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Geno-geographic origin of Y-specific STR haplotypes of a sample of Caucasian-Mestizo and African descent male individuals from Colombia

Y-STR haplotypes in Colombia

Origen geno-geográfico de haplotipos STR de cromosoma Y en una muestra cauásico-mestiza y afrodescendiente de Colombia

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Juan J. Yunis: dirigió el proyecto.

Emilio J. Yunis: codirigió el proyecto.

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Todos los autores participaron en el análisis de resultados y en la escritura de artículo.

Introduction. The Y chromosome STR haplotypes have been widely used in population studies to establish the origin of diverse populations.

Objective. We analyzed Y chromosome STR haplotypes (8 loci) in 134 Caucasian-mestizo and 137 African descent Colombian unrelated individuals to correlate the geographical origin with historical data as well as the genetic relationships and possible admixture patterns.

Materials and methods. 134 samples of African descent and 137 Caucasian-mestizo samples analyzed for Y chromosome STR haplotypes by PCR followed by acrylamide electrophoresis.

Results. No evidence of populations sub-structure was found for the African descent. 2.59% of the haplotypes were shared between the two groups with the possible existence of Caucasian gene flow towards Afro-descendants.

Conclusion. The Caucasian-Mestizo Colombian population is grouped with other populations of the Iberian Peninsula and Europe, while the Afro-Colombian population is grouped with other African populations reported.

Key words: Haplotypes, Y chromosome, genetics, Colombia.

Introducción. Los haplotipos STR de cromosoma Y han sido ampliamente utilizados en estudios poblacionales para establecer el origen de diversas poblaciones.

Objetivo. Se analizaron haplotipos STR de cromosoma Y (8 loci) en 134 Afrodescendientes y Caucásico-mestizos no relacionados de Colombia para correlacionar el origen geográfico con los datos históricos, así como las relaciones genéticas y posibles patrones de mezcla.

Materiales y métodos. Se analizaron haplotipos STR de cromosoma Y mediante PCR segudios de electroforesis en acrilamida de 134 muestras de afrodescendientes y 137 muestras de caucásicos mestizos.

Resultados. No se encontró evidencia de subestructuración de la población afrodescendiente. El 2,59% de los haplotipos eran compartidos en los dos grupos analizados con la posible existencia de flujo génico de caucásico-mestizos hacia los afrodescendientes.

Conclusión. La población caucásico-mestiza colombiana se agrupa con otras poblaciones de la peninsula Ibérica y Europa, mientras que la población afrodescendiente colombiana se agrupa con otras poblaciones africanas reportadas.

Palabras clave: haplotipos, cromosoma Y, genética, Colombia.

In recent years, the analysis of Y chromosome polymorphisms has become an important tool for anthropological studies as well as in regular forensic casework (1-6). A set of 8 Y-chromosome specific Short Tandem repeats found in the non-recombinant portion of the Y chromosome, the tetranucleotide STR DYS19, DYS385, DYS389-I, DYS389-II, DYS390, DYS391 and DYS393 and the trinucleotide DYS392 conforms the minimal haplotype for forensic casework (7). Several populations from Europe, Asia, and the United States have been typed for the Y chromosome minimal haplotype and their haplotype frequencies have been reported to the Y chromosome reference database (YHRD) (8-17). In South America several populations of non-Amerindian origin have been analyzed for the minimal Y-STR haplotype. Among them, the population from Buenos Aires Argentina (15), a population from Medellin (18), another from Valle del Cauca (Builes JJ, Castañeda SP, Espinal C, Aguirre D, Gómez MV, Villamarin D, *et a*l. Analysis of 16 Y-chromosomal STRs in a Valle (Colombia) population sample. Proceedings of the 21st International ISFG Congress held in Ponta Delgada, The Azores, Portugal between 13 and 16 September 2005. Antonio Amorim FC-RaNM, editor. Amsterdam, The Nederlands: Elsevier.; 2006. 891 p.), and those reported by us (19). Additional studies have been carried out with the newest platforms (YFiler® Applied Biosystems Palo Alto CA, PowerPlex Y® and PowerPlex Y23® Promega Corporation, Madison WI) in some Colombian populations (20-24). Most of these studies have focused their attention on non-Amerindian admixed populations (Mestizo populations)(20-24). None of the studies carried out in regions with strong presence of African descendant populations, selected unmixed African descendant individuals for analysis (20,22,24).Other studies have not analyzed the Y chromosome minimal STR haplotype (25). Due to different genetic background,

migrations, culture and admixture processes, additional studies are required to better understand the genetic composition within different regions of each country and between different countries within a continent.

In Colombia, the majority of the population is considered Caucasian-Mestizo composed mainly of Spanish descent and in a minor degree of other European, Arab and Jewish populations. The African Descent population is located mainly in the Pacific coast, Caribbean coast and islands. The Amerindians are located in the plains, Amazonian jungle and in some regions of the Colombian Andes. However, different degrees of admixture have been found in different regions based on blood group analysis (26). In the Pacific and Caribbean coast different degrees of admixture between African descent and Caucasian Mestizos is present. On the other side, Amerindian admixture is stronger in the southwest and decreases toward the north section of the Andes were the Caucasian component is stronger.

In the present study, we have analyzed the Y-chromosome haplotype data from 271 unrelated individual, 134 of African Descent and 137 Caucasian Mestizos of Colombia in order to determine Y chromosome haplotype origin of the two population groups tested. Additional analysis was carried out in order to determine the degree of genetic admixture between these groups and Amerindian populations of Colombia studied by us (unpublished results) and to correlate our findings with previously defined populations of Europe and North America.

Material and Methods

One hundred and thirty four (134) unrelated individuals of African descent, collected between 1994-1995 after proper informed consent was obtained in four different towns (Quibdo n=40, the state capital, Condoto n=34, Istmina n=30, and Tado n=30) of the Choco department in the Pacific region of Colombia, as part of a research

project at the Universidad Nacional de Colombia to genetically characterize different ethnic groups, were analyzed. In addition, a total of 177 Caucasian Mestizo individuals from the East-central Andean Region of Colombia (Cundinamarca n=40, Boyaca n=40, and Santander n=40 departments, as well as Bogotá n=50, the capital city) divided as 45 alleged father/son pairs with known exclusion of paternity, 40 alleged father/son pairs with proven paternity (minimum of 12 STR's and W > 99.99%) and 7 unrelated individuals were analyzed for a total of 137 unrelated Caucasian-Mestizo individuals were provided by Servicios Medicos Yunis Turbay y cia. after proper informed consent was obtained in order to stablish Y STR haplotype frequencies that could be used for forensic stimations by many laboratories in Colombia and abroad.Generally, a sample size between 120-150 individuals is considered an adecuate sample size for forensic frequency estimations.

Whole blood was obtained from all individuals to participate in genetic populations studies. Genomic DNA was extracted by the Quick light DNA isolation kit (Lifecodes Corporation) following manufacturers recommendations or by Chelex 100 as described previously (19,27)

PCR amplification and typing

The tetranucleotide STR DYS19, DYS385, DYS389-I, DYS389-II, DYS390, DYS391 and DYS393 and the trinucleotide DYS392 were analyzed as follow: Primers sequences for DYS19, DYS390, DYS391 and DYS392 were those described previously (3-5). PCR amplifications were carried out as single loci for DYS19, and DYS392 with identical conditions as described (3). A multiplex reaction was performed for DYS390, DYS391 and DYS393 as follow: Initial denaturation at 94 ^{o}C for 3 minutes; 5 cycles of 94 ^{o}C for 15 seconds, 58 ^{o}C for 20 sec., 72 ^{o}C for 20 sec., and 30 cycles of 94 ^{o}C for 15 sec.; 54 ^{o}C 20 sec.; 72 ^{o}C 20 sec. For DYS389-I and

DYS389-II, Fluorescent labeled primers (FITC) and amplification conditions were identical as previously described (6). For DYS385, Fluorescent labeled primers (TMR) and amplification conditions were identical as described by Schneider et al.(28).

The PCR products were resolved in 4% Acrylamide/bis-acrylamide denaturing gels for DYS19, DYS390, DYS391, DYS392 and DYS393 with silver nitrate staining. For DYS389 and DYS385, typing was carried out in 4% Acrylamide/bis-acrylamide denaturing gels and analyzed in a Hitachi FMBIO II with Internal Lane Standards (CXR 400, Promega Corporation, Madison, WI).

Allele designations were made according to the reference tables with the aid of sequenced allelic ladders provided by Dr. Angel Carracedo, Instituto de Medicina Legal, Universidad de Santiago de Compostela (Galicia, Espain) and Dr. Daniel Corach, Servicio de Huellas Digitales Genéticas, Universidad de Buenos Aires, (Buenos Aires Argentina) and from reference samples provided by Dr. Lutz Roewer, Institute of Legal Medicine, Medical Faculty (Charité), Humboldt-University (Berlin, Germany). Proficiency testing for these STR's has been carried out for ystr.charite.de; GEP-ISFG WG; Collaborative Testing Service (CTS, Sterling, VA); and Sociedad Argentina de Genética Forense (SAGF).

Data analysis

Haplotype and allele frequencies, as well as haplotype diversity, based on eight-locus Y-STR haplotypes were determined with the aid of ARLEQUIN (29). The haplotype frequencies and other parameters of forensic interest have been published earlier (19).

The genetic structure among the African descent population was analyzed with consideration for the molecular difference between individual haplotypes (AMOVA),

in addition to differences in haplotype frequencies, resulting in estimates of Φ_{st} (or R_{st}), an F_{st} analogue (excluding DYS385) (28). Significance levels of the genetic-variance components as well as Φ_{st} values were estimated by use of 100,000 permutations.

Haplotype sharing between the Caucasian-Mestizo and African descent individuals from Colombia were determined in order to obtain an approximation to the degree of admixture between these groups. For this purpose, numbers of shared haplotypes between the Caucasian Mestizo and African descent populations of Colombia were determined for each of the haplotypes identified.

Genetic distances (Da) were calculated according to Nei (30) with Populations (Populations Version 1.2.23. Software. http://www.cnrs-gif.fr/pge). A Multi-Dimensional Scaling (MDS) analysis (excluding DYS385) was carried out with the genetic distance matrix obtained previously with the aid of the Statistica for Windows (StatSoft, Inc. Tulsa, Ok). This analysis was carried out for the Caucasian Mestizo and the African descent (as a whole) populations of Colombia and those previously described by others (31-33); as well as for 698 African descent individuals and 225 individuals from Medellin Colombia reported in ystr.org. In addition, 254 unrelated individuals belonging to 20 Amerindian tribes of Colombia (Yunis J.J, Campo D.S., Camelo, Z., Acevedo, L., and Yunis, E.J., unpublished results) were included in the MDS analysis.

Results

The haplotype frequencies for the African descent and the Caucasian-Mestizo populations of Colombia have been published before (19). For the African descent population, a total of 110 haplotypes were found out of 134 unrelated individuals

tested. Ninety-six out of 110 haplotypes were single instance haplotypes, one haplotype was shared between 6 individuals, 6 haplotypes were shared by three individuals and seven haplotypes were shared by two individuals each (table 1).
The genetic structure (excluding DYS385) of the African descent population collected in four different towns of the Choco Department of Colombia as determined by estimates of Φ_{st} (or R_{st}) and showed that only 2.6% (p 0.054) of the genetic variation was due to differences between groups (towns) and 97.4% variation was found within the sample collected in each town (100,000 permutations).
For the Caucasian-Mestizo population of Colombia, from a total of 177 individuals analyzed of which 45 pairs corresponded to excluded alleged Father/son pairs by previous analysis of autosomic STR's, 40 pairs of Alleged father/son whose probability of paternity was > 99.99% and 7 unrelated individuals, for a total of 137 unrelated individuals were tested. All 40 pairs of compatible paternity by autosomic STR's were also compatible for Y-STR haplotypes. No mutations were detected among these 40 pairs. One out of 45 pairs of excluded paternity by autosomic STR's had an identical Y-STR haplotype. From the 137 unrelated individuals, a total of 122 haplotypes were determined. One hundred and fourteen of the 122 haplotypes were single instance haplotypes, one haplotype was present in five unrelated individuals, 2 haplotypes in four individuals and 5 haplotypes in two unrelated individuals each.
Numbers of shared haplotypes between the Caucasian Mestizo and African descent populations of Colombia were determined for each of the 232 haplotypes identified. Only six haplotypes were found to be shared between these two ethnic groups (2.59%) as shown in table 2. Three out of these six haplotypes are the most frequent haplotypes in Caucasian Mestizos.

The multidimensional scaling analysis is presented in figure 1. The Caucasian mestizo population from Colombia clustered with most of the Caucasian populations included in the analysis. The closest genetic distances (data not shown) were found with the populations form Andalucia (Spain), Medellin (Colombia), and Galician (Spain) populations. On the other hand, the African descent population of Colombia clustered with the African American and Surinam populations. The Amerindian population of Colombia clustered with other Amerindian populations including the Inuit population of Greenland.

Discussion

The analysis of Y-STR haplotypes found in the non-recombinant portion of the Y chromosome (minimal haplotype, DYS19, DYS389-I, DYS389-II, DYS390, DYS391, DYS392, DYS393 and DYS385) were used to determine genetic affinities between groups of males individuals and to detect population structure (34) in 271 unrelated individuals of Colombia, of which 134 were of African descent and 137 were Caucasian mestizos.

The first immigrants from Spain and Portugal founded the first non-Amerindian villages in present day Colombia in the early 16th Century (35). The ancestors of the African descent population of Colombia were brought as slaves between 1580 and 1650 (36) from Senegal, Ivory Coast, Mali and the west coast of Guinea. From the Caribbean coast of Colombia, a large proportion of this population were transferred to Popayan, the slave capital located in the southwest section of the country (Cauca department) to be distributed later mainly to the Choco, Antioquia, Cauca and Valle del Cauca departments to work in field and mines.

The Amerindian population that inhabited present day Colombia started to be eliminated since the beginning of the conquest. The major Amerindian cultures that

existed at the arrival of the Spanish conquerors, located in the Andean region of Colombia, do not longer exist. The present day Amerindian tribes are located in the Amazonian and Orinoquian regions, and some groups in the south and north sections of the country. Based on the 1993 census, around 1.5% of the population was Amerindian (37).

The Colombian Andes has been a strong barrier against free demographic flow within Colombia. In the southwest of Colombia, the Andes divide itself into three mountain ranges (east, central and west Andes) that extend towards the north covering most of the central portion of the territory (38). Thus, the Andes define five different geographic regions named the Pacific (were the Choco department is located), the Atlantic, the Andean, the Orinoquian and the Amazonian regions. For centuries, demographic growth of different population groups occurred within each region with some degree of isolation generating subcultures within the country. Most of the demographic flow between regions started in the middle to late 20th century, with some genetic exchange between some regions. Previous studies of the population found in the East-central Andean region of Colombia based on Blood groups analysis, have shown a little contribution of African descent individuals to the genetic makeup of this region (26). In that report, the African descent component was between 2.5% and 5.9% in the three departments where the Caucasian-Mestizo sample was collected for the present study. Our results based on Y-chromosome haplotypes showed that only 2.59% (6 out of 232) of the haplotypes were shared between Caucasian Mestizos and African descent individuals (table 2). Most of the shared Y-STR haplotypes are the most common haplotypes among the Caucasian mestizo population implying a genetic flow from male Caucasians into African descent individuals. Thus, our results corroborate previous findings of African

admixture for the east-central region of the Colombian Andes. On the other hand, a simmilar result was obtained for a sample of 225 unrelated individuals from Medellin (18)(Central-west Andes) when compared to the African descent population reported here. Out of 281 haplotypes (110 African descent and 171 from Medellin), only ten haplotypes were shared between these two groups (2.81%). Once again, the haplotypes shared between these groups are among the most frequent haplotypes found in the Caucasian population. However, based on blood groups, near 15% of the population of Antioquia is of African descent. Although the demographics for the 225 individuals from Medellin was not known, it is possible that it was composed mainly of Caucasian-mestizo individuals (predominant group in the city) and not from other sections of the department like the west section were the African descent component is somewhat stronger. The genetic affinity of the Medellin sample based on genetic distances with other Caucasian populations in the multidimensional scaling analysis provides further support for this assumption.

