) - A7.1 (112) - H4 (135)- DYS391 (166) - DYS392 (110) - DYS438 (162) - DYS437 (186). The cumulative haplotype diversity (*HD*) was 0.996, and the probability of finding the identical haplotype in a pair of random, unrelated males is 0.37% (calculated as $[1-HD]\times100$).

Table 5 A-C outlines the haplotypes found in the Caucasian, African American, and SE Hispanic populations. The haplotypes are sequentially listed and do not necessarily correlate across population groups. Haplotype correlations between populations are summarized in Table 4. As shown in Table 5-A, the Arlequin software discerned 161 different haplotypes in the Caucasian population ($n = 225$). The haplotype diversity for the Caucasian population was calculated to be 0.991. DYS439 was the most diverse locus ($D = 0.634$), while DYS436 was the least diverse ($D = 0.044$) in the Caucasian population. The average gene diversity over all ten loci was 0.453782 +/- 0.247855, as calculated by Arlequin.

In a total sample size of 206 individuals, Arlequin detected 153 different haplotypes in the African American population (Table 5-B), and the haplotype diversity was computed as 0.995. The most diverse locus in the African American population was DYS19 ($D = 0.747$), and DYS435 was the least diverse ($D = 0.075$). Average gene diversity over all analyzed loci was calculated to be 0.436780 +/- 0.239818.

As shown in Table 5-C, a total of 87 different haplotypes were detected in the SE Hispanic population ($n = 94$). The haplotype diversity was calculated as 0.998. DYS19 exhibited the most diversity in the Hispanic population ($D = 0.721$), whereas DYS436 showed the least diversity ($D = 0.063$). Arlequin software calculated the average gene diversity across all loci as 0.526424 +/- 0.284347.

Base pair ranges observed when developing the Y-STR database are listed in Table 1. There were two instances in which the allele at the DYS435 locus overlapped with allele sizes observed at DYS439. One allele each of 126 bp and 134 bp was observed at the DYS435 locus. The base pair sizes observed at DYS439 and DYS435 ranged from 108-136 and 126-151, respectively. The next largest locus, DYS19, ranges from 181-205 bp. It may be necessary to redesign the DYS435 primers to increase the amplicon lengths so that they do not interfere with the DYS19 locus, yet would avoid any overlap with the DYS439 locus.

Linkage disequilibrium is the nonrandom association of alleles at different loci. Because all of the NIST Y-STR 10-plex loci are on the same chromosome and are transmitted without recombination, they are all linked by structural constraints. The NIST Y-STR 10-plex loci were tested for linkage disequilibrium with Arlequin software using a significance level of 0.05. The results of this test, as shown in Table 6, demonstrate that the Y-STR loci exhibit varying amounts of linkage disequilibrium, which may be a function of the size of the database. Linkage disequilibrium may be influenced by the location of a microsatellite on a chromosome. However, chromosomal location does not appear to explain the observed differences in linkage disequilibrium in

this study. For example, in the Caucasian population, both DYS392 and DYS437 exhibited linkage disequilibrium to seven loci in the NIST 10-plex, yet DYS437 is located near the centromere, and DYS392 is located distally on the *q* arm (Figure 6). As a second example, the chromosomal location of DYS435 is very near DYS437, yet DYS437 shows a significant amount of linkage to other 10-plex loci, whereas DYS435 shows substantially less linkage to other loci in the NIST 10-plex. The extent of linkage disequilibrium can also be largely influenced by the amount of recombination, yet because the Y-chromosome is inherited in an unmodified form from father to son(s), recombination cannot explain the observed differences in linkage. Rather, variations in linkage disequilibrium may be due to the Y-STR loci changing at different evolutionary rates and being under different mutational pressures. The loci which have an increased propensity for mutations tend to be more polymorphic and thus, may exhibit a lesser degree of linkage disequilibrium with other Y-STR loci. Conversely, loci under decreased mutational pressure may appear to be coinherited.

Locus duplication or triplication has previously been reported at several Y-STR loci, including DYS19, DYS385, DYS390, and DYS436 (10, 12, 13, 15). Three instances of locus multiplication were observed in the present study, two at DYS19 (Figure 4) and one at DYS435 (Figure 5). In each of these three cases, two peaks of approximately even heights were observed, and in each instance of locus duplication the alleles were separated by only one repeat. Locus duplication may be explained by one of two mechanisms. Although implausible, the appearance of two peaks may occur if the primer is complimentary to the DNA in a manner in which it is able to bind in two places

that are four base pairs apart. However, if this mechanism were to occur, one would expect the appearance of two peaks to occur more frequently than it did in the present study. Rather, locus multiplication likely occurs due to a transposable element, such as an insertion sequence, or replicative transposition. During the transposition of an insertion sequence, a sequence of host DNA (for example, a Y-STR) is duplicated at the site of insertion (21). In replicative transposition, the transposable element (a Y-STR) is duplicated, generating two copies of the original element (21). In either scenario, the transposition must be followed by a mutational event, thereby producing two alleles of different sizes.

### *5.4 Validation of the Y-STR 10-Plex for Forensic Casework*

Before applying the Y-STR 10-plex to forensic casework specimens, it was necessary to carry out several experiments to examine how the 10-plex performed on mixed samples, as well to determine the sensitivity and male-specificity of the system.

To examine the sensitivity of the Y-STR 10-plex, varying amounts of male DNA (BS #1) ranging from 0.25 ng to 2.5 ng were added to the PCR reaction. All ten Y-STR systems were visible down to 0.75 ng, although the peak heights were quite low at 0.75 ng input DNA. Two loci (DYS438 and DYS437) dropped out at 0.5 ng male DNA, and at 0.25 ng no loci were detected.

In an effort to determine if the 10-plex is specific to males, 25 females were subjected to Y-STR testing. There were three instances in which a "hump" (as compared to a well-defined peak) appeared on the electropherograms in the VIC™-labeled loci:

~136 bp (153 RFU [relative fluorescence units]), 136 bp (116 RFU), and 103 bp (120 RFU). However, their morphology was very different than the Y-STR peaks observed in male samples, thus leading to the conclusion that the humps were merely artifacts rather than something derived from the X-chromosome and/or autosomal regions.

Male:female (ATCC 45514:9947A) mixture studies were performed in the following ratios using a total of 2.5 ng DNA: 100:0, 90:10, 75:25, 50:50, 25:75, 10:90, and 0:100. In all cases there was no detectable interference from the female DNA. Mixture studies of male:male DNA samples (BS #1:ATCC 45514) were performed in the same ratios using 2.5 ng total DNA. The electropherogram was interpreted as a single source sample at a 90:10 ratio, but at 75:25, alleles from a second individual began to appear (Figure 7). However, in a mixture of 50:50, Y-STR profiles from two males were able to be distinguished. In the male:male mixtures, the ability to differentiate the Y-STR haplotype from the minor component of the mixture depends on the peak positions of the alleles. If an allele falls in a stutter position, it may be difficult to determine whether the peak is a minor component of the mixture or if it truly is stutter. Clearly, this may impair the ability of an analyst to unambiguously differentiate between Y-STR haplotypes in the absence of reference samples.

# CHAPTER 6

# DISCUSSION

The ability of Y-chromosome short tandem repeats (Y-STRs) to selectively detect and differentiate between male DNA samples makes them a beneficial addition to the well-established autosomal STR systems (15). However, before implementing a male identification system in a forensics laboratory, the procedure must be validated to determine its efficacy and reliability, as required by the DNA Advisory Board Quality Assurance Standards (16). The current study was undertaken to begin validation of the National Institute of Standards and Technology (NIST) Y-STR 10-plex for use in forensic and paternity laboratories at Orchid GeneScreen Dallas. This validation study included optimization of the polymerase chain reaction (PCR), analysis of father-son pairs, development of a Y-STR database, sensitivity, male-specificity, and mixture studies.

Many of the DNA samples encountered in forensic casework have been exposed to adverse conditions which results in their degradation. Thus, it is advantageous to have a method of Y-STR testing that requires very little template DNA for the amplification process. In optimizing the PCR conditions for the NIST Y-STR 10-plex, it was determined that the optimal amount of DNA to incorporate into each reaction is ~2.5 ng. This amount of DNA is comparable to the quantity of DNA used by Prinz, et al. in their

validation of a Y-STR multiplex consisting of four loci (17). However, other groups working with Y-STR multiplexes incorporate up to 20 ng of template DNA in the PCR reaction mix (12). This suggests that the working range of the NIST Y-STR 10-plex is comparable to that of other Y-STR multiplexes.

In paternity testing, differences at genetic markers result in an exclusion of biological paternity. Spontaneous mutations which result in a father-son pair having different Y-STR haplotypes may erroneously lead to an exclusion of paternity. Thus, it is clear that having an accurate understanding of the mutational rates and processes is essential in correctly interpreting Y-STR haplotypes. Based on 4,999 male germline transmissions, Kayser and Sajantila have estimated the locus specific mutation rate between 0 and $8.58 \times 10^{-3}$, with the average mutation rate for Y-STRs being approximately $2.8 \times 10^{-3}$ (13). This is comparable with the mutation rates reported for autosomal loci (13). However, in the current analysis of 42 father-son pairs, a single mutation at the H4 locus was observed and the mutation rate was estimated at 0.0238 +/– 0.046 (95% confidence interval). This rate is approximately 8.5 times higher than Kayser and Sajantila's estimate. Two possibilities may explain this high mutation rate. First, it is almost certain that the mutation rate in the present analysis is skewed due to the low sample size, and that the mutation was merely a stochastic event. Second, it is possible that the H4 locus is simply more prone to mutational processes than other Y-STR loci. In either scenario, analysis of additional father-son pairs would facilitate either confirming or rejecting these hypotheses. Knowledge of mutation rates at the H4 and other Y-STR

loci is an important consideration in developing interpretational guidelines for the laboratory.

Instances of locus multiplication (allele duplication or triplication, for example) have been previously reported by several groups at DYS19, DYS385, DYS390, and DYS436 (10, 12, 13, 15). The current study has demonstrated one instance of locus multiplication at DYS435, which has not before been reported, and two instances of allele duplication at the DYS19 locus. Previous studies have estimated the frequency of allele duplication at the DYS19 locus to be 0.12%, based on analysis of 7,772 individuals (13). The frequency of DYS19 duplication in the current study ($n = 525$) was calculated at 0.38% (0.00381 +/– 0.00526, 95% confidence interval), roughly three times greater than Kayser and Sajantila's estimate. The variability in locus duplication frequencies may be explained by differences in sample sizes. Because of the potential for locus multiplication to occur, analysts must take care when interpreting Y-STR haplotypes so that a single source sample is not erroneously interpreted as a mixed profile.

Analysis revealed that the haplotype diversity across the Caucasian, African American, and Southeastern Hispanic populations was 0.996, which is comparable to haplotype diversity statistics published by other groups studying Y-STRs (12). This high degree of haplotype diversity suggests that the NIST Y-STR system provides the power of discrimination necessary to effectively discriminate among male samples. Future analysis involving discerning the number of haplotypes that differ by only one allele may provide additional insight into the level of diversity that the NIST 10-plex provides. However, before the NIST Y-STR 10-plex can be applied to forensic casework samples,

a standard male reference must be sequenced and an allelic ladder must be developed. Sequencing a standard male reference sample is needed to determine the number of repeating units of DNA at each Y-STR locus and the corresponding base pair size. This information can then be incorporated into Genotyper® macros and aid in the accurate sizing of unknown and reference samples. The allelic ladder serves as a standard for performing Y-STR analysis on unknown samples, and thus, must be well characterized. The allelic ladder consists of a mixture of the common alleles present in a population and provides a reference DNA size for each allele.

Due to the haploid nature of the Y-chromosome, each Y-STR locus is less informative than an autosomal STR with similar allele diversity (12). As a consequence, examining an increased number of Y-chromosome microsatellites is necessary to achieve the same level of discrimination as when investigating autosomal STRs. The product rule cannot be applied to Y-STRs, and thus, estimation of allele and haplotype frequencies is dependent on the size of population databases. The current study analyzed a total of 525 individuals. In order to strengthen the statistical analysis of Y-STR haplotyping, more males must be characterized. To examine Y-chromosome haplotypes across many populations, an international collaborative effort is needed. A number of internet resources such as STRBase (3, 18) and the Y-STR Haplotype Reference Database (19) have been established to facilitate on-going research involving both Y-chromosome and autosomal STRs.

In conclusion, Y-chromosome short tandem repeats can provide valuable information in cases of sexual assault and questioned paternity of male children. Y-STRs

can be used not only to genetically exclude a suspect or alleged father from contributing the male component of a sample, but can also be used to exclude all of his paternal relatives. The NIST Y-STR 10-plex analyzes ten loci on the Y-chromosome, thus providing an increased power of discrimination relative to other available Y-STR systems. The current validation study has demonstrated that the NIST Y-STR 10-plex can effectively discriminate between male samples. Therefore, the 10-plex would be an excellent addition to the autosomal STRs already employed in the Orchid GeneScreen Dallas forensic and paternity laboratories.

# CHAPTER 7

## TABLES AND FIGURES

Table 1. Summary of Y-STR 10-plex loci. Base pair sizes are those observed in the current study.

| Locus | Dye | Color | Repeat Motif | Base Pair Sizes |
|---|---|---|---|---|
| DYS436 | FAM | Blue | GTT | 79-91 |
| DYS439 | FAM | Blue | AGAT | 108-136 |
| DYS435 | FAM | Blue | TGGA | 126-151 |
| DYS19 | FAM | Blue | TAGA | 181-205 |
| A7.1 | VIC | Green | ATAG | 96-120 |
| H4 | VIC | Green | TAGA | 123-147 |
| DYS391 | VIC | Green | TCTA | 158-178 |
| DYS392 | NED | Yellow | TAT | 98-122 |
| DYS438 | NED | Yellow | TTTTC | 135-167 |
| DYS437 | NED | Yellow | TCTA | 174-198 |

Table 2. Primer sequences (14) developed by NIST for the Y-STR 10-plex.

| Locus | Primer Sequences |
|---|---|
| DYS19 | Forward (20 pmol): **FAM**-CTACTGAGTTTCTGTTATAGT<br>Reverse (20 pmol): ATGGCATGTAGTGAGGACA |
| DYS391 | Forward (10 pmol): **TET**-CTATTCATTCAATCATACACCCATAT<br>Reverse (10 pmol): ACATAGCCAAATATCTCCTGGG |
| DYS392 | Forward (20 pmol): **HEX**-AAAAGCCAAGAAGGAAAACAAA<br>Reverse (20 pmol): AAACCTACCAATCCCATTCCTT |
| DYS435 | Forward (5 pmol): GGGTTGTCCAGAGAAACAGC<br>Reverse (5 pmol): **FAM**-CCCCCTCCTCTCGTCTATCT |
| DYS436 | Forward (10 pmol): CCAGGAGAGCACACACAAAA<br>Reverse (10 pmol): **FAM**-ACGAGCTGCGTTAGAGGTGA |
| DYS437 | Forward (10 pmol): GACTATGGGCGTGAGTGCAT<br>Reverse (10 pmol): **HEX**-AGACCCTGTCATTCACAGATGA |
| DYS438 | Forward (10 pmol): **HEX**-TGGGGAATAGTTGAACGGTAA<br>Reverse (10 pmol): GGAGGTTGTGGTGAGTCGAG |
| DYS439 | Forward (10 pmol): **FAM**-ACATAGGTGGAGACAGATAGATGAT<br>Reverse (10 pmol): GCCTGGCTTGGAATTCTTTT |
| A7.1 | Forward (10 pmol): GAGGAATCTGACACCTCTGACA<br>Reverse (10 pmol): **TET**-TCCATATCATCTATCCTCTGCCTA |
| H4 | Forward (20 pmol): **TET**-ATGCTGAGGAGAATTTCCAA<br>Reverse (20 pmol): CTATTCATCCATCTAATCTATCCATT |

Table 3-A. Allele frequencies in the Caucasian population ($n = 225$). All sizes are in base pairs. Gene diversity ($D$) for each locus was calculated as described in "Materials and Methods".

| DYS436 | | | DYS439 | | | DYS435 | | | DYS19 | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** |
| 79 | 1 | 0.0044 | 108 | 1 | 0.0044 | 142 | 5 | 0.0222 | 181 | 2 | 0.0088 |
| 82 | 1 | 0.0044 | 112 | 1 | 0.0044 | 146 | 214 | 0.9511 | 185 | 7 | 0.0308 |
| 85 | 220 | 0.9778 | 116 | 3 | 0.0133 | 151 | 6 | 0.0267 | 188 | 1 | 0.0044 |
| 88 | 2 | 0.0089 | 120 | 66 | 0.2933 | | | | 189 | 163 | 0.7181 |
| 91 | 1 | 0.0044 | 124 | 115 | 0.5111 | | | | 193 | 40 | 0.1762 |
| | | | 125 | 1 | 0.0044 | | | | 197 | 6 | 0.0264 |
| | | | 128 | 32 | 0.1422 | | | | 201 | 7 | 0.0308 |
| | | | 132 | 6 | 0.0267 | | | | 205 | 1 | 0.0044 |
| **D =** | 0.044 | | **D =** | 0.634 | | **D =** | 0.095 | | **D =** | 0.453 | |

| A7.1 | | | H4 | | | DYS391 | | |
|---|---|---|---|---|---|---|---|---|
| **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** |
| 104 | 9 | 0.0400 | 123 | 1 | 0.0044 | 158 | 4 | 0.0178 |
| 108 | 56 | 0.2489 | 127 | 5 | 0.0222 | 162 | 105 | 0.4667 |
| 112 | 145 | 0.6444 | 131 | 75 | 0.3333 | 166 | 109 | 0.4844 |
| 116 | 15 | 0.0667 | 135 | 130 | 0.5778 | 169 | 1 | 0.0044 |
| | | | 139 | 12 | 0.0533 | 170 | 6 | 0.0267 |
| | | | 143 | 1 | 0.0044 | | | |
| | | | 147 | 1 | 0.0044 | | | |
| **D =** | 0.519 | | **D =** | 0.554 | | **D =** | 0.549 | |

| DYS392 | | | DYS438 | | | DYS437 | | |
|---|---|---|---|---|---|---|---|---|
| **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** |
| 98 | 1 | 0.0044 | 146 | 11 | 0.0489 | 178 | 1 | 0.0044 |
| 101 | 1 | 0.0044 | 151 | 51 | 0.2267 | 182 | 45 | 0.2000 |
| 103 | 2 | 0.0089 | 157 | 12 | 0.0533 | 186 | 145 | 0.6444 |
| 104 | 54 | 0.2400 | 162 | 141 | 0.6267 | 190 | 33 | 0.1467 |
| 107 | 14 | 0.0622 | 167 | 10 | 0.0444 | 194 | 1 | 0.0044 |
| 110 | 124 | 0.5511 | | | | | | |
| 112 | 2 | 0.0089 | | | | | | |
| 113 | 25 | 0.1111 | | | | | | |
| 116 | 2 | 0.0089 | | | | | | |
| **D =** | 0.625 | | **D =** | 0.551 | | **D =** | 0.525 | |

Table 3-B. Allele frequencies in the African American population ($n = 206$). All sizes are in base pairs. Gene diversity ($D$) for each locus was calculated as described in "Materials and Methods".

| DYS436 | | | DYS439 | | | DYS435 | | | DYS19 | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** |
| 79 | 1 | 0.0049 | 116 | 2 | 0.0097 | 126 | 1 | 0.0048 | 185 | 3 | 0.0146 |
| 82 | 7 | 0.0340 | 120 | 52 | 0.2524 | 142 | 1 | 0.0048 | 189 | 37 | 0.1796 |
| 85 | 195 | 0.9466 | 124 | 104 | 0.5049 | 146 | 199 | 0.9614 | 193 | 72 | 0.3495 |
| 88 | 3 | 0.0146 | 128 | 43 | 0.2087 | 151 | 6 | 0.0290 | 197 | 49 | 0.2379 |
| | | | 132 | 5 | 0.0243 | | | | 201 | 44 | 0.2136 |
| | | | | | | | | | 205 | 1 | 0.0049 |
| **D =** | 0.103 | | **D =** | 0.640 | | **D =** | 0.075 | | **D =** | 0.747 | |

| A7.1 | | | H4 | | | DYS391 | | |
|---|---|---|---|---|---|---|---|---|
| **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** |
| 96 | 1 | 0.0049 | 123 | 2 | 0.0097 | 162 | 162 | 0.7864 |
| 104 | 4 | 0.0194 | 124 | 1 | 0.0049 | 164 | 1 | 0.0049 |
| 108 | 83 | 0.4029 | 127 | 17 | 0.0825 | 166 | 39 | 0.1893 |
| 112 | 104 | 0.5049 | 131 | 119 | 0.5777 | 170 | 3 | 0.0146 |
| 116 | 12 | 0.0583 | 135 | 62 | 0.3010 | 178 | 1 | 0.0049 |
| 120 | 2 | 0.0097 | 139 | 5 | 0.0243 | | | |
| **D =** | 0.582 | | **D =** | 0.571 | | **D =** | 0.347 | |

| DYS392 | | | DYS438 | | | DYS437 | | |
|---|---|---|---|---|---|---|---|---|
| **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** |
| 98 | 1 | 0.0049 | 140 | 6 | 0.0291 | 174 | 1 | 0.0049 |
| 101 | 2 | 0.0097 | 146 | 4 | 0.0194 | 178 | 16 | 0.0777 |
| 103 | 1 | 0.0049 | 151 | 20 | 0.0971 | 182 | 147 | 0.7136 |
| 104 | 166 | 0.8058 | 157 | 140 | 0.6796 | 186 | 29 | 0.1408 |
| 107 | 10 | 0.0485 | 162 | 34 | 0.1650 | 190 | 8 | 0.0388 |
| 109 | 1 | 0.0049 | 167 | 2 | 0.0097 | 194 | 4 | 0.0194 |
| 110 | 24 | 0.1165 | | | | 198 | 1 | 0.0049 |
| 113 | 1 | 0.0049 | | | | | | |
| **D =** | 0.336 | | **D =** | 0.503 | | **D =** | 0.465 | |

Table 3-C. Allele frequencies in the SE Hispanic population ($n = 94$). All sizes are in base pairs. Gene diversity ($D$) for each locus was calculated as described in "Materials and Methods".

| DYS436 | | | DYS439 | | | DYS435 | | | DYS19 | | |
|---|---|---|---|---|---|---|---|---|---|---|---|
| **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** |
| 79 | 1 | 0.0106 | 112 | 2 | 0.0213 | 134 | 1 | 0.0106 | 185 | 20 | 0.2128 |
| 82 | 2 | 0.0213 | 116 | 6 | 0.0638 | 146 | 88 | 0.9362 | 189 | 42 | 0.4468 |
| 85 | 91 | 0.9681 | 120 | 22 | 0.2340 | 149 | 1 | 0.0106 | 193 | 20 | 0.2128 |
| | | | 124 | 49 | 0.5213 | 151 | 4 | 0.0426 | 197 | 7 | 0.0745 |
| | | | 128 | 14 | 0.1489 | | | | 201 | 5 | 0.0532 |
| | | | 136 | 1 | 0.0106 | | | | | | |
| **D =** | 0.063 | | **D =** | 0.654 | | **D =** | 0.123 | | **D =** | 0.709 | |

| A7.1 | | | H4 | | | DYS391 | | |
|---|---|---|---|---|---|---|---|---|
| **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** |
| 104 | 3 | 0.0319 | 123 | 2 | 0.0213 | 158 | 5 | 0.0532 |
| 108 | 37 | 0.3936 | 127 | 4 | 0.0426 | 162 | 55 | 0.5851 |
| 112 | 49 | 0.5213 | 131 | 43 | 0.4574 | 166 | 33 | 0.3511 |
| 116 | 4 | 0.0426 | 135 | 37 | 0.3936 | 170 | 1 | 0.0106 |
| 120 | 1 | 0.0106 | 139 | 8 | 0.0851 | | | |
| **D =** | 0.577 | | **D =** | 0.633 | | **D =** | 0.537 | |

| DYS392 | | | DYS438 | | | DYS437 | | |
|---|---|---|---|---|---|---|---|---|
| **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** | **Size** | **# found** | **Freq** |
| 101 | 1 | 0.0106 | 135 | 1 | 0.0106 | 182 | 54 | 0.5745 |
| 103 | 1 | 0.0106 | 140 | 1 | 0.0106 | 186 | 31 | 0.3298 |
| 104 | 39 | 0.4149 | 146 | 9 | 0.0957 | 190 | 9 | 0.0957 |
| 107 | 4 | 0.0426 | 151 | 23 | 0.2447 | | | |
| 110 | 35 | 0.3723 | 157 | 20 | 0.2128 | | | |
| 113 | 8 | 0.0851 | 162 | 39 | 0.4149 | | | |
| 116 | 2 | 0.0213 | 167 | 1 | 0.0106 | | | |
| 119 | 2 | 0.0213 | | | | | | |
| 122 | 2 | 0.0213 | | | | | | |
| **D =** | 0.686 | | **D =** | 0.721 | | **D =** | 0.558 | |

Table 3-D. Y-STR allele frequencies in three populations ($n$ = 525): Caucasian, African American, and SE Hispanic. All sizes are in base pairs. Gene diversity ($D$) for each locus was calculated as described in "Materials and Methods".

**DYS436**

| Size | # found | Freq |
|---|---|---|
| 79 | 3 | 0.006 |
| 82 | 10 | 0.019 |
| 85 | 506 | 0.964 |
| 88 | 5 | 0.010 |
| 91 | 1 | 0.002 |

**D =** 0.071

**DYS439**

| Size | # foun | Freq |
|---|---|---|
| 108 | 1 | 0.002 |
| 112 | 3 | 0.006 |
| 116 | 11 | 0.021 |
| 120 | 140 | 0.267 |
| 124 | 268 | 0.510 |
| 125 | 1 | 0.002 |
| 128 | 89 | 0.170 |
| 132 | 11 | 0.021 |
| 136 | 1 | 0.002 |

**D =** 0.640

**DYS435**

| Size | # found | Freq |
|---|---|---|
| 126 | 1 | 0.002 |
| 134 | 1 | 0.002 |
| 142 | 6 | 0.011 |
| 146 | 501 | 0.952 |
| 149 | 1 | 0.002 |
| 151 | 16 | 0.030 |

**D =** 0.092

**DYS19**

| Size | # found | Freq |
|---|---|---|
| 181 | 2 | 0.004 |
| 185 | 30 | 0.057 |
| 188 | 1 | 0.002 |
| 189 | 242 | 0.459 |
| 193 | 132 | 0.250 |
| 197 | 62 | 0.118 |
| 201 | 56 | 0.106 |
| 205 | 2 | 0.004 |

**D =** 0.699

**A7.1**

| Size | # found | Freq |
|---|---|---|
| 96 | 1 | 0.002 |
| 104 | 16 | 0.030 |
| 108 | 176 | 0.335 |
| 112 | 298 | 0.568 |
| 116 | 31 | 0.059 |
| 120 | 3 | 0.006 |

**D =** 0.562

**H4**

| Size | # foun | Freq |
|---|---|---|
| 123 | 5 | 0.010 |
| 124 | 1 | 0.002 |
| 127 | 26 | 0.050 |
| 131 | 237 | 0.451 |
| 135 | 229 | 0.436 |
| 139 | 25 | 0.048 |
| 143 | 1 | 0.002 |
| 147 | 1 | 0.002 |

**D =** 0.602

**DYS391**

| Size | # found | Freq |
|---|---|---|
| 158 | 9 | 0.017 |
| 162 | 322 | 0.613 |
| 164 | 1 | 0.002 |
| 166 | 181 | 0.345 |
| 169 | 1 | 0.002 |
| 170 | 10 | 0.019 |
| 178 | 1 | 0.002 |

**D =** 0.505

**DYS392**

| Size | # found | Freq |
|---|---|---|
| 98 | 2 | 0.004 |
| 101 | 4 | 0.008 |
| 103 | 4 | 0.008 |
| 104 | 259 | 0.493 |
| 107 | 28 | 0.053 |
| 109 | 1 | 0.002 |
| 110 | 183 | 0.349 |
| 112 | 2 | 0.004 |
| 113 | 34 | 0.065 |
| 116 | 4 | 0.008 |
| 119 | 2 | 0.004 |
| 122 | 2 | 0.004 |

**D =** 0.629

**DYS438**

| Size | # foun | Freq |
|---|---|---|
| 135 | 1 | 0.002 |
| 140 | 7 | 0.013 |
| 146 | 24 | 0.046 |
| 151 | 94 | 0.179 |
| 157 | 172 | 0.328 |
| 162 | 214 | 0.408 |
| 167 | 13 | 0.025 |

**D =** 0.693

**DYS437**

| Size | # found | Freq |
|---|---|---|
| 174 | 1 | 0.002 |
| 178 | 17 | 0.032 |
| 182 | 246 | 0.469 |
| 186 | 205 | 0.390 |
| 190 | 50 | 0.095 |
| 194 | 5 | 0.010 |
| 198 | 1 | 0.002 |

**D =** 0.619

Table 4. Haplotypes found in the Caucasian, African American, and SE Hispanic populations ($n$ = 525). Each haplotype is listed, followed by the number of occurrences(haplotype frequency) in each population.

| Hap | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 | Cauc. | Af. Amer | SE Hisp. |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1 | 85 | 124 | 146 | 189 | 116 | 131 | 166 | 113 | 162 | 182 | 1(0.00444 | 0 | 0 |
| 2 | 85 | 120 | 146 | 189 | 108 | 131 | 62 | 104 | 151 | 190 | 3(0.0133) | (0.00485 | 2(0.0213) |
| 3 | 85 | 132 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 186 | 1(0.00444 | 0 | 0 |
| 4 | 85 | 124 | 146 | 189 | 116 | 131 | 166 | 110 | 162 | 182 | 1(0.00444 | 0 | 0 |
| 5 | 85 | 120 | 146 | 193 | 104 | 131 | 162 | 107 | 151 | 182 | 1(0.00444 | 0 | 0 |
| 6 | 85 | 124 | 151 | 193 | 112 | 135 | 166 | 110 | 162 | 186 | 1(0.00444 | 0 | 0 |
| 7 | 85 | 124 | 146 | 189 | 108 | 131 | 166 | 110 | 162 | 182 | 1(0.00444 | 0 | 2(0.0213) |
| 8 | 85 | 124 | 146 | 193 | 112 | 131 | 166 | 107 | 151 | 186 | 1(0.00444 | 0 | 0 |
| 9 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 | 15(0.0667 | 3(0.0146) | 2(0.0213) |
| 10 | 85 | 124 | 146 | 185 | 104 | 135 | 162 | 104 | 151 | 182 | 1(0.00444 | 0 | 0 |
| 11 | 85 | 120 | 146 | 185 | 104 | 135 | 162 | 98 | 151 | 182 | 1(0.00444 | 0 | 0 |
| 12 | 85 | 128 | 146 | 189 | 112 | 135 | 162 | 110 | 157 | 182 | 1(0.00444 | 0 | 0 |
| 13 | 85 | 120 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 | 7(0.0311) | (0.00485 | 1(0.0106) |
| 14 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 167 | 186 | 1(0.00444 | 0 | 0 |
| 15 | 85 | 128 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 | 5(0.0222) | 0 | 0 |
| 16 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 167 | 190 | 1(0.00444 | 0 | 0 |
| 17 | 85 | 116 | 146 | 197 | 112 | 135 | 166 | 104 | 157 | 182 | 1(0.00444 | 0 | 0 |
| 18 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 113 | 162 | 186 | 7(0.0311) | 0 | 1(0.0106) |
| 19 | 85 | 124 | 146 | 185 | 108 | 127 | 162 | 116 | 157 | 182 | 1(0.00444 | 0 | 0 |
| 20 | 85 | 128 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 186 | 2(0.00889 | 0 | 1(0.0106) |
| 21 | 85 | 132 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 | 1(0.00444 | 0 | 0 |
| 22 | 91 | 124 | 146 | 189 | 112 | 139 | 162 | 110 | 162 | 186 | 1(0.00444 | 0 | 0 |
| 23 | 85 | 124 | 146 | 189 | 108 | 135 | 166 | 104 | 146 | 182 | 1(0.00444 | 0 | 0 |
| 24 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 186 | 2(0.00889 | (0.00485 | 1(0.0106) |
| 25 | 85 | 124 | 146 | 189 | 116 | 135 | 166 | 113 | 162 | 186 | 1(0.00444 | 0 | 1(0.0106) |
| 26 | 85 | 120 | 146 | 193 | 112 | 131 | 162 | 107 | 151 | 186 | 1(0.00444 | 0 | 0 |
| 27 | 85 | 120 | 146 | 189 | 108 | 135 | 162 | 110 | 162 | 186 | 1(0.00444 | 0 | 0 |
| 28 | 85 | 120 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 186 | 4(0.0178) | 0 | 0 |
| 29 | 85 | 124 | 142 | 189 | 112 | 131 | 166 | 110 | 162 | 186 | 1(0.00444 | 0 | 0 |
| 30 | 85 | 124 | 146 | 185 | 108 | 135 | 162 | 104 | 151 | 182 | 1(0.00444 | 0 | 0 |
| 31 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 104 | 151 | 190 | 5(0.0222) | 0 | 0 |
| 32 | 85 | 124 | 146 | 189 | 112 | 135 | 162 | 113 | 162 | 186 | 1(0.00444 | 0 | 0 |
| 33 | 85 | 128 | 146 | 189 | 112 | 147 | 166 | 110 | 162 | 186 | 1(0.00444 | 0 | 0 |
| 34 | 85 | 124 | 146 | 189 | 112 | 131 | 162 | 104 | 146 | 186 | 1(0.00444 | 0 | 0 |
| 35 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 104 | 151 | 182 | 1(0.00444 | 0 | 0 |
| 36 | 85 | 124 | 146 | 189 | 112 | 131 | 162 | 110 | 162 | 182 | 1(0.00444 | 0 | 0 |
| 37 | 85 | 124 | 146 | 193 | 108 | 135 | 162 | 104 | 151 | 190 | 1(0.00444 | 0 | 0 |
| 38 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 190 | 1(0.00444 | 0 | 0 |
| 39 | 85 | 124 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 186 | 6(0.0267) | (0.00485 | 0 |

Table 4, continued. Haplotypes found in the Caucasian, African American, and SE Hispanic population ($n$ = 525).

| Hap. | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 | Cauc. | Af. Amer. | SE Hisp. |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 40 | 85 | 120 | 146 | 189 | 108 | 135 | 166 | 110 | 167 | 186 | 1(0.00444) | 0 | 0 |
| 41 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 104 | 151 | 194 | 1(0.00444) | 0 | 0 |
| 42 | 82 | 124 | 146 | 193 | 116 | 135 | 166 | 104 | 151 | 182 | 1(0.00444) | 0 | 0 |
| 43 | 85 | 128 | 146 | 189 | 116 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 44 | 85 | 124 | 146 | 189 | 104 | 123 | 162 | 104 | 146 | 186 | 1(0.00444) | 0 | 0 |
| 45 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 107 | 151 | 182 | 3(0.0133) | 2(0.00971) | 0 |
| 46 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 151 | 182 | 1(0.00444) | 1(0.00485) | 0 |
| 47 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 113 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 48 | 85 | 128 | 146 | 189 | 108 | 131 | 162 | 104 | 146 | 186 | 1(0.00444) | 0 | 0 |
| 49 | 85 | 124 | 146 | 189 | 112 | 139 | 166 | 110 | 162 | 186 | 3(0.0133) | 0 | 1(0.0106) |
| 50 | 85 | 120 | 146 | 189 | 112 | 135 | 162 | 110 | 167 | 186 | 2(0.00889) | 0 | 0 |
| 51 | 85 | 128 | 146 | 189 | 112 | 131 | 166 | 107 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 52 | 85 | 120 | 146 | 193 | 112 | 135 | 166 | 110 | 162 | 186 | 2(0.00889) | 1(0.00485) | 1(0.0106) |
| 53 | 85 | 128 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 190 | 1(0.00444) | 0 | 0 |
| 54 | 85 | 124 | 146 | 193 | 112 | 135 | 170 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 55 | 85 | 124 | 146 | 201 | 108 | 135 | 166 | 104 | 157 | 178 | 1(0.00444) | 0 | 0 |
| 56 | 85 | 120 | 142 | 193 | 108 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 57 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 104 | 151 | 186 | 1(0.00444) | 0 | 0 |
| 58 | 85 | 124 | 146 | 201 | 108 | 135 | 162 | 104 | 151 | 182 | 1(0.00444) | 0 | 0 |
| 59 | 85 | 120 | 146 | 188 | 112 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 60 | 85 | 124 | 146 | 189/193 | 104 | 131 | 162 | 104 | 151 | 182 | 2(0.00889) | 0 | 0 |
| 61 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 113 | 162 | 182 | 1(0.00444) | 0 | 0 |
| 62 | 85 | 124 | 146 | 201 | 112 | 127 | 162 | 104 | 157 | 182 | 1(0.00444) | 3(0.0146) | 0 |
| 63 | 85 | 120 | 146 | 193 | 112 | 135 | 162 | 104 | 151 | 186 | 1(0.00444) | 0 | 0 |
| 64 | 85 | 108 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 182 | 1(0.00444) | 0 | 0 |
| 65 | 85 | 120 | 146 | 197 | 108 | 135 | 162 | 104 | 151 | 190 | 1(0.00444) | 0 | 0 |
| 66 | 85 | 124 | 146 | 197 | 108 | 131 | 162 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 67 | 85 | 124 | 146 | 189 | 108 | 139 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 68 | 85 | 124 | 146 | 189 | 116 | 135 | 166 | 112 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 69 | 85 | 120 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 182 | 1(0.00444) | 0 | 0 |
| 70 | 85 | 128 | 146 | 189 | 112 | 131 | 166 | 107 | 162 | 190 | 1(0.00444) | 0 | 0 |
| 71 | 85 | 124 | 146 | 189 | 112 | 135 | 170 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 72 | 85 | 120 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 73 | 85 | 128 | 146 | 189 | 112 | 139 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 74 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 112 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 75 | 85 | 124 | 146 | 201 | 108 | 135 | 162 | 104 | 157 | 182 | 1(0.00444) | 0 | 0 |
| 76 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 104 | 151 | 190 | 3(0.0133) | 0 | 0 |
| 77 | 85 | 128 | 146 | 189 | 108 | 135 | 162 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 78 | 85 | 120 | 146 | 189 | 108 | 131 | 162 | 104 | 146 | 186 | 2(0.00889) | 0 | 0 |
| 79 | 85 | 120 | 146 | 185 | 112 | 135 | 162 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 80 | 85 | 120 | 146 | 197 | 108 | 135 | 166 | 104 | 156 | 182 | 1(0.00444) | 0 | 0 |

Table 4, continued. Haplotypes found in the Caucasian, African American, and SE Hispanic population ($n = 525$).

| Hap. | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 | Cauc. | Af. Amer. | SE Hisp. |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 81 | 85 | 124 | 146 | 193 | 112 | 135 | 162 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 82 | 85 | 124 | 146 | 193 | 112 | 139 | 162 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 83 | 85 | 128 | 146 | 189 | 112 | 143 | 162 | 110 | 167 | 186 | 1(0.00444) | 0 | 0 |
| 84 | 85 | 120 | 146 | 193 | 112 | 127 | 162 | 107 | 151 | 182 | 1(0.00444) | 0 | 0 |
| 85 | 85 | 120 | 146 | 189 | 112 | 135 | 170 | 113 | 167 | 186 | 1(0.00444) | 0 | 0 |
| 86 | 85 | 112 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 182 | 1(0.00444) | 0 | 0 |
| 87 | 85 | 120 | 146 | 197 | 116 | 135 | 166 | 104 | 157 | 182 | 1(0.00444) | 0 | 0 |
| 88 | 85 | 128 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 186 | 2(0.00889) | 0 | 2(0.0213) |
| 89 | 85 | 124 | 151 | 189 | 112 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 90 | 85 | 124 | 146 | 193 | 112 | 135 | 166 | 113 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 91 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 107 | 162 | 190 | 1(0.00444) | 0 | 0 |
| 92 | 85 | 120 | 146 | 189 | 104 | 131 | 162 | 104 | 146 | 182 | 1(0.00444) | 0 | 1(0.0106) |
| 93 | 85 | 124 | 146 | 193 | 108 | 131 | 162 | 104 | 151 | 190 | 1(0.00444) | 0 | 0 |
| 94 | 85 | 132 | 146 | 197 | 108 | 131 | 162 | 104 | 151 | 182 | 1(0.00444) | 0 | 0 |
| 95 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 104 | 151 | 186 | 1(0.00444) | 0 | 0 |
| 96 | 85 | 124 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 186 | 3(0.0133) | 1(0.00485) | 0 |
| 97 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 110 | 162 | 186 | 2(0.00889) | 2(0.00971) | 0 |
| 98 | 85 | 128 | 146 | 193 | 104 | 131 | 170 | 104 | 151 | 186 | 1(0.00444) | 0 | 0 |
| 99 | 85 | 128 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 182 | 1(0.00444) | 0 | 0 |
| 100 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 113 | 167 | 186 | 1(0.00444) | 0 | 0 |
| 101 | 85 | 124 | 146 | 201 | 116 | 131 | 162 | 107 | 151 | 186 | 1(0.00444) | 0 | 0 |
| 102 | 85 | 128 | 146 | 189 | 112 | 139 | 162 | 113 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 103 | 85 | 128 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 182 | 1(0.00444) | 0 | 0 |
| 104 | 85 | 128 | 151 | 193 | 104 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 105 | 85 | 120 | 146 | 193 | 108 | 131 | 169 | 104 | 151 | 190 | 1(0.00444) | 0 | 0 |
| 106 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 113 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 107 | 85 | 124 | 146 | 193 | 116 | 131 | 166 | 110 | 157 | 190 | 1(0.00444) | 0 | 0 |
| 108 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 109 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 182 | 1(0.00444) | 0 | 1(0.0106) |
| 110 | 88 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 111 | 85 | 124 | 151 | 189 | 112 | 135 | 166 | 113 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 112 | 85 | 128 | 146 | 189 | 112 | 135 | 166 | 113 | 162 | 190 | 1(0.00444) | 0 | 0 |
| 113 | 85 | 124 | 146 | 189 | 108 | 131 | 166 | 110 | 162 | 186 | 3(0.0133) | 0 | 1(0.0106) |
| 114 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 110 | 146 | 182 | 1(0.00444) | 0 | 0 |
| 115 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 157 | 186 | 1(0.00444) | 0 | 0 |
| 116 | 85 | 124 | 146 | 193 | 112 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 1(0.00485) | 1(0.0106) |
| 117 | 85 | 124 | 146 | 193 | 112 | 135 | 162 | 110 | 162 | 182 | 1(0.00444) | 0 | 0 |
| 118 | 85 | 120 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 190 | 1(0.00444) | 0 | 0 |
| 119 | 85 | 128 | 142 | 189 | 112 | 135 | 166 | 113 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 120 | 85 | 124 | 146 | 189 | 112 | 139 | 166 | 113 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 121 | 85 | 120 | 146 | 189 | 108 | 135 | 158 | 104 | 151 | 190 | 1(0.00444) | 0 | 0 |

Table 4, continued. Haplotypes found in the Caucasian, African American, and SE Hispanic population ($n$ = 525).

| Hap. | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 | Cauc. | Af. Amer. | SE Hisp. |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 122 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 110 | 167 | 186 | 1(0.00444) | 0 | 0 |
| 123 | 85 | 132 | 151 | 189 | 108 | 135 | 158 | 107 | 151 | 190 | 1(0.00444) | 0 | 0 |
| 124 | 85 | 124 | 146 | 193 | 112 | 135 | 166 | 116 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 125 | 85 | 132 | 146 | 189 | 112 | 131 | 162 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 126 | 85 | 120 | 146 | 181 | 112 | 135 | 162 | 104 | 151 | 182 | 1(0.00444) | 0 | 0 |
| 127 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 151 | 190 | 1(0.00444) | 0 | 0 |
| 128 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 107 | 151 | 190 | 1(0.00444) | 0 | 0 |
| 129 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 110 | 146 | 186 | 1(0.00444) | 0 | 0 |
| 130 | 85 | 120 | 146 | 189 | 112 | 127 | 162 | 101 | 151 | 190 | 1(0.00444) | 0 | 0 |
| 131 | 85 | 128 | 146 | 193 | 108 | 135 | 162 | 107 | 151 | 190 | 1(0.00444) | 0 | 0 |
| 132 | 85 | 124 | 146 | 189 | 112 | 127 | 162 | 103 | 151 | 190 | 1(0.00444) | 0 | 0 |
| 133 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 104 | 151 | 182 | 1(0.00444) | 0 | 0 |
| 134 | 85 | 120 | 146 | 181 | 108 | 131 | 162 | 104 | 151 | 182 | 1(0.00444) | 0 | 0 |
| 135 | 79 | 120 | 146 | 189 | 112 | 135 | 162 | 113 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 136 | 85 | 120 | 146 | 205 | 112 | 135 | 162 | 104 | 157 | 182 | 1(0.00444) | 0 | 0 |
| 137 | 85 | 125 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 138 | 85 | 124 | 142 | 189 | 116 | 131 | 162 | 104 | 146 | 190 | 1(0.00444) | 0 | 0 |
| 139 | 85 | 124 | 151 | 193 | 108 | 131 | 162 | 104 | 146 | 186 | 1(0.00444) | 0 | 0 |
| 140 | 85 | 128 | 146 | 189 | 112 | 139 | 166 | 113 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 141 | 85 | 124 | 146 | 189 | 116 | 135 | 162 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 142 | 85 | 124 | 146 | 189 | 112 | 131 | 158 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 143 | 85 | 116 | 146 | 201 | 112 | 135 | 162 | 104 | 157 | 182 | 1(0.00444) | 0 | 0 |
| 144 | 85 | 120 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 182 | 1(0.00444) | 0 | 0 |
| 145 | 85 | 128 | 142 | 201 | 112 | 139 | 162 | 104 | 151 | 186 | 1(0.00444) | 0 | 0 |
| 146 | 85 | 120 | 146 | 189 | 116 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 147 | 85 | 132 | 146 | 193 | 112 | 135 | 162 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 148 | 85 | 124 | 146 | 189 | 112 | 131 | 162 | 110 | 162 | 186 | 2(0.00889) | 0 | 1(0.0106) |
| 149 | 85 | 120 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 150 | 85 | 124 | 146 | 189 | 112 | 131 | 170 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 151 | 85 | 128 | 146 | 189 | 112 | 135 | 162 | 113 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 152 | 85 | 120 | 146 | 189 | 116 | 135 | 166 | 110 | 162 | 190 | 1(0.00444) | 0 | 0 |
| 153 | 85 | 128 | 146 | 185 | 108 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 154 | 85 | 120 | 146 | 189 | 116 | 131 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 155 | 88 | 120 | 146 | 189 | 112 | 131 | 166 | 110 | 167 | 186 | 1(0.00444) | 0 | 0 |
| 156 | 85 | 124 | 146 | 189 | 108 | 139 | 162 | 104 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 157 | 85 | 124 | 146 | 189 | 116 | 135 | 166 | 110 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 158 | 85 | 124 | 146 | 193 | 108 | 131 | 166 | 110 | 162 | 182 | 1(0.00444) | 0 | 0 |
| 159 | 85 | 116 | 146 | 185 | 112 | 135 | 158 | 104 | 151 | 182 | 1(0.00444) | 0 | 0 |
| 160 | 85 | 128 | 146 | 193 | 112 | 131 | 162 | 103 | 151 | 190 | 1(0.00444) | 0 | 0 |
| 161 | 85 | 124 | 146 | 189 | 108 | 135 | 166 | 113 | 162 | 186 | 1(0.00444) | 0 | 0 |
| 162 | 85 | 128 | 146 | 201 | 112 | 131 | 166 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |

Table 4, continued. Haplotypes found in the Caucasian, African American, and SE Hispanic population ($n = 525$).

| Hap. | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 | Cauc. | Af. Amer. | SE Hisp. |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 163 | 85 | 124 | 146 | 197 | 108 | 131 | 162 | 104 | 157 | 182 | 0 | 5(0.0243) | 0 |
| 164 | 85 | 124 | 146 | 197 | 108 | 135 | 162 | 104 | 157 | 174 | 0 | 1(0.00485 | 0 |
| 165 | 85 | 116 | 146 | 197 | 112 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 166 | 85 | 124 | 146 | 193 | 112 | 131 | 166 | 110 | 162 | 186 | 0 | 1(0.00485 | 0 |
| 167 | 85 | 124 | 146 | 197 | 116 | 131 | 162 | 104 | 151 | 182 | 0 | 1(0.00485 | 0 |
| 168 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 146 | 190 | 0 | 1(0.00485 | 0 |
| 169 | 85 | 128 | 146 | 193 | 108 | 131 | 163 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 170 | 85 | 128 | 146 | 201 | 112 | 131 | 162 | 104 | 157 | 182 | 0 | 3(0.0146) | 1(0.0106) |
| 171 | 85 | 120 | 146 | 201 | 108 | 131 | 162 | 104 | 157 | 182 | 0 | 2(0.00971 | 0 |
| 172 | 85 | 124 | 146 | 197 | 112 | 131 | 162 | 104 | 157 | 182 | 0 | 6(0.0291) | 0 |
| 173 | 85 | 128 | 151 | 201 | 112 | 131 | 162 | 104 | 157 | 178 | 0 | 1(0.00485 | 0 |
| 174 | 85 | 116 | 146 | 201 | 112 | 139 | 166 | 104 | 157 | 182 | 0 | 1(0.00485 | 1(0.0106) |
| 175 | 85 | 128 | 146 | 189 | 112 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 176 | 85 | 120 | 146 | 201 | 112 | 131 | 162 | 104 | 157 | 182 | 0 | 3(0.0146) | 0 |
| 177 | 85 | 120 | 146 | 193 | 112 | 131 | 166 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 178 | 85 | 120 | 146 | 193 | 112 | 135 | 162 | 104 | 140 | 194 | 0 | 1(0.00485 | 0 |
| 179 | 85 | 128 | 146 | 193 | 112 | 135 | 162 | 104 | 157 | 182 | 0 | 2(0.00971 | 0 |
| 180 | 85 | 124 | 146 | 197 | 112 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 1(0.0106) |
| 181 | 85 | 128 | 151 | 197 | 108 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 182 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 157 | 178 | 0 | 2(0.00971 | 0 |
| 183 | 85 | 128 | 146 | 197 | 112 | 127 | 162 | 104 | 157 | 186 | 0 | 1(0.00485 | 0 |
| 184 | 85 | 120 | 146 | 197 | 108 | 123 | 166 | 104 | 140 | 194 | 0 | 1(0.00485 | 0 |
| 185 | 85 | 120 | 146 | 193 | 108 | 135 | 162 | 104 | 157 | 182 | 0 | 3(0.0146) | 1(0.0106) |
| 186 | 85 | 120 | 146 | 193 | 112 | 123 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 187 | 85 | 124 | 146 | 201 | 112 | 131 | 162 | 104 | 157 | 182 | 0 | 6(0.0291) | 0 |
| 188 | 85 | 128 | 146 | 197 | 116 | 131 | 162 | 104 | 157 | 182 | 0 | 2(0.00971 | 0 |
| 189 | 85 | 128 | 146 | 193 | 108 | 131 | 162 | 104 | 151 | 182 | 0 | 1(0.00485 | 0 |
| 190 | 85 | 124 | 146 | 201 | 116 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 191 | 85 | 124 | 146 | 193 | 112 | 135 | 164 | 110 | 162 | 186 | 0 | 1(0.00485 | 0 |
| 192 | 85 | 124 | 146 | 193 | 108 | 135 | 162 | 104 | 157 | 182 | 0 | 4(0.0194) | 0 |
| 193 | 85 | 128 | 146 | 193 | 108 | 131 | 162 | 104 | 157 | 178 | 0 | 1(0.00485 | 0 |
| 194 | 85 | 124 | 146 | 201 | 112 | 131 | 162 | 104 | 157 | 178 | 0 | 5(0.0243) | 0 |
| 195 | 85 | 132 | 146 | 193 | 112 | 131 | 162 | 104 | 157 | 178 | 0 | 1(0.00485 | 0 |
| 196 | 85 | 120 | 146/151 | 197 | 112 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 197 | 85 | 120 | 146 | 189 | 112 | 135 | 162 | 110 | 151 | 186 | 0 | 1(0.00485 | 0 |
| 198 | 85 | 124 | 146 | 201 | 108 | 131 | 162 | 103 | 162 | 178 | 0 | 1(0.00485 | 0 |
| 199 | 85 | 124 | 151 | 193 | 108 | 131 | 166 | 104 | 140 | 182 | 0 | 1(0.00485 | 0 |
| 200 | 85 | 124 | 146 | 201 | 108 | 124 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 201 | 85 | 128 | 146 | 197 | 108 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 202 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 104 | 140 | 190 | 0 | 1(0.00485 | 0 |
| 203 | 85 | 128 | 146 | 197 | 108 | 139 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |

Table 4, continued. Haplotypes found in the Caucasian, African American, and SE Hispanic population ($n$ = 525).

| Hap. | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 | Cauc. | Af. Amer. | SE Hisp. |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 204 | 85 | 124 | 146 | 193 | 112 | 127 | 162 | 104 | 157 | 182 | 0 | 2(0.00971 | 0 |
| 205 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 104 | 157 | 182 | 0 | 3(0.0146) | 0 |
| 206 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 157 | 182 | 0 | 5(0.0243) | 0 |
| 207 | 85 | 128 | 146 | 193 | 108 | 131 | 162 | 104 | 146 | 194 | 0 | 1(0.00485 | 0 |
| 208 | 85 | 120 | 151 | 201 | 108 | 131 | 162 | 107 | 151 | 194 | 0 | 1(0.00485 | 0 |
| 209 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 104 | 151 | 190 | 0 | 1(0.00485 | 1(0.0106) |
| 210 | 88 | 124 | 146 | 197 | 108 | 135 | 166 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 211 | 85 | 120 | 146 | 193 | 112 | 135 | 162 | 104 | 146 | 190 | 0 | 1(0.00485 | 0 |
| 212 | 85 | 120 | 146 | 197 | 112 | 131 | 162 | 104 | 157 | 182 | 0 | 2(0.00971 | 0 |
| 213 | 85 | 128 | 146 | 193 | 112 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 214 | 85 | 120 | 146 | 193 | 112 | 131 | 162 | 107 | 151 | 190 | 0 | 1(0.00485 | 0 |
| 215 | 85 | 124 | 146 | 193 | 108 | 135 | 166 | 104 | 140 | 182 | 0 | 1(0.00485 | 0 |
| 216 | 85 | 120 | 146 | 197 | 112 | 127 | 162 | 104 | 157 | 182 | 0 | 2(0.00971 | 0 |
| 217 | 85 | 120 | 146 | 189 | 120 | 131 | 170 | 110 | 162 | 186 | 0 | 1(0.00485 | 0 |
| 218 | 85 | 120 | 146 | 189 | 120 | 131 | 170 | 109 | 162 | 186 | 0 | 1(0.00485 | 0 |
| 219 | 85 | 124 | 146 | 201 | 112 | 127 | 162 | 107 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 220 | 88 | 124 | 146 | 193 | 108 | 127 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 221 | 85 | 124 | 146 | 197 | 112 | 135 | 162 | 104 | 162 | 182 | 0 | 1(0.00485 | 0 |
| 222 | 85 | 128 | 146 | 193 | 108 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 223 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 110 | 162 | 186 | 0 | 1(0.00485 | 0 |
| 224 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 225 | 85 | 120 | 146 | 189 | 108 | 131 | 162 | 104 | 157 | 186 | 0 | 1(0.00485 | 0 |
| 226 | 85 | 128 | 146 | 189 | 108 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 227 | 85 | 128 | 146 | 193 | 108 | 135 | 162 | 101 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 228 | 85 | 124 | 146 | 201 | 112 | 131 | 166 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 229 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 162 | 182 | 0 | 1(0.00485 | 0 |
| 230 | 85 | 120 | 146 | 193 | 108 | 135 | 170 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 231 | 85 | 124 | 146 | 193 | 104 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 232 | 85 | 120 | 146 | 193 | 108 | 135 | 166 | 104 | 157 | 182 | 0 | 2(0.00971 | 1(0.0106) |
| 233 | 85 | 128 | 146 | 185 | 104 | 131 | 162 | 104 | 151 | 182 | 0 | 1(0.00485 | 0 |
| 234 | 85 | 120 | 146 | 197 | 116 | 131 | 162 | 104 | 157 | 182 | 0 | 2(0.00971 | 0 |
| 235 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 104 | 157 | 182 | 0 | 2(0.00971 | 0 |
| 236 | 85 | 128 | 146 | 189 | 112 | 131 | 162 | 110 | 162 | 186 | 0 | 1(0.00485 | 0 |
| 237 | 85 | 124 | 146 | 201 | 112 | 139 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 238 | 85 | 124 | 146 | 193 | 108 | 131 | 166 | 104 | 157 | 178 | 0 | 1(0.00485 | 0 |
| 239 | 82 | 128 | 146 | 193 | 116 | 135 | 166 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 240 | 85 | 128 | 146 | 193 | 112 | 131 | 162 | 104 | 162 | 182 | 0 | 1(0.00485 | 0 |
| 241 | 85 | 128 | 146 | 189 | 112 | 139 | 162 | 110 | 167 | 186 | 0 | 1(0.00485 | 0 |
| 242 | 85 | 124 | 146 | 201 | 108 | 127 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 243 | 85 | 124 | 146 | 197 | 112 | 127 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 244 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 104 | 157 | 182 | 0 | 2(0.00971 | 0 |

