Genetic Genealogy in Context: Italy

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Types of DNA Inheritance

There are four types of DNA which follow unique inheritance patterns: mitochondrial DNA, the Y chromosome, the X chromosome and autosomal DNA. Here we review each of these inheritance patterns.

Inheritance of Mitochondrial DNA

Both males and females inherit mitochondrial DNA from their mother, but only females pass it on to their children. Therefore, it represents an individual's direct-line maternal ancestry and can be used to address questions of shared maternal ancestry. Occasionally, mutations are introduced into mitochondrial DNA before it is passed on to a subsequent generation which can help delineate unique and distinguishable mitochondrial lineages. These lineages are grouped according to their common hierarchical mutations into haplogroups, some of which are geographically or ethnically specific. When two individuals share the same mitochondrial DNA signature it indicates that they share a common direct-line maternal ancestress.

Inheritance of Sex Chromosomes (Y and X)

The Y chromosome is the male sex chromosome. Each male inherits his Y-chromosome from his father in a direct paternal line of ancestry. Therefore, Y-DNA follows the same inheritance pattern as surnames in many western civilizations. This quality makes it particularly useful for answering questions regarding paternity or shared paternal ancestry. As with mitochondrial DNA, occasional mutations distinguish unique Y-chromosome lineages and haplogroups some of which are geographically and ethnically specific. Though both mitochondrial and Y-DNA haplogroups have letter designations, they do not coincide geographically or ethnically. For example, whereas mitochondrial haplogroup A is commonly found in Native American populations, Y-DNA haplogroup A is most commonly found in African populations. When two individuals share a common Y-DNA signature it indicates that they share a common direct-line paternal ancestor.

The X chromosome is the female sex chromosome. Males inherit one X chromosome from their mother and a Y chromosome from their father. Meanwhile, females inherit one X chromosome from their mother and one from the father. Since males only have one X chromosome from their mother which does not recombine at significant levels, the X chromosome they pass on to their daughters is inherited intact from their own mother. Therefore, females in fact inherit one X chromosome from their mother and one from their paternal grandmother. Before a female passes an X-chromosome on to her children, her two X-chromosomes may undergo a process called recombination. Recombination can result in new chromosomes which are composed of portions of the two chromosomes carried by an individual. Therefore, an individual's maternal X chromosome may have come from one or more of his/her maternal ancestors. Alternatively, an individual's maternal X chromosome may not include any DNA from particular individuals who theoretically could have contributed to the makeup of the chromosome. Percentages of inherited DNA cannot be assigned or applied to individual chromosomes. Some

chromosomes will be inherited intact from a grandparent with no recombination between other copies. The same is true of the X chromosome.

When two individuals share DNA on the X chromosome, the possibilities for the origin of their shared DNA are limited to a subset of their respective ancestors. X-DNA inheritance cannot pass through two successive generations of males.

Autosomal Inheritance

Each individual inherits half of their autosomal DNA from each of their parents. Beyond that, the amount of DNA shared in common with specific ancestors is only approximate due to a random process called recombination which shuffles the DNA each generation. Each individual will inherit about 25% from each grandparent; about 12.5% form each great-grandparent; and approximately half the previous amount for each subsequent generation. Eventually there will be some ancestors who do not contribute significantly to an individual's genetic makeup due to the random nature of recombination and autosomal inheritance. Therefore, some documented distant relatives, may not share autosomal DNA segments with each other. This typically begins to occur at the level of second cousins once removed (seven generational steps). Individuals who are more closely related than the level of second cousins once removed should share at least some autosomal DNA with each other.