In a separate report, based on Y chromosome microsatellites and biallelic markers, 94% of male haplotypes found in Antioquia were of European ancestry, 5% of African and 1% Amerindian while around 90% of the mtDNA gene pool was of Amerindian heritage (37) suggesting a Caucasian/Amerindian sex bias. These results are likely true for the Caucasian-mestizo population of Antioquia based on the demographics of the sample selected for that study. However, the genetic composition of Antioquian population should be somewhat different and the 15% African admixture could probably be a real estimate when considering all the population of that region.

On the other hand, it is likely that a similar Caucasian/Amerindian sex bias will be found for the Caucasian-Mestizo population since historical data indicates a limited

female immigration from Spain during the conquest and colony periods (39,40). Thus, the genetic pool was limited to male European Caucasians and Amerindian mtDNA. This admixture process would have generated the Caucasian/Amerindian sex bias based on Y-Chromosomes and mtDNA as mentioned earlier but with various degrees of admixture at the autosomal level differentiating the present day population from the Amerindian and Spanish populations as has been demonstrated for the Caucasian mestizo population of Colombia and different populations from Spain and Portugal based on autosomic STR analysis (41-43). In that regard, in another report (24), the analysisi of 305 unrelated individuals from the departments located in the the Caribean region of Colombia showed that most of the Y-chromosome haplotypes present in that region of Colombia, are also of Spanish/European origin that clustered near the populations reported by us and others published in YHRD database. A similar scenario has been found for mestizo samples analyzed in Cartagena (20), Cundinamarca and Boyacá (23), Valle del Cauca, Cauca and Nariño (22).

The genetic structure of the African descent population collected in four different towns of the Choco Department of Colombia based on the AMOVA showed that only 2.6% of the genetic variation was due to differences between groups (towns) and 97.4% variation was found within the sample collected in each town. As we have previously reported, these results are likely due to a process of genetic 'homogenization' carried out by the slavers in Colombia (36). In that process, in order to avoid emancipation movements among the slaves, individuals speaking the same language were separated. A similar process has been reported for African descent populations of Venezuela and Brazil (44).

The African populations have shown the highest genetic heterogeneity based on mtDNA and other autosomic markers so far studied. However, the results obtained in our study based on Y-Chromosome STR showed a lower haplotype diversity and discrimination power for the African descent population of Colombia than those obtained for the Caucasian mestizo population (table 1). Out of 110 haplotypes identified in the African Descent population, only 96 were single instance haplotypes as compared to 114 out of 122 in the Caucasian mestizo populations. Thus, several haplotypes among the African descent population might be Identical by descent (IBD) leading to a lower haplotype diversity and lower discrimination power. These results are probably true for the population of the Choco department due to several factors. First, a genetic gene pool that has remained unchanged for the last 350 years and the absence of new genetic flow from African populations and other ethnic groups limiting the effective size of Y-chromosomes in this African descent population of Colombia. However, a different picture should be expected in regions were the Caucasian/African admixture process has been stronger as in the Atlantic and the southwest sections of the country.

A multidimensional scaling analysis based on Nei genetic distances was carried out for the Colombian populations described here and several European, American, Asian, Arabs and Amerindian populations in order to determine their genetic affinities. Although the sample size analyzed here could seem small, 134 haplotypes for African descent and 137 haplotypes for the Caucasian Mestizo sample, these values are similar to those used in other studies (23-25) to determine the genetic relationship between populations (see footnote of figure 1 for sample sizes from multiple populations analyzed around the globe). The results showed clustering of the Caucasian mestizo population of Colombia with other Caucasian populations of

Europe and America (Medellin and Buenos Aires). The closest genetic distances were with the population of Andalucia, followed by Medellin and Galicia. Our results are in agreement with historical data indicating that most of the Spanish colonizers to the new world came from Andalucia, Extremadura, and Castilla in that order during the XVI and XVII centuries (45). The Caucasian Mestizo population seems to be genetically distant from the Valencian and Catalunian populations of Spain (both of them belonging to the Aragon province during 16th century) corroborating historical data indicating a little contribution of this province to the colonizing force. On the other hand, our sample seem to be genetically distant from the Basque population, a result likely due to the fact that the Basque immigrants to present day Colombia are mainly located in the west section of Colombia (45).

The African descent population clustered with other African descent populations such as the African-American and the population from Surinam. Based on autosomal STR analyzed for the African descent population of Colombia (41-43) no statistically significant differences were found with the African American population. Our results with Y-STR haplotypes are in agreement with our previous findings, as well as historical data suggesting a common origin in the West coast of Africa for both the African American and Afro-Colombian individuals. The grouping of the population from Surinam, and admixed population, is probably due to the strong African descent component (predominant group) and a lesser extent, the components derived from Caucasian (Netherlands) and Asian (India and Indonesia) populations of Surinam.

