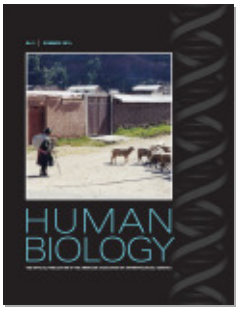
Genetic Profile of the Kichwas (Quichuas) from Ecuador by Analysis of STR Loci

Fabricio González Andrade, Dora Sánchez



Human Biology, Volume 76, Number 5, October 2004, pp. 723-730 (Article)

Published by Wayne State University Press
DOI: 10.1353/hub.2005.0007

Genetic Profile of the Kichwas (Quichuas) from Ecuador by Analysis of STR Loci

FABRICIO GONZÁLEZ-ANDRADE[1] AND DORA SÁNCHEZ[1]

Abstract Allele frequency data for the STR systems CSF1PO, TPOX, TH01, D13S317, D16S539, D5S818, and D7S820 were determined in a population sample of unrelated, healthy Amerindian Kichwa individuals. All loci met Hardy–Weinberg expectations, and there was no evidence for association of alleles among the seven loci.

Ecuador has three main ethnic groups: Caucasian mestizos, Amerindian natives (more than 100 multiethnic and pluricultural groups), and descendants of African slaves (Sánchez 1998). During the last five centuries, three or more diverse populations came into contact, interacted, and mixed. They are, in chronological order, Amerindians (a fairly homogeneous Asian-derived group), Europeans (mostly Spanish), and Africans (who were brought to Latin America as slaves) (Sáns 2000). Amerindian populations of the Andes are classically divided into two linguistics groups: Quechua and Aymara speakers (Rodríguez-Delfín et al. 2001). Quechua, with an *e*, is the language spoken today by the descendants of Incas, especially in Peru and Bolivia (Gené et al. 1998, 2000). But this is not a reality in Ecuador, because most of the indigenous people speak Quichua (Kichwa), with an *i*, which is an anthropological language variation, derived from the admixture process after the Inca period (SIDENPE 2003).

The Kichwa (the ancestral name) or Quichua (a name derived from the mixture of natives and Spanish) population has almost 3 million inhabitants (20% of the total Ecuadoran population). They are the most representative Amerindian group in Ecuador. Most of them live in the mountains, in the Andean region, from Carchi province in the north to Loja province in the south. Some of them are distributed in the Amazonia region, in the provinces of Pastaza, Napo, Sucumbíos, and Orellana (Figure 1). They number approximately 70,000 persons and speak a Kichwa language distinguishable from highlands Quichua dialects (CODENPE 2003). Everyone speaks Spanish as a second language.

[1]Laboratory of Molecular Genetics, Metropolitan Hospital, Quito, Ecuador.

Human Biology, October 2004, v. 76, no. 5, pp. 723–730.

KEY WORDS: KICHWAS, QUICHUAS, ECUADOR, AMERINDIANS, DNA TYPING, SHORT TANDEM REPEAT, CSF1PO, TPOX, TH01, D13S317, D16S539, D5S818, D7S820, POPULATION GENETICS.

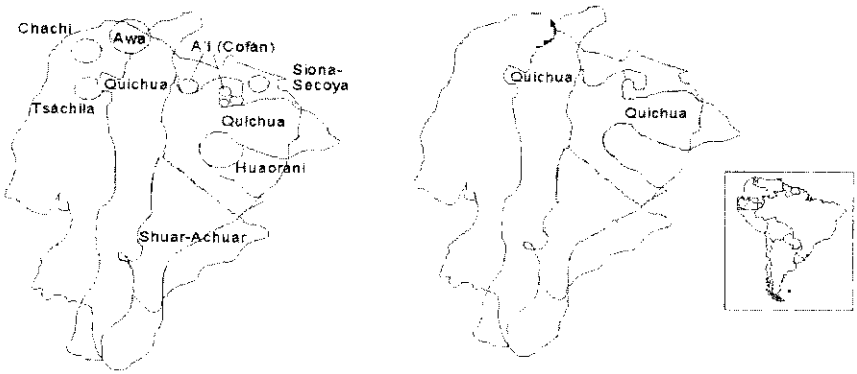


Figure 1. Location of Kichwas in Ecuador.