Although two first cousins will have both inherited 25% of their DNA from each of their common grandparents (50% in total) they will inherit a different 25%. Therefore, first cousins typically only share about 12.5% of their DNA with each other. This shared DNA will consist of large segments of DNA inherited from their common grandparents. When two individuals share a segment of autosomal DNA, then they share at least one common ancestor. The exact nature of the relationship between two genetic cousins will affect the number of segments they share in common, the length of those segments, and the total amount of shared DNA. By analyzing the positions of the segments on the respective chromosomes, and the nature of the population to which two individuals may belong it is sometimes possible to estimate the level of their relationship. Most relationships within the range of parents to second cousins will share unique and identifiable amounts of shared DNA. Eventually these amounts become more variable across relationship levels. A third cousin might share as much DNA with an individual as a fourth cousin, and a fifth cousin might share as much DNA with an individual as an eighth cousin. A test subject and one of their autosomal DNA matches could be related through any of their respective ancestral lines.

Genetic Genealogy in Context: Italy

Though genetic genealogy evidence can certainly help in resolving genealogical research obstacles, at the end of the day genetic genealogy is just genealogy and requires the consideration of historical, geographical, and cultural context. From my personal experience working on genetic genealogy cases in Italy, following are a few hints and tips to consider.

Prevalence and availability of genetic genealogy testing in Italy

I have found that the prevalence of DNA testing among current residents of Italy is quite low (as of November 2022). Rather, the majority of an Italian test taker's genetic cousins are likely to be descended from migrants who left Italy and settled in other areas of the world.

A researcher is most likely to find genetic cousins from Italy who still reside in Italy at the following companies:

- MyHeritage: They launched their DNA testing service in 2019 but they have seen rapid growth throughout Europe, particularly among younger populations.
- 23andMe has been selling kits in Italy for several years, and I have occasionally found residents of Italy who have tested there.

AncestryDNA opened up sales of their DNA kits to Italy in early 2022. As such, most Italian genetic cousins in their database are descendants of immigrants. While Family Tree DNA has sold kits in Italy for several years, it is one of the smallest databases and has few Italian testers in our experience. As such, most Italian testers perform autosomal DNA testing, and few have performed Y-DNA or mitochondrial DNA testing.

Ethnicity admixture estimates

Many DNA test takers perform genetic genealogy testing in order to learn more about their ethnicity percentages. Individuals with recent Italian ancestry are sometimes surprised with their results showing connections to surrounding populations.

- Individuals with ancestry in Northern Italy (including Piedmont, Lombardy, Liguria, Emilia Romagna, Tuscany, Veneto and Trentino-Alto) often have admixture estimates from France, Germany, and Northwestern Europe.
- Individuals with ancestry in Southern Italy (including Campagnia, Apulia, Bacilicata, Calabria, Sardinia, and Sicily) often has admixture from the Mediterranean and North Africa.
- Individuals with ancestry in Southern Italy sometimes have admixture from the Balkans and Greece.

As companies continue to refine their ethnicity estimates, it is likely that these admixture regions will become more refined over time to center more on corresponding regions in Italy.

Y-DNA haplogroups in Italy

While few individuals from Italy have taken Y-DNA or mtDNA tests at Family Tree DNA, 23andMe does report Y-DNA and mtDNA haplogroups and academic studies have also explored the distribution of common haplogroups in the country.

Most Italian men belong to Y-DNA haplogroup R1b (approximately 40%) but there are also large percentages of men belonging to haplogroup J2 (15%) and E1b1b (15%). In general, haploroup R1b is more common in Northern Italy (higher than 40% in most Northern Italian regions) and at lower levels in Southern Italy (lower than 39% in most Southern Italian regions). Meanwhile haplogroups J3 and E1b1b are found at higher levels in Southern Italy and at lower levels in Northern Italy. Other regional peculiarities include the following:[2]

- Haplogroup I1 is found at high percentages in Friuli-Venezia Giulia (22%) Trentino-Alto (21%) and Molise (14%)
- Haplogroup I2a is found at a high percentage in Sardinia (38%)
- Haplogroup R1a is found at a high percentage in Friuli-Venezia Giulia (13%)

[2] "Genetic history of the Italians," *Eupedia*, https://www.eupedia.com/genetics/italian_dna.shtml, accessed November 2022.

- Haplogroup G is found at high percentages in Molise (24%), and at moderate levels (10%-16%) in many other regions including Umbