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HUMAN X-CROMOSOME NON-CODING VARIATION IN LATIN AMERICAN POPULATIONS: A REVIEW

VARIACIÓN NO CODIFICANTE DEL CROMOSOMA X HUMANO EN POBLACIONES LATINOAMERICANAS: UNA REVISIÓN

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ABSTRACT

The human X-chromosome non-coding markers, such as short tandem repeats (STRs), single nucleotide polymorphisms (SNPs), insertion-deletions (INDELs) and *Alu* insertions, are useful for revealing relationships among populations and for the identification of individuals. In the last decades, a number of studies have been performed to determine the genetic structure of Latin American populations by using X-chromosome markers. These studies provided useful information regarding the genetic composition of these populations and their relationship with Native American, Asian and European populations. One of the most interesting findings achieved by X-chromosome studies is the bias in the sex ratio of individuals that gave rise to the current Latin American populations, as it was previously observed through the analysis of uniparental markers, and which is undoubtedly evidenced in the differential inheritance of X-chromosome in comparison to autosomes. Besides, the genetic drift process that affected Native American populations is more pronounced in X-chromosome markers than in autosomes. The present review summarizes our current knowledge concerning X-chromosome non-coding polymorphisms studied in Latin American populations.

Key words: genetic diversity, INDEL, SNP, STR, Alu insertion

RESUMEN

Los marcadores no codificantes del cromosoma X humano, como las repeticiones cortas en tándem (STR), los polimorfismos de un solo nucleótido (SNP), las inserciones-deleciones (INDEL) y las inserciones Alu, son útiles para revelar la relación existente entre poblaciones, y también para la identificación de personas. En las últimas décadas, se han realizado una serie de estudios para determinar la estructura genética de las poblaciones latinoamericanas, utilizando marcadores de cromosoma X. Estos estudios proporcionaron información útil sobre la composición genética de estas poblaciones y su relación con las poblaciones nativas americanas, asiáticas y europeas. Uno de los hallazgos más interesantes logrados en estos estudios es el sesgo en la proporción de sexos de los individuos que originaron las poblaciones latinoamericanas actuales, tal como se observó previamente a través del análisis de marcadores uniparentales, y que queda evidenciado por la herencia diferencial del cromosoma X en comparación con los autosomas. Además, el proceso de deriva genética que afectó a las poblaciones nativas americanas actuó de manera más pronunciada en los marcadores del cromosoma X que en los autosomas. La presente revisión resume nuestro conocimiento actual sobre los polimorfismos no codificantes del cromosoma X estudiados en poblaciones latinoamericanas.

Palabras clave: diversidad genética, INDEL, SNP, STR, inserción Alu

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INTRODUCTION

In the last decade, the human X-chromosome gained significant importance in the study of population and forensic genetics. This is due to the intrinsic properties of this chromosome, such as haplotype accessibility in males (because of its hemizygosity), lower mutation and recombination rates, and faster genetic drift due to smaller effective population size (intermediate between autosomes and Y-chromosomes) (Schaffner, 2004). For this reason, the non-coding regions of the X-chromosome exhibit a more important differentiation among populations than autosomes (Ramachandran *et al.*, 2004). Moreover, X-chromosome markers are disproportionately influenced by sex, giving an excellent opportunity for analyzing demographic differences between men and women (Casto *et al.*, 2010). In this sense, the X-chromosome helps to understand a sex bias which occurred in the past in South American populations, as Native and African components are revealed in X chromosome in a higher amount than autosome chromosomes (Wang *et al.*, 2008; Hedrick, 2017; Resano and Moral, 2018; Ongaro *et al.*, 2021).

The X-chromosome contains several types of non-coding polymorphisms distributed along its sequence. The combination of different markers can be a useful tool for analyzing microevolutionary processes that occurred in populations in the past (Pereira *et al.*, 2006; Santos-Lopes *et al.*, 2007; Di Santo Meztler *et al.*, 2018, among other reports). The most important types of X-chromosome polymorphisms are short tandem repeats (STRs), single nucleotide polymorphisms (SNPs), insertion-deletions (INDELs) and *Alu* insertions. STRs -also called microsatellites- are a type of repetitive DNA markers usually comprising tandem repeats of di-, tri-, tetra- or pentanucleotide units (less than 10 bp long). They are considered hypervariable markers because they usually present a high number of alleles within populations. Their high variability makes STRs very suitable for paternity and identity tests. Therefore, several efforts have been made for generating databases of validated X-STRs with high discriminatory power and low internal variation of the repetitive sequence (Willems *et al.*, 2014; Gomes *et al.*, 2020).

SNPs are changes of only one nucleotide in a particular position in the genome (e.g., cytosine or guanine, alternatively). They originate as biallelic markers when a point mutation occurs in a genome, converting one nucleotide into another, but tri- and tetra-allelic SNPs can also be found. A block of a few tens of kilobases in the X-chromosome can carry around 50 to 100 SNPs, therefore they are very informative markers, and easy to genotype, even from degraded DNA samples, given the short length of amplicons (Schaffner, 2004; Kidd *et al.*, 2006).

INDELs are positions in an alignment between two DNA sequences where an insertion or deletion has occurred. SNPs and INDELs offer many advantages for population studies: they are widely spread throughout the genome (including the X-chromosome), most of them derive from a single mutation event, mutation rates in both types of markers are much lower than those of repetitive markers, and they can show significant differences in allele frequencies among geographically distant populations (Li *et al.*, 2008; Pereira *et al.*, 2009; Ribeiro-Rodrigues *et al.*, 2009; Casto *et al.*, 2010; Ali *et al.* 2022).

Alu insertions are fragments of DNA of about 300 bp length, and are classified into 12 major subfamilies that appeared at different times during primate evolution (Kapitonov and Jurka, 1996). They are the most abundant and extensively studied class of short interspersed nuclear elements (SINEs) present in the genome, representing about 11% of the human genome (Carroll *et al.*, 2001).

The presence or absence of *Alu* elements at a particular locus gives rise to genetic polymorphisms. Usually, *Alu* polymorphisms are selectively neutral and, as their location hardly changes or rearranges, they are considered to be derived from one unique event in which the absence of the insertion is the ancestral state for *Alu* markers (Batzer *et al.*, 1994). These characteristics make the human *Alu* insertion polymorphisms a good tool for studying the genetic diversity and the evolutionary relationships among human populations (Stoneking *et al.*, 1997).

Genetic and genome studies focusing on the process of colonization of the American continent, and in the composition of Latin American populations were for several years mostly based on Y-chromosome and mitochondrial variability. More recently, these studies were focused on X-chromosome variation, and provided useful information regarding the genetic composition of Latin American populations and their relationship with American, Asian and European populations. This information has not been previously reviewed in detail. Therefore, the aim of the present review is to summarize our current knowledge concerning the X-chromosome markers in Latin American populations and their significance in terms of genetic relationships between these populations and their evolution. In the next sections we will refer in detail to the main findings concerning different X-chromosome polymorphisms in Latin American populations. A summary of the reports included in this work in reference to X-chromosome variation in Latin American populations is listed in Table 1.

Table 1. Summary of the articles included in this work listed by the first author. Type of polymorphisms, analyzed populations, and main contributions are detailed.

Reference	Type/s of polymorphism/s	Population/s analyzed	Main contributions
AguilarVelázquez et al. 2022	STRs	Mestizo (admixed) individuals from Mexico	female genotypes and male haplotypes with 100% discriminatory capacity
Amorim et al. 2011	STRs	Amerindians and admixed populations from the Central Valley of Costa Rica and Southern Brazil	Lower genetic diversity and higher proportion of linkage disequilibrium in Native than admixed populations
Baeta et al. 2021	STRs	Native American and Mestizo groups of Central America	X-chromosome database for Central American populations
Battilana et al. 2006	*ALU* insertions	Native American, Northern Arctic, and several other populations as outgroups	Importance of genetic flow and of dispersive processes (genetic drift and founder effects) for the differentiation of Native American populations
Bobillo et al. 2011	STRs	Argentinians	High power of exclusion, and usefulness for kinship tests and human identification
Bryc et al. 2010	SNPs	populations from Puerto Rico, the Dominican Republic, Ecuador, and Colombia	Significant sex bias in admixture proportions. Lower linkage disequilibrium in populations of higher African ancestry
Cainé et al. 2010	STRs	Brazilians (non-Amerindians) from Rio Grande do Sul and Amerindians from Brazilian population	Pattern of variation consistent with the known historical migrations from Portugal to Brazilian region of Santa Catarina
Caputo et al. 2021	INDELs	Argentinean individuals living in urban areas	Differential ancestry contribution between autosomal and X-markers. More important Native American and African contribution in X-chromosome
Casals et al. 2022	STRs	El Salvador, Central America	Gender-biased admixture pattern, with the lowest *FST* values for El Salvador
Casto et al. 2010	SNPs	CEPH-HGDP populations (including Native American and Colombian populations)	Selection and demographic processes are partially responsible for allele frequency differences
Catanesi et al. 2007	STRs	Gran Chaco Amerindians	Genetic structure in agreement with the geographic distribution of the populations
Córdoba et al. 2012	INDELs	Colombians	European and Amerindian contribution to the current gene pool of the studied population
Cortés-Trujillo et al. 2019	STRs	Different geographic regions of Mexico	Homogeneity of variation, and usefulness for constructing a global Mexican population database (Fst *p*-value >0.05) for forensic casework
Di Santo Meztler et al. 2018	*ALU* insertions, INDELs and SNPs	Northeast Argentina	INDEL and SNP variation differentiate populations within the country, while Alu-insertions are helpful in ancestry studies
Di Santo Meztler et al. 2019	STR	Northeast Argentina	A high level of genetic variability, with differentiation between Native Americans and urban populations, and also between Native populations
Ferragut et al. 2019	*ALU* insertions and STRs	north-western Argentina	A high Native American component for Salta population. Helpful for forensic purposes in north-western Argentina
Flores-Espinoza et al. 2021	INDELs and STRs	Ecuadorian Mestizo populations	Significant levels of linkage disequilibrium between the studied markers
Freitas et al. 2010a	INDELs	Brazilian Amazon	Statistical parameters agree with estimations from other populations. Usefulness of panels for forensic use
Freitas et al. 2010b	INDELs	Brazilian Amazon and urban populations	Construction of haplotypes show different admixture models for Amerindians and urban populations
García et al. 2019	STRs	Argentinians from all regions	High gene diversity. Provide a database for Argentinians, with highly discriminatory genetic information
Gayá-Vidal, M.et al. 2010	*ALU* insertions	Native Americans from the Bolivian Altiplano	A high genetic similarity for the two studied populations, with a big differentiation from the Quechua-speakers from Peru

Table 1. (continues) Summary of the articles included in this work listed by the first author. Type of polymorphisms, analyzed populations, and main contributions are detailed.

