

Review

Genealogy: The Tree Where History Meets Genetics

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Abstract: Although biological relationships are a universal reality for all human beings, the concepts of “family” and “family bond” depend on both the geographic region and the historical moment to which they refer. However, the concept of “family” can be determinant in a large variety of societies, since it can influence the lines of succession, inheritances and social relationships, as well as where and with whom an individual is buried. The relation between a deceased person and other members of a community, other individuals of the same necropolis, or even with those who are buried in the same tomb can be analysed from the genetic point of view, considering different perspectives: archaeological, historical, and forensic. In the present work, the concepts of “family” and “kinship” are discussed, explaining the relevance of genetic analysis, such as nuclear and lineage markers, and their contribution to genealogical research, for example in the heritage of surnames and Y-chromosome, as well as those cases where some discrepancies with historical record are detected, such as cases of adoption. Finally, we explain how genetic genealogical analyses can help to solve some cold cases, through the analysis of biologically related relatives.



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1. Introduction

Although biological relationships are a reality for all human beings, the “family” concept depends on the region and the historical period to which it refers. In this article, we explain how this concept has varied through different periods and societies. Associated with the concepts of family, kinship, and clan, there is another interesting point, mainly in prehistoric societies: with what criteria were people buried? According to biological ties? With their families? Burial rituals and possible relations between buried people in the same grave, or the same necropolis, are some of the fields genetics can help to understand. For example, in cases where children were found buried with adults, without sharing biological information between them, only a genetic study can propose a new path in genealogical studies: adoption.

In this sense, the genetic approach helps to explain how biological sciences can assist new perspectives on human genealogy, not only on the study of buried individuals, but also in the case of the surname heritage, or in the forensic field. Indeed, despite population movements being increasingly significant and fast, their genetic genealogy still allows individuals to be related, not only to search for missing relatives but also to investigate, for example, cold crimes.

2. “Family”: The Evolution of the Concept

Since the beginning of history in society, it is thought that humans have always been organized into family groups (Maynes and Waltner 2012). If, on the one hand, kinship relationships

are a practically universal human experience, on the other hand, the concept of “family” and “family bond” are not universal concepts, either in space or in time (Johnson and Paul 2016).

In various societies, the concept of family is restricted to the closest blood relatives, such as mother, father, siblings, grandparents, uncles, and cousins, with whom stable contact is maintained over time. Already, the concept of “relative” is extended to a variable number of individuals and may even include neighbours with a special bond in certain communities (Maynes and Waltner 2012; Johnson and Paul 2016).

On the other hand, the concepts of family and kinship can structure marriage patterns and possible heirs and can even affect where and/or with whom an individual is buried (Harper and Tung 2012). The following pages describe the concept of family and kinship in some of the most significant historical periods in Western European history.

2.1. *The Concept of Family and Relationship throughout Western European History*

2.1.1. Prehistory

In the 19th century, the line of research of the archaeologist Johann Bachofen focused on the hypothesis that the first humans were organized in matriarchal family groups. According to Bachofen, it was in later periods of humanity that the father figure replaced the mother figure as the leader of society (Allen 1999), centred on “Man, the hunter” as a central figure in the most primitive human societies and abandoning in a certain way the family and maternal concept (Maynes and Waltner 2012). Already with the Neolithic Revolution and the transition to agriculture, it is likely that life in “family” changed substantially, as groups began to seek stability and prosperity (Maynes and Waltner 2012). In addition, in the case of agriculture, some archaeological evidence suggests that the different marital ties and the formation of new family clans may have facilitated the hunter-gatherer transition to a sedentary lifestyle. Ceramics dating back to 5000 BC have been found in East Africa, from very distant regions, suggesting the possibility of complementary cross-marriages between men from pastoral cultures and hunter-gatherer women (Maynes and Waltner 2012). Indeed, a recent investigation by Haas et al. (2020) has demonstrated what could have been a common practice in early periods—female hunters.

Moving to the Bronze Age, deposits dating from the Late Bronze Age do not demonstrate significant differences between women and men, signifying a complete transition to agriculture, already from childhood (Whitehouse 2006).

(a) The case of Çatalhöyük (Konya, Anatolia, Turkey)—the family society

Çatalhöyük (Konya, Anatolia, modern Turkey) is the oldest known human settlement of particularly significant size: approximately 9000 years ago, between 3000 and 8000 people lived there. Various excavations have been carried out, uncovering an early agricultural settlement (7000 BC), unusually well preserved. Archaeologists believe that the population was mainly engaged in subsistence agriculture, but the presence of shells and obsidian suggests long-distance trade (Fielder and King 2004; Gutrie 2005). One of the most outstanding features of Çatalhöyük seems to be the absence of complex social organization or centralized political authority. Indeed, it seems to have been a community where social life was organized on a domestic scale, given that public spaces, administrative buildings, or overly elaborate houses have not been detected. The most widely accepted theory is that the community of Çatalhöyük was centred on the family (Fielder and King 2004; Gutrie 2005). There is no evidence of the existence of social classes since residences with different characteristics have never been found nor has evidence of social differences based on gender been found, since paleo-diet studies revealed a similar diet in men compared to in women, indicating a probable similar social status (Fielder and King 2004; Gutrie 2005).

2.1.2. Ancient Period

(a) Ancient Egypt (3100–30 BC)

In ancient Egyptian society, there were several family typologies: mono- or polygamous. Within monogamy, it was common to observe married couples without descendants,

couples with descendants (designated “nuclear family”), as well as married couples with descendants living with other relatives (almost always, grandparents and/or uncles and cousins) (Allen 2009). This “nuclear family” was the centre of society in Ancient Egypt, and many of the Egyptian divinities were ordered in similar family groups (Brewer et al. 2004): father, mother, and son/daughter (s) (Goucher and Walton 1998). The lineage was traced through both paternal and maternal lines. It is noteworthy that the word to designate “mother” was also used for “grandmother”; just as the word for “father” was also similar to that used for “grandfather”. Likewise, the words “son”, “grandson”, and “nephew” (the same in the female case) had the same meaning (Brewer et al. 2004), as well as “uncle” and “brother” (the same in the female case) also being designated by the same word. Possibly the most difficult thing to understand nowadays is the case of “sister” and “wife” that in ancient Egypt seemed sometimes to be used as synonyms, demonstrating the narrowing and importance of marital ties (Brewer et al. 2004).

Despite the antiquity of this society, there seems to be an idea of relatives very close what is currently used in Biology, particularly concerning the maternal lineage and paternal lineage. According to Allen (2009), both the family and the social organization in Ancient Egypt had a particular focus on women, whatever their place in society—widows, mothers, daughters, etc.—their social status was exceptional. The position of women in ancient Egypt is a fundamental criterion to distinguish ancient Egyptian society (and other African ones) from other contemporary Semitic societies, such as those of Mesopotamia, Babylon, and Indo-Europeans (Diop 1971). According to Diop (1971), “the couples represented in Egyptian monuments are united by tenderness, friendship and life in common, contrary to what was observed in the Eurasian world of this period: Greece, Rome and Near East”.

Another curious point is the fact that no records of infanticides of girls have been found, for example, by abandonment, contrary to what has been registered for other ancient societies, such as Greek or Roman societies. All children born in Egypt were considered legitimate, as they were always the result of maternal lineage (Allen 2009).

(b) Ancient Greece (1200–146 BC)

The “family” was considered the social and economic centre of life in Ancient Greece (Moulton 1998). The most important function of the wife was to provide the husband with legitimate male descendants and to inherit the material assets of her father, following the line of inheritance within the “family”. A curious issue is that the wife was in her husband’s house as a “loan” since she would always be considered part of her father’s house “family”, even if she already had a family of her own (Moulton 1998). On the other hand, the size of the “family” was very important in ancient Greece. The family nucleus was preferably reduced to avoid too many heirs, as well as excessive gifts in the case of daughters. Thus, two or three offspring would be the most desirable, and if possible, males (Moulton 1998). Indeed, almost always in the case of daughters, it was possible to “expose (εκθέσει) unwanted descendants in public places, a practice only prohibited in the City-States of Thebes and Ephesus” (Moulton 1998).

The issue of marriages within the same biological family seemed not to be prohibited, since in cases of the death of the father without a male heir, the unmarried daughter was encouraged to marry the closest male relative so that the inheritance was safeguarded within the family nucleus (Moulton 1998; Carr 2017). The Greek family nucleus could also be made up of other single relatives, such as sisters or aunts, and widowed or aged relatives, frequently paternal grandparents (Carr 2017).

(c) Jewish Culture

Some historians point to the relationship between the emergence of monotheism in the ancient Middle East and the “creation of patriarchy”, that is, the elevation of the father to a place of power and authority in the family and other social and political institutions. This line of thought is based on religious texts, especially in the Old Testament, that originated in a specific historical background, probably around 2000 BC. In effect, the Judaic tribes

who wrote the books were transitioning from the society of semi-nomadic tribal clans to agricultural settlements and then to monarchical states (Maynes and Waltner 2012).

The predominant family form showed a strong emphasis on the male line of descent. For example, the Jewish woman was expected to marry and move from the home of her father and her lineage to that of her husband. In Israel, to keep women and their children within the line of their husbands, it was considered opportune and convenient for men to marry the widows of their brothers. Men could have multiple wives and concubines, while women could only have one husband (Maynes and Waltner 2012). In this sense, Jews were similar to other Near Eastern cultures of the time. Marriage was conceived as a partnership between husband and wife.

On the other hand, although the Hebrew Scriptures regulated marriage, inheritance, and definitions of legitimacy, not all those contents were adapted to the circumstances in which the Jews found themselves, for example, in the medieval world. Jews living in Christian Europe, or in the Byzantine Empire, were monogamous since Christian law prohibited polygamy. By contrast, Jews living in Muslim territories practised polygamy (Mitchell 2007). In general, marriages under Jewish law were endogamous, always within their communities, or even within their own families. Although intergenerational marriage within a family (such as an uncle-niece) was relatively rare, it was not prohibited. In Muslim territory, the marriage of very close relatives occurred regularly between Muslims and Jews. Since Jews were generally not allowed to own land, Western feudal laws of primogeniture (inheritance by the eldest son) were not relevant. When this occurred, the need to maintain the integrity of family properties encouraged marriage between relatives, such as cousins (Mitchell 2007).

One of the most unusual aspects of Judaism is that both tradition and religion are transmitted through the maternal line. Unlike what happened in other contemporary cultures, all children were highly valued, and the births of both boys and girls were celebrated. Girls and women were not as restricted in their activities as in Christian and Muslim culture (Mitchell 2007).

(d) Ancient Rome (753 BC–476 AD)

As in ancient Greek society, the “family” was always the social unit in Rome. While in Greece the figure of the husband-father was decisive, the “Roman family” presented a practically equitable relationship within marriage (Moulton 1998). Around 200 BC, there are already marriage records where the social status of the woman was not altered by the fact of getting married (*liberum matrimonium* or free marriage) (Moulton 1998). On the other hand, as in Greece, women continued to be linked to the authority of her father (*paterfamilias*) and her biological family, at least until his death, unless the *paterfamilias* himself granted her liberation from his authority. Another difference from Greece is the fact that marriage between relatives was prohibited in Roman society. It would be considered incestuous and not recognized by the law. Nonetheless, the main objective of marriage continued to be legitimate offspring, if possible male, thus ensuring paternal inheritance (Moulton 1998).

(e) Islamic Culture (632 AD)

An example of the close relationship between religious belief and the establishment of the “family” is Islam, which emerged more or less in 632 AD. Before the rise of Islam, future Islamic societies were formerly Bedouin or Arab. According to the traditional view, women in Bedouin and Arab culture were “non-legal people” who were considered property of their parents. Parents could demand that baby girls be killed outright, as they would not be useful in the future. It was also normal to sell daughters to other men, as wives or as concubines. Moreover, daughters could not inherit (Mitchell 2007). Today, many historians believe that the Prophet would be determined to modernize (from a 7th century perspective) Arab culture and concept of the family, mainly to improve the status of women (Mitchell 2007). The Qur’an, the record of the Prophet’s revelations, contains many passages that treat women as the religious equivalents of men (Maynes and Waltner

2012). As Islam spread from the Arabian Peninsula, it settled in a region where women had great independence, and many tribes followed matrilineal principles, where, for example, wives remained with their families of origin and their children. For the first time, an Arab Prophet declared that women could be full members of the Islamic community, the umma (Mitchell 2007). Dramatic changes occurred in Islamic marriage between the time of the Prophet's life and the time when the codification of the sacred writings was completed, in the 10th or 11th century. After the 8th century, there is no longer evidence that women, elite or not, were involved in covenants or conditions for their marriages, just as marriage to a widow was no longer acceptable (Maynes and Waltner 2012). Patriarchal practices inherited from the Jewish, Greco-Roman, Christian period and Persian cultural legacies in the areas of Muslim influence intervened in the development of Islam, contributing new realities of life and family formation (Mitchell 2007; Maynes and Waltner 2012). When it comes to the offspring, in general, the children belong to the husband. Given the traditions of a tribal organization that formed the basis of Arab culture, long before the Prophets, children were not necessarily associated only with their biological parents. The extended kinship between men operated as a social network and as political connections. In fact, for example, the Kurdish general Salah ad-Din ("Saladin") himself was raised by his uncle and succeeded him as sultan in Egypt (Maynes and Waltner 2012). In this way, the children were raised in large extended kinship environments, where full and half-siblings socialized freely. Girls from wealthy families were confined to women's rooms. In general, descendants of marriages, or formal cohabitation arrangements, were considered legitimate under Sharia; therefore, Muslim families with significant economic power tended to be enormous, with dozens of descendants (Mitchell 2007).

2.1.3. Western Medieval Period (476 AD–1453 AD)

The literature indicates that kinship models according to genealogy-genealogical models, emerged for the first time in Western European society, during the Medieval Period (Johnson and Paul 2016). For example, as early as the 11th century, Christian scholars were dedicated to formalizing the genealogy of the ancestors and family of Jesus Christ as a tree (Klapisch-Zuber 1991, 2000), later adopted as a "family tree", and already in the 16th century, the representation of families according to the concept of the genealogical tree was quite popular throughout Europe (Johnson and Paul 2016).

(a) Christian Western Europe-The "Old Europe"

The so-called "Old Europe" society was a society structured to some extent around lineages and/or kinship ties (Casey and Hernández-Franco 1997). From Ireland to Genoa, Tuscany or Naples, from Portugal, Castile and Aragon to Poland or Lithuania, the family structure adopted both forms of organization. Anthropologists and historians have demonstrated over the centuries these relations of consanguinity and filiation that can be established within the family. On the other hand, the "patriarchal family", characteristic of feudal Europe, has prevailed for a long time, where the lord held the power over his lands, regarding both food and the way of life, as well as the application of justice, death, and burial site.

In this society, the idea of lineage was perceived as a symbolic form of ordered bloodlines of unilineal descent, which had its origin in a reputed founder and which was transmitted through the trunk to successive generations in the lineage. Lineages were not only a trait that granted rank within the social structure but also allowed the family name and house to be maintained for generations—their name, their assets, and their distinctive deeds.

According to Casey and Hernández-Franco (1997), the family was a plastic concept, since belonging to a lineage depended largely on oral tradition before the middle of the 17th century, when "christening records began to be available tracing the grandparents". The Catholic Church itself in the West has contributed in some way to the importance of the family in society, implementing norms for the constitution of what could be called "the true family". Marriage should be exogamous, thus hindering cases of incest and allowing

the circulation of inheritances and wealth between different families. In addition, at the end of the 11th century, a ritual among the betrothed, the wedding, began to become more frequent, being mandatory from the 16th century through the Council of Trent. In general, the wife after marriage becomes a member of the family of her husband.

On the other hand, the concept of “house societies” developed by Lévi-Strauss (1984, 1987, 1999) is quite relevant in medieval society, where kinship may not be biological, but based on the social relations of those who cohabit in the same home. The “House” is considered a significant space that would serve as a link for a social formation between individuals. This type of relationship between individuals could be studied throughout the generations since there was a record (symbols, marks, and designs) in different objects belonging to the individuals (clothing, war material, household objects, etc.) (Johnson and Paul 2016), and later, those belonging to the same family, or “House”, are included in the genealogical trees.

The role of men and women in the medieval Christian family depended mostly on the territory where they lived, since territories under German law, or under the influence of Roman law, had different perspectives on the role that the father and mother should play, men and women, in society, mainly regarding business. In general, adult males were expected to spend much of their time outside the family unit, participating in warfare, agriculture, herding, or administration. Adult women, especially when married, ran the household. They oversaw not only the cooking, manufacturing, cleaning, babysitting, and accounts but much of the farming as well (Murray 2017).

Families could include a large number of children who were not necessarily biologically related in their home. Thus, “children” could include not only legitimate progeny, but also cousins, stepbrothers, or even children that the father had had in previous relationships (Murray 2017).

2.1.4. Modern Period

It was not until the 20th century where the concept of relative, kinship, and family began to undergo important changes in literature. In the early 1960s, research on “kinship” faced considerable resistance from researchers, as they considered this research as “too biological” (Johnson and Paul 2016). Indeed, researcher Schneider (1972, 1980) published various criticisms during this period, claiming that the concept of “kinship” was an inter-cultural system. He insisted that the West was overly concerned with “natural kinship”, prioritizing biological classification over social models (Schneider 1972, 1980; Johnson and Paul 2016). To emphasize the rejection of the strict “biological” concept of the 20th century, researchers begin to study kinships as “relationships” or “relatives”, trying to distance the “biological kinships” based on reproduction (from the Latin “*genitrix*” (mother) and “*genitor*” (life generator)). Similarly, these 20th century Western approaches to kinship may not be appropriate to analyse the social relationship in non-European contexts, where their application may hide the way the family relationship is understood and perceived (Schneider 1980; Astuti 2009; Holmes 2009; Viveiros de Castro 2009; Lambek 2011). For example, family and relatives in some tribes of Amazonia are based on the strict concept of “nonbiological theory of life” (Viveiros de Castro 2009). In their vision, the soul, or spirit, is a shared substance that connects all individuals (human and non-human), while a person’s body is built through interaction with other individuals. Such interactions with other bodies form the basis of the family in these Amazonian societies. In Papua New Guinea, the Kamea conceptualize “parent–child” relation as an “inherently disembodied” bond, since family relationships are based on interactions with other individuals in the environment where they live (Bamford 2004); like the Reites, also in Papua New Guinea, they believe in non-genealogical kinship (Leach 2004). Finally, the Vevo, in Madagascar, distinguish genealogical relationships from social relationships, emphasizing that non-biological parent–child relationships would be crucial for family life (Astuti 2009).

3. The Concept of Family, Clan, and Kinship

According to [Johnson and Paul \(2016\)](#), “family” is interpreted in the current Western world as a fundamental human institution and construction, which forms the basic social unit of collective action, beyond the individual. On the other hand, according to [Maynes and Waltner \(2012\)](#), families are small groups formed by people, linked by culturally recognized marriage ties, or similar forms of conjugal lives, descendants, adopted or not, that typically share a common space (house) over some time. This joint residence is necessary for a time, varying according to the stages of the family circle and respective society. Family ties are not broken simply by leaving the space shared by the family, although family relationships can change. However, the fact of inhabiting the same space does not imply belonging to the same “family” ([Maynes and Waltner 2012](#)).

The term ‘clan’ is derived from the Gaelic word *clann* meaning ‘children of’ and came into medieval English usage approximately in the 15th century, to describe the kin-based character of early Irish and Scottish Highlands societies ([Burnham 2015](#)). According to [Burnham \(2015\)](#), members of Scottish clans were not necessarily all blood related, regardless of a strong idea of family. The *lineage* and *clan* concepts are not well defined, depending mostly on the society and culture in which they are rooted. An approximation to the possible explanation could be that members of a lineage allege to know the genealogical connections interlinking all members of the group, and these links are viewed in terms of generation and relative birth order ([Burnham 2015](#)). By contrast, persons are members of a clan because they are the descendants either of their fathers or their mothers, and the terms ‘patriclan’ and ‘matriclan’ are often used. However, according to [Burnham \(2015\)](#), clan members in many societies recognize a founding clan ancestor, who is often of mythical or non-human status, and this is also related to totemism ([Lévi-Strauss 1962](#)). The reason for clanship is related more to ethnicity than biological kinship, and in some cases, “clan” is also applied to territorial groups which are recruited based on both unilineal descent and long-term co-residence. According to [Burnham \(2015\)](#), “clan” is often used in a symbolic way to “refer to any group of persons who act toward each other in a particularly close and mutually supportive way”.

In turn, “kinship” is almost always considered a biological relationship between people, except for adopted individuals who, in that case, are also considered to belong to the bloodline.

The Concept of Family, Kinship, and Burial Place

If, on the one hand, the concept of family and kinship can affect where and with whom an individual is buried, reverse thinking according to [Harper and Tung \(2012\)](#) is also possible. Indeed, where and with whom an individual is buried can inform about the family status, as well as of his/her descendants (both biological and non-biological), regarding a community, other individuals in the same necropolis, or even the same grave. In this way, funeral and burial rituals may represent a possible biological kinship affiliation ([Harper and Tung 2012](#)). According to [Harper and Tung \(2012\)](#), funeral rites function as an identity for both the person who buries and the deceased individual.

The study of funeral ceremonies is based mainly on written records, where this practice is described according to the family tradition and/or society. However, in prehistoric societies, such as Neolithic or Bronze Age societies, among others, the study between the funeral ritual, the type of burial or cremation, and the possible relationships between individuals tends to be local, given the lack of written information to determine if a certain procedure was common in several places or typical of a specific community. In most cases, these studies are carried out by specialized teams of archaeologists and anthropologists ([Stojanowski and Schillaci 2006](#)), who, through finds of different objects and burial typologies, address various questions related to possible kinship networks ([Alt and Vach 1995, 1998; Stojanowski and Schillaci 2006](#)).

Recently, molecular studies, more specifically DNA genetic analysis, have begun to be used considerably to identify kinship relationships within the same burial area ([Keyser-](#)

Tracqui et al. 2003), since some anthropological indicators, such as cranial and dental similarities, etc., are frankly less precise than the genetic data. However, only the genetic study of a population allows the detection of burial patterns of biologically unrelated individuals (such as adoption), although there are records, both written and oral, that indicate a possible kinship between them.

The study of mass graves, where the genetic study indicates a total absence of close kinship or of any type of common biological lineage among the buried individuals, undoubtedly opens other avenues of investigation beyond adoption. Hypotheses, such as wars and conflicts, diseases, or a place where unidentified people were buried, are examples of cases where, in principle, DNA analysis would be useful to confirm the absence of biological patterns among buried people. However, in addition, it could suggest another kind of situation, where the burials are a reflection of the fact that community bonds were stronger than biological or family ties; thus, it could provide interesting data about social behaviour (Palomo-Díez et al. 2015).

For example, in the Pre-Bell Beaker period (initial Chalcolithic) (3700–4000 B.P) in the Central Iberian Peninsula, the main burial pattern consisted of individual inhumations, even though many collective burials have been observed, such as those in El Tomillar (Ávila, Spain), Los Areneros (Segovia, Spain), or Los Cercados (Valladolid, Spain) (Palomo-Díez 2015). However, during the Bell Beaker period in the same region, collective inhumations are not known, and they reappear in the Bronze Age, in places such as Cueva de la Revilla (Burgos, Spain) or Los Rompizales (Burgos, Spain) with a high number of individuals inhumed together; other simultaneous burials also appeared with a fewer number of individuals, such as Los Tolmos (Soria, Spain) (Palomo-Díez et al. 2018). Moreover, we must consider that appearances are sometimes deceptive. For example, in the case of Los Tolmos, the find of a three-person burial, constituted by two adults and a child, seemed to point to a modern classic family (father, mother, and child). Nevertheless, genetic evidence revealed that the infant's and one of the adults' skeletons were related by mother–child kinship, but the third inhumed person was an adult female (not a male as expected), and she was not biologically related to the other (Esparza et al. 2017; Palomo-Díez et al. 2018). This case demonstrates that genetics could be a great tool to unravel these kinds of issues.

4. The Genetic Approach

There is a wide range of historical, anthropological, and archaeological questions that DNA analysis can help answer. Examples of this are the phylogenetic study of plants, research on biogeographic origins and population movements, the determination of the molecular sex of adult and infant individuals (Mulligan 2006), or the family relationships between individuals buried together or separately. According to Johnson and Paul (2016), the archaeological studies of kinship reflect the growing interest in understanding what the concept of “family” has meant throughout history and if human beings have followed the same rituals and concepts in distinct periods. Alternatively, in cases of soldiers whose identification is not known given the time passed after their deaths, or of victims of war crimes and torture, the application of DNA analysis acquires a leading role (Hummel 2003; Zupanič Pajnič et al. 2016; Friš et al. 2019; Gomes et al. 2019; Obal et al. 2019; Palomo-Díez et al. 2019; Marshall et al. 2020).

At a historical level, the use of genetic research has been recognized in numerous cases: to give a few examples, the case of the alleged slave son of Thomas Jefferson (Foster et al. 1998), and the presumed son of Louis XVI of France (Jehaes et al. 1998; Olalde et al. 2014). Finally, one of the most publicized contributions of genetic analysis on the remains in a particularly degraded state has been the investigations in which the corpses of the Russian Romanov royal family, found in a mass grave, were analysed and identified (Gill et al. 1994; Gilgenkrantz 2009; Coble et al. 2009).

In both archaeological and forensic cases, genetic studies almost always focus on identifying the donor of the biological sample or determining its biogeographic origin, describing its external phenotype and relating two or more samples. In each analysis, a set

of DNA polymorphisms, also called genetic markers, are characterized, almost always located in non-coding loci (physical places) of the human genome (Butler 2005; Pinheiro 2010). Each variant within the same genetic marker is called an allele (Butler 2005). In practically all genetic studies, the objective is to obtain the genetic profile of the individual. This could be defined as a set of heritable characteristics (alleles), for a certain number of genetic markers, detectable in any sample that belongs to it. In other words, for the same set of genetic markers, a sample of blood and lung tissue from the same individual must present the same genetic information.

4.1. Nuclear Information

Nuclear DNA (nuDNA) present in all living nucleated human cells is organized into chromosomes (organized by pairs). It is divided into autosomal DNA (auDNA), with 22 pairs of chromosomes, and heterosomal DNA, made up of the sex-responsible chromosomes: two X chromosomes (in women) or an X chromosome and a Y chromosome (in men).

(a) Transmission of markers located on autosomal chromosomes

Both the father and the mother transmit to all their descendants, sons, and daughters, one chromosome from each autosomal homologous pair. That is, each offspring receives 22 autosomal chromosomes via the father and another 22 homologues via the mother (Figure 1).

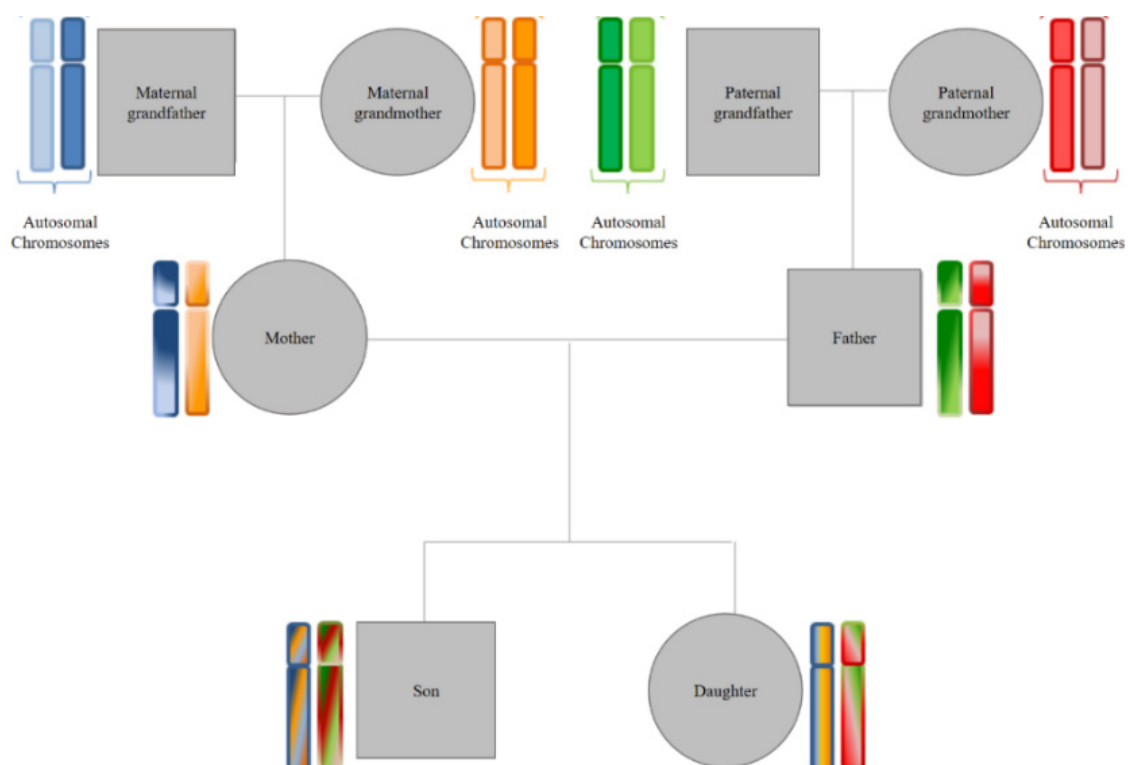


Figure 1. Autosomal chromosome transmission from the maternal and paternal sides. The recombination of the genetic information can be visualized by the mixed colours in the progeny's chromosomes. Source: (Gomes 2020).

(b) X chromosome transmission

X chromosome transmission takes place differently in mother and father. On the one hand, a father transmits an X chromosome exclusively to his daughters, without recombination; on the other hand, a mother transmits to all her descendants a copy of one of her X chromosomes, and there may be recombination between her homologous X chromosomes (Figure 2).

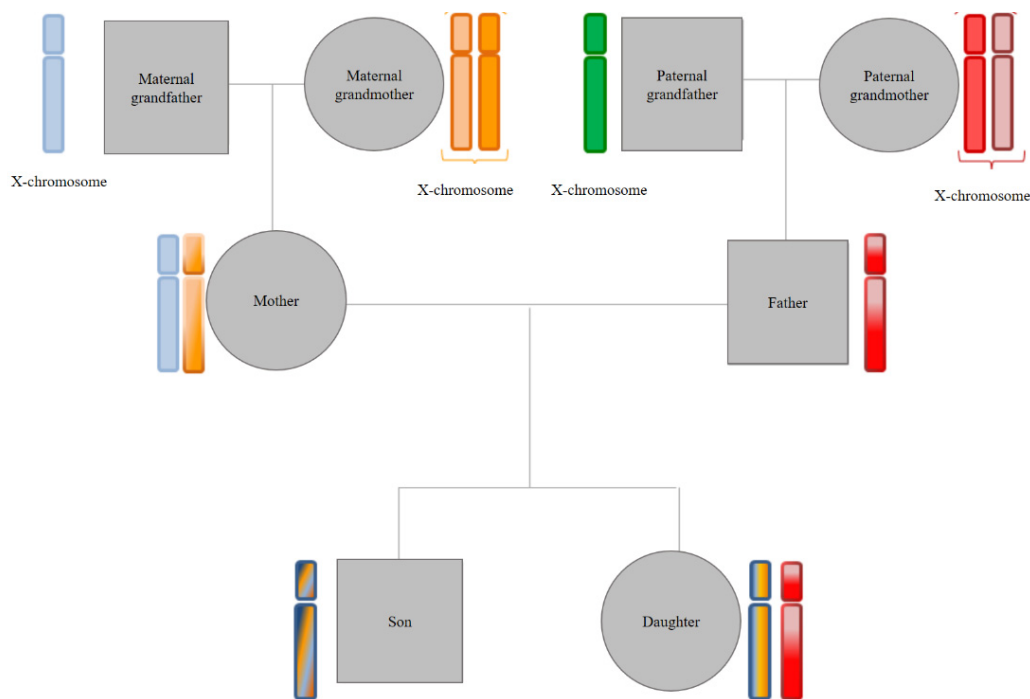


Figure 2. X-chromosome transmission from the maternal and paternal sides. The recombination of the genetic information can be visualized by the mixed colours in the progeny's chromosomes. Source: (Gomes 2020).

4.2. Lineage Markers

Lineage markers include the Y chromosome and mitochondrial DNA (mtDNA), which are transferred directly from generation to generation, either from mother to child in the case of mtDNA, or from father to son in the case of Y-chromosome.

Y-chromosome markers and mtDNA sequence information are treated as linked markers with the entire profile inherited as a block.

The advantages of lineage markers are their capacity for the estimation of the biogeographical origin or exclusion in paternity testing or identification of missing persons. Their principal disadvantage is their null capacity for establishing direct relationships between individuals. Nonetheless, they are a great tool to trace an individual's membership to a certain lineage or to discard it.

Lineage marker characteristics mean that they cannot be used to individualize or identify a specific individual, but these same characteristics bring a rich pool of possibilities:

- The study of families
- The study of lineages
- The analysis of the biogeographical origin
- The possibility of obtaining information from degraded samples or minimal signs; in the worst cases, these markers could provide the only available information when DNA is in very poor conditions.

(a) Y chromosome

The Y chromosome is a small acrocentric chromosome (Skaletsky et al. 2003). Two types of regions can be distinguished in this chromosome, fundamentally: non-recombinant region and pseudoautosomal region (PAR1 and PAR2) (Figure 3).

three male-line descendants of three sons of John Carr (Jefferson's nephews) and compared with samples from two of Sally Hemings' male descendants (Thomas Woodson and Eston Hemings Jefferson). The results pointed to a match between descendants of Eston Hemings Jefferson and relatives of Thomas Jefferson but not with descendants of Thomas Woodson, and it was concluded that according to this result "Thomas Jefferson was the biological father of Eston Hemings Jefferson". Nevertheless, this statement was not correct, and later, in a letter, the authors clarified that Thomas Jefferson's brother could also have been the father of Sally Hemings' later children, because both Thomas Jefferson and his brother shared the same Y-chromosome haplotype.

The absence of recombination in most Y-chromosomes provides the advantage of inferring paternal biogeography ancestry. When a mutation occurs, it is inherited by all males in this family along the paternal line.

The analysis of the genetic structure of populations through Y chromosome mutations is one of the main applications of this chromosome, and it could theoretically be related to population events in a specific geographic and temporal context. In addition, the polymorphism of Y-chromosomes has been used to trace the biogeographic ancestry of populations to resolve important questions about population origins and migration history worldwide.

(b) mtDNA

Mitochondrial DNA is the DNA portion that is in the mitochondrial organelles, and it consists of a closed circular double-nucleotide chain structure (Figure 4).

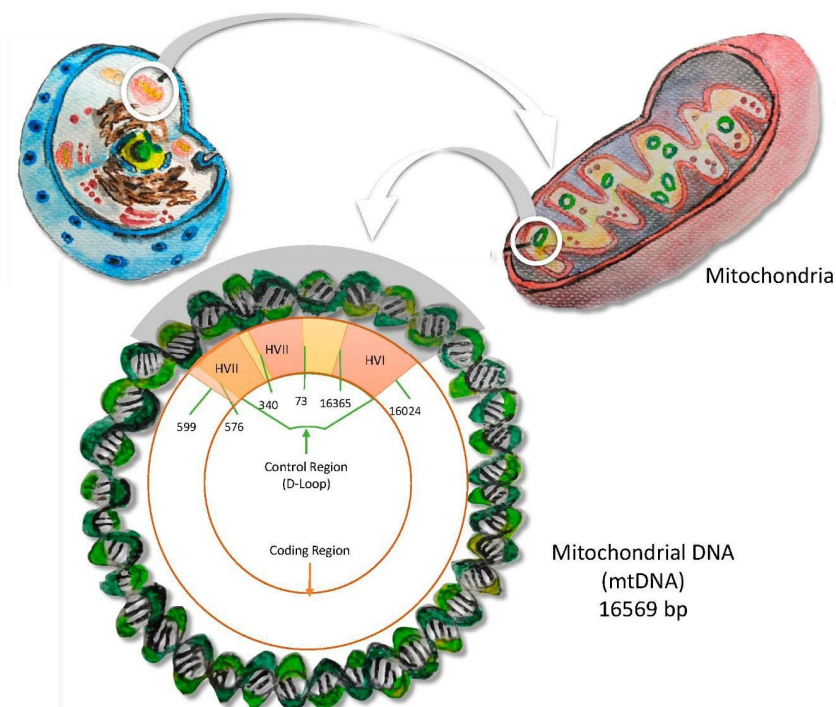


Figure 4. mtDNA location and structure.

To be precise, human mtDNA is located in the mitochondrial matrix, and it is a double-chain circular molecule divided into two different regions (Figure 4): a coding region and a non-coding region, also named: control region, D-Loop, or regulation region (Pinheiro 2010; Crespillo and Barrio 2018) (Figure 4). This Control Region is subdivided into three regions: Hypervariable Region I (HVI), II (HVII), and III (HVIII). HVI and HVII display nucleotides more or less prone to mutation, denominated hotspots (Pinheiro 2010).

In genealogical and kinship studies, the most interesting mtDNA region is the non-coding region, because it contains the most genetic polymorphisms, but from a phylogenetic perspective, it is necessary to study the full mitochondrial genome.

The mtDNA displays a series of interesting characteristics (Crespillo and Barrio 2018):

1. mtDNA is inherited through the maternal lineage, from the mother to all her descendants.
2. This molecule presents a recombination absence.
3. The cell contains a high number of mtDNA molecules, which allow a greater probability of results obtained from problematic samples, such as human skeletal remains.
4. The mtDNA shows specific geographical and ethnic distributions, involving the possibility of assigning a particular origin to an individual.
5. The small size and the circular structure of mtDNA support less degradation of the molecule. Furthermore, the higher copy number compared with nuclear DNA means there are many more mtDNA molecules available for replication. This characteristic is fundamental when working with critical samples with low template DNA.

As occurred with the Y-chromosome, mtDNA also can be organized in haplotypes and haplogroups. The haplotype of a specific sample is defined by the set of variations compared to the reference sequence (rCRS) (Andrews et al. 1999) and which are inherited as a block; it provides information about the maternal lineage since it is shared by all the individuals maternally linked.

mtDNA is inherited nearly inalterable through multiple generations, which has allowed maternal lineages preserved throughout human history to be established. This characteristic enables phylogenetic studies among different human populations. It has been possible to establish genetic mtDNA differences among these human populations; therefore, through mtDNA analysis, it is possible to infer an approximation of the biogeographical origin of an individual. In consequence, the haplogroup is the set of haplotypes that contain certain specific polymorphisms and have a common origin. The specific combinations of this polymorphism in specific positions are typical of certain geographical areas: Europeans, Asians, and Africans, and the different clades are subdivided into other subclades (Van Oven and Kayser 2009). The result is a specific mtDNA geographical distribution.

To track the biogeographical origin of an individual, it is necessary to compare the results with population databases, such as EMPOP (www.empop.org), SWGDAM (Scientific Working Group on DNA Analysis and Methods), mtDB (<http://www.mtodb.igp.uu.se/>) or MITOMAP (<http://www.mitomap.org>) for contemporary populations; or AmtDB (<https://amtdb.org/>) for ancient human populations.

Applications of mtDNA Testing

In comparison with nuclear markers, mtDNA does not allow us to identify individuals and only is useful to establish maternal lineages. Another disadvantage is its low discrimination power. However, its higher mutation rate, and other own characteristics of this molecule, such as matrilineal inheritance and no recombination, mean that mtDNA can provide interesting supplementary data in many archaeological cases:

- When facing low template DNA samples, such as ancient human skeletal remains.
- When the objective is to detect kinship through the maternal lineage.
- To approximate the biogeographical origin.

Not only mitochondrial DNA but all kinds of genetic tools have allowed us to establish or discard kinships. Indeed, despite finding two or more individuals buried together or sharing the same surname, genetic analysis can discard the biological relationship whenever the sharing of genetic information is not observed. This raises other hypotheses, such as adoption.

5. Adoption and Genealogy

Adoption is understood as a process whereby a person assumes the parenting of another person, usually a child, from that person's biological or legal parent or parents. Nevertheless, the "adoption" concept has changed in the course of history.

The modern form of adoption emerged in the United States in the middle 19th century, when the American Civil War resulted in unprecedented overcrowding of orphanages and founding homes. As a consequence, the Orphan Train movement emerged (1859), which eventually shipped an estimated 2,000,000 children from the urban centres of the East to the rural region; but the children were generally indentured rather than adopted (O'Connor 2004).

Nevertheless, different forms of adoption practice have appeared throughout history. The oldest well-documented adoption practice dates to ancient Rome (Lindsay 2009).

Although there is no record of adoption in most ancient chronological periods, it is very important to take into account that it does not mean the adoption phenomena did not exist, and this would have consequences when approaching an archaeological find. For example, in the cases of a collective (simultaneous) burial, it has been assumed that the inhumed persons belonged to the same biological family, but it has been demonstrated that this is not necessarily true. Burials have been found with what seemed to be a complete family nucleus, but where nonetheless the deceased did not maintain family ties (Palomo-Díez et al. 2018).

There is a wide register of adoption cases in the Roman world. To study this, onomastic evidence is useful, and the main features of Roman nomenclature after adoption are explored. A general observation, in this case, points to the practice being most relevant among the elite and the imperial family, and many times adoptions were used to serve political rather than familial ends because adoption was considered as a mode of succession. Many of Rome's emperors were adopted sons. When the family patriarch was about to die without a male heir, an heir could be provided from another family through adoption.

Adoptions have been detected in both eastern and western cultures throughout history, but with different objectives. While the Western idea of adoption is focused on extending family lines; evidence suggests the goal of this practice in oriental cultures was to ensure the continuity of cultural and religious practices.

One of the applications of lineage markers is to track the membership of living descendants of certain famous people with the purpose of their identification (Romanov Family or Columbus, for example). Many times, these searches are performed in comparison with surnames or family names. In these cases, it is necessary to take into account that the existence of an adoption event could alter the analysis, because the surnames lineage may not match biological lineage. Similar issues would occur in the case of children descended from extramarital affairs.

After the decline of the Roman Empire, in the late Middle Ages (1300–1500), the rules began to change, bloodlines were paramount, and the adoption practice was denounced. For example, France's Napoleonic Code made adoption difficult, requiring adopters to be over the age of 50, sterile, older than the adopted person by at least 15 years, and to have fostered the adoptee for at least six years (Brodzinsky and Schechter 1994). English Common Law outright forbids adoption. For this reason, child abandonment rose with the fall of the empire, and many of the foundlings were left on the doorstep of the Church. Then, the oblation concept emerged, whereby children were dedicated to lay life within monastic institutions and reared within a monastery, creating a system through which abandoned children did not have legal, social, or moral disadvantages. As a result, the Church took the role of adopter of these abandoned and orphaned children. As the number of abandoned children grew, the church began to regulate the practice leading to the first official orphanages in Europe.

Before the industrial revolution, it was usual to notice the adoption of children of single mothers, because there was a negative stigma against them, and it was a way to avoid the shame associated with an illegitimate child. Also in this era, some mothers or

families may have placed their child for adoption in such cases as a poor family situation, poverty, or sickness.

Nowadays, transracial, transcultural, and transnational adoptions have increased substantially as have special needs adoption. A typical situation could be the “combined” families, formed by a couple with descendants from past couples. However, this case could be assumed as a special kind of adoption, where one or both of the members of the couple “adopts” the partner’s descendants. In these cases, the cultural concept of family is not concordant with the biological concept of kinship. Regarding same-sex marriage, in the same way as divorce, the simple couple relationship makes no sense analysed from a genetic point of view, unless the couple’s descendants are studied. In this case, we can find cases where only one of the members of the couple presents a biological relationship with the descendants of the family nucleus, or even in the case of same-sex marriage with adopted descendants, without any biological bond with descendants.

From the forensic point of view, many difficulties can be imagined when identifying a person whose biological parents or descendants are unknown. To solve it, many databases have been created to register personal DNA profiles.

6. Surnames Heritage

Y-chromosome testing is useful for several different applications of human genetics, including forensic evidence examination, paternity testing, historical investigations, biogeographical origin, human migrations, and genealogical research.

In many modern societies, the transmission of the father’s surname is linked with the biological inheritance of the male Y chromosome. The possible correlation between a specific surname and a specific haplotype or haplogroup required a series of conditions: (a) the surname must have a unique origin, (b) there should be no illegitimacy, which would introduce chromosomes from other surname groups, and (c) chromosomes associated with different surnames must have been unrelated at the time of surname establishment (Jobling et al. 2004). This link may be disrupted in cases of adoption, a surname change, a maternal surname transmission, a (hidden) baby exchange, and also in the case of independently co-founded surnames or an extra-pair paternity event. For example, the rate of non-biological paternity is very small (1–2%), estimated in different populations (Anderson 2006; Sasse et al. 1994; Greeff and Erasmus 2015; Boattini et al. 2015; Larmuseau et al. 2016). Moreover, genetic drift could affect the probability of survival or multiplication of a particular lineage within a surname over time.

Y-haplogroups had their origin long before the use of surnames in human populations (Jobling and Tyler-Smith 2003; Underhill et al. 2000). The fact that two men do not share their Y-haplogroup necessarily implies that they do not share paternal ancestry. The fast-mutating DNA polymorphisms Y-STRs can be used to attribute a person to a family lineage. On the other hand, it should not be forgotten that the Y chromosome variants began to be distributed throughout the populations before the introduction of surnames. In addition, a very old mutation in time can be blurred, becoming frequent in individuals who do not carry the same surname.

The first study about the heredity of surnames and Y-STR was performed by Bryan Sykes with his surname (Sykes and Irven 2000). In this case, Sykes used 4 Y-STRs. This number of Y-STRs is small and could lead to false family relationships being established.

The correlation of Y-chromosome with surnames could be influenced by surname frequency and/or the specific geographical origin of the surnames. King and Jobling (2009) investigated males with 150 different British surnames. They observed a better correlation in rare surnames. More frequent surnames can have multiple founders. The same results have been obtained with Spanish, Dutch, and Japanese surnames (King et al. 2006; López-Parra et al. 2005; Martínez-Cadenas et al. 2016; Claerhout et al. 2020; Ochiai et al. 2021). By contrast, common Irish surnames displayed very strong Y chromosome co-ancestry levels (McEvoy and Bradley 2006). These differences can be explained by a higher recent population drift than in Britain. For example, the Great Famine in the

mid-19th century reduced the Irish population by a fifth (King and Jobling 2009). This event may have favoured the disappearance of individuals who had different surnames, with men surviving who shared the same surname by chance, that is, an effect of genetic drift. Thus, the Y-chromosome could have suffered from the effect of natural selection due to the surname (Calafell and Lamurseen 2017).

Recently, Claerhout et al. (2020) have analysed 2401 men from Belgium and the Netherlands. They found the highest surname match frequency in the Belgium samples. The lower surname match frequency for the Dutch subset could be because the Dutch heredity surname traditions came into use later than in Belgium. Thus, the age of hereditary surname practices is an important factor for the correlation between surnames and Y-chromosome. The Y-chromosome displays strong geographic differentiation, probably because of a higher female migration rate linked to traditional patrilocal marriages (Wilkins 2006). They observed a strong negative exponential correlation between the geographical distance and the correlation between surname and Y-chromosome.

Regarding the number of markers, Claerhout et al. (2020) observed that the greater the number of markers, including RM Y-STRs, the smaller the probability of establishing false family relationships.

Concerning surname types, Martínez-Cadenas et al. (2016) suggest that does not influence the correlation between Y chromosome and surname. However, Claerhout et al. (2020) observed significant differences.

There are numerous studies about the correlation between surnames and Y-STRs and/or Y-SNPs. For example, Martínez-Gonzalez et al. (2012) carried out a study on the surname Colon. One of the objectives of the work was to determine the origin of the famous discoverer who carried this surname (Columbus). They analysed the samples of men with the surname Colon and surnames similar to Colon from Catalonia, Valencia, the Balearic Islands, southern France, and northern Italy. They obtained haplotype diversity in Italian men who shared the surname Colomba, equal to that of the general population. These researchers raised the possibility that the cause was that this surname was given to orphaned children until 1825 in the orphanage at the Ospedale Maggiore in Milan because a dove figured prominently in the crest of the Ospedale Maggiore. Thus, these men would share the surname but not the paternal lineage. About the origin of Christopher Columbus, with the Y-chromosome haplotype, they obtained four lineages with a cumulative frequency of such lineages of 71%, 87%, and 82% in the Catalan, Valencian, and the Balearic Coloms, respectively, but only 18% and 0% in the Ligurian and Lombard Colombos. Therefore, identification would be much more likely for a Catalan than for a Ligurian Columbus (Martínez-Gonzalez et al. 2012).

Zeng et al. (2019) have inferred the history of the surname Ye based on Y-SNPs. Among samples with the surname Ye, O-F492 showed the highest frequency. Most of the Ye males belong to three major branches, deriving from the node O-FGC66168. Three sub-haplogroups of the O-Z23494 displayed star-like phylogenetic structures. Zeng et al. considered that would reflect population expansions after the Yongjia chaos in AD 311 in the Jin Dynasty. Therefore, the authors associated the frequency of these subhaplogroups with the first of the three massive migrations from north to south in China. In this way, the analysis of markers of Y-chromosome allowed the history of the surname to be inferred and detected signals of population stratification and migrations (Larmuseau et al. 2014).

Theoretically, the correlation between surnames and markers of Y-chromosome could eventually provide a label of regional and family relationships. This could be useful for family searching to identify the surname of a criminal from his DNA left at the crime scene. Claerhout et al. (2020) developed a surname prediction model using Y chromosome markers. This model uses logistic regression to calculate the probability of a positive surname match. This model estimated a surname prediction sensitivity equal to 72%, showing sensitivity in the ability of the model to identify true positives.

In this regard, the case of the killer of Marianne Vaatstra is an example of the use of genetic genealogy). In 30 May 1999, Marianne Vaatstra, a 16 year-old girl, was raped and

murdered in The Netherlands. Traces of semen were found in and on her body. In this case, police thought that the perpetrator probably came from the region of the murder. They collected 6600 samples from local men. When police analysed Y-STR in the first set of 81 volunteers, they found two Y-STRs haplotype matches with the semen trace found in the body of the victim. Autosomal STR profiling excluded both men as likely suspects who had different surnames, but police found that they shared the same paternal ancestor at a time before the Dutch were forced to have their surnames registered. Thus, they decided to select samples from volunteers with these two surnames, and also, they decided to use RM Y-STRs to exclude distant male relatives, decreasing the suspect pool. Surprisingly, one of the volunteers with a Y-STR profile match showed an autosomal STR profile match with the semen trace. It is a clear example of how genetic genealogists could help to resolve a case.

Similarly, the analysis of surnames and Y-chromosome markers enjoys great interest in the general population. Thus, companies have emerged that, in addition to analysing the Y-chromosome markers, have developed projects for each surname, allowing the formation of communities of men who share the surname and are interested in knowing their origins.

7. Familial DNA Genealogy and Criminal Investigation

According to [Mateen et al. \(2020\)](#), familial DNA analysis is “the strategy in which biological family members’ DNA is investigated to provide investigative clues for the identification of the unknown individual”. Instead of searching for direct DNA matches, familial DNA analysis is used to find biological relationships. Instead of looking for direct coincidences, this search focuses on similar matches taking into account the same loci, allowing biological kinships to be identified among the compared individuals. This kind of investigation is used mainly when there are no reference samples to compare with samples obtained from the crime scene ([Bieber et al. 2006](#); [Debus-Sherrill and Field 2018](#); [Fortier et al. 2020](#); [Mateen et al. 2020](#)). Although the comparison with relatives of those possibly implicated in crime scenes has always been a common practice, “familial” DNA genealogy for forensic purposes was officially used for the first time, in the UK, in 2002. However, it was the famous “Golden State Killer case”, in the USA, that brought this kind of investigation into the spotlight, in 2018. As explained below, one of the fundamental allies, in this case, was the search for genetic profiles in a genealogy genetic database, which, until then, was not an official practice in the methodology of security forces.

Thus, the importance of genetic databases is explained, as well as the possible impact of their use on an ethical level, in both privacy and citizen security.

7.1. Genealogical Databases

DNA databases have provided significant investigation leads in solving, for example, cold crimes. Nevertheless, this kind of analysis created a class of problems that have not been solved so far, mainly related to ethical issues and unclear legal aspects of familial DNA analysis ([Mateen et al. 2020](#)). Another problem is the partial profiles obtained from the crime scene that may not match a non-direct relative that could be in the database, for example, a second cousin. When a familial DNA partial match is obtained, it is advisable to analyse lineage markers, such as Y-haplotype and mtDNA.

7.1.1. GEDmatch

The genetic database GEDmatch (<https://www.gedmatch.com/>) was founded in 2010 as a non-scientific website by two genealogists Curtis Rogers and John Olson. GEDmatch operates a series of comparisons with other uploaded profiles and provides additional functionalities. The website permits the user to search for matches with profiles that have been tested on other platforms and by different testing companies. Currently, this platform accepts SNP profiles from over 20 “direct-to-consumer providers” and receives raw data from both microarrays and whole-genome sequencing. It is also possible to upload ancient DNA profiles, whenever it accomplishes some criteria (i.e., number or density of genetic

markers) (Kling et al. 2021). In 2019, the company VEROGEN, a private forensic genomics company (Kling et al. 2021), acquired GEDmatch. In December 2020, a law enforcement portal known as GEDmatch Pro (<https://pro.gedmatch.com/>) was introduced on this platform (Kling et al. 2021). In January 2021, Verogen updated the site policy, allowing unidentified human remains to be compared against the entire database. It is not evident how the division between “offenders and unidentified remains” will be implemented (Kling et al. 2021).

7.1.2. FamilyTreeDNA

Another genealogical database is FamilyTreeDNA (FTDNA), created in 2000, one of the first companies to offer DNA test kits directly to the public. This company provides not only standard autosomal testing but also mtDNA and Y-chromosome analysis. According to the platform, it is not possible to create a family tree (a genealogy); although the information provided can be used by genealogists to search for common ancestors. One of the biggest advantages of this page is their privacy commitment with the clients. Nevertheless, in the FamilyTreeDNA Privacy Statement, the company informs that they may use the data provided “in order to comply with the law and requests from government bodies (. . .) if that information is relevant to a court subpoena or to a law enforcement authority or other government investigation, provided this is permissible under applicable data protection law” (<https://www.familytreedna.com/legal/privacy-statement>, accessed on 29 September 2021).

7.1.3. 23AndMe

The Internet and the 23andMe web page itself display a large amount of information concerning this company. 23andMe is a privately held personal genomics and biotechnology company based in Sunnyvale, California (USA). It made a name for itself in 2007 by offering tests to determine the biogeographical origin of its clients and relatives. The idea was soon copied by other companies, and today, it has become a normal service. In addition, it was a pioneer in carrying out genetic tests from saliva samples collected thanks to a simple commercial “kit” that anyone could use and that was sent to the client’s home. Clients deposited a saliva sample in a container, from which DNA was extracted to genotype SNP polymorphisms.

For this idea (The Retail DNA Test), “Time” magazine named it in 2008 the “invention of the year”, ahead of the electric car “Tesla Roadster” or the Large Hadron Collider (TIME 2008).

A short time later, in 2010, the company had problems with the Food and Drug Administration (FDA), since it seemed that genetic diagnostic tests lacked approval by the FDA, which they achieved in 2015 (Herper 2013; Pollack 2015).

23andMe performs genetic tests that relate the biogeographic origin of a person with the genetic composition of the population of his ancestors. In addition, they relate these factors of biogeographic origin with other genetic factors that can condition health: thus, the reports provide relevant data on “genetic risk” (the existence of genetic variants that increase the predisposition to suffer a disease), on pharmacogenetics (variant genetics that increase or decrease the ability to metabolically process a medication), and on the possible status of an individual as a carrier of a genetic disease (that is, that carries a genetic polymorphism causing or related to a disease in their genome).

The “ancestry” detected by the 23andMe studies allows us to relate our DNA with its possible origin in more than 2000 regions of the planet grouped into six large categories (European, East Asian and Native American, Western Asian and North African, Sub-Saharan African, Central and South Asian, Melanesian, Recent Ancestry in the Americas) and in many cases descending to country level.

To obtain genetic data, 23andMe uses information from autosomal DNA, mitochondrial DNA and Y chromosome, obtained with the Illumina HumanOmni Express-24 chip, now designated as the Infinium Core-24 Kit.