Table 4, continued. Haplotypes found in the Caucasian, African American, and SE Hispanic population ($n$ = 525).

| Hap. | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 | Cauc. | Af. Amer. | SE Hisp. |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 245 | 85 | 120 | 146 | 189 | 108 | 131 | 166 | 104 | 157 | 186 | 0 | 2(0.00971) | 0 |
| 246 | 85 | 124 | 146 | 193 | 108 | 135 | 162 | 98 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 247 | 85 | 120 | 146 | 201 | 112 | 131 | 166 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 248 | 85 | 132 | 146 | 201 | 112 | 131 | 162 | 104 | 157 | 178 | 0 | 1(0.00485) | 0 |
| 249 | 85 | 132 | 146 | 193 | 112 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 250 | 85 | 128 | 146 | 201 | 108 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 251 | 85 | 128 | 126 | 193 | 108 | 135 | 166 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 252 | 85 | 128 | 146 | 198 | 108 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 253 | 85 | 128 | 146 | 197 | 112 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 254 | 85 | 124 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 182 | 0 | 1(0.00485) | 0 |
| 255 | 85 | 120 | 151 | 193 | 112 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 256 | 79 | 124 | 146 | 201 | 108 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 257 | 85 | 124 | 146 | 189 | 104 | 135 | 166 | 110 | 162 | 182 | 0 | 1(0.00485) | 0 |
| 258 | 82 | 124 | 146 | 193 | 116 | 135 | 162 | 104 | 151 | 182 | 0 | 1(0.00485) | 0 |
| 259 | 85 | 132 | 146 | 197 | 112 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 260 | 85 | 120 | 146 | 197 | 108 | 131 | 178 | 104 | 151 | 198 | 0 | 1(0.00485) | 0 |
| 261 | 88 | 124 | 146 | 193 | 108 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 262 | 85 | 124 | 146 | 201 | 112 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 263 | 85 | 128 | 146 | 201 | 108 | 127 | 162 | 104 | 157 | 186 | 0 | 1(0.00485) | 0 |
| 264 | 85 | 128 | 146 | 193 | 96 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 265 | 85 | 120 | 146 | 189 | 108 | 131 | 166 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 266 | 85 | 120 | 146 | 193 | 112 | 131 | 162 | 107 | 146 | 182 | 0 | 1(0.00485) | 0 |
| 267 | 85 | 124 | 146 | 205 | 112 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 268 | 85 | 120 | 146 | 193 | 108 | 127 | 162 | 107 | 151 | 182 | 0 | 1(0.00485) | 0 |
| 269 | 85 | 124 | 146 | 201 | 108 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 270 | 85 | 120 | 146 | 197 | 108 | 131 | 162 | 107 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 271 | 85 | 128 | 146 | 201 | 108 | 131 | 162 | 104 | 157 | 178 | 0 | 1(0.00485) | 0 |
| 272 | 85 | 128 | 146 | 193 | 112 | 127 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 273 | 85 | 124 | 146 | 185 | 108 | 131 | 162 | 104 | 151 | 182 | 0 | 1(0.00485) | 0 |
| 274 | 85 | 124 | 146 | 197 | 108 | 131 | 162 | 104 | 167 | 182 | 0 | 1(0.00485) | 0 |
| 275 | 85 | 124 | 146 | 201 | 112 | 131 | 162 | 104 | 162 | 182 | 0 | 1(0.00485) | 0 |
| 276 | 85 | 124 | 146 | 193 | 112 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 277 | 85 | 128 | 146 | 189 | 108 | 135 | 166 | 113 | 162 | 186 | 0 | 1(0.00485) | 1(0.0106) |
| 278 | 85 | 124 | 146 | 197 | 112 | 131 | 162 | 104 | 162 | 182 | 0 | 1(0.00485) | 0 |
| 279 | 85 | 132 | 146 | 193 | 116 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485) | 0 |
| 280 | 85 | 124 | 146 | 193 | 108 | 131 | 162 | 104 | 162 | 182 | 0 | 1(0.00485) | 0 |
| 281 | 85 | 128 | 146 | 189 | 116 | 131 | 166 | 110 | 162 | 190 | 0 | 1(0.00485) | 0 |
| 282 | 82 | 120 | 146 | 197 | 112 | 135 | 166 | 104 | 151 | 182 | 0 | 1(0.00485) | 0 |
| 283 | 85 | 124 | 146 | 189 | 116 | 139 | 166 | 107 | 162 | 186 | 0 | 1(0.00485) | 0 |
| 284 | 85 | 128 | 146 | 197 | 108 | 131 | 166 | 104 | 162 | 182 | 0 | 1(0.00485) | 0 |
| 285 | 85 | 128 | 146 | 197 | 108 | 131 | 162 | 104 | 157 | 182 | 0 | 2(0.00971) | 0 |

Table 4, continued. Haplotypes found in the Caucasian, African American, and SE Hispanic population ($n$ = 525).

| Hap. | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 | Cauc. | Af. Amer. | SE Hisp. |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 286 | 85 | 124 | 146 | 197 | 108 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 287 | 82 | 124 | 146 | 197 | 112 | 135 | 162 | 104 | 157 | 186 | 0 | 2(0.00971 | 0 |
| 288 | 82 | 120 | 146 | 197 | 112 | 127 | 162 | 104 | 151 | 182 | 0 | 1(0.00485 | 0 |
| 289 | 85 | 128 | 146 | 189 | 108 | 131 | 166 | 110 | 162 | 182 | 0 | 1(0.00485 | 0 |
| 290 | 85 | 124 | 146 | 197 | 112 | 131 | 162 | 101 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 291 | 82 | 128 | 146 | 193 | 112 | 135 | 166 | 104 | 151 | 182 | 0 | 1(0.00485 | 0 |
| 292 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 104 | 151 | 182 | 0 | 1(0.00485 | 0 |
| 293 | 85 | 120 | 146 | 189 | 108 | 131 | 166 | 110 | 162 | 182 | 0 | 1(0.00485 | 0 |
| 294 | 85 | 124 | 146 | 201 | 112 | 131 | 162 | 104 | 162 | 178 | 0 | 1(0.00485 | 0 |
| 295 | 85 | 128 | 146 | 197 | 112 | 131 | 162 | 104 | 157 | 178 | 0 | 1(0.00485 | 0 |
| 296 | 85 | 124 | 146 | 193 | 108 | 131 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 297 | 85 | 124 | 146 | 193 | 116 | 127 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 298 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 107 | 151 | 182 | 0 | 1(0.00485 | 0 |
| 299 | 85 | 120 | 146 | 197 | 112 | 131 | 166 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 300 | 85 | 124 | 142 | 193 | 108 | 135 | 162 | 104 | 157 | 182 | 0 | 1(0.00485 | 0 |
| 301 | 85 | 120 | 146 | 201 | 104 | 135 | 162 | 104 | 140 | 190 | 0 | 1(0.00485 | 0 |
| 302 | 85 | 120 | 146 | 185 | 112 | 135 | 162 | 122 | 157 | 182 | 0 | 0 | 1(0.0106) |
| 303 | 85 | 124 | 146 | 197 | 112 | 135 | 166 | 104 | 151 | 182 | 0 | 0 | 1(0.0106) |
| 304 | 85 | 128 | 146 | 197 | 112 | 131 | 166 | 113 | 162 | 186 | 0 | 0 | 1(0.0106) |
| 305 | 85 | 120 | 146 | 185 | 104 | 135 | 162 | 104 | 151 | 182 | 0 | 0 | 1(0.0106) |
| 306 | 85 | 124 | 146 | 189 | 116 | 135 | 166 | 107 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 307 | 85 | 124 | 146 | 185 | 112 | 135 | 162 | 104 | 151 | 182 | 0 | 0 | 1(0.0106) |
| 308 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 146 | 182 | 0 | 0 | 2(0.0213) |
| 309 | 85 | 124 | 146 | 185 | 112 | 131 | 162 | 119 | 157 | 182 | 0 | 0 | 1(0.0106) |
| 310 | 85 | 128 | 146 | 193 | 112 | 135 | 162 | 110 | 162 | 186 | 0 | 0 | 1(0.0106) |
| 311 | 85 | 120 | 146 | 189 | 120 | 135 | 162 | 110 | 162 | 186 | 0 | 0 | 1(0.0106) |
| 312 | 85 | 120 | 146 | 197 | 116 | 131 | 162 | 104 | 146 | 186 | 0 | 0 | 1(0.0106) |
| 313 | 85 | 124 | 146 | 189 | 108 | 131 | 158 | 104 | 135 | 190 | 0 | 0 | 1(0.0106) |
| 314 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 110 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 315 | 85 | 124 | 146 | 185 | 108 | 131 | 162 | 104 | 146 | 186 | 0 | 0 | 1(0.0106) |
| 316 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 113 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 317 | 85 | 124 | 146 | 185 | 108 | 135 | 162 | 119 | 157 | 186 | 0 | 0 | 1(0.0106) |
| 318 | 85 | 128 | 146 | 185 | 108 | 131 | 158 | 104 | 151 | 182 | 0 | 0 | 1(0.0106) |
| 319 | 85 | 124 | 146 | 185 | 112 | 123 | 162 | 104 | 151 | 182 | 0 | 0 | 2(0.0213) |
| 320 | 85 | 128 | 149 | 193 | 112 | 127 | 162 | 104 | 146 | 190 | 0 | 0 | 1(0.0106) |
| 321 | 79 | 124 | 146 | 189 | 108 | 139 | 162 | 104 | 146 | 182 | 0 | 0 | 1(0.0106) |
| 322 | 85 | 124 | 146 | 193 | 112 | 139 | 162 | 104 | 157 | 182 | 0 | 0 | 1(0.0106) |
| 323 | 85 | 120 | 146 | 189 | 108 | 135 | 166 | 110 | 151 | 186 | 0 | 0 | 1(0.0106) |
| 324 | 85 | 112 | 146 | 189 | 112 | 135 | 162 | 107 | 162 | 186 | 0 | 0 | 1(0.0106) |
| 325 | 85 | 120 | 146 | 193 | 108 | 135 | 162 | 104 | 151 | 190 | 0 | 0 | 1(0.0106) |
| 326 | 85 | 112 | 146 | 185 | 112 | 135 | 162 | 110 | 157 | 182 | 0 | 0 | 1(0.0106) |

Table 4, continued. Haplotypes found in the Caucasian, African American, and SE Hispanic population ($n = 525$).

| Hap. | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 | Cauc. | Af. Amer. | SE Hisp. |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 327 | 85 | 116 | 146 | 189 | 112 | 131 | 162 | 104 | 146 | 182 | 0 | 0 | 1(0.0106) |
| 328 | 85 | 120 | 146 | 201 | 108 | 127 | 166 | 104 | 151 | 186 | 0 | 0 | 1(0.0106) |
| 329 | 85 | 124 | 146 | 189 | 108 | 131 | 170 | 110 | 167 | 186 | 0 | 0 | 1(0.0106) |
| 330 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 110 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 331 | 85 | 120 | 146 | 185 | 108 | 127 | 162 | 113 | 157 | 182 | 0 | 0 | 1(0.0106) |
| 332 | 85 | 128 | 146 | 189 | 116 | 131 | 166 | 110 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 333 | 85 | 124 | 146 | 201 | 108 | 131 | 162 | 104 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 334 | 85 | 124 | 146 | 185 | 108 | 131 | 162 | 110 | 151 | 182 | 0 | 0 | 1(0.0106) |
| 335 | 85 | 124 | 151 | 193 | 108 | 135 | 162 | 104 | 157 | 182 | 0 | 0 | 1(0.0106) |
| 336 | 85 | 124 | 146 | 193 | 112 | 131 | 166 | 110 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 337 | 85 | 128 | 146 | 193 | 108 | 131 | 162 | 101 | 151 | 190 | 0 | 0 | 1(0.0106) |
| 338 | 85 | 120 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 339 | 85 | 116 | 146 | 185 | 112 | 131 | 158 | 104 | 151 | 182 | 0 | 0 | 1(0.0106) |
| 340 | 85 | 128 | 146 | 193 | 108 | 131 | 166 | 110 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 341 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 107 | 151 | 190 | 0 | 0 | 1(0.0106) |
| 342 | 82 | 124 | 146 | 197 | 112 | 131 | 162 | 104 | 151 | 182 | 0 | 0 | 1(0.0106) |
| 343 | 85 | 124 | 146 | 185 | 112 | 135 | 162 | 122 | 157 | 182 | 0 | 0 | 1(0.0106) |
| 344 | 85 | 120 | 146 | 185 | 108 | 135 | 162 | 113 | 157 | 182 | 0 | 0 | 1(0.0106) |
| 345 | 85 | 136 | 146 | 201 | 112 | 131 | 162 | 104 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 346 | 85 | 124 | 146 | 193 | 108 | 135 | 162 | 110 | 162 | 186 | 0 | 0 | 1(0.0106) |
| 347 | 85 | 116 | 146 | 185 | 108 | 139 | 158 | 104 | 151 | 182 | 0 | 0 | 1(0.0106) |
| 348 | 85 | 120 | 146 | 189 | 108 | 131 | 162 | 110 | 162 | 182 | 0 | 0 | 2(0.0213) |
| 349 | 85 | 116 | 151 | 197 | 112 | 135 | 162 | 107 | 151 | 186 | 0 | 0 | 1(0.0106) |
| 350 | 85 | 124 | 146 | 189 | 112 | 131 | 162 | 104 | 151 | 182 | 0 | 0 | 1(0.0106) |
| 351 | 85 | 120 | 146 | 197 | 108 | 131 | 166 | 104 | 157 | 182 | 0 | 0 | 1(0.0106) |
| 352 | 85 | 124 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 353 | 85 | 124 | 146 | 185 | 108 | 131 | 162 | 113 | 157 | 182 | 0 | 0 | 1(0.0106) |
| 354 | 82 | 124 | 134 | 193 | 112 | 131 | 162 | 103 | 140 | 182 | 0 | 0 | 1(0.0106) |
| 355 | 85 | 120 | 146 | 189 | 112 | 139 | 166 | 110 | 162 | 186 | 0 | 0 | 1(0.0106) |
| 256 | 85 | 116 | 146 | 185 | 108 | 131 | 162 | 104 | 151 | 182 | 0 | 0 | 1(0.0106) |
| 257 | 85 | 124 | 146 | 185 | 104 | 135 | 166 | 104 | 151 | 182 | 0 | 0 | 1(0.0106) |
| 358 | 85 | 124 | 151 | 189 | 112 | 131 | 166 | 110 | 162 | 186 | 0 | 0 | 1(0.0106) |
| 359 | 85 | 124 | 146 | 189 | 108 | 139 | 166 | 110 | 162 | 182 | 0 | 0 | 1(0.0106) |
| 360 | 85 | 124 | 146 | 185 | 108 | 131 | 158 | 110 | 157 | 182 | 0 | 0 | 1(0.0106) |
| 361 | 85 | 124 | 151 | 189 | 112 | 131 | 162 | 116 | 146 | 182 | 0 | 0 | 1(0.0106) |
| 362 | 85 | 128 | 146 | 193 | 112 | 135 | 166 | 104 | 151 | 186 | 0 | 0 | 1(0.0106) |
| 363 | 85 | 124 | 146 | 189 | 112 | 135 | 162 | 116 | 157 | 182 | 0 | 0 | 1(0.0106) |
| 364 | 85 | 128 | 146 | 193 | 112 | 127 | 162 | 104 | 157 | 186 | 0 | 0 | 1(0.0106) |
| 365 | 85 | 120 | 146 | 193 | 112 | 139 | 162 | 104 | 156 | 190 | 0 | 0 | 1(0.0106) |

Table 5-A. Haplotypes found in the Caucasian population. All sizes are in base pairs. Haplotype number does not necessarily correspond to the haplotype numbers listed in Table 4.

| Hap. | Freq. | td. Dev | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 116 | 131 | 166 | 113 | 162 | 182 |
| 2 | 0.0133 | 0.0077 | 85 | 120 | 146 | 189 | 108 | 131 | 162 | 104 | 151 | 190 |
| 3 | 0.0044 | 0.0044 | 85 | 132 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 186 |
| 4 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 116 | 131 | 166 | 110 | 162 | 182 |
| 5 | 0.0044 | 0.0044 | 85 | 120 | 146 | 193 | 104 | 131 | 162 | 107 | 151 | 182 |
| 6 | 0.0044 | 0.0044 | 85 | 124 | 151 | 193 | 112 | 135 | 166 | 110 | 162 | 186 |
| 7 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 108 | 131 | 166 | 110 | 162 | 182 |
| 8 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 112 | 131 | 166 | 107 | 151 | 186 |
| 9 | 0.0667 | 0.0167 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 |
| 10 | 0.0044 | 0.0044 | 85 | 124 | 146 | 185 | 104 | 135 | 162 | 104 | 151 | 182 |
| 11 | 0.0044 | 0.0044 | 85 | 120 | 146 | 185 | 104 | 135 | 162 | 98 | 151 | 182 |
| 12 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 135 | 162 | 110 | 157 | 182 |
| 13 | 0.0311 | 0.0116 | 85 | 120 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 |
| 14 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 167 | 186 |
| 15 | 0.0222 | 0.0098 | 85 | 128 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 |
| 16 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 167 | 190 |
| 17 | 0.0044 | 0.0044 | 85 | 116 | 146 | 197 | 112 | 135 | 166 | 104 | 157 | 182 |
| 18 | 0.0311 | 0.0116 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 113 | 162 | 186 |
| 19 | 0.0044 | 0.0044 | 85 | 124 | 146 | 185 | 108 | 127 | 162 | 116 | 157 | 182 |
| 20 | 0.0089 | 0.0063 | 85 | 128 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 186 |
| 21 | 0.0044 | 0.0044 | 85 | 132 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 |
| 22 | 0.0044 | 0.0044 | 91 | 124 | 146 | 189 | 112 | 139 | 162 | 110 | 162 | 186 |
| 23 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 108 | 135 | 166 | 104 | 146 | 182 |
| 24 | 0.0089 | 0.0063 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 186 |
| 25 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 116 | 135 | 166 | 113 | 162 | 186 |
| 26 | 0.0044 | 0.0044 | 85 | 120 | 146 | 193 | 112 | 131 | 162 | 107 | 151 | 186 |
| 27 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 108 | 135 | 162 | 110 | 162 | 186 |
| 28 | 0.0178 | 0.0088 | 85 | 120 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 186 |
| 29 | 0.0044 | 0.0044 | 85 | 124 | 142 | 189 | 112 | 131 | 166 | 110 | 162 | 186 |
| 30 | 0.0044 | 0.0044 | 85 | 124 | 146 | 185 | 108 | 135 | 162 | 104 | 151 | 182 |
| 31 | 0.0222 | 0.0098 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 104 | 151 | 190 |
| 32 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 135 | 162 | 113 | 162 | 186 |
| 33 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 147 | 166 | 110 | 162 | 186 |
| 34 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 131 | 162 | 104 | 146 | 186 |
| 35 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 104 | 151 | 182 |
| 36 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 131 | 162 | 110 | 162 | 182 |
| 37 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 108 | 135 | 162 | 104 | 151 | 190 |
| 38 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 190 |
| 39 | 0.0267 | 0.0108 | 85 | 124 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 186 |
| 40 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 108 | 135 | 166 | 110 | 167 | 186 |
| 41 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 104 | 151 | 194 |

Table 5-A, continued. Haplotypes found in the Caucasian population.

| Hap. | Freq. | td. Dev | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 42 | 0.0044 | 0.0044 | 82 | 124 | 146 | 193 | 116 | 135 | 166 | 104 | 151 | 182 |
| 43 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 116 | 135 | 166 | 110 | 162 | 186 |
| 44 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 104 | 123 | 162 | 104 | 146 | 186 |
| 45 | 0.0133 | 0.0077 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 107 | 151 | 182 |
| 46 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 151 | 182 |
| 47 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 113 | 162 | 186 |
| 48 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 108 | 131 | 162 | 104 | 146 | 186 |
| 49 | 0.0133 | 0.0077 | 85 | 124 | 146 | 189 | 112 | 139 | 166 | 110 | 162 | 186 |
| 50 | 0.0089 | 0.0063 | 85 | 120 | 146 | 189 | 112 | 135 | 162 | 110 | 167 | 186 |
| 51 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 131 | 166 | 107 | 162 | 186 |
| 52 | 0.0089 | 0.0063 | 85 | 120 | 146 | 193 | 112 | 135 | 166 | 110 | 162 | 186 |
| 53 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 190 |
| 54 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 112 | 135 | 170 | 110 | 162 | 186 |
| 55 | 0.0044 | 0.0044 | 85 | 124 | 146 | 201 | 108 | 135 | 166 | 104 | 157 | 178 |
| 56 | 0.0044 | 0.0044 | 85 | 120 | 142 | 193 | 108 | 135 | 166 | 110 | 162 | 186 |
| 57 | 0.0044 | 0.0044 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 104 | 151 | 186 |
| 58 | 0.0044 | 0.0044 | 85 | 124 | 146 | 201 | 108 | 135 | 162 | 104 | 151 | 182 |
| 59 | 0.0044 | 0.0044 | 85 | 120 | 146 | 188 | 112 | 135 | 166 | 110 | 162 | 186 |
| 60 | 0.0089 | 0.0063 | 85 | 124 | 146 | 189/193 | 104 | 131 | 162 | 104 | 151 | 182 |
| 61 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 113 | 162 | 182 |
| 62 | 0.0044 | 0.0044 | 85 | 124 | 146 | 201 | 112 | 127 | 162 | 104 | 157 | 182 |
| 63 | 0.0044 | 0.0044 | 85 | 120 | 146 | 193 | 112 | 135 | 162 | 104 | 151 | 186 |
| 64 | 0.0044 | 0.0044 | 85 | 108 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 182 |
| 65 | 0.0044 | 0.0044 | 85 | 120 | 146 | 197 | 108 | 135 | 162 | 104 | 151 | 190 |
| 66 | 0.0044 | 0.0044 | 85 | 124 | 146 | 197 | 108 | 131 | 162 | 110 | 162 | 186 |
| 67 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 108 | 139 | 166 | 110 | 162 | 186 |
| 68 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 116 | 135 | 166 | 112 | 162 | 186 |
| 69 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 182 |
| 70 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 131 | 166 | 107 | 162 | 190 |
| 71 | 0.0089 | 0.0063 | 85 | 124 | 146 | 189 | 112 | 135 | 170 | 110 | 162 | 186 |
| 72 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 186 |
| 73 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 139 | 166 | 110 | 162 | 186 |
| 74 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 112 | 162 | 186 |
| 75 | 0.0044 | 0.0044 | 85 | 124 | 146 | 201 | 108 | 135 | 162 | 104 | 157 | 182 |
| 76 | 0.0133 | 0.0077 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 104 | 151 | 190 |
| 77 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 108 | 135 | 162 | 110 | 162 | 186 |
| 78 | 0.0089 | 0.0063 | 85 | 120 | 146 | 189 | 108 | 131 | 162 | 104 | 146 | 186 |
| 79 | 0.0044 | 0.0044 | 85 | 120 | 146 | 185 | 112 | 135 | 162 | 110 | 162 | 186 |
| 80 | 0.0044 | 0.0044 | 85 | 120 | 146 | 197 | 108 | 135 | 166 | 104 | 156 | 182 |
| 81 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 112 | 135 | 162 | 110 | 162 | 186 |
| 82 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 112 | 139 | 162 | 110 | 162 | 186 |

Table 5-A, continued. Haplotypes found in the Caucasian population.

| Hap. | Freq. | td. Dev | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 83 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 143 | 162 | 110 | 167 | 186 |
| 84 | 0.0044 | 0.0044 | 85 | 120 | 146 | 193 | 112 | 127 | 162 | 107 | 151 | 182 |
| 85 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 112 | 135 | 170 | 113 | 167 | 186 |
| 86 | 0.0044 | 0.0044 | 85 | 112 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 182 |
| 87 | 0.0044 | 0.0044 | 85 | 120 | 146 | 197 | 116 | 135 | 166 | 104 | 157 | 182 |
| 88 | 0.0089 | 0.0063 | 85 | 128 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 186 |
| 89 | 0.0044 | 0.0044 | 85 | 124 | 151 | 189 | 112 | 135 | 166 | 110 | 162 | 186 |
| 90 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 112 | 135 | 166 | 113 | 162 | 186 |
| 91 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 107 | 162 | 190 |
| 92 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 104 | 131 | 162 | 104 | 146 | 182 |
| 93 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 108 | 131 | 162 | 104 | 151 | 190 |
| 94 | 0.0044 | 0.0044 | 85 | 132 | 146 | 197 | 108 | 131 | 162 | 104 | 151 | 182 |
| 95 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 104 | 151 | 186 |
| 96 | 0.0133 | 0.0077 | 85 | 124 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 186 |
| 97 | 0.0089 | 0.0063 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 110 | 162 | 186 |
| 98 | 0.0044 | 0.0044 | 85 | 128 | 146 | 193 | 104 | 131 | 170 | 104 | 151 | 186 |
| 99 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 182 |
| 100 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 113 | 167 | 186 |
| 101 | 0.0044 | 0.0044 | 85 | 124 | 146 | 201 | 116 | 131 | 162 | 107 | 151 | 186 |
| 102 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 139 | 162 | 113 | 162 | 186 |
| 103 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 182 |
| 104 | 0.0044 | 0.0044 | 85 | 128 | 151 | 193 | 104 | 135 | 166 | 110 | 162 | 186 |
| 105 | 0.0044 | 0.0044 | 85 | 120 | 146 | 193 | 108 | 131 | 169 | 104 | 151 | 190 |
| 106 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 113 | 162 | 186 |
| 107 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 116 | 131 | 166 | 110 | 157 | 190 |
| 108 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 110 | 162 | 186 |
| 109 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 182 |
| 110 | 0.0044 | 0.0044 | 88 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 |
| 111 | 0.0044 | 0.0044 | 85 | 124 | 151 | 189 | 112 | 135 | 166 | 113 | 162 | 186 |
| 112 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 135 | 166 | 113 | 162 | 190 |
| 113 | 0.0133 | 0.0077 | 85 | 124 | 146 | 189 | 108 | 131 | 166 | 110 | 162 | 186 |
| 114 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 110 | 146 | 182 |
| 115 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 157 | 186 |
| 116 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 112 | 135 | 166 | 110 | 162 | 186 |
| 117 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 112 | 135 | 162 | 110 | 162 | 182 |
| 118 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 190 |
| 119 | 0.0044 | 0.0044 | 85 | 128 | 142 | 189 | 112 | 135 | 166 | 113 | 162 | 186 |
| 120 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 139 | 166 | 113 | 162 | 186 |
| 121 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 108 | 135 | 158 | 104 | 151 | 190 |
| 122 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 110 | 167 | 186 |
| 123 | 0.0044 | 0.0044 | 85 | 132 | 151 | 189 | 108 | 135 | 158 | 107 | 151 | 190 |

Table 5-A, continued. Haplotypes found in the Caucasian population.

| Hap. | Freq. | td. Dev | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 124 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 112 | 135 | 166 | 116 | 162 | 186 |
| 125 | 0.0044 | 0.0044 | 85 | 132 | 146 | 189 | 112 | 131 | 162 | 110 | 162 | 186 |
| 126 | 0.0044 | 0.0044 | 85 | 120 | 146 | 181 | 112 | 135 | 162 | 104 | 151 | 182 |
| 127 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 151 | 190 |
| 128 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 107 | 151 | 190 |
| 129 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 112 | 131 | 162 | 110 | 146 | 186 |
| 130 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 112 | 127 | 162 | 101 | 151 | 190 |
| 131 | 0.0044 | 0.0044 | 85 | 128 | 146 | 193 | 108 | 135 | 162 | 107 | 151 | 190 |
| 132 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 127 | 162 | 103 | 151 | 190 |
| 133 | 0.0044 | 0.0044 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 104 | 151 | 182 |
| 134 | 0.0044 | 0.0044 | 85 | 120 | 146 | 181 | 108 | 131 | 162 | 104 | 151 | 182 |
| 135 | 0.0044 | 0.0044 | 79 | 120 | 146 | 189 | 112 | 135 | 162 | 113 | 162 | 186 |
| 136 | 0.0044 | 0.0044 | 85 | 120 | 146 | 205 | 112 | 135 | 162 | 104 | 157 | 182 |
| 137 | 0.0044 | 0.0044 | 85 | 125 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 186 |
| 138 | 0.0044 | 0.0044 | 85 | 124 | 142 | 189 | 116 | 131 | 162 | 104 | 146 | 190 |
| 139 | 0.0044 | 0.0044 | 85 | 124 | 151 | 193 | 108 | 131 | 162 | 104 | 146 | 186 |
| 140 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 139 | 166 | 113 | 162 | 186 |
| 141 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 116 | 135 | 162 | 110 | 162 | 186 |
| 142 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 131 | 158 | 110 | 162 | 186 |
| 143 | 0.0044 | 0.0044 | 85 | 116 | 146 | 201 | 112 | 135 | 162 | 104 | 157 | 182 |
| 144 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 182 |
| 145 | 0.0044 | 0.0044 | 85 | 128 | 142 | 201 | 112 | 139 | 162 | 104 | 151 | 186 |
| 146 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 116 | 135 | 166 | 110 | 162 | 186 |
| 147 | 0.0044 | 0.0044 | 85 | 132 | 146 | 193 | 112 | 135 | 162 | 110 | 162 | 186 |
| 148 | 0.0089 | 0.0063 | 85 | 124 | 146 | 189 | 112 | 131 | 162 | 110 | 162 | 186 |
| 149 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 186 |
| 150 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 112 | 131 | 170 | 110 | 162 | 186 |
| 151 | 0.0044 | 0.0044 | 85 | 128 | 146 | 189 | 112 | 135 | 162 | 113 | 162 | 186 |
| 152 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 116 | 135 | 166 | 110 | 162 | 190 |
| 153 | 0.0044 | 0.0044 | 85 | 128 | 146 | 185 | 108 | 135 | 166 | 110 | 162 | 186 |
| 154 | 0.0044 | 0.0044 | 85 | 120 | 146 | 189 | 116 | 131 | 166 | 110 | 162 | 186 |
| 155 | 0.0044 | 0.0044 | 88 | 120 | 146 | 189 | 112 | 131 | 166 | 110 | 167 | 186 |
| 156 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 108 | 139 | 162 | 104 | 162 | 186 |
| 157 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 116 | 135 | 166 | 110 | 162 | 186 |
| 158 | 0.0044 | 0.0044 | 85 | 124 | 146 | 193 | 108 | 131 | 166 | 110 | 162 | 182 |
| 159 | 0.0044 | 0.0044 | 85 | 116 | 146 | 185 | 112 | 135 | 158 | 104 | 151 | 182 |
| 160 | 0.0044 | 0.0044 | 85 | 128 | 146 | 193 | 112 | 131 | 162 | 103 | 151 | 190 |
| 161 | 0.0044 | 0.0044 | 85 | 124 | 146 | 189 | 108 | 135 | 166 | 113 | 162 | 186 |

Table 5-B. Haplotypes found in the African American population. All sizes are in base pairs. Haplotype number does not necessarily correspond to the haplotype numbers listed in Table 4.

| Hap. | Freq. | Std. Dev | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1 | 0.0049 | 0.0049 | 85 | 128 | 146 | 201 | 112 | 131 | 166 | 104 | 157 | 182 |
| 2 | 0.0049 | 0.0049 | 85 | 120 | 146 | 193 | 112 | 135 | 166 | 110 | 162 | 186 |
| 3 | 0.0243 | 0.0107 | 85 | 124 | 146 | 197 | 108 | 131 | 162 | 104 | 157 | 182 |
| 4 | 0.0049 | 0.0049 | 85 | 124 | 146 | 197 | 108 | 135 | 162 | 104 | 157 | 174 |
| 5 | 0.0049 | 0.0049 | 85 | 116 | 146 | 197 | 112 | 131 | 162 | 104 | 157 | 182 |
| 6 | 0.0146 | 0.0084 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 |
| 7 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 112 | 131 | 166 | 110 | 162 | 186 |
| 8 | 0.0049 | 0.0049 | 85 | 124 | 146 | 197 | 116 | 131 | 162 | 104 | 151 | 182 |
| 9 | 0.0049 | 0.0049 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 186 |
| 10 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 146 | 190 |
| 11 | 0.0049 | 0.0049 | 85 | 128 | 146 | 193 | 108 | 131 | 163 | 104 | 157 | 182 |
| 12 | 0.0146 | 0.0084 | 85 | 128 | 146 | 201 | 112 | 131 | 162 | 104 | 157 | 182 |
| 13 | 0.0097 | 0.0068 | 85 | 120 | 146 | 201 | 108 | 131 | 162 | 104 | 157 | 182 |
| 14 | 0.0291 | 0.0117 | 85 | 124 | 146 | 197 | 112 | 131 | 162 | 104 | 157 | 182 |
| 15 | 0.0097 | 0.0068 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 107 | 151 | 182 |
| 16 | 0.0049 | 0.0049 | 85 | 128 | 151 | 201 | 112 | 131 | 162 | 104 | 157 | 178 |
| 17 | 0.0049 | 0.0049 | 85 | 116 | 146 | 201 | 112 | 139 | 166 | 104 | 157 | 182 |
| 18 | 0.0049 | 0.0049 | 85 | 128 | 146 | 189 | 112 | 135 | 162 | 104 | 157 | 182 |
| 19 | 0.0146 | 0.0084 | 85 | 120 | 146 | 201 | 112 | 131 | 162 | 104 | 157 | 182 |
| 20 | 0.0049 | 0.0049 | 85 | 120 | 146 | 193 | 112 | 131 | 166 | 104 | 157 | 182 |
| 21 | 0.0049 | 0.0049 | 85 | 120 | 146 | 193 | 112 | 135 | 162 | 104 | 140 | 194 |
| 22 | 0.0097 | 0.0068 | 85 | 128 | 146 | 193 | 112 | 135 | 162 | 104 | 157 | 182 |
| 23 | 0.0049 | 0.0049 | 85 | 124 | 146 | 197 | 112 | 135 | 162 | 104 | 157 | 182 |
| 24 | 0.0049 | 0.0049 | 85 | 128 | 151 | 197 | 108 | 131 | 162 | 104 | 157 | 182 |
| 25 | 0.0097 | 0.0068 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 157 | 178 |
| 26 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 112 | 135 | 166 | 110 | 162 | 186 |
| 27 | 0.0049 | 0.0049 | 85 | 128 | 146 | 197 | 112 | 127 | 162 | 104 | 157 | 186 |
| 28 | 0.0049 | 0.0049 | 85 | 120 | 146 | 197 | 108 | 123 | 166 | 104 | 140 | 194 |
| 29 | 0.0146 | 0.0084 | 85 | 120 | 146 | 193 | 108 | 135 | 162 | 104 | 157 | 182 |
| 30 | 0.0049 | 0.0049 | 85 | 120 | 146 | 193 | 112 | 123 | 162 | 104 | 157 | 182 |
| 31 | 0.0291 | 0.0117 | 85 | 124 | 146 | 201 | 112 | 131 | 162 | 104 | 157 | 182 |
| 32 | 0.0097 | 0.0068 | 85 | 128 | 146 | 197 | 116 | 131 | 162 | 104 | 157 | 182 |
| 33 | 0.0049 | 0.0049 | 85 | 128 | 146 | 193 | 108 | 131 | 162 | 104 | 151 | 182 |
| 34 | 0.0049 | 0.0049 | 85 | 124 | 146 | 201 | 116 | 131 | 162 | 104 | 157 | 182 |
| 35 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 112 | 135 | 164 | 110 | 162 | 186 |
| 36 | 0.0194 | 0.0096 | 85 | 124 | 146 | 193 | 108 | 135 | 162 | 104 | 157 | 182 |
| 37 | 0.0049 | 0.0049 | 85 | 128 | 146 | 193 | 108 | 131 | 162 | 104 | 157 | 178 |
| 38 | 0.0243 | 0.0107 | 85 | 124 | 146 | 201 | 112 | 131 | 162 | 104 | 157 | 178 |
| 39 | 0.0049 | 0.0049 | 85 | 132 | 146 | 193 | 112 | 131 | 162 | 104 | 157 | 178 |
| 40 | 0.0049 | 0.0049 | 85 | 120 | 146/151 | 197 | 112 | 135 | 162 | 104 | 157 | 182 |
| 41 | 0.0049 | 0.0049 | 85 | 120 | 146 | 189 | 112 | 135 | 162 | 110 | 151 | 186 |

Table 5-B, continued. Haplotypes found in the African American population.

| Hap. | Freq. | Std. Dev | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 42 | 0.0049 | 0.0049 | 85 | 124 | 146 | 201 | 108 | 131 | 162 | 103 | 162 | 178 |
| 43 | 0.0049 | 0.0049 | 85 | 124 | 151 | 193 | 108 | 131 | 166 | 104 | 140 | 182 |
| 44 | 0.0049 | 0.0049 | 85 | 124 | 146 | 201 | 108 | 124 | 162 | 104 | 157 | 182 |
| 45 | 0.0049 | 0.0049 | 85 | 128 | 146 | 197 | 108 | 135 | 162 | 104 | 157 | 182 |
| 46 | 0.0049 | 0.0049 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 104 | 140 | 190 |
| 47 | 0.0049 | 0.0049 | 85 | 128 | 146 | 197 | 108 | 139 | 162 | 104 | 157 | 182 |
| 48 | 0.0097 | 0.0068 | 85 | 124 | 146 | 193 | 112 | 127 | 162 | 104 | 157 | 182 |
| 49 | 0.0146 | 0.0084 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 104 | 157 | 182 |
| 50 | 0.0243 | 0.0107 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 157 | 182 |
| 51 | 0.0049 | 0.0049 | 85 | 128 | 146 | 193 | 108 | 131 | 162 | 104 | 146 | 194 |
| 52 | 0.0049 | 0.0049 | 85 | 120 | 151 | 201 | 108 | 131 | 162 | 107 | 151 | 194 |
| 53 | 0.0049 | 0.0049 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 104 | 151 | 190 |
| 54 | 0.0049 | 0.0049 | 88 | 124 | 146 | 197 | 108 | 135 | 166 | 104 | 157 | 182 |
| 55 | 0.0049 | 0.0049 | 85 | 120 | 146 | 193 | 112 | 135 | 162 | 104 | 146 | 190 |
| 56 | 0.0097 | 0.0068 | 85 | 120 | 146 | 197 | 112 | 131 | 162 | 104 | 157 | 182 |
| 57 | 0.0049 | 0.0049 | 85 | 128 | 146 | 193 | 112 | 131 | 162 | 104 | 157 | 182 |
| 58 | 0.0049 | 0.0049 | 85 | 120 | 146 | 193 | 112 | 131 | 162 | 107 | 151 | 190 |
| 59 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 108 | 135 | 166 | 104 | 140 | 182 |
| 60 | 0.0097 | 0.0068 | 85 | 120 | 146 | 197 | 112 | 127 | 162 | 104 | 157 | 182 |
| 61 | 0.0049 | 0.0049 | 85 | 120 | 146 | 189 | 120 | 131 | 170 | 110 | 162 | 186 |
| 62 | 0.0049 | 0.0049 | 85 | 120 | 146 | 189 | 120 | 131 | 170 | 109 | 162 | 186 |
| 63 | 0.0049 | 0.0049 | 85 | 124 | 146 | 201 | 112 | 127 | 162 | 107 | 157 | 182 |
| 64 | 0.0049 | 0.0049 | 88 | 124 | 146 | 193 | 108 | 127 | 162 | 104 | 157 | 182 |
| 65 | 0.0049 | 0.0049 | 85 | 124 | 146 | 197 | 112 | 135 | 162 | 104 | 162 | 182 |
| 66 | 0.0049 | 0.0049 | 85 | 120 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 |
| 67 | 0.0049 | 0.0049 | 85 | 128 | 146 | 193 | 108 | 131 | 162 | 104 | 157 | 182 |
| 68 | 0.0049 | 0.0049 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 110 | 162 | 186 |
| 69 | 0.0049 | 0.0049 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 104 | 157 | 182 |
| 70 | 0.0049 | 0.0049 | 85 | 120 | 146 | 189 | 108 | 131 | 162 | 104 | 157 | 186 |
| 71 | 0.0049 | 0.0049 | 85 | 128 | 146 | 189 | 108 | 131 | 162 | 104 | 157 | 182 |
| 72 | 0.0049 | 0.0049 | 85 | 128 | 146 | 193 | 108 | 135 | 162 | 101 | 157 | 182 |
| 73 | 0.0049 | 0.0049 | 85 | 124 | 146 | 201 | 112 | 131 | 166 | 104 | 157 | 182 |
| 74 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 162 | 182 |
| 75 | 0.0049 | 0.0049 | 85 | 120 | 146 | 193 | 108 | 135 | 170 | 104 | 157 | 182 |
| 76 | 0.0146 | 0.0084 | 85 | 124 | 146 | 201 | 112 | 127 | 162 | 104 | 157 | 182 |
| 77 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 104 | 135 | 162 | 104 | 157 | 182 |
| 78 | 0.0097 | 0.0068 | 85 | 120 | 146 | 193 | 108 | 135 | 166 | 104 | 157 | 182 |
| 79 | 0.0049 | 0.0049 | 85 | 124 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 186 |
| 80 | 0.0049 | 0.0049 | 85 | 128 | 146 | 185 | 104 | 131 | 162 | 104 | 151 | 182 |
| 81 | 0.0097 | 0.0068 | 85 | 120 | 146 | 197 | 116 | 131 | 162 | 104 | 157 | 182 |
| 82 | 0.0097 | 0.0068 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 104 | 157 | 182 |

Table 5-B, continued. Haplotypes found in the African American population.

| Hap. | Freq. | Std. Dev | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 83 | 0.0049 | 0.0049 | 85 | 128 | 146 | 189 | 112 | 131 | 162 | 110 | 162 | 186 |
| 84 | 0.0049 | 0.0049 | 85 | 124 | 146 | 201 | 112 | 139 | 162 | 104 | 157 | 182 |
| 85 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 108 | 131 | 166 | 104 | 157 | 178 |
| 86 | 0.0049 | 0.0049 | 82 | 128 | 146 | 193 | 116 | 135 | 166 | 104 | 157 | 182 |
| 87 | 0.0049 | 0.0049 | 85 | 128 | 146 | 193 | 112 | 131 | 162 | 104 | 162 | 182 |
| 88 | 0.0049 | 0.0049 | 85 | 128 | 146 | 189 | 112 | 139 | 162 | 110 | 167 | 186 |
| 89 | 0.0049 | 0.0049 | 85 | 124 | 146 | 201 | 108 | 127 | 162 | 104 | 157 | 182 |
| 90 | 0.0049 | 0.0049 | 85 | 124 | 146 | 197 | 112 | 127 | 162 | 104 | 157 | 182 |
| 91 | 0.0097 | 0.0068 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 104 | 157 | 182 |
| 92 | 0.0097 | 0.0068 | 85 | 120 | 146 | 189 | 108 | 131 | 166 | 104 | 157 | 186 |
| 93 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 108 | 135 | 162 | 98 | 157 | 182 |
| 94 | 0.0049 | 0.0049 | 85 | 120 | 146 | 201 | 112 | 131 | 166 | 104 | 157 | 182 |
| 95 | 0.0049 | 0.0049 | 85 | 132 | 146 | 201 | 112 | 131 | 162 | 104 | 157 | 178 |
| 96 | 0.0049 | 0.0049 | 85 | 132 | 146 | 193 | 112 | 135 | 162 | 104 | 157 | 182 |
| 97 | 0.0049 | 0.0049 | 85 | 128 | 146 | 201 | 108 | 135 | 162 | 104 | 157 | 182 |
| 98 | 0.0049 | 0.0049 | 85 | 128 | 126 | 193 | 108 | 135 | 166 | 104 | 157 | 182 |
| 99 | 0.0049 | 0.0049 | 85 | 128 | 146 | 198 | 108 | 131 | 162 | 104 | 157 | 182 |
| 100 | 0.0049 | 0.0049 | 85 | 128 | 146 | 197 | 112 | 131 | 162 | 104 | 157 | 182 |
| 101 | 0.0049 | 0.0049 | 85 | 124 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 182 |
| 102 | 0.0049 | 0.0049 | 85 | 120 | 151 | 193 | 112 | 131 | 162 | 104 | 157 | 182 |
| 103 | 0.0049 | 0.0049 | 79 | 124 | 146 | 201 | 108 | 135 | 162 | 104 | 157 | 182 |
| 104 | 0.0049 | 0.0049 | 85 | 124 | 146 | 189 | 104 | 135 | 166 | 110 | 162 | 182 |
| 105 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 151 | 182 |
| 106 | 0.0049 | 0.0049 | 82 | 124 | 146 | 193 | 116 | 135 | 162 | 104 | 151 | 182 |
| 107 | 0.0049 | 0.0049 | 85 | 132 | 146 | 197 | 112 | 135 | 162 | 104 | 157 | 182 |
| 108 | 0.0049 | 0.0049 | 85 | 120 | 146 | 197 | 108 | 131 | 178 | 104 | 151 | 198 |
| 109 | 0.0049 | 0.0049 | 88 | 124 | 146 | 193 | 108 | 135 | 162 | 104 | 157 | 182 |
| 110 | 0.0049 | 0.0049 | 85 | 124 | 146 | 201 | 112 | 135 | 162 | 104 | 157 | 182 |
| 111 | 0.0049 | 0.0049 | 85 | 128 | 146 | 201 | 108 | 127 | 162 | 104 | 157 | 186 |
| 112 | 0.0049 | 0.0049 | 85 | 128 | 146 | 193 | 96 | 135 | 162 | 104 | 157 | 182 |
| 113 | 0.0049 | 0.0049 | 85 | 120 | 146 | 189 | 108 | 131 | 166 | 104 | 157 | 182 |
| 114 | 0.0049 | 0.0049 | 85 | 120 | 146 | 193 | 112 | 131 | 162 | 107 | 146 | 182 |
| 115 | 0.0049 | 0.0049 | 85 | 124 | 146 | 205 | 112 | 131 | 162 | 104 | 157 | 182 |
| 116 | 0.0049 | 0.0049 | 85 | 120 | 146 | 193 | 108 | 127 | 162 | 107 | 151 | 182 |
| 117 | 0.0097 | 0.0068 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 110 | 162 | 186 |
| 118 | 0.0049 | 0.0049 | 85 | 124 | 146 | 201 | 108 | 131 | 162 | 104 | 157 | 182 |
| 119 | 0.0049 | 0.0049 | 85 | 120 | 146 | 197 | 108 | 131 | 162 | 107 | 157 | 182 |
| 120 | 0.0049 | 0.0049 | 85 | 128 | 146 | 201 | 108 | 131 | 162 | 104 | 157 | 178 |
| 121 | 0.0049 | 0.0049 | 85 | 128 | 146 | 193 | 112 | 127 | 162 | 104 | 157 | 182 |
| 122 | 0.0049 | 0.0049 | 85 | 124 | 146 | 185 | 108 | 131 | 162 | 104 | 151 | 182 |
| 123 | 0.0049 | 0.0049 | 85 | 124 | 146 | 197 | 108 | 131 | 162 | 104 | 167 | 182 |

Table 5-B, continued. Haplotypes found in the African American population.

| Hap. | Freq. | Std. Dev | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 124 | 0.0049 | 0.0049 | 85 | 124 | 146 | 201 | 112 | 131 | 162 | 104 | 162 | 182 |
| 125 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 112 | 135 | 162 | 104 | 157 | 182 |
| 126 | 0.0049 | 0.0049 | 85 | 124 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 186 |
| 127 | 0.0049 | 0.0049 | 85 | 128 | 146 | 189 | 108 | 135 | 166 | 113 | 162 | 186 |
| 128 | 0.0049 | 0.0049 | 85 | 124 | 146 | 197 | 112 | 131 | 162 | 104 | 162 | 182 |
| 129 | 0.0049 | 0.0049 | 85 | 132 | 146 | 193 | 116 | 135 | 162 | 104 | 157 | 182 |
| 130 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 108 | 131 | 162 | 104 | 162 | 182 |
| 131 | 0.0049 | 0.0049 | 85 | 128 | 146 | 189 | 116 | 131 | 166 | 110 | 162 | 190 |
| 132 | 0.0049 | 0.0049 | 82 | 120 | 146 | 197 | 112 | 135 | 166 | 104 | 151 | 182 |
| 133 | 0.0049 | 0.0049 | 85 | 124 | 146 | 189 | 116 | 139 | 166 | 107 | 162 | 186 |
| 134 | 0.0049 | 0.0049 | 85 | 128 | 146 | 197 | 108 | 131 | 166 | 104 | 162 | 182 |
| 135 | 0.0049 | 0.0049 | 85 | 120 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 186 |
| 136 | 0.0097 | 0.0068 | 85 | 128 | 146 | 197 | 108 | 131 | 162 | 104 | 157 | 182 |
| 137 | 0.0049 | 0.0049 | 85 | 124 | 146 | 197 | 108 | 135 | 162 | 104 | 157 | 182 |
| 138 | 0.0097 | 0.0068 | 82 | 124 | 146 | 197 | 112 | 135 | 162 | 104 | 157 | 186 |
| 139 | 0.0049 | 0.0049 | 82 | 120 | 146 | 197 | 112 | 127 | 162 | 104 | 151 | 182 |
| 140 | 0.0049 | 0.0049 | 85 | 128 | 146 | 189 | 108 | 131 | 166 | 110 | 162 | 182 |
| 141 | 0.0049 | 0.0049 | 85 | 124 | 146 | 197 | 112 | 131 | 162 | 101 | 157 | 182 |
| 142 | 0.0049 | 0.0049 | 82 | 128 | 146 | 193 | 112 | 135 | 166 | 104 | 151 | 182 |
| 143 | 0.0049 | 0.0049 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 104 | 151 | 182 |
| 144 | 0.0049 | 0.0049 | 85 | 120 | 146 | 189 | 108 | 131 | 166 | 110 | 162 | 182 |
| 145 | 0.0049 | 0.0049 | 85 | 124 | 146 | 201 | 112 | 131 | 162 | 104 | 162 | 178 |
| 146 | 0.0049 | 0.0049 | 85 | 128 | 146 | 197 | 112 | 131 | 162 | 104 | 157 | 178 |
| 147 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 108 | 131 | 162 | 104 | 157 | 182 |
| 148 | 0.0049 | 0.0049 | 85 | 124 | 146 | 193 | 116 | 127 | 162 | 104 | 157 | 182 |
| 149 | 0.0049 | 0.0049 | 85 | 120 | 146 | 193 | 108 | 131 | 162 | 107 | 151 | 182 |
| 150 | 0.0049 | 0.0049 | 85 | 120 | 146 | 197 | 112 | 131 | 166 | 104 | 157 | 182 |
| 151 | 0.0049 | 0.0049 | 85 | 124 | 142 | 193 | 108 | 135 | 162 | 104 | 157 | 182 |
| 152 | 0.0049 | 0.0049 | 85 | 120 | 146 | 201 | 104 | 135 | 162 | 104 | 140 | 190 |
| 153 | 0.0049 | 0.0049 | 85 | 120 | 146 | 189 | 108 | 131 | 162 | 104 | 151 | 190 |

Table 5-C. Haplotypes found in the SE Hispanic population. All sizes are in base pairs. Haplotype number does not necessarily correspond to the haplotype numbers listed in Table 4.

| Hap. | Freq. | Std. Dev | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 1 | 0.0106 | 0.0106 | 85 | 120 | 146 | 185 | 112 | 135 | 162 | 122 | 157 | 182 |
| 2 | 0.0106 | 0.0106 | 85 | 124 | 146 | 197 | 112 | 135 | 166 | 104 | 151 | 182 |
| 3 | 0.0106 | 0.0106 | 85 | 128 | 146 | 197 | 112 | 131 | 166 | 113 | 162 | 186 |
| 4 | 0.0106 | 0.0106 | 85 | 120 | 146 | 185 | 104 | 135 | 162 | 104 | 151 | 182 |
| 5 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 116 | 135 | 166 | 107 | 162 | 182 |
| 6 | 0.0106 | 0.0106 | 85 | 124 | 146 | 185 | 112 | 135 | 162 | 104 | 151 | 182 |
| 7 | 0.0213 | 0.0150 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 104 | 146 | 182 |
| 8 | 0.0106 | 0.0106 | 85 | 124 | 146 | 185 | 112 | 131 | 162 | 119 | 157 | 182 |
| 9 | 0.0106 | 0.0106 | 85 | 128 | 146 | 193 | 112 | 135 | 162 | 110 | 162 | 186 |
| 10 | 0.0106 | 0.0106 | 85 | 120 | 146 | 189 | 120 | 135 | 162 | 110 | 162 | 186 |
| 11 | 0.0106 | 0.0106 | 85 | 120 | 146 | 197 | 116 | 131 | 162 | 104 | 146 | 186 |
| 12 | 0.0106 | 0.0106 | 85 | 120 | 146 | 189 | 104 | 131 | 162 | 104 | 146 | 182 |
| 13 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 108 | 131 | 158 | 104 | 135 | 190 |
| 14 | 0.0106 | 0.0106 | 85 | 120 | 146 | 193 | 112 | 135 | 166 | 110 | 162 | 186 |
| 15 | 0.0213 | 0.0150 | 85 | 120 | 146 | 189 | 108 | 131 | 162 | 104 | 151 | 190 |
| 16 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 110 | 162 | 182 |
| 17 | 0.0106 | 0.0106 | 85 | 124 | 146 | 185 | 108 | 131 | 162 | 104 | 146 | 186 |
| 18 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 113 | 162 | 182 |
| 19 | 0.0106 | 0.0106 | 85 | 124 | 146 | 185 | 108 | 135 | 162 | 119 | 157 | 186 |
| 20 | 0.0106 | 0.0106 | 85 | 128 | 146 | 185 | 108 | 131 | 158 | 104 | 151 | 182 |
| 21 | 0.0213 | 0.0150 | 85 | 124 | 146 | 185 | 112 | 123 | 162 | 104 | 151 | 182 |
| 22 | 0.0106 | 0.0106 | 85 | 128 | 149 | 193 | 112 | 127 | 162 | 104 | 146 | 190 |
| 23 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 112 | 131 | 162 | 110 | 162 | 186 |
| 24 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 186 |
| 25 | 0.0106 | 0.0106 | 79 | 124 | 146 | 189 | 108 | 139 | 162 | 104 | 146 | 182 |
| 26 | 0.0106 | 0.0106 | 85 | 124 | 146 | 193 | 112 | 139 | 162 | 104 | 157 | 182 |
| 27 | 0.0106 | 0.0106 | 85 | 128 | 146 | 201 | 112 | 131 | 162 | 104 | 157 | 182 |
| 28 | 0.0106 | 0.0106 | 85 | 120 | 146 | 189 | 108 | 135 | 166 | 110 | 151 | 186 |
| 29 | 0.0106 | 0.0106 | 85 | 112 | 146 | 189 | 112 | 135 | 162 | 107 | 162 | 186 |
| 30 | 0.0106 | 0.0106 | 85 | 120 | 146 | 193 | 108 | 135 | 162 | 104 | 151 | 190 |
| 31 | 0.0106 | 0.0106 | 85 | 116 | 146 | 201 | 112 | 139 | 166 | 104 | 157 | 182 |
| 32 | 0.0106 | 0.0106 | 85 | 112 | 146 | 185 | 112 | 135 | 162 | 110 | 157 | 182 |
| 33 | 0.0106 | 0.0106 | 85 | 116 | 146 | 189 | 112 | 131 | 162 | 104 | 146 | 182 |
| 34 | 0.0106 | 0.0106 | 85 | 128 | 146 | 189 | 108 | 135 | 166 | 113 | 162 | 186 |
| 35 | 0.0106 | 0.0106 | 85 | 120 | 146 | 201 | 108 | 127 | 166 | 104 | 151 | 186 |
| 36 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 108 | 131 | 170 | 110 | 167 | 186 |
| 37 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 108 | 135 | 162 | 110 | 162 | 182 |
| 38 | 0.0106 | 0.0106 | 85 | 120 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 |
| 39 | 0.0106 | 0.0106 | 85 | 120 | 146 | 185 | 108 | 127 | 162 | 113 | 157 | 182 |
| 40 | 0.0106 | 0.0106 | 85 | 128 | 146 | 189 | 116 | 131 | 166 | 110 | 162 | 182 |
| 41 | 0.0106 | 0.0106 | 85 | 124 | 146 | 201 | 108 | 131 | 162 | 104 | 162 | 182 |

Table 5-C, continued. Haplotypes found in the SE Hispanic population.

| Hap. | Freq. | Std. Dev | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 42 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 112 | 131 | 166 | 110 | 162 | 182 |
| 43 | 0.0106 | 0.0106 | 85 | 124 | 146 | 185 | 108 | 131 | 162 | 110 | 151 | 182 |
| 44 | 0.0106 | 0.0106 | 85 | 124 | 151 | 193 | 108 | 135 | 162 | 104 | 157 | 182 |
| 45 | 0.0106 | 0.0106 | 85 | 124 | 146 | 193 | 112 | 131 | 166 | 110 | 162 | 182 |
| 46 | 0.0106 | 0.0106 | 85 | 128 | 146 | 193 | 108 | 131 | 162 | 101 | 151 | 190 |
| 47 | 0.0106 | 0.0106 | 85 | 120 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 182 |
| 48 | 0.0106 | 0.0106 | 85 | 124 | 146 | 197 | 112 | 135 | 162 | 104 | 157 | 182 |
| 49 | 0.0106 | 0.0106 | 85 | 116 | 146 | 185 | 112 | 131 | 158 | 104 | 151 | 182 |
| 50 | 0.0106 | 0.0106 | 85 | 128 | 146 | 193 | 108 | 131 | 166 | 110 | 162 | 182 |
| 51 | 0.0106 | 0.0106 | 85 | 120 | 146 | 193 | 108 | 135 | 162 | 104 | 157 | 182 |
| 52 | 0.0106 | 0.0106 | 85 | 124 | 146 | 193 | 112 | 131 | 162 | 107 | 151 | 190 |
| 53 | 0.0106 | 0.0106 | 82 | 124 | 146 | 197 | 112 | 131 | 162 | 104 | 151 | 182 |
| 54 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 108 | 131 | 166 | 110 | 162 | 186 |
| 55 | 0.0106 | 0.0106 | 85 | 124 | 146 | 185 | 112 | 135 | 162 | 122 | 157 | 182 |
| 56 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 113 | 162 | 186 |
| 57 | 0.0213 | 0.0150 | 85 | 124 | 146 | 189 | 112 | 135 | 166 | 110 | 162 | 186 |
| 58 | 0.0213 | 0.0150 | 85 | 128 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 186 |
| 59 | 0.0106 | 0.0106 | 85 | 120 | 146 | 185 | 108 | 135 | 162 | 113 | 157 | 182 |
| 60 | 0.0106 | 0.0106 | 85 | 136 | 146 | 201 | 112 | 131 | 162 | 104 | 162 | 182 |
| 61 | 0.0106 | 0.0106 | 85 | 124 | 146 | 193 | 108 | 135 | 162 | 110 | 162 | 186 |
| 62 | 0.0106 | 0.0106 | 85 | 116 | 146 | 185 | 108 | 139 | 158 | 104 | 151 | 182 |
| 63 | 0.0213 | 0.0150 | 85 | 120 | 146 | 189 | 108 | 131 | 162 | 110 | 162 | 182 |
| 64 | 0.0106 | 0.0106 | 85 | 116 | 151 | 197 | 112 | 135 | 162 | 107 | 151 | 186 |
| 65 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 112 | 131 | 162 | 104 | 151 | 182 |
| 66 | 0.0106 | 0.0106 | 85 | 120 | 146 | 197 | 108 | 131 | 166 | 104 | 157 | 182 |
| 67 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 112 | 135 | 162 | 110 | 162 | 182 |
| 68 | 0.0106 | 0.0106 | 85 | 124 | 146 | 185 | 108 | 131 | 162 | 113 | 157 | 182 |
| 69 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 108 | 131 | 162 | 104 | 151 | 190 |
| 70 | 0.0106 | 0.0106 | 82 | 124 | 134 | 193 | 112 | 131 | 162 | 103 | 140 | 182 |
| 71 | 0.0106 | 0.0106 | 85 | 120 | 146 | 189 | 112 | 139 | 166 | 110 | 162 | 186 |
| 72 | 0.0106 | 0.0106 | 85 | 116 | 146 | 185 | 108 | 131 | 162 | 104 | 151 | 182 |
| 73 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 116 | 135 | 166 | 113 | 162 | 186 |
| 74 | 0.0106 | 0.0106 | 85 | 128 | 146 | 189 | 108 | 135 | 166 | 110 | 162 | 186 |
| 75 | 0.0106 | 0.0106 | 85 | 124 | 146 | 185 | 104 | 135 | 166 | 104 | 151 | 182 |
| 76 | 0.0106 | 0.0106 | 85 | 124 | 151 | 189 | 112 | 131 | 166 | 110 | 162 | 186 |
| 77 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 108 | 139 | 166 | 110 | 162 | 182 |
| 78 | 0.0106 | 0.0106 | 85 | 120 | 146 | 193 | 108 | 135 | 166 | 104 | 157 | 182 |
| 79 | 0.0106 | 0.0106 | 85 | 124 | 146 | 185 | 108 | 131 | 158 | 110 | 157 | 182 |
| 80 | 0.0106 | 0.0106 | 85 | 124 | 151 | 189 | 112 | 131 | 162 | 116 | 146 | 182 |
| 81 | 0.0106 | 0.0106 | 85 | 128 | 146 | 193 | 112 | 135 | 166 | 104 | 151 | 186 |
| 82 | 0.0213 | 0.0150 | 85 | 124 | 146 | 189 | 108 | 131 | 166 | 110 | 162 | 182 |

Table 5-C, continued. Haplotypes found in the SE Hispanic population.

| Hap. | Freq. | Std. Dev | Y436 | Y439 | Y435 | Y19 | A7.1 | H4 | Y391 | Y392 | Y438 | Y437 |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| 83 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 112 | 135 | 162 | 116 | 157 | 182 |
| 84 | 0.0106 | 0.0106 | 85 | 128 | 146 | 193 | 112 | 127 | 162 | 104 | 157 | 186 |
| 85 | 0.0106 | 0.0106 | 85 | 124 | 146 | 189 | 112 | 139 | 166 | 110 | 162 | 186 |
| 86 | 0.0106 | 0.0106 | 85 | 120 | 146 | 193 | 112 | 139 | 162 | 104 | 156 | 190 |
| 87 | 0.0106 | 0.0106 | 85 | 124 | 146 | 193 | 112 | 135 | 166 | 110 | 162 | 186 |

Table 6. Significant linkage disequilibrium in Caucasian (A), African American (B), and SE Hispanic (C) populations. "+" indicates significant linkage disequilibrium (significance level = 0.05), and "-" indicates that there is not significant linkage disequilibrium. "*" represents a cell in which the locus is analyzed against itself.

**A)**

| Locus | DYS436 | DYS439 | DYS435 | DYS19 | A7.1 | H4 | DYS391 | DYS392 | DYS438 | DYS437 |
|---|---|---|---|---|---|---|---|---|---|---|
| DYS436 | * | - | - | - | - | - | - | - | - | - |
| DYS439 | - | * | - | - | - | - | + | + | + | + |
| DYS435 | - | - | * | - | - | - | - | - | - | - |
| DYS19 | - | - | - | * | + | - | + | + | + | + |
| A7.1 | - | - | - | + | * | - | + | + | + | + |
| H4 | - | - | - | - | - | * | + | + | + | + |
| DYS391 | - | + | - | + | + | + | * | + | + | + |
| DYS392 | - | + | - | + | + | + | + | * | + | + |
| DYS438 | - | + | - | + | + | + | + | + | * | + |
| DYS437 | - | + | - | + | + | + | + | + | + | * |

**B)**

| Locus | DYS436 | DYS439 | DYS435 | DYS19 | A7.1 | H4 | DYS391 | DYS392 | DYS438 | DYS437 |
|---|---|---|---|---|---|---|---|---|---|---|
| DYS436 | * | - | - | - | - | + | - | - | - | - |
| DYS439 | - | * | - | - | - | - | - | - | - | + |
| DYS435 | - | - | * | - | - | - | - | - | - | - |
| DYS19 | - | - | - | * | + | + | + | + | + | + |
| A7.1 | - | - | - | + | * | - | + | - | - | - |
| H4 | + | - | - | + | - | * | - | - | - | + |
| DYS391 | - | - | - | + | + | - | * | + | + | + |
| DYS392 | - | - | - | + | - | - | + | * | + | + |
| DYS438 | - | - | - | + | - | - | + | + | * | + |
| DYS437 | - | + | - | + | - | + | + | + | + | * |

**C)**

| Locus | DYS436 | DYS439 | DYS435 | DYS19 | A7.1 | H4 | DYS391 | DYS392 | DYS438 | DYS437 |
|---|---|---|---|---|---|---|---|---|---|---|
| DYS436 | * | - | - | - | - | - | - | - | + | - |
| DYS439 | - | * | - | - | - | - | - | - | - | - |
| DYS435 | - | - | * | - | - | - | - | + | - | - |
| DYS19 | - | - | - | * | - | - | + | + | + | + |
| A7.1 | - | - | - | - | * | - | - | - | - | - |
| H4 | - | - | - | - | - | * | - | - | + | + |
| DYS391 | - | - | - | + | - | - | * | - | + | + |
| DYS392 | - | - | + | + | - | - | - | * | + | + |
| DYS438 | + | - | - | + | - | + | + | + | * | + |
| DYS437 | - | - | - | + | - | + | + | + | + | * |

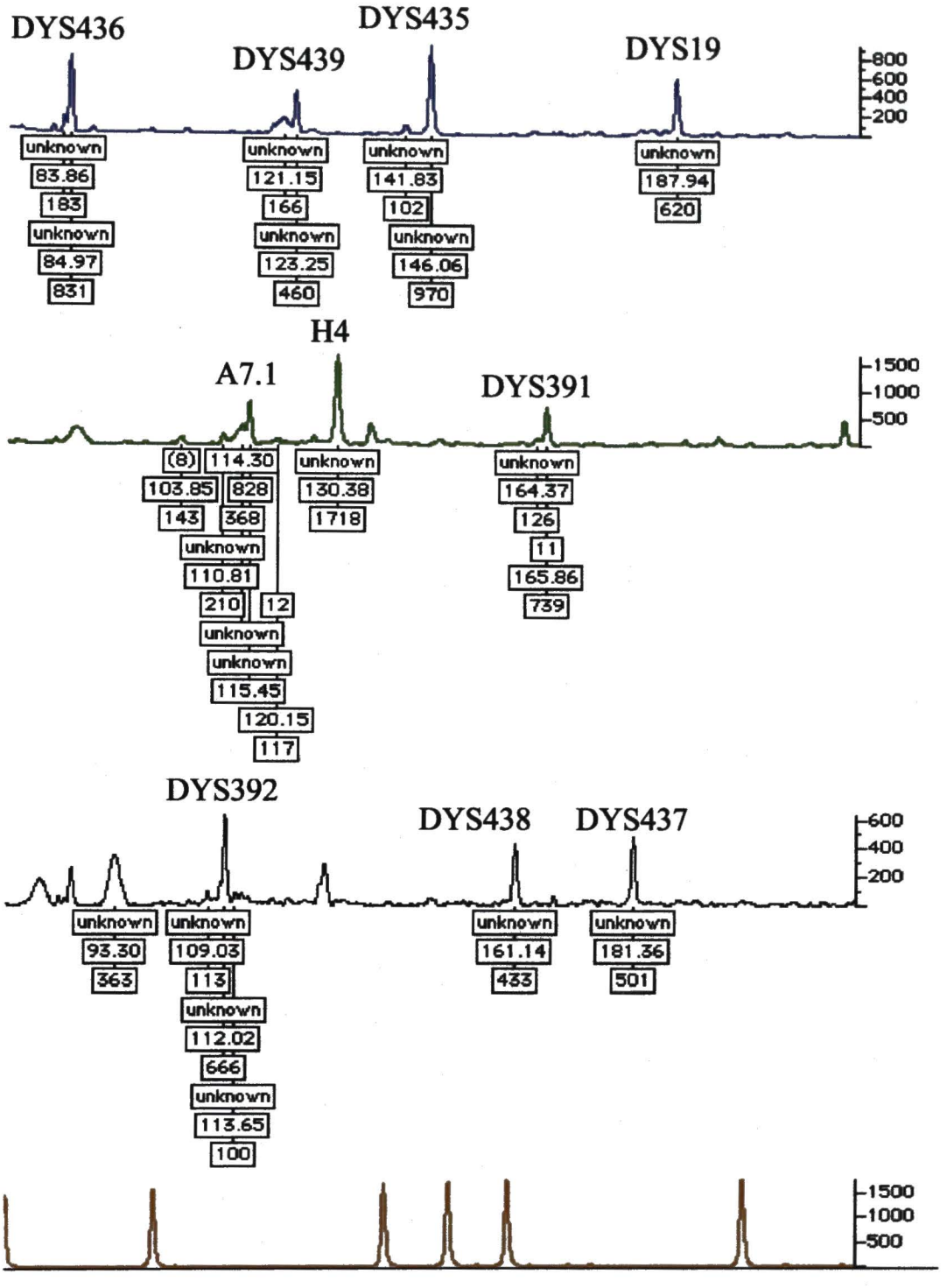



Figure 1. Y-STR 10-plex using PCR conditions established by NIST. A magnesium chloride concentration of 2 mM and an annealing temperature of 55° C were used.

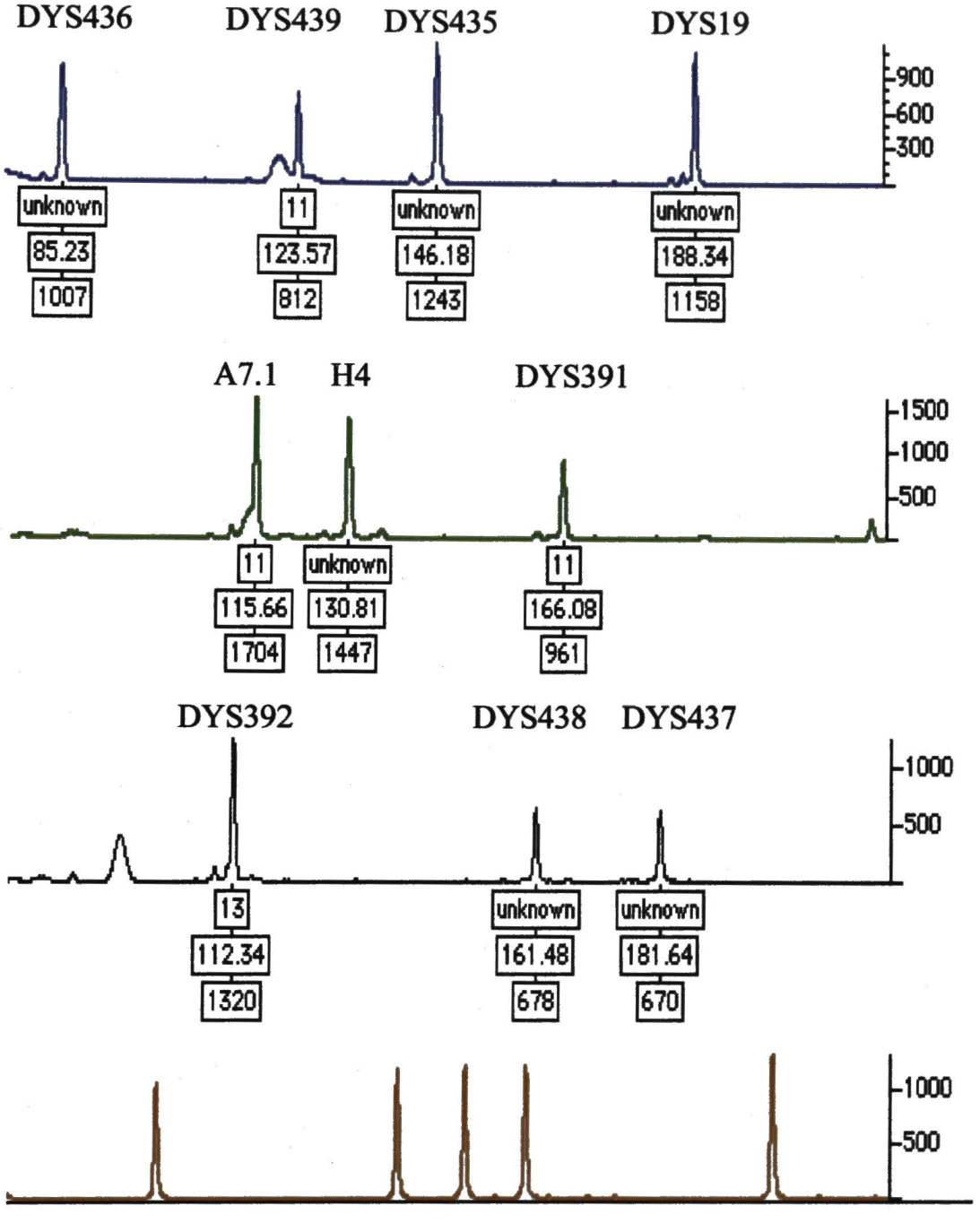



Figure 2. Addition of bovine serum albumin (BSA) to the PCR reaction. The PCR conditions included 2 mM $MgCl_2$, BSA, and a 55° C annealing temperature.