The Arab populations included in the Multidimensional Scaling analysis are very distant from the Caucasian Mestizo population of Colombia. This result is probably due to the fact that the Arab admixture of the Colombian population has been mainly with Lebanese and Palestinian populations and not with Arab populations of North

Africa. In addition, the Lebanese and Palestinian presence is stronger in the Atlantic region of Colombia and in a lesser extent in the north Andean region of the country; therefore the Arab ancestry may not be well represented in the sample studied. Previously, a Jewish ancestry of around 14% for the population of Antioquia was proposed based on 6 loci Y-STR haplotypes (DYS19, DYS388, DYS390, DYS391, DYS392, DYS393) (38). Although our studies did not include the DYS388 loci analyzed in that report, we evaluated the presence of the Cohen modal haplotype based on the remaining 5 STRs analyzed in our sample and in the sample reported for Medellin by Builes et al. The Cohen haplotype (14; 23; 10; 11; 12) was found only in two individuals (2/114, 1.75%) of our sample and in three individuals from Medellin (3/171 haplotypes, 1.75%). In addition, when considering all the haplotypes used to obtain the 14% Jewish ancestry reported by Thomas (46), the number of potential Jewish haplotypes in our sample was 3.5% (4/114) and 3.5% in the sample from Medellin (6/171). Therefore, additional studies are required in order to properly establish the Jewish genetic influence in the population of Colombia.

Taken together, our results showed a limited contribution of Caucasian Y-STR haplotypes in the Afro-colombian population of the Choco Department as well as a limited contribution of Afro-Colombian Y-STR haplotypes in the Caucasian-Mestizo population of the east-central Andean region of Colombia corroborating previous studies with autosomal markers.

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Conflict of interest

All authors declare that there is no conflict of interest. Servicios Mèdicos Yunis Turbay y Cia provided some of the samples, materials and reagents, and the fluorescent genetic analyzer Hitachi FMBIOII that was used in the present study study.

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References

1. **Hammer M**, **Zegura S.** The role of the Y chromosome in human evolution studies. Evol Anthropol. 1996;5:116-34. http://dx.doi.org/10.1002/(SICI)1520-6505(1996)5:4<116::AID-EVAN2>3.0.CO;2-E

2. **Jobling MA**, **Pandya A**, **Tyler-Smith C**. The Y chromosome in forensic analysis and paternity testing. Int J Legal Med. 1997;110:118-24. http://dx.doi.org/10.1007/s004140050050

3. **Kayser M**, **Caglia A**, **Corach D**, **Fretwell N**, **Gehrig C**, **Graziosi G, *et al*.** Evaluation of Y-chromosomal STRs: a multicenter study. Int J Legal Med. 1997;110:125-33. http://dx.doi.org/10.1007/s004140050051

4. **de Knijff P, Kayser M, Caglia A, Corach D, Fretwell N, Gehrig C, et al.** Chromosome Y microsatellites: population genetic and evolutionary aspects. Int J Legal Med. 1997;110:134-49. http://dx.doi.org/10.1007/s004140050052

5. **Appendix.** Reference Tables to: evaluation of Y-chromosomal STRs: A multicenter study (Kayser et al.) and Chromosome Y microsatellites: population genetic and evolutionary aspects (de Knijff et al.)(1997). Int J Legal Med. 1997;110:141-9. http://dx.doi.org/10.1007/s004140050053

6. **Schultes T**, **Hummel S**, **Herrmann B.** Amplification of Y-Chromosomal STRs from ancient skeletal material. Hum Genet. 1999;104:164-6. http://dx.doi.org/10.1007/s004390050930

7. **Gusmao L**, **Brion M**, **Gonzales-Neira A**, **Lareu M**, **Carracedo A**. Y chromosome 26 specific polymorphisms in forensic analysis. Leg Med (Tokyo). 1999;1:55-60. http://dx.doi.org/10.1016/S1344-6223(99)80013-3

8. **Pérez-Lezaun A**, **Calafell F**, **Comas D**, **Mateu E**, **Bosch E**, **Martinez-Arias R**, ***et al*.** Sex specific migration patterns in central Asian population, revealed by analysis of Y chromosome short tandem repeats and mtDNA. Am J Hum Genet. 1999;45:265-70. http://dx.doi.org/10.1086/302451

9. **Roewer L**, **Kayser M**, **de Knijff P**, **Anslinger K**, **Betz A**, **Caglia A**, ***et al*.** A new method for the evaluation of matches in no recombining genomes: application to Y-chromosomal short tandem repeat (STR) haplotypes in European males. Forensic Sci Int. 2000;114:31-43. http://dx.doi.org/10.1016/S0379-0738(00)00287-5

10. **Nata M**, **Brinkmann B**, **Rolf B.** Y-Chromosomal STR haplotypes in a population from north west Germany. Int J Legal Med. 1999;112:406-8. http://dx.doi.org/10.1007/s004140050026