In addition, the company has the CLIA (Clinical Laboratory Improvement Amendments) accreditation and the CAP (College of American Pathologists) certification, thus guaranteeing that its laboratories meet the highest quality standards.

Finally, the 23andMe database allows very versatile searches, such as finding people who share the same DNA in many parts of the world, relating our genotype with that of historical figures or even finding traces of Neanderthal origin in our DNA.

Since the approval of the General Data Protection Regulation (GDPR) in the European Union (EU), on 25 May 2018, 23andMe is subject to this regulation in the EU market, since it applies to companies that come from outside the EU. European regulation allows individuals to control personal information and is mandatory above the national laws of the EU states. Consequently, the company has denied justice access to profiles in its databases on six occasions. However, it reserves the right to disclose *“any and all Personal Information to law enforcement agencies or others if required to do so by law or in the good faith belief that such preservation or disclosure is reasonably necessary”*.

7.1.4. The Golden State Killer Case

On 24 April 2018, a former police officer named Joseph James DeAngelo was charged with eight counts of first-degree murder, based on DNA evidence. The researchers had identified members of DeAngelo’s family in the database of a genetic genealogy company. The indictment connected DeAngelo to various crimes that the California police had been trying to solve since 1973 and that had resulted in different designations (the “Visalia Ransacker”, the East Area Rapist, the Night Stalker, etc.), albeit in 2013 novelist Michelle McNamara coined the term “The Golden State Killer”.

For legal reasons in the State of California, DeAngelo could not be charged with the violations in the 1970s, but he was charged in August 2018 with 13 cases of kidnapping. On 29 January 2020, he was again charged with multiple murders and kidnappings. Subsequently, DeAngelo negotiated with the prosecutor to plead guilty to other homicides and rapes, of which he had not been formally charged, to avoid the death penalty.

On 21 August 2020, DeAngelo was sentenced to life imprisonment without the possibility of parole. Today, it is known that he committed across California at least 13 murders, 50 rapes, and 120 burglaries between 1973 and 1986. In all these cases, DNA played a central role. The identification of DeAngelo had begun around January 2018 when a team of police officers led by Paul Holes and FBI attorney Steve Kramer uploaded the killer’s genetic profile, collected with a simple rape sampling test, to the GEDmatch company database. The website application identified between ten and twenty people who had the same great-great-grandfather as the now-infamous Golden State Killer. A team of five researchers plus genealogist Barbara Rae-Venter used this information to build a large family tree. From this tree, they identified two suspects: one was discarded by a DNA test leaving only DeAngelo as the main suspect. To clear up the question, a DNA sample was surreptitiously taken from DeAngelo’s car door handle in April 2018, and a second sample was later taken from his garbage can. Both samples matched the profile of the Golden State Killer.

The arrest of DeAngelo sparked controversy over the secondary use of personally identifiable information. The issue was summed up by “The Atlantic” journalist Sarah Zhang (Zhang 2019): “Police officers were uploading crime-scene DNA to genealogical databases without any formal oversight, and prominent genealogists disagreed bitterly on how far they should be let in. The debate became so toxic that genealogy groups on Facebook banned any discussion of law enforcement. Decades-old accusations—unrelated to genealogy—were dragged up to discredit vocal members. People were blocked. Friendships ended. At a genealogy conference in June, the different sides ignored each other from opposite ends of the bar”.

The controversy had been unleashed by the GEDmatch company. This company was not strictly speaking a genotyping company but allowed the profiles provided by genotyping companies, such as FamilyTreeDNA or 23andMe, to be uploaded to its database.

From a certain point on, the police began to upload profiles of a robbery with violence to carry out searches. This was against the GEDmatch rules which stated that this could only be performed in homicide and sexual assault cases. The company had done so, Zhang explains, at the request of a detective to one of the GEDmatch co-founders. When the situation was made public, GEDmatch decided that the users themselves would decide, so that out of 1.3 million profiles, only 163,000 made their data available to law enforcement.

Genetic genealogy companies—which use DNA to build family trees—divided on the matter: FamilyTreeDNA or Parabon NanoLabs chose to help the law, while 23andMe and AncestryDNA did not allow the courts to upload profiles. Things became even worse when it emerged that FamilyTreeDNA had been secretly sharing information with the FBI (Haag 2019). The trade-off between the right to information privacy and legal assistance remains controversial.

7.1.5. Privacy and Security Implications

There are a considerable number of ethical concerns with the increasing search for our own familial DNA analysis. The DNA profile of a person who is once registered into the database raises important concerns, since this information may have an impact on the family and relatives of the person (Mateen et al. 2020). As demonstrated by the Golden State Killer case, the family structure and how they manage their relationship can be seriously affected by familial DNA analysis. In the case of the accessible genealogy database, people have agreed to have their DNA searched to find “possible relatives”. In general, people are not well informed about the importance of informed consent. According to Beauchamp and Childress (2001), everyone has the right to place their DNA in a database, with informed consent authorizing the familial search. In the Jurisprudence Section presentation at the American Academy Of Forensic Sciences Annual Meeting in Baltimore, Maryland, on 21 February 2019 (Wickenheiser 2019), Don Shelton explained how a suspect searched on a forensic genealogy database “does not have legal standing to argue on behalf of his relatives. Only the relatives themselves are in a position to dispute the use of their profiles, as they have the autonomy to permit their profiles to be used to uncover a potential criminal”.

As Williams and Johnson (2005), explains in their investigation, familial DNA searching is usually based on partial matches, which may increase the number of individuals for investigation. Modern forensic databases considerably over-represent individuals of particular ethnic groups (for example, in the United States, African Americans and Latinos), and those communities are disproportionately exposed to mistakes related to familial searching (Greely et al. 2006; Wickenheiser 2019). As a result, members of these groups are more often targeted by familial searching (Greely et al. 2006; McGlynn 2019).

In their work, Wickenheiser (2019) propose some measures to improve the security in familial searches, such as “restricting access to specifically authorized individuals; providing sufficient checks and balances for access, and strict use of the data that is accessed; data should only be shared in a very controlled manner to provide investigative leads. There should be penalties for misuse to safeguard data”. Nevertheless, the main problem lies in informed consent, which must explain all implications a match between profiles can lead to.

8. Conclusions

Although the concept of family has varied with history and culture, it is not an institution devoid of rules. In each place and at each moment, there have always been rules for forming a family. Those rules have always been, at least in large part, linked to biological kinship. Although previously biological kinship was linked to written records, whether they were sagas, baptismal certificates, or any other document of a cultural context, the arrival of DNA analysis and the genetic polymorphism existing in all living beings have made it possible to find parallels between documentary records and genetic inheritance. Thus, while nuclear DNA genetic polymorphisms allow first- and second-degree kinship

to be established with sufficiently high probabilities, the emergence of so-called “lineage markers” in the mid-1990s (mitochondrial DNA but especially polymorphisms of the Y chromosome) has come to be a powerful tool for establishing links between individuals, family groups, and lineages.

The tool has been even more useful in countries of Western Christian cultural heritage, in which the lineage was established by the transmission of a surname of patrilineal heritage. As a result, Y chromosome polymorphisms have been able to help clarify the veracity of written records.

Furthermore, genetic tools applied to genealogy have originated other applications of interest, from the construction of databases of genealogical interest to the use of those same databases in forensic investigation or the determination of the ethnic composition of individuals and human groups. The interest aroused by genealogy and the knowledge of one’s origins has, for the first time in history, made genealogy a first-rate business activity, hence the creation of commercial companies that exploit genealogy globally. In addition, the construction of databases by these same companies has exceeded traditional genealogical research based on written documents, allowing the establishment of unknown relationships of individuals with people to whom no document relates them but who share the same genetic markers stored in the database.

However, the possibilities raised are not without problems, arising in the field of data protection, in this case genetic information stored in databases and its use for purposes for which it had not been foreseen, such as police investigation. Undoubtedly, in the future, we will see closer legal regulation of this genetic information throughout the world, which must also be homogeneous, since genetic profiles of individuals from very different countries appear in the databases.

Finally, current DNA techniques make it possible to obtain the profiles of ancient human remains that are tens, hundreds, or thousands of years old. This opens unprecedented possibilities for historical genealogy and will be able to solve historical problems that have been unapproachable until now.

It is clear, therefore, that genotyping tools have produced a true revolution in genealogy that does not replace traditional research tools but rather complements them and opens radically new possibilities.

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