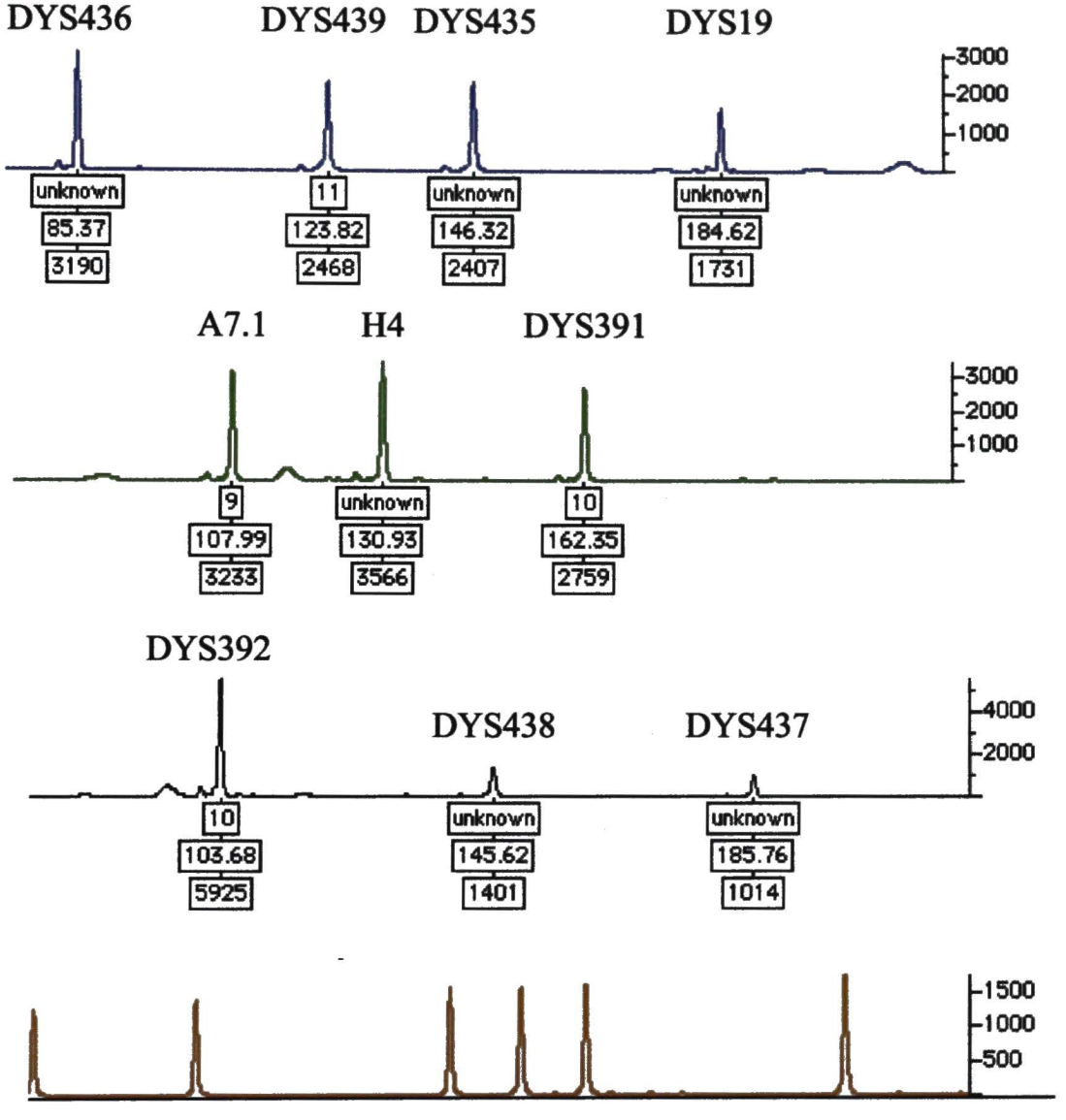



Figure 3. Increasing the annealing temperature to 60° C. The PCR reaction mix included 2 mM $MgCl_2$ and BSA.

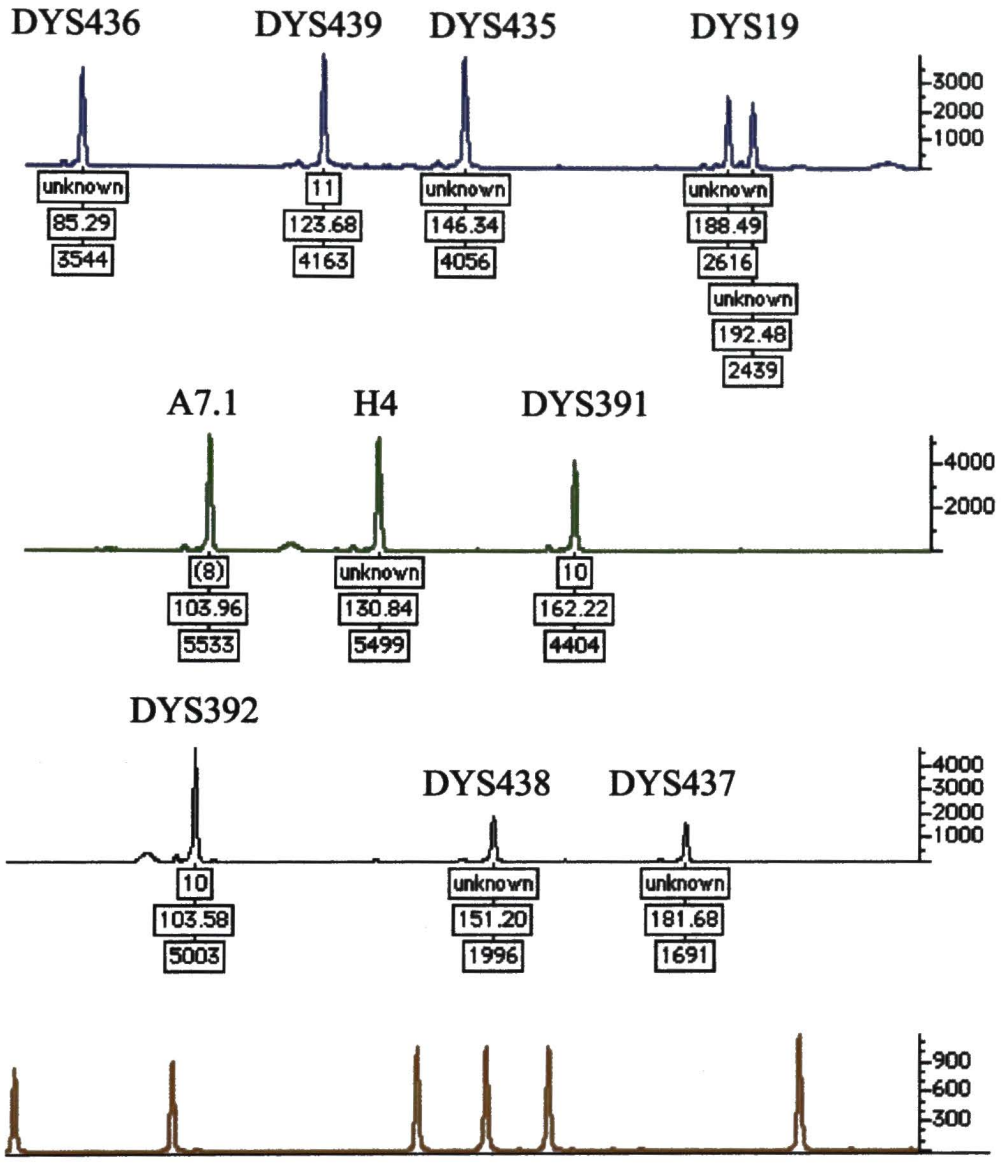



Figure 4. Duplication at the DYS19 locus. The PCR conditions included 2 mM $MgCl_2$, BSA, and a 60° C annealing temperature.

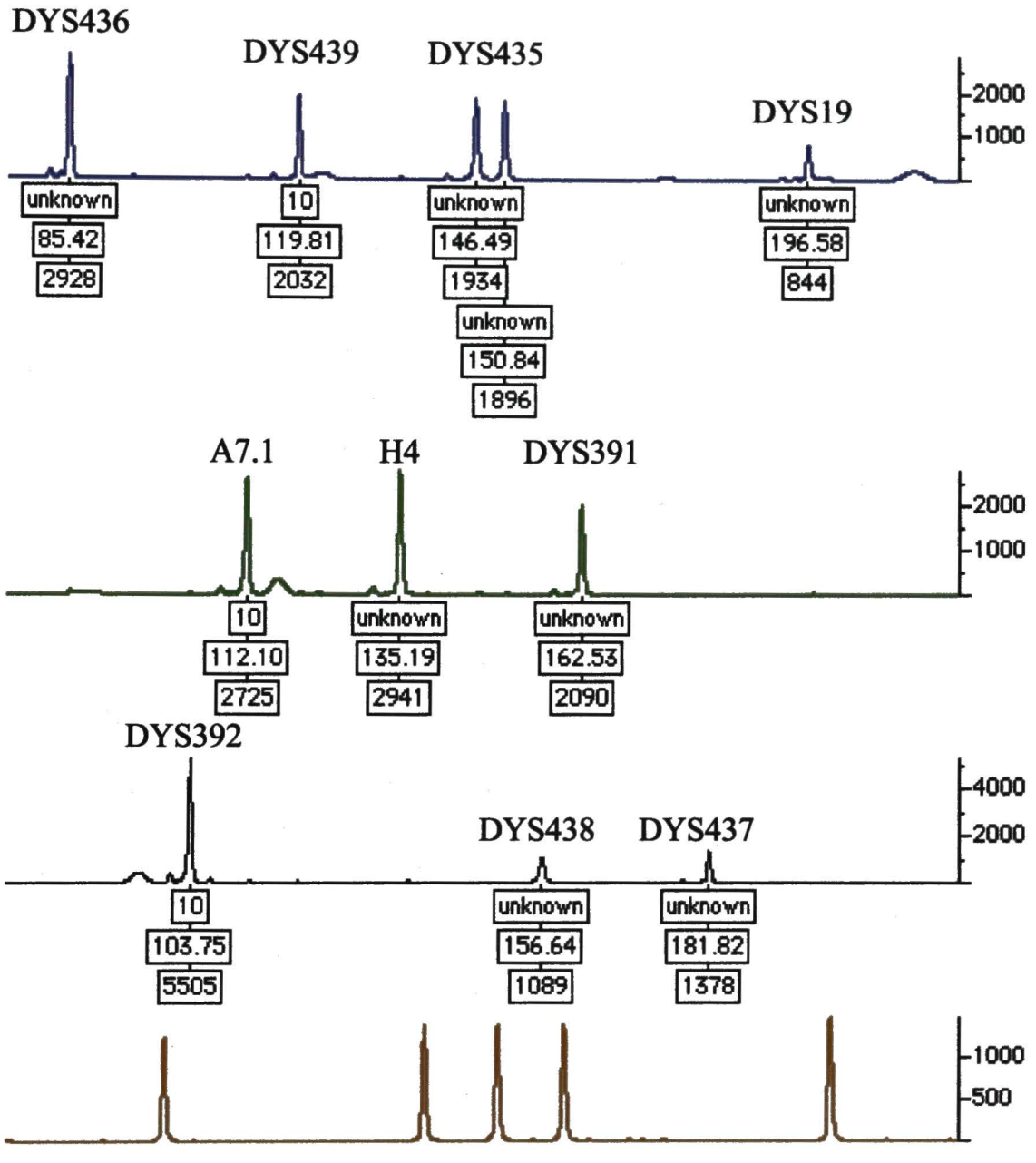



Figure 5. Duplication at the DYS435 locus. The PCR conditions included 2 mM $MgCl_2$, BSA, and a 60° C annealing temperature.

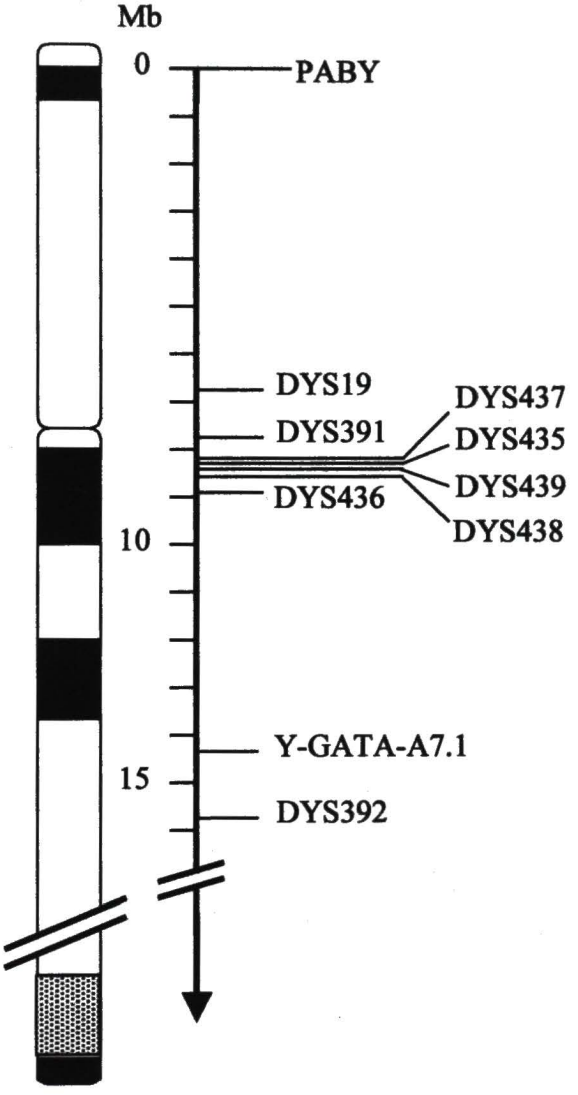



Figure 6. Physical locations of microsatellites on the Y-chromosome (12). The approximate positions of the microsatellites are shown on a scale (in Mb) with reference to the PABY (pseudoautosomal boundary).

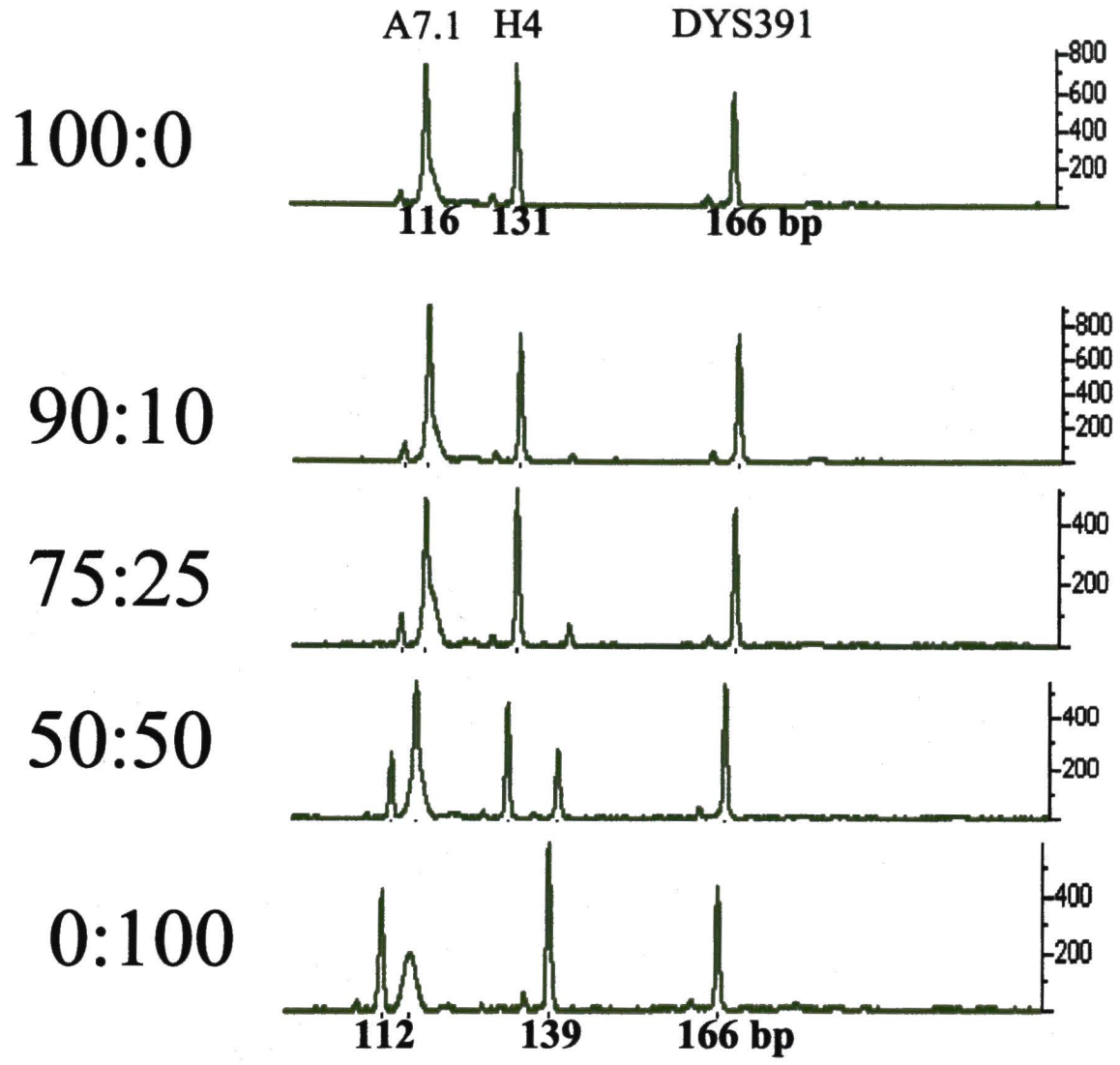



Figure 7. Male:male mixture studies (BS #1: ATCC 45514). ATCC 45514 and BS #1 were mixed in varying ratios using a total of 2.5 ng DNA. Only the loci labeled with the VIC dye are shown.

# CHAPTER 8

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