Reference	Type/s of polymorphism/s	Population/s analyzed	Main contributions
Glesmann et al. 2013	STRs	Native American population from Argentinian Chaco	High homozygote proportion and high linkage disequilibrium found in Wichí
Gusmão et al. 2009	STRs	Argentina , Brazil, Colombia, Costa Rica, Portugal, and Spain	High discrimination power for all populations. High mean exclusion chance in duos and trios of paternity test
Homburger et al. 2015	SNPs	Colombia, Ecuador, Peru, Chile, and Argentina	Differentiation among different South American urban populations concerning Native ancestry
Ibarra et al. 2014	INDELs	Colombian populations	A highly diverse genetic background in all admixed populations. X-chromosome contributions from all continental sources
Kehdy et al. 2015	SNPs	Brazilian populations	Sex-biased mating pattern, between males with predominant European ancestry and women with predominant African or Amerindian ancestry
Leite et al. 2009	STRs	non-Amerindian Brazilians, and Amerindians from Brazil, Argentina, and Paraguay	Demographic histories have significant effects on linkage disequilibrium levels. Provide a first approach to the X-chromosome ancestry of Brazilians
Martinez et al. 2019	INDELs	Brazilian population	High diversity and a high forensic efficiency for the São Paulo population
Martins et al. 2009	STRs	Brazilian population	High power of discrimination in males and females. Usefulness in human identification
Martins et al. 2010	STRs	Southeastern region of Brazil	Differences between Brazilians and other populations, and also among different Brazilian populations. High potential for forensic purposes
Martins et.al 2017	STRs	Mato Grosso, Brazil	Usefulness of Decaplex, no need for a specific forensic database, except for the population of Rio de Janeiro
Medina-Mora et al. 2021	STRs	Mexico	Highly informative markers for use in forensic casework
Moreno-Estrada et al. 2013	SNPs	Greater Antilles, Honduras, Colombia, and three Native South American populations	A higher proportion of Native American ancestry on the X chromosome than on the autosomes
Núñez et al. 2013	STRs	Nicaragua	Usefulness of the Decaplex for forensic purposes. Warn on a possible stratification among Latin American populations, as they are not homogeneous
Ongaro et al. 2021	SNPs	American	Complexity in the post-colonial admixture dynamics
Penna et al. 2012	STRs	South Brazilian population	Particular allele frequencies, highly informative markers
Pereira and Pena 2006	STRs	Brazilian geographical regions, and several populations from the world	Strong linkage disequilibrium between certain pairs of markers useful for constructing haplotypes
Pereira et al. 2006	*ALU* insertions	populations from 7 regions of the world	Worldwide frequency distribution of a new polymorphic Alu insertion
Pico et al. 2008	STRs	Colombian population	High discrimination power, and high mean exclusion chance in duos and trios. Usefulness in forensic and kinship analysis
Resano et al. 2016	*ALU* insertions	Argentinean population	X-chromosome variation helps revealing demographic histories. Importance of Native American and African ancestry components
Resque et al. 2010	INDELs	African, European, Native American, and Brazilian populations	Usefulness for admixture estimations in Brazilian populations, and, in general, for a colonization model with sex-biased admixture
Ribeiro Rodrigues et al. 2008	STRs	Admixed Brazilian populations	Usefulness for paternity investigations and complex forensic cases
Ribeiro-Rodrigues et al. 2011	STRs	Brazilian populations	High power of discrimination, potential use in forensic and paternity casework

Table 1. (continues) Summary of the articles included in this work listed by the first author. Type of polymorphisms, analyzed populations, and main contributions are detailed.

Reference	Type/s of polymorphism/s	Population/s analyzed	Main contributions
Ribeiro-Rodrigues et al. 2009	INDELs	African, European, Native American, and Brazilian populations	Usefulness to measure ancestry proportions in three-hybrid populations
Rojas et al. 2010	STRs	Native American and admixed Colombian populations	Sex bias in admixture proportions, involving predominantly Native American women and European men
Santos-Lopes et al. 2007	*ALU* insertions and STRs	populations from seven regional groups worldwide	Identification of a region of low recombination rate in the long arm of the human X chromosome
Tavares et al. 2008	STRs	Brazilian population	High discrimination power, and high exclusion chance in duos and trios. Useful for forensic casework
Wang et.al 2010	STRs	Native Americans	Low diversity and high linkage disequilibrium in Native American communities
Wang et.al 2008	STRs	Mestizo populations	Marked variation in ancestry both within and between mestizo populations
Yang et.al.2010	STRs	Native American populations along American continent	A north to south gradient of decreasing population diversity

STRs

The information available on X-chromosome STRs mostly focuses on the production of local databases that render good power of discrimination for forensic purposes and in cases in which paternity testing is deficient (refer to Szibor et al., 2003; Szibor, 2007; Ribeiro-Rodrigues *et al.*, 2008; Tavares *et al.*, 2008; Gusmão *et al.*, 2009; Martins *et al.*, 2009; Cainé *et al.*, 2010; Bobillo *et al.*, 2011; García *et al.*, 2019; Medina-Mora *et al.*, 2021; among others). For Latin American populations, several reports refer to X-STR polymorphisms mainly on admixed communities and, to a lesser extent, on native tribes. Among the first efforts analyzing X-chromosome variability, Wang *et al.* (2008) provided interesting results on the variation of a high number of STRs in 13 Latin American populations, including 29 of the markers located in the X-chromosome. This work showed the existence of a sex bias in the ancestry components of the chromosomes with different mode of inheritance, with a much higher Native American and African proportion of genetic variation in the X-chromosome than in the autosomes. This bias is present in many admixed Latin American populations, and is a consequence of the predominance of Native and/or African women, and European men giving rise to the first admixed populations at the time of colonization of the continent, given that women contribute double of X-chromosomes than men in each generation (Wang *et al.*, 2008, Hedrick, 2017).

Another important contribution on X-STRs was the study of a set of 10 markers designed by Gusmão *et al.* (2009) for comparing 11 South American populations, together with data from four European and one African population. Pairwise genetic distances showed a relationship within Argentina (Buenos Aires, Misiones, Río Negro, Córdoba, Entre Ríos, and Paraná), and within Brazil (Mato Grosso do Sul, São Paulo, Rio de Janeiro) for the populations included in the analysis. Costa Rica and Antioquia (Colombia) populations showed the shortest genetic distance with Argentinian populations, while the European populations were positioned between the Argentinian and Brazilian ones. However, particularly for Argentina, significant distances were found by Bobillo *et al.* (2011) using the same set of STRs, when comparing Argentinian with Spanish and Portuguese populations, likely because of the admixture process with Native Americans and other non-European communities living in Argentina until present. In the same way, an analysis of 12 X-STRs included in the *Investigator Argus X-12* kit (Qiagen, Germany) suggested a Native American component for the Argentinian population from Salta province. The sign of a Native American contribution was the finding of alleles which were only described for populations of South American origin in four of the 12 markers (DXS10079, DXS10134, DXS7132, and DXS10148) which are then interpreted as variants of Native American origin (Ferragut *et al.*, 2019). On the other hand, the same set of 12 X-STRs (*Investigator Argus*

X-12) was used for analyzing admixed populations from all provinces of Argentina, which notably did not show significant differences among them when they were grouped geographically in four regions (García *et al.*, 2019).

Concerning populations from Brazil, they are in a closer relationship with Africans than those of other South American countries, given the past history of intense slavery in this country. When 16 populations covering the five geopolitical regions of Brazil were analyzed using 12 X-STRs, significant values of genetic structure were found for the whole country, mainly as a consequence of differentiation from north to south of the Brazilian territory (Ribeiro-Rodrigues *et al.*, 2011). Other reports on X-STRs from different regions of Brazil complemented this information pointing out the high variability (Pereira and Pena, 2006), proven by a number of unique haplotypes and a particular diversity of each region (Martins *et al.*, 2010). It is noteworthy that some X-STR markers presented population-specific alleles such as a single-base deletion in one of the repetitive motifs, giving rise to incomplete alleles (for example, alleles named 16.3, 17.3, and 18.3 instead of alleles 17, 18, and 19 for DXS7132). These alleles were described only for individuals who were born in Brazil, and thus considered to be of a Native American origin, as they were not reported for Europeans, Asians or Africans (Gomes *et al.*, 2009; Gusmão *et al.*, 2009; Chr-XSTR.org). These singularities are due to miscegenation of different groups, being an example the population from Rio Grande do Sul, where immigrants came mainly from Portugal, Italy and Germany (Penna *et al.*, 2012), while in the Mato Grosso region the initial immigration came from Spanish and Portuguese individuals (Martins *et al.*, 2017). Similar findings were reported for populations of the north of Argentina, where important differences on native contribution (between 30% and 72%) were reported for Catamarca, Tucumán and Salta, three provinces from the northwest of the country (Wang *et al.*, 2008). Moreover, genetic distances for 10 STRs in populations of Northeast Argentina revealed the influence of both the different geographical origin of immigrants and the particular processes of settlement, separating the locality of Misión Nueva Pompeya in the province of Chaco from other populations of the same region (provinces of Corrientes and Misiones), and even from the capital city of the province of Chaco (Di Santo Meztler *et al.*, 2019).

Another factor adding variation among populations within regions of South America is the relatively elevated level of genetic drift that characterizes Amerindian populations, as it has been reported by Santos-Lopes *et al.* (2007). These authors identified seven X-STRs located on Xq21, and four of them, together with one *Alu* insertion polymorphism (spanning a 47 kb interval), resulted in complete linkage disequilibrium, thus providing a highly informative X-chromosome haplotype for inferring human phylogeography. They performed a worldwide analysis of this linked block (four STRs and one *Alu* marker) in 667 males from the CEPH Human Genome Diversity Panel, representing 52 different populations from Africa, Europe, Middle East, Central and South Asia, East Asia, Oceania and America (the Karitiana Native population was excluded from the analysis), and found the lowest haplotype diversity among Americans: 0.839 (the highest in Africa: 0.992), and the highest molecular variance among populations from America (15.29%) in contrast to the lowest among Eurasian populations (1.06%) (Santos-Lopes *et al.*, 2007). The results reported for America in their work gave clear evidence of the genetic drift process that affected Native American populations which are isolated from other communities. Several other reports on STR markers also give support to the high influence of genetic drift causing a reduction in diversity, not only on the X-chromosome (Catanesi *et al.*, 2007), but also on different regions of the genome of native populations (Dejean *et al.*, 2004, Catanesi *et al.*, 2007, Demarchi *et al.*, 2009).

About Colombian population, a study on 10 X-STRs in the Department of Santander resulted in significant differences in comparison with Spanish, Peruvian, and Argentinian data (Pico *et al.*, 2008). These differences are likely due to Native American contribution, which is higher for X-chromosome markers than for autosomes, as is the case of other Latin American countries (Rojas *et al.*, 2010).

Other reports refer to Latin American populations from Central America. In a study on an admixed sample of Nicaraguan individuals, significant genetic distances were found for Native American tribes (Colla, Toba, Kichwa and Waorani), as well as for populations from Asia and Africa (Núñez *et al.*, 2013). On the other hand, Nicaraguans resulted closer to an Argentinian sample from Río Negro province, to Costa Rica, and to the Colombian department of Antioquia (Núñez *et al.*, 2013).

Another report which included admixed populations from Guatemala, El Salvador, Nicaragua, and Panama, showed that the Panama population resulted more closely related to Colombian population than to the other three Central American populations (Baeta *et al.*, 2021). In the same analysis, a Native American group from Guatemala showed signs of genetic drift and isolation, as it commonly happens with South American native communities (Baeta *et al.*, 2021).

Concerning Mexico, an analysis of five X-STRs on the admixed population from the western part of the country reported significant genetic distances in a

comparison with data from Argentina (Bobillo *et al.*, 2011), African-Americans (Diegoli *et al.*, 2014), and Brazilians (Martins *et al.*, 2017), pointing out that these results are the consequence of an important multiethnic colonization process which occurred in that part of Mexico (Medina *et al.*, 2021). This work also reported a sex bias in the genetic composition of the X-chromosome of Mexican people. Another report spanning along different regions of Mexico, performed a forensic validation of the Argus kit (Qiagen), and found certain homogeneity among subpopulations from North, South, East, and West Mexico (Cortés-Trujillo and Zuñiga-Chiquette, 2019). More recently, a study of seven X-STRs on 105 individuals from Monterrey city was performed through massive parallel sequencing (MPS), together with a group of markers located on autosomes and Y-chromosome. MPS provides a greater amount of information than other analyses, although this technique is so far infrequently used for analyzing Latin American populations (Aguilar-Velázquez *et al.*, 2022). In this study, the number of alleles reported for the population of Monterrey was higher than that of other populations from Latin America (Peru), Europe (Spain and France) and Asia (Tibet).

Other works that present X-STRs data for different Latin American countries show a similar bias in gender admixture, with X-chromosome showing a larger Native and African component than autosomes, as already described (Casals *et al.*, 2022). Particularly, El Salvador did not show any regional division within the country (Casals *et al.*, 2022).

In regard to South American Native populations, the available information on X-STRs refers predominantly to communities from Colombia, Brazil and Argentina, and, to a lesser extent, to those from other countries (Chile, Bolivia, Venezuela). Besides the phylogeographic analysis reported by Santos-Lopes *et al.* (2007), an integrative analysis from Yang *et al.* (2010) collected data from six communities from the northwest of South America (including Zenu, Wayuu, Kogi, Embera, Waunana, and Arhuaco, together with two more from Central America), four Andean (Inga, Quechua, Aymara, and Huilliche), and four southeastern (Ticuna, Aché, Guarani, Kaingang). This report compared the variation of different portions of the genome, including 38 X-STRs, and evidenced an intermediate level of diversity for X-STRs, with an important correlation between genetic variation from autosomes and the X-chromosome (77%), in contrast to what was observed between other compartments, such as the Y-chromosome and the mitochondrial DNA (1%). Besides, this work confirmed the gradient in diversity previously reported from the north to the south of the continent (Yang *et al.*, 2010).