11. **Pestoni C**, **Cal ML**, **Lareu MV**, **Rodriguez-Calvo MS**, **Carracedo A.** Y chromosome STR haplotypes: genetic and sequencing data of the Galician population (NW Spain). Int J Legal Med. 1999;112:15-21. http://dx.doi.org/10.1007/s004140050191

12. **Junge A**, **Madea B.** Population studies of the Y-chromosome specific polymorphism DYS19, DYS389I + II, DYS390 and DYS393 in a Western German population (Bonn area). Forensic Sci Int. 1999;101:195-201. http://dx.doi.org/10.1016/S0379-0738(99)00029-8

13. **Anslinger K**, **Keil W**, **Weichhold G**, **Eisenmenger W.** Y-chromosomal STR haplotypes in a population sample from Bavaria. Int J Legal Med. 2000;113:189-92. http://dx.doi.org/10.1007/s004140050296

14. **Hidding M**, **Schmitt C**. Haplotype frequencies and population data of nine Y-chromosomal STR polymorphisms in a German and a Chinese population. Forensic Sci Int. 2000;113:47-53. http://dx.doi.org/10.1016/S0379-0738(00)00261-9

15. **Corach D**, **Filgueira Risso L**, **Marino M**, **Penacino G**, **Sala A**. Routine Y-STR typing in forensic casework. Forensic Sci Int. 2001;118:131-5. http://dx.doi.org/10.1016/S0379-0738(00)00483-7

16. **Hou YP**, **Zhang J**, **Li YB**, **Wu J**, **Zhang SZ**, **Prinz M**. Allele sequences of six new Y-STR loci and haplotypes in the Chinese Han population. Forensic Sci Int. 2001;118:147-52. http://dx.doi.org/10.1016/S0379-0738(01)00383-8

17. **Mohyuddin A**, **Ayub Q**, **Qamar R**, **Zerjal T**, **Helgason A**, **Mehdi SQ**, ***et al***. Y-chromosomal STR haplotypes in Pakistani populations. Forensic Sci Int. 2001;118:141-6. http://dx.doi.org/10.1016/S0379-0738(01)00382-6

18. **Builes JJ**, **Bravo ML**, **Gomez C**, **Espinal C**, **Aguirre D**, **Gomez A**, ***et al*.** Y-chromosome STRs in an Antioquian (Colombia) population sample. Forensic Sci Int. 2006;164:79-86. http://dx.doi.org/10.1016/j.forsciint.2005.10.005

19. **Yunis JJ**, **Acevedo LE**, **Campo DS**, **Yunis EJ**. Population data of Y-STR minimal haplotypes in a sample of Caucasian-Mestizo and African descent individuals of Colombia. Forensic Sci Int. 2005;151:307-13. http://dx.doi.org/10.1016/j.forsciint.2005.02.005

20. **Builes JJ**, **Martinez B**, **Gomez A**, **Caraballo L**, **Espinal C**, **Aguirre D**, ***et al*.** Y chromosome STR haplotypes in the Caribbean city of Cartagena (Colombia). Forensic Sci Int. 2007;167:62-9. http://dx.doi.org/10.1016/j.forsciint.2005.12.015

21. **Gaviria AA**, **Ibarra AA**, **Palacio OD**, **Posada YC**, **Triana O**, **Ochoa LM**, ***et al*.** Y-chromosome haplotype analysis in Antioquia (Colombia). Forensic Sci Int. 2005;151:85-91. http://dx.doi.org/10.1016/j.forsciint.2004.07.006

22. **Avila J**, **Briceno I**, **Gomez A**. Genetic population analysis of 17 Y-chromosomal STRs in three states (Valle del Cauca, Cauca and Narino) from Southwestern Colombia. J Forensic Leg Med. 2009;16:204-11. http://dx.doi.org/10.1016/j.jflm.2008.12.002

23. **Rojas KM**, **Roa M**, **Briceño I**, **Guaneme C**, **Gómez A**. Polimorfismos de 17 marcadores STR del cromosoma-Y en una muestra poblacional del altiplano cundiboyacense. Colombia Médica. 2011;42:88-97.

24. **Romero RE**, **Briceno I**, **Lizarazo Rdel P**, **Willuweit S**, **Roewer L**, **Gomez A**. A Colombian Caribbean population study of 16 Y-chromosome STR loci. Forensic Sci Int Genet. 2008;2:e5-8. http://dx.doi.org 10.1016/j.fsigen.2007.09.005

25. **Rojas W**, **Campo O**, **García J**, **Soto I**, **Duque C**, **Bedoya G**, et al. Coancestría de apellidos y linajes del cromosoma Y en el noroeste de Colombia: Una herramienta útil para establecer migración entre poblaciones. Revista Colombiana de Antropología. 2012;48:49-79.

26. **Sandoval C**, **De la Hoz A**, **Yunis E**. Estructura genética de la población colombiana. Revista de la Facultad de Medicina. 1993;41:3-14.

27. **Walsh PS**, **Metzger DA**, **Higuchi R**. Chelex 100 as a medium for simple extraction of DNA for PCR-based typing from forensic material. Biotechniques. 1991;4:506-13.

28. **Schneider PM**, **Meuser S**, **Waiyawuth W**, **Seo Y**, **Rittner C.** Tandem repeat structure of the duplicated Y-chromosomal STR locus DYS385 and frequency

studies in the German and three Asian populations. Forensic Sci Int. 1998;97:61-70. http://dx.doi.org/10.1016/S0379-0738(98)00146-7

29. **Schneider S**, **Roessli D**, **Excoffier L.** ARLEQUIN, ver 2000. A software for population genetics data analysis. Geneva: Genetics and Biometry laboratory, University of Geneva; 2000.

30. **Nei M**, **Tajima F**, **Tateno Y**. Accuracy of estimated phylogenetic trees from molecular data. II. Gene frequency data. J Mol Evol. 1983;19:153-70.

31. **Bianchi NO**, **Catanesi CI**, **Bailliet G**, **Martinez-Marignac VL**, **Bravi CM**, **Vidal-Rioja LB**, ***et al*.** Characterization of ancestral and derived Y-chromosome haplotypes of New World native populations. Am J Hum Genet. 1998;6:1862-71. http://dx.doi.org/10.1086/302141

32. **Bosch E**, **Calafell F**, **Comas D**, **Oefner PJ**, **Underhill PA**, **Bertranpetit J**. High-resolution analysis of human Y-chromosome variation shows a sharp discontinuity and limited gene flow between northwestern Africa and the Iberian Peninsula. Am J Hum Genet. 2001;68:1019-29. http://dx.doi.org/10.1086/319521

33. **González-Neira A**, **Gusmão L**, **Brión M**, **Lareu MV**, **Amorim A**, **Carracedo A**. Distribution of Y-chromosome STR defined haplotypes in Iberia. Forensic Sci Int. 2000;110:117-26. http://dx.doi.org/10.1016/S0379-0738(00)00156-0

34. **Kayser M**, **Krawczak M**, **Excoffier L**, **Dieltjes P**, **Corach D**, **Pascali V, *et al*.** An extensive analysis of Y-chromosomal microsatellite haplotypes in globally dispersed human populations. Am J Hum Genet. 2001;68:990-1018. http://dx.doi.org/10.1086/319510

35. **Friede J**. Historia extensa de Colombia “descubrimiento y conquista del Nuevo Reino de Granada". Volumen II. Bogotá: Editorial Lerner; 1965.

36. **Gutierrez I**. Historia del negro en Colombia. Bogotá: Nueva América; 1986. p. 17-23.

37. **Departamento Administrativo Nacional de Estadística, DANE.** Los grupos étnicos de Colombia en el censo de 1993. Bogotá: DANE; 1998.

38. **Carvajal-Carmona LG, Soto ID, Pineda N, Ortiz-Barrientos D, Duque C, Ospina-Duque J, *et al*.** Strong Amerind/white sex bias and a possible Sephardic contribution among the founders of a population in northwest Colombia. Am J Hum Genet. 2000;67:1287-95. http://dx.doi.org/10.1016/S0002-9297(07)62956-5

39. **Mörner M.** La mezcla de razas en la historia de América latina. Buenos Aires: Editorial Paidos; 1989.

40. **Sánchez Albornoz C.** Historia de América Latina. Barcelona: Crítica ed.; 1990.

41. **Yunis JJ**, **Garcia O**, **Uriarte I**, **Yunis EJ**. Population data on 6 short tandem repeat loci in a sample of Caucasian-Mestizos from Colombia. Int J Legal Med. 2000;113:175-8. http://dx.doi.org/10.1007/s004140050293

42. **Yunis JJ**, **Garcia O**, **Baena A**, **Arboleda G**, **Uriarte I**, **Yunis E**. Population frequency for the short tandem repeat loci D18S849, D3S1744, and D12S1090 in Caucasian-Mestizo and African descent populations of Colombia. J Forensic Sci. 2000;45:429-31.

43. **Yunis JJ**, **Baena A**, **Garcia O**, **Uriarte I**, **Yunis EJ**. Population data of F13AO1, FES/FPS, VWA, CSF1PO, TPOX and THO1 short tandem repeat loci in a sample of African descent individuals of Colombia. Forensic Sci Int. 2001;117:235-6. http://dx.doi.org/10.1016/S0379-0738(00)00414-X

44. **Bortolini MC**, **da Silva-Júnior WA**, **Weimer T de A**, **Zago MA**, **de Guerra DC**, **Schneider MP**, et al. Protein and hypervariable tandem repeat diversity in eight

African-derived South American populations: inferred relationships do not coincide. Hum Biol. 1998;70:443-61.

45. **Agudelo L**. Genesis del pueblo antioqueño. Bogotá: Editorial Era Cósmica; 1986.

46. **Thomas MG**, **Skorecki K**, **Ben-Ami H**, **Parfitt T**, **Bradman N**, **Goldstein DB**. Origins of Old Testament priests. Nature. 1998;394:138-40. http://dx.doi.org/10.1038/28083

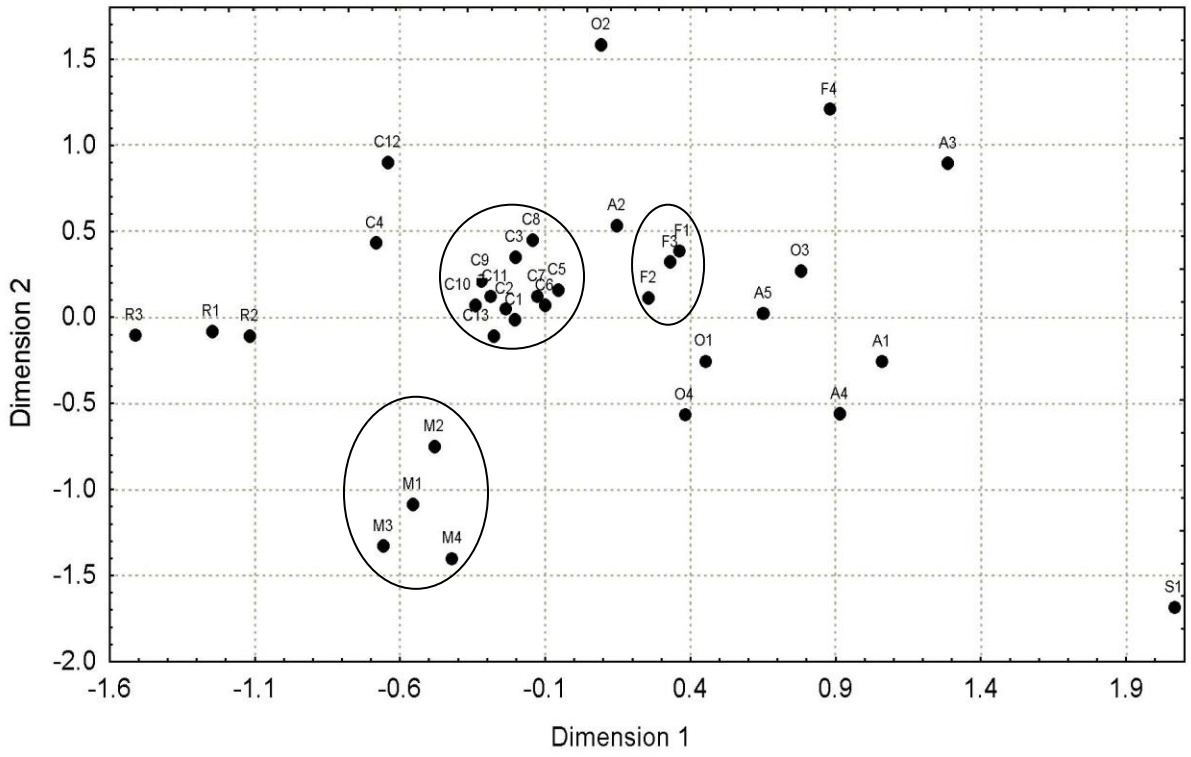

Figure 1.

Scatter plot of a Multidimensional Scaling analysis (MDS) based on Nei genetic distances (Da) for the Colombian Caucasian mestizo population and African descent individuals and several published populations (see text for reference). C1: Caucasian mestizos from Colombia (n=137); C2: Medellin (n=225); C3: Dutch (88); C4: Basques (n=30); C5: Germans (n=88); C6: Swiss (n=64); C7: Italians (n=100); C8: Valencia (Spain) (n=28); C9: Portugal (n=55); C10: Galicia (Spain) (n=53); C11: Andalucia (Spain) (n=93); C12: Cataluña (Spain) (n=14); C13: Buenos Aires metropolitans (n=100); A1: Han Chinese (n=36); A2: Mongolians (n=40); A3: Indians (n=25); A4: Taiwanese aborigines (n=30); A5: Indonesians (n=56); O1: Papua New Guineans (n=26); O2: Roro New Guineans (n=12); O3: Tolain from New Britain (n=16); O4: Trobriand Islanders (n=59); S1: West Samoan (n=10) ; M1: Colombian Amerindians (n= 254); M2: South American Natives (n= 46); M3: Native Americans

(n=89); M4: Inuit (n=62); F1: African descent from Colombia (n=134); F2: Surinamese (n=54); F3: African American (n=698); F4: Central African pygmies (n=31); R1: Morrocan Arabs (n=44); R2: North central Moroccan Berbers (n=20); R3: South Moroccan Berbers (n=40). Circled are most of the Caucasian populations (code leter C), African descent populations (code letter F) and Amerindian populations (code letter M).

Table 1. Y-STR haplotype sharing statistics between Caucasian Mestizo and African descent populations of Colombia.

		Caucasian Andean Region	African Descent Choco
No. of Individuals		137	134
No. of Haplotypes		122	110
Discrimination (%)		89.05	82.08
Haplotype Class			
Single	No.	114	96
	Proportion	0.934	0.8727
Multiple	No.	8	14
	Proportion	0.066	0.1272
Haplotype Diversity		0.9971	0.9955
Haplotype Diversity S.E.		0.0016	0.0019

Table 2. Haplotypes shared between Caucasian Mestizo and African descent populations of Colombia

Caucasian Mestizo	n =	African Descent	n =	Haplotype							
HC26	1	HN6	1	13	14	30	24	9	11	13	13,14
HC45	4	HN20	1	14	13	29	24	10	13	13	11,14
HC47	1	HN21	1	14	13	29	24	10	13	13	11,15
HC51	5	HN25	1	14	13	29	24	11	13	13	11,14
HC63	1	HN32	1	14	13	30	23	10	13	13	11,14
HC80	4	HN40	1	14	14	30	24	11	13	13	11,14

Haplotypes are order according to DYS19, DYS389-I, DYS389-II, DYS390, DYS391, DYS392, DYS393 and DYS385.