Mestizos are the most representative and largest group in Ecuador. They are descendants of Spanish (Caucasian) and Amerindian peoples. This is a new hybrid group, like Mulatos, who are a mixture of Caucasians and blacks, or Zambos, who are a mixture of Amerindians and blacks. These three groups sometimes appear differentiated in censuses and had different legal rights in the past (González-Andrade et al. 2003). Amerindians were easily assimilated in this new class into the society. Sometimes the union between a conqueror and an Amerindian had political or religious reasons: the domination of a territory or the conversion to Catholicism. Actually, Mestizos mostly live in urban regions. We think that different degrees of genetic admixture, mainly with Amerindians, also exist. This situation has several variations in the cities of the Andes and the cities of the Pacific coast (González-Andrade et al. 2002).

The State of Ecuador has recognized different indigenous nationalities, each with their own identity and language, as shown in Table 1. Ethnicity means cultural practices and moral values that distinguish groups and communities. Individuals from different ethnic groups see themselves as different from other social and native groups. This concept has two dimensions: cultural and social characteristics (language, religion, faith, location, etc.) and a sense of shared identity and tradition.

Indigenous nationality means a joining of thousand-year-old peoples before the Ecuadoran state was established. Indigenous nationals share a historical identity, language, and culture, live in a certain territory, have their own institutions and traditional forms of social, economic, and political organization, and practice their own authority.

Kichwa nationality is a product of several groups who are culturally different but have the same language, such as the Karanki, Natabuela, Otavalo, Kayambi, Kitu-kara, Panzaleo, Chibuleo, Kichwa del Tungurahua, Salasaka, Puruhá, Kañari, and Saraguro.

Few genetic studies have been carried out in Ecuadoran native populations,

Table 1. Indigenous Nationalities of Ecuador

Name	*Language*	*Population (Approximate Number of People)*	*Location*	*Neighboring Country with Similar Group*
Awá	Awapít	900–1,000	Along Mira and San Juan rivers in Carchi and Esmeraldas provinces	Colombia
Secoya	Paicoca	200	Along the Shushufindi, Aguarico, and Cuyabeno rivers	Peru
Huaoroani (Aucas)	Hao Tiriro	2,000	Between the Napo River in the north and the Curaray River in the south	None
Epera	Sia Pedee (Wamuna or Epena)	200	In Esmeraldas province, around Borbón, San Francisco, and Tablillo	Colombia
Siona	Paicoca	400 secoyas (Ankuteres and Piojes)	Along the Shushufindi, Aguarico, and Cuyabeno rivers	Colombia
A'i Cofan	A'ingae	500	Along the upper Aguarico River and the Guanúes and San Miguel rivers, and on the Ecuadorian-Colombian border	Colombia
Shiwiar	Shiwar Chicham	600	In Pastaza province, along the Corrientes River	Peru
Zápara	Zapara	150	Along the Conambo, Pindoyaco, and Curaray rivers	Peru
Kichwa	Kichwa	3,000,000 in the Andean region; 70,000 in Amazonia	In the highlands from Carchi province in the north to Loja province in the south; in Amazonia, in Pastaza, Napo, Sucumbíos, and Orellana provinces	Peru
Chachi (Cayapas)	Cha'palaa	4,000	In Esmeraldas province, along the Cayapas, Santiago, Dabable, and Rosario rivers.	None
Tsa'Chila (Colorados)	Tsa'fiqui	2,000	In Santo Domingo de los Colorados	None
Shuar	Shuar Chicham	70,000	In Zamora Chinchipe and Morona Santiago provinces	Peru
Achuar	Achuar Chicham	3,000	In Pastaza and Morona Santiago	Peru
Afro-Ecuadorans	Spanish	500,000	In Esmeraldas and Carchi provinces	None

Sources: CODENPE (2003) and our data.

Table 2. Previous Genetic Studies in Ecuadoran Populations

Group Studied	*Genetic Marker Analyzed*	*Number of Individuals Analyzed*	*Reference*
Chachi (Cayapas)	HLA class II	100	Titus-Trachtenberg et al. (1994)
Chachi (Cayapas)	HLA class II	100	Trachtenberg et al. (1995)
Chachi (Cayapas)	*HLA*DQA1*	40	Zimmermann et al. (1995)
Chachi (Cayapas)	HLA class I (B)	13	Garber et al. (1995)
Huaoranis	*HLA*DRB1*	6	Blagitko et al. (1997)
Chachi (Cayapas)	mtDNA	204	Rickards et al. (1999)
African descendants	Blood subgroups and serum markers	255	Martínez-Labarga et al. (1999)
Mestizo population	STRs (CSF1PO, TPOX, TH01, F13A01, VWA, D13S317, D16S539, D5S818, D7S820, LPL, HPRTB, F13B)	400	González-Andrade et al. (2003)
Black population	STRs (CSF1PO, TPOX, TH01, F13A01, VWA, D13S317, D16S539, D5S818, D7S820, LPL, HPRTB, F13B)	104	González-Andrade et al. (2004)

as shown in Table 2. Extensive work may be required in this field to demonstrate the different genetic admixture processes between original individuals. Some groups, such as the Cayapas, have been studied principally. There is a general lack of information on the Ecuadoran population from a genetic point of view, and no previous publications on the distribution of short tandem repeats (STRs) are available.

In this paper we report the allele frequency distribution of seven STR loci that have proven to be extremely useful for forensic casework, human identification, and population genetics—CSF1PO, TPOX, TH01, D13S317, D5S818, D16S539, and D7S820—in a population sample of Amerindian Kichwas (Quichuas) from Ecuador.

Materials and Methods

Population Sample. Whole blood was obtained by venipuncture into vacutainer tubes containing EDTA. The subjects were healthy, unrelated Kichwas, of both sexes, who had been born and who now live in the highlands of Ecuador. The number of chromosomes analyzed was 150.

DNA Extraction. DNA was extracted using the Wizard Genomic DNA Purification Kit System (Promega Corp. 1998), and the quantity was estimated by ultraviolet absorbance (Gene Quant Calculator, Pharmacia, Uppsala, Sweden).

Typing. The multiplex analysis of CSF1PO, TPOX, and TH01 was performed using the CTT Multiplex System kit, and the analysis of D13S317, D5S818, D16S539, and D7S820 was performed using the Gamma STR Multiplex System (Promega Corp. 1998), following the manufacturer's recommendations in both cases.

Amplification using the PCR was performed in a Techne Thermal Cycler, model Genius, following the manufacturer's recommendations. The PCR products were typed by vertical electrophoresis on 0.40-mm-thick 4% denaturing polyacrylamide gels (19:1 acrylamide:bisacrylamide, 7 *M* urea) with silver staining. Electrophoresis was carried out in a Gibco BRL Sequencing System, model SA (Gibco, United States) at 200 V for 1 min, followed by 200 V for 2 min at the end, at a fixed temperature of 50°C. The recommendations of the DNA Commission of the International Society for Forensic Haemogenetics for analysis of STR systems were followed (Bär et al. 1997; ISFG 1992).

Analyses of Data. Evaluation of Hardy–Weinberg expectations was carried out using the exact test, and determination of further statistical parameters of forensic interest was carried out by using the computer program HWE-Analysis, version 3.3 (Christoph Puers, Institute for Legal Medicine, University of Münster), as previously described (Martínez-Jarreta et al. 1999; González-Andrade et al. 2002).

Results and Discussion

The distributions of observed allele frequencies for the seven loci (CSF1PO, TPOX, TH01, D13S317, D5S818, D16S539, and D7S820) and the results of the different analytical procedures for testing the correspondence of the genotype frequencies with Hardy–Weinberg expectations are summarized in Table 3. This table shows the forensic value of the analyzed systems expressed as various statistical parameters.

All loci met Hardy–Weinberg expectations, and there was no evidence of association of alleles among the seven loci. The forensic efficiency parameters revealed the high forensic efficiency of the seven STR loci analyzed. The data presented in this study will allow the calculation of matching probabilities in forensic casework if Ecuadoran Kichwa individuals are considered as a source of DNA evidence.

Some forensic values of interest were calculated. The cumulative power of discrimination was calculated to be 0.68212, and the cumulative mean exclusion chance was 0.99627. Statistical parameters of genetic variability, such as heterozygosity and the number of alleles, were also calculated. The heterozygosity ranged from 0.5800 to 0.8235. The number of alleles observed ranged from 5 to 7.

In Ecuador, ethnic differences have been associated with differences in

Table 3. Genetic Distribution of Seven STR Loci in the Kichwa Population

Allele	*CSF1PO* ($n = 150$)	*TPOX* ($n = 150$)	*TH01* ($n = 150$)	*D13S317* ($n = 102$)	*D16S539* ($n = 104$)	*D5S818* ($n = 95$)	*D7S820* ($n = 104$)
6	–	–	0.4800	–	–	–	–
7	–	–	0.3200	–	–	0.1110	–
8	0.0030	0.5130	0.0070	0.0690	0.0140	–	0.0050
9	0.0070	–	0.0230	0.2450	0.1920	0.1160	0.0029
9.3	–	–	0.1330	–	–	–	–
10	0.3130	0.0030	0.1230	0.1570	0.29 30	0.0420	0.2600
11	0.2930	0.3600	0.0370	0.0930	0.2640	0.5740	0.3700
12	0.3130	0.1230	–	0.1720	0.1830	0.1320	0.2880
13	0.0570	0.0030	–	0.1760	0.0480	0.0260	0.0480
14	0.0130	–	–	0.0880	0.0050	–	–
χ^2	0.3744	0.8728	0.2032	0.9518	0.6680	0.8948	0.7000
G test	0.3408	0.9080	0.5280	0.9366	0.5464	0.9486	0.1956
Exact test	0.4956	0.8192	0.3928	0.8956	0.4214	0.9548	0.1090
Observed heterozygosity	0.7333	0.5800	0.6400	0.8235	0.7308	0.6421	0.6442
Expected heterozygosity	0.7165	0.5936	0.6497	0.8377	0.7749	0.6288	0.7126
Mean exclusion chance	0.4552	0.3065	0.3876	0.66663	0.55157	0.41383	0.45228
Mean exclusion probability	0.4543	0.2833	0.3547	0.67073	0.55331	0.32687	0.44803
Polymorphism information content	0.6591	0.5114	0.5871	0.81208	0.73358	0.59417	0.65486
Probability of match	0.1401	0.2462	0.1906	0.05498	0.09578	0.17429	0.14127
Power of discrimination	0.8599	0.7538	0.8094	0.94502	0.90422	0.82571	0.85873
Minimal allele frequency	0.0184	0.0167	0.0173	0.0284	0.0260	0.0268	0.0254

Number of random shuffles performed: 5,000.

power and wealth; ethnic groups are discriminated against. There has not been a complete census of all ethnic nationalities and groups. Only maternal language has been considered as a tool to mark differences between native groups in Ecuador. Genetics should be a most efficient tool to establish a real biological history of Ecuadoran populations.

Kichwa nationality appears to be due to a process of quichuization of several peoples who had their own characteristics. This nationality arose from contacts between ethnic groups in the pre-Inca period, and they adopted a Quichua-like maternal language. Quichua was the Inca language.

Kichwas live near the Andean region, which has ecological features of the

paramo region. They adopted a micro vertical system of production, characterized by cultivation in microclimatic areas. In their political organization the *curaca* or *cacique* was the most important authority. Commercial interchanges facilitated the movement of language. The *mindalas* transport exotic products, cultural manifestations, and rituals into neighboring zones and even farther in a synergy of peoples and populations.

Inca invasion produced cultural changes in original peoples, but they did not suffer a rupture of their structure; they adopted a new way of production. During the conquest period the original peoples experienced a strong rupture of ancestral societies, with an imposition of Spanish language. Quichuas survived this period, and they call their language Kichwa, an ancestral and millenarian name, a product of a mixture of aboriginal language, pre-Inca language, and Quichua from Incas.

The information summarized in this report allows further comparison between worldwide populations and may also provide data for statistical evaluation in some areas such as forensics.

Received 13 August 2003; revision received 12 April 2004.

Literature Cited

Bär, W., B. Brinkmann, B. Budowle et al. 1997. DNA recommendations: Further report of the DNA Commission of the ISFH regarding the use of short tandem repeat systems. *Forensic Sci. Int.* 87(3):181–184.

Blagitko, N., C. O'Huigin, F. Figueroa et al. 1997. Polymorphism of the HLA-DRB1 locus in Colombian, Ecuadorian, and Chilean Amerinds. *Hum. Immunol.* 54:74–81.

CODENPE (Consejo de Desarrollo de las Nacionalidades y Pueblos del Ecuador). 2003. *Nacionalidades y Pueblos del Ecuador, Quito*. Quito, Ecuador: CODENPE.

Garber, T., L. Butler, E. Trachtenberg et al. 1995. HLA-B alleles of the Cayapa of Ecuador: New B39 and B15 alleles. *Immunogenetics* 42:19–27.

Gené, M., M. Fuentes, E. Huguet et al. 1998. Quechua Amerindian population characterized by HLA-DQα, YNZ22, 3APO B, HUMTH01, and HUMVWAA31A polymorphism. *J. Forensic Sci.* 43(2):403–405.

Gené, M., P. Moreno, N. Borrego et al. 2000. Population study of Aymara Amerindians for the PCR-DNA polymorphisms HUMTH01, HUMVWAA31A, D3S1358, D8S1179, D18S51, D19S253, YNZ22, and HLA-DQα. *Int. J. Legal Med.* 113:126–128.

González-Andrade, F., D. Sánchez, and B. Martínez-Jarreta. 2002. Evaluation of 1,495 cases of disputed paternity in Ecuador (South America) resolved with STR-PCR polymorphisms. *Proc. Int. Assoc. Forensic Sci.* 1:225–230.

González-Andrade, F., D. Sánchez, and B. Martínez-Jarreta. 2003. Genetic profile of the Ecuadorian Mestizo population by using the Power-Plex 16 system kit. *Int. J. Forensic Sci. Int.* 135(1):64–66.

González-Andrade, F., D. Sánchez, and B. Martínez-Jarreta. 2004. Genetic profile of the Ecuadorian Black population (Ecuador, South America) by using the Power Plex16 system kit. *Intl. Congr. Ser.* 1261:166–168.

ISFG (International Society for Forensic Haemogenetics). 1992. Recommendations of the DNA Commission of the International Society for Forensic Haemogenetics relating to the use of PCR-based polymorphisms. *Forensic Sci. Int*. 55(1):1–3.

Martínez-Jarreta, B., P. Díaz Roche, and E. Abecia. 1999. Genetic variation at six STR loci (CTT, FFV) in Aragon (north Spain). *Forensic Sci. Int*. 100(1–2):87–92.

Martinez-Labarga, C., O. Rickards, R. Scacchi et al. 1999. Genetic population structure of two African-Ecuadorian communities of Esmeraldas. *Am. J. Phys. Anthropol*. 109(2):159–174.

Promega Corporation. 1998. *Gene Print STR Systems for Silver Stain Detection: Technical Manual*. TMD no. 004. Foster City, WI: Promega Corp.

Rickards, O., C. Martínez-Labarga, J. Kum et al. 1999. mtDNA history of the Cayapa Amerinds of Ecuador: Detection of additional founding lineages for the native American populations. *Am. J. Hum. Genet*. 65:519–530.

Rodríguez-Delfin, L., V. Rubin-de-Celis, and M. Zago. 2001. Genetic diversity in an Andean population from Peru and regional migration patterns of Amerindians in South America: Data from Y chromosome and mitochondrial DNA. *Hum. Hered*. 51:97–106.

Sánchez, D. 1998. Human identification in Ecuador. In *Memoires of 22nd Symposium of Biology in Ecuador,* C. Cerón, ed. Quito: Central University of Ecuador, 79–80.

Sánchez, D., F. González-Andrade, and B. Martínez Jarreta. 2003. Population genetics of 12 STR loci in a sample of Mestizos from Ecuador (South America). *J. Forensic Sci*. 48(2):453–454.

Sáns, M. 2000. Admixture studies in Latin America: From the 20th century to the 21st century. *Hum. Biol*. 72(1):155–177.

SIDENPE. 2003. *Sistema Integrado de Indicadores Sociales del Ecuador: Frente Social*. Available at http://www.sidenpe.gov.ec

Titus-Trachtenberg, E.A., O. Rickards, G.F. De Stefano et al. 1994. Analysis of HLA class II haplotypes reveals a novel HLA-DRB1 allele in the Cayapa Indians of Ecuador: Evidence for convergent evolution and balancing selection at position 86. *Am. J. Hum. Genet*. 55:160–167.

Trachtenberg, E., A. Erlich, O. Rickards et al. 1995. HLA class II linkage disequilibrium and haplotype evolution in the Cayapa Indians of Ecuador. *Am. J. Hum. Genet*. 57:415–424.

Zimmermann, P., P. Phadke, A. Lee et al. 1995. Migration of a novel **DQA1* allele from African origin to North and South America. *Hum. Immunol*. 42:233–240.