Amorim *et al.* (2011) analyzed X-STR variation on three native communities from Colombia (Kogi, Wayuu, and Zenu), in comparison to data from admixed populations from Costa Rica (Central Valley) and Brazil (Southern Gaucho), and described one haplotype block between Xp22.22 and Xp22.3, and a second one between Xp11.4 and Xq21.1, which were only observed in the Native populations. This particular pattern of linkage disequilibrium remarks that a process of genetic drift occurred in isolated Amerindian communities (Amorim *et al.*, 2011). This increase in linkage disequilibrium values in X-chromosome markers was also reported for Amerindians from other South American countries (Leite *et al.*, 2009; Wang *et al.*, 2010; Glesmann *et al.*, 2013; Caputo *et al.*, 2021).

In the Gran Chaco region of Argentina, a study on five X-STRs showed a genetic structure for Native groups according to their geographic location, being Ayoreo and Lengua from Paraguay separated from Chorote and Wichí from the Argentinian province of Salta, and in a more southern position, from Mocoví in the province of Santa Fe (Catanesi *et al.*, 2007). In another work, a more extensive analysis of the 10 STRs reported elsewhere (Gusmão *et al.*, 2009) for the Argentinian Wichí and Mocoví communities showed high percentages of homozygosity and linkage disequilibrium. A conspicuous random reduction in the genetic variation resulted not only in significant differences when they were compared to Argentinian urban populations, but also when both native communities were compared to each other, being a typical picture of the action of genetic drift (Glesmann *et al.*, 2013).

The relatively high mutation rate of STR polymorphisms make X-STRs very helpful, not only in solving paternity cases when the alleged father is missing, but also in population studies. These genetic markers proved their usefulness in analyzing the processes of genetic flow and genetic drift that occurred within South America from the start point of colonization, ≈500 years before present.

SNPs

In general, linkage disequilibrium tends to form blocks of sequences extending from a few kilobases to tens of them, with a very low recombination rate. This is particularly frequent along the non-recombining region of the X-chromosome, given the absence of recombination with Y-chromosome, thus having a low recombination rate in comparison to autosomes, with a tendency to form linked haplotypes (Szibor, 2007; Gabriel 2002). Thus, a block of a few tens of kb in the X-chromosome, which probably contains between 50

and 100 SNPs, can be very informative (Schaffner, 2004).

It is widely known that admixed Latin American populations show a sex bias in genetic information other than autosomal, given the origin of admixed populations during colonization by Spaniards. This bias has been demonstrated for the X-chromosome in an analysis of arrays including more than 500,000 SNPs along the whole genome (Bryc *et al.*, 2010), where the contribution resulted clearly biased to Native American and/or African ancestry. This imbalance in sex contribution has been described in different reports for populations from The Caribe -Mexico, Ecuador, Colombia, Puerto Rico, República Dominicana, Cuba, Honduras, Haití (Moreno-Estrada *et al.*, 2013), supporting a more important contribution of native women to the current Latin American heritage.

Particularly for Argentina, and in a previous report, Seldin *et al.* (2007) determined the contribution to Argentinian´s genome of three parental populations (Native American, European, and African) by selecting a set of SNPs for drawing up ancestral informative markers (AIMs). Eight out of 78 SNPs were located in the X-chromosome, including rs3747295, rs1978240, rs734329, rs5981813, rs992864, rs2380316, rs1867024, and rs762656. The global results showed a major European contribution (78%), followed by Native American (, 19.4%), and African (2.5%). Particularly, the X-SNPs showed a wide range of F_{ST} distances (from 0.00 to 0.92) between Native Americans and European Americans, including low F_{ST} values in some cases (e.g. rs1867024 -0.02-, rs992864 -0.05-, and rs1978240 -0.08-), intermediate F_{ST} values in some others (e.g. rs5981813 -0.31- and rs3747295 -0.33-), while other SNPs resulted with high F_{ST} values (e.g. rs2380316 -0.65-, rs734329 -0.67-, and rs762656 -0.68-). In a comparison with Argentinians, these AIMs resulted more similar to Europeans (Seldin *et al.*, 2007).

In fact, the variation observed in X-SNPs in CEPH-HGDP database is higher than in autosome SNPs, which could be explained as an effect of genetic drift, acting more intensely on X-chromosome variants (Li *et al.*, 2008). More than 16,000 SNPs from the non-pseudoautosomal region of X-chromosome were analyzed in 51 populations of the CEPH-HGDP, including Native American populations from Mexico (Pima and Maya), Colombia (Piapoco and Curripaco), and Brazil (Karitiana and Surui) (Casto *et al.*, 2010). An analysis of variance using SNPs showed a lower within-population component of the variation for X-chromosome than for autosomes. This is related to selective sweeps on X-chromosome that can occur in males, thus reducing its variability to a greater extent than for autosomes. Therefore, interpopulation differentiation can be more informative in the X-chromosome, and that is why it is important to consider its variation in population studies.

In the case of Brazilians, not only a Native component is present in their genome but also an African one, as it was reported for three populations (Salvador, Bambuí and Pelotas) on autosome and X-chromosome SNPs (Kehdy *et al.*, 2015). The analysis of 5,729 X-SNPs plus 331,790 autosomal SNPs showed a much higher African and Native component in the X-chromosome than in autosomes. This is consistent with a major contribution of African and Native X-chromosomes to current Brazilian populations than autosomes, according to its lower recombination rate and the important contribution of non-European women to current Latin American populations. Similar findings were reported by Homburger *et al.* (2015) using 694,834 SNPs in 437 admixed individuals from five countries (Colombia, Ecuador, Peru, Chile, and Argentina), to explore the population structure and demographic history of South American Latinos. By applying ancestry-specific Principal Component Analysis, they found that most of the European ancestry in South American Latinos was from the Iberian Peninsula, even though many individuals trace their ancestry back to Southern Europe, especially within Argentina (Homburger *et al.*, 2015).

In another study on the Hispanic Community Health Study/Study of Latinos (HCHS/SOL), which included people living in the United States of America, a different contribution of ancestry for 2,233 X-chromosome SNPs was reported according to the procedence of individuals. This study revealed a higher African contribution in Caribbean people from Cuba, Dominicana and Puerto Rico, and a lower African contribution in continental countries of South America and Mexico (Conomos *et al.*, 2016).

A study on different kinds of X-chromosome markers in three populations of Northeast Argentina was carried out with 15 X SNPs (rs6639398, rs5986751, rs5964206, rs9781645, rs2209420, rs1299087, rs318173, rs933315, rs1991961, rs4825889, rs1781116, rs1937193, rs1781104, rs149910, and rs652) (Di Santo Meztler *et al.*, 2018). In 198 individuals from three different locations in Northeast Argentina (Corrientes, Posadas, and Eldorado), the SNPs showed significant F_{ST} values for the comparison between Posadas and Corrientes, and also a genetic structure was found within the population of Eldorado. For this reason, it was necessary to separate Eldorado samples according to the origin of the reported grandparents into: donors who knew that their four grandparents were German and/or Swiss, and donors who did not know the origin of all four grandparents (Di Santo Meztler *et al.*, 2018). Thus, X-SNP variation was informative for differentiation within a region of the country.

According to Backenroth and Carmi (2019), sex-

biased admixture does not allow testing for Hardy–Weinberg (H-W) equilibrium on X-chromosome. Therefore, in American populations, although more than five generations have passed since colonization, some of the X-SNPs have not reached equilibrium or have been affected by lower and more recent gene flows.

Very recently, in an analysis of populations from different Latin American countries (Argentina, Barbados, Brasil, Chile, Colombia, Dominicana, Ecuador, Mexico, Perú, Puerto Rico) using SNPs located in autosomes and in the X-chromosome, Ongaro *et al.* (2021) tested different statistical methods of analysis for obtaining more evidence of the higher contribution from Native American females in X-chromosome and, inversely, the higher proportion of European male contribution in autosomes. A more important unbalanced contribution was reported for populations from Chile, Colombia and Ecuador, and less remarkable for those from Mexico and Peru (Ongaro *et al.*, 2021).

In general, the SNP polymorphisms mostly used for populational studies are biallelic, and thus it is considered that they emerged from a single mutation event in the past of the history of humanity. When it is possible to recognize the ancestral allele, SNPs can help to differentiate the origin of the populations under study, giving the possibility to differentiate the three main ancestors of current South American populations.

INDELS

In an extensive and pivotal study, Weber *et al.* (2002) identified and characterized 2,000 human diallelic INDELs distributed throughout the human genome in Europeans, Africans, Japanese, and Native Americans, where new alleles were generally lower in frequency than old alleles. The accuracy and stability of this kind of marker enable researchers to identify predecessors and descendants in historically demographic studies (Yang *et al.*, 2016).

There are several reports concerning X-chromosome INDELs polymorphisms in populations from Brazil, Colombia, Ecuador, and Argentina. Ribeiro-Rodrigues *et al.* (2009) analyzed 13 X-chromosome INDELs by PCR multiplex in different populations of Africans, Europeans, and Native Americans, revealing high inter-population variability. These authors analyzed 663 samples, including 151 African, 149 European, 176 Native American, and 187 Brazilian (from Belém), and found that these INDELs exhibited high differentiation between ancestral populations, thus allowing its use to measure ancestry proportions in three-hybrid populations, as it was the case of Belém and other Latin American populations. The estimated proportions of X-chromosomes in an admixed population from the Brazilian Amazon region showed a predominant Amerindian contribution (41%), followed by European (32%) and African (27%) contributions. The observed 41% Amerindian contribution based on X-linked data was consistent with the expected value based on previous mtDNA and Y-chromosome information (Santos *et al.*, 1999). The Native American and African ancestry proportions in the Belém population, based on X-chromosome information, resulted in higher percentages than those previously estimated for autosomal contributions, in accordance with the sex-biased mating pattern already mentioned for Latin American admixed populations (e.g., Marrero *et al.*, 2007; Corach *et al.*, 2010, Caputo *et al.*, 2021, Flores-Espinoza *et al.*, 2021). These results are in agreement with those obtained by Wang *et al.* (2008), in an extensive analysis of 13 mestizo and 26 Amerindian populations from Latin America, where Native American and African contributions were always higher for X-chromosome than for autosomes.

In addition, Freitas *et al.* (2010a) developed a 33-INDELs marker panel for the X-chromosome in a single PCR multiplex reaction, followed by capillary electrophoresis, to genotype a sample of 351 individuals (249 males and 102 females) of a mixed population also from the Brazilian Amazon. They chose physically close markers in an attempt to identify INDEL blocks with pronounced linkage disequilibrium in allele transmission. Using these criteria, they selected six possible marker blocks. After correcting for multiple analyses, only one marker (MID3756) showed significant H-W disequilibrium. Their results demonstrated that the measurement of statistical parameters for forensic use in this population is compatible with prior estimates from other populations using current X-STR panels. The 33 X-INDELs were also analyzed in 185 individuals from indigenous tribes of the Brazilian Amazon, in order to define linkage disequilibrium between the blocks. Their results demonstrated that among the markers, only twelve pairs had significant linkage disequilibrium. These authors were able to identify seven linked groups containing two and three polymorphisms each (Freitas *et al.*, 2010b). Their results for Native American tribes differ from those previously found for the population of Belém, since the Amerindians have low intra-population genetic diversity, and different mixing models can create different patterns of linkage disequilibrium according to the population studied.

Also in 2010, Resque *et al.* analyzed a sample of 461 individuals of European, African, and Native American populations using X-chromosome markers and identified four linkage groups. The data obtained

were used to describe the ancestral contribution of populations from four different geographical regions of Brazil: north (146 from Pará state), northeast (235 from Ceará and Pernambuco states), southeast (226 from São Paulo and Minas Gerais), and south (138 from Rio Grande do Sul). These authors selected three possible marker blocks based on the following criteria: (i) physical location on the X-chromosome, with a maximum distance of 225 Kb between them; (ii) heterozygosity in parental populations >30%; and (iii) allele length variation of 2-30 base pairs. The expected heterozygosity in parental populations was larger in Africans (36.3%) and Europeans (28%) than in Native Americans (17.1%). These results were likely due to the known population reduction effect in indigenous people during the migration to the American continent. Among the admixed Brazilian populations, heterozygosity is slightly larger in samples from the northeast (41.2%) and the southeast (39.6%) than in those from the north (32.5%) and south (29.5%). In the north region, a large Native American ancestry was observed (42%). The northeast and southeast regions had low Native American contribution (27% in both of them). In the south region, there was a large European contribution (46%). Overall, the results by Resque *et al.* (2010) showed that, considering X-chromosome markers, European ancestry averaged 39%, African ancestry averaged 29%, and Native American ancestry averaged 32%. The estimates obtained were compatible with expectations for a colonization model with biased admixture between European men and Native American and African women, so the authors concluded that the 24 X-INDELs panel described by them could be a useful tool for studying interethnic admixture in Brazilian populations.

In 2019, Martinez *et al.* studied a population sample of 500 unrelated Brazilian individuals from São Paulo using 32 X-INDELs, and found no deviations from the H-W equilibrium, except for the MID1361 marker, and observed lower genetic distances when they were compared to the Colombian admixed and European populations than to Native American, Asian, or African populations. Ancestry analysis revealed 41.8% European, 31.6% African, and 26.6% Native American contributions. In conclusion, the 32 X-INDEL markers presented high variability in the São Paulo population and a high forensic efficiency. The results of genetic distance analysis and ancestry estimates showed a closer proximity of this population to European than to Native American or African populations.

On the other hand, Rojas *et al.* (2010), in one of the largest surveys of admixture carried out so far in a Latin American country, evaluated ancestry in over 1,700 individuals from 24 Colombian populations using biparental (autosomal and X-chromosome), maternal (mtDNA), and paternal (Y-chromosome) markers. They found that autosomal ancestry varies markedly both within and between regions, confirming the great genetic diversity of the Colombian population. The X-chromosome, mtDNA, and Y-chromosome data obtained showed that there is a pattern across regions indicative of admixture involving predominantly Native American women and European and African men. In particular, eight X-linked STRs were genotyped in 385 males as previously described in detail in Collins-Schramm *et al.* (2002). These data showed that, for all populations, Native American ancestry on the X-chromosome is higher than on the autosomes. The higher Native and African ancestry estimated from X-chromosome markers, compared with autosomes, agrees with the sex-biased admixture at the origin of the populations examined inferred from the mtDNA and Y-chromosome data (Bedoya *et al.*, 2006; Wang *et al.*, 2008). Moreover, Córdoba *et al.* (2012) analyzed the genetic composition of 306 residents (all males) from Cauca (Colombia) using different variants, including 34 autosomal, nine in the X-chromosome, six mitochondrial, and eight in the Y-chromosome. These authors showed that the European (average 57.3%) and Amerindian (average 32%) populations have contributed in greater proportion to the current gene pool than the African ones (10.7%). Regarding the individual mixture, these authors observed that the African component was very homogeneous in all individuals, with greater variation in the Amerindian and European contributions. In addition, Ibarra *et al.* (2014) examined 11 urban admixed populations and a Native American group called Pastos, for 32 X-INDELs to deeply understand the genetic background of Colombia. They studied 869 unrelated individuals and found a highly diverse genetic background comprising all admixed populations, harbouring important X-chromosome contributions from all continental source populations. Indeed, Colombia is genetically sub-structured, with different proportions of European and African influxes depending on the regions. The samples from the North Pacific and Caribbean coasts have a high African ancestry, showing the highest levels of diversity. The sample from the South Andean region showed the lowest diversity and significantly higher proportion of Native American ancestry than those from the North Pacific and Caribbean coasts, Central-West and Central-East Andean regions, and the Orinoquian region. The results of the admixture analysis using X-chromosome markers suggest that the high proportion of African ancestry in the North Pacific coast was primarily male driven. These men have joined to females with higher

Native American and European ancestry (likely classic colonial asymmetric mating type). This high proportion of male-mediated African contributions is atypical of colonial settings, suggesting that the admixture occurred during a period when African people were no longer enslaved. In the remaining regions, the African contribution was primarily female mediated, whereas the European counterpart was primarily male driven and the Native American ancestry contribution was not gender biased.

Concerning Argentina, Di Santo Meztler *et al.* (2019) analyzed five X-INDELs (MID193, MID1705, MID3754, MID3756 and MID1540), together with other types of markers, from three Argentinian northeast cities already mentioned above. They reported no deviations from H-W equilibrium for Posadas and Corrientes, but deviations for Eldorado were given by an internal substructuring with two groups of different origin, one showing higher similarity with European countries, and the other more similar to Posadas and Corrientes populations.

More recently, Caputo *et al.* (2021) compared data obtained using 33 X-INDELS previously reported by Freitas *et al.* (2010a) with data from 30 autosomal-INDELS chosen from previous reports (Santos *et al.*, 2009, 2015; Pereira *et al.*, 2012; Ramos *et al.*, 2016) to analyze their potential use for ancestry determination in Argentinian populations. They were able to quantify the different contribution of Europeans, Native Americans and Africans in three regions of Argentina, being the north the region that showed much higher non-European components (38.81% European, 55.88% Native, and 5.31% African for X-INDELS), while the Centre showed intermediate values (52.96% European, 37.98% Native, and 9.06% African for X-INDELS) and the south was the region with the highest European component (64.76 European, 29.42% Native, and 5.82% African for X-INDELS). Concerning pairwise differences within populations, they found an important diversity in all three regions of the country, given by individual differences in African and Native American components (Caputo *et al.*, 2021). These authors proposed that the time since admixture and the model of admixture used for the analysis are factors involved in the differences observed when comparing ancestries from X-INDELS and autosomal-INDELs.

INDELs are considered as a unique event of insertion or, alternatively, deletion of a particular sequence of nucleotides in a precise region of the genome. Therefore, they are very helpful for recognizing different continental ancestries within a population, especially for those located in the X-chromosome, where selective sweeps have a deeper effect than in the autosomes.

ALU INSERTIONS

At present, there are only a few studies published concerning *Alu* insertion polymorphisms in the X-chromosome in Latin American populations. Pereira and Pena (2006) described a polymorphic *Alu* insertion embedded in a LINE 1 (L1) retrotransposon (*DXS225*, flanked by two microsatellites) on Xq21.3, and genotyped 684 males from the CEPH Human Genome Diversity Panel. Although this *Alu* insertion was found in all regions of the globe (namely Africa, Middle East, Central Asia, Oceania, Europe and America) only one of the five Amerindian populations studied (Karitiana) showed the insertion allele. Therefore, it is assumed that this allele has been introduced in this population by European and/or African admixture since the Karitiana had contact with European and African descendants in the early 20th century, and that it was absent from pre-Columbian Native Americans (Pereira *et al.*, 2006). In addition, the combination on the X-chromosome of a unique event polymorphism in linkage disequilibrium with two fast evolving microsatellites could make *DXS225* a valuable tool to evaluate genetic diversity of human populations and a highly informative tool for studies on human evolution. A year later, the same research group (Santos-Lopes *et al.*, 2007) increased the resolution power of their X-chromosome molecular analysis, as it has been explained in the STRs section. Through the combination of an X-*Alu* polymorphism of very low mutation rate with X-STRs of much higher mutation rate, these authors were able to support a single origin of modern man in Africa, and to identify a series of founder effects which occurred during migrations to occupy the other continents (Santos-Lopes *et al.*, 2007).

In 2010, Gayá-Vidal *et al.* investigated autosomal and X-chromosome *Alu* insertions in 192 individuals from two Amerindian populations of the Bolivian Altiplano, Aymaras and Quechuas (the two main Andean linguistic groups). Their study provided the first data on 14 X-*Alu* polymorphic elements in Native Americans, to establish if the *Alu* variation among South Amerindians exhibits some geographical or linguistic structure. These authors analyzed 18 autosomal and 14 X-*Alu* insertions. The two Bolivian samples showed a high genetic similarity for both sets of markers and were clearly differentiated from the two Peruvian Quechua samples available in the literature (Battilana *et al.*, 2006). Thus, no clear geographical or linguistic structure was found for the *Alu* variation among South Amerindians, which indicates that languages may not be congruent with the genetic features of the populations. With few exceptions (two polymorphisms), X-*Alu* insertions showed lower heterozygosity proportions compared to *Alu* insertions located in autosomes, with average values of 0.12 vs.

0.20 in Bolivian populations (Gayá-Vidal *et al.*, 2010).

Concerning Argentinians, three reports refer to X-*Alu* variabiliy. Resano *et al.* (2016), analyzed 10 *Alu* insertions from the X-chromosome in the population from the city of Bahía Blanca (province of Buenos Aires), and found that seven of these elements were polymorphic. In a comparison of the data obtained with those from eight different populations from Africa, Europe and America, the population of Bahía Blanca resulted genetically closer to Europeans and north Africans (average genetic distances 0.106 and 0.113 respectively) than to Native Americans (0.163) and Sub-Saharans (0.247). In addition, admixture analysis indicated a similar proportion of Native American (0.472) and European (0.479) parental contribution, with minor contribution of Sub-Saharan African component (0.049). In this way, these results reported higher Native American and African contributions than previous studies.

On the other hand, Di Santo Meztler *et al.* (2018) studied 10 *Alu* insertions, five INDELs, and 15 SNPs to gain insight into the genetic composition of North-East Argentina populations from Posadas, Corrientes, and Eldorado cities (total: 198 unrelated individuals). No deviations from H-W equilibrium were observed for Posadas and Corrientes, but Eldorado showed significant values because of an internal heterogeneity (i.e. genetic substructuring). Indeed, two groups of different origin were distinguished, one showing higher similarity with Europeans and the other with more similarity to people from Posadas and Corrientes. Of particular interest, *Alu* insertions compared to INDELs and SNPs, demonstrated the most differences with data from other countries and continents, and could be of use in ancestry studies for these populations. On the contrary, INDELs and SNPs were more informative for differentiation within the country. The genetic diversity found in this work between the studied populations was similar to those referenced in the bibliography (Callinan *et al.*, 2003; Athanasiadis *et al.*, 2007; Gayá-Vidal *et al.*, 2010), except for two *Alu* insertions that were monomorphic in some cases, and for the *Alu* insertion Ya5DP13 which was invariant, similarly to previous data for European populations (Callinan *et al.*, 2003; Athanasiadis *et al.*, 2007).

A third study included the analysis of nine X-chromosome Alu insertions in populations from the northern Argentinian province of Salta, showing non-significant differentiation between rural and urban populations within the province, and a close relationship with two Bolivian Amerindian populations (Ferragut *et al.*, 2019).

In accordance with the lower rate of mutation of *Alu* insertions compared to other markers, they can be found as monomorphic within certain communities, and they can show an absence of marked differences among closely related populations from South America. Therefore, X-*Alu* insertions are useful in ancestry studies of populations distantly related (i.e. from different countries or continents), while X-STRs, X-INDELs, and X-SNPs are more informative for differentiation within a region or country.

CONCLUSIONS AND FUTURE PROSPECTS

X-chromosome non-coding markers represent an important source of variability among populations, showing in many cases an important genetic differentiation when comparing populations of different ethnic origin. Its particular mode of inheritance makes a difference between X- and autosome variation in Central and South American populations, showing a more important Native and African contribution on X-chromosome markers than on those located in autosomes. Besides, within population variation is also important, and this advantage is taken for identification purposes, mainly for paternity cases when daughters are involved. For these reasons, the contributions on forensic markers located on the X-chromosome is currently growing, and the characterization of the different types of X-chromosome polymorphisms became very important especially when the populations under study are the result of genetic admixture.

Overall, the present review demonstrates that a bulk of information is already available for Latin American countries, although new techniques of analysis (as MPS) are still underutilized. Moreover, for some regions (for example, populations from the Patagonia, in the southernmost part of South America) and countries (for example, Perú and Paraguay) additional studies on X-chromosome non-coding variation are needed to provide a full picture of the human genetic diversity along Latin America.

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Catanesi C.I.: literature search, manuscript writing, preparation of Table 1, manuscript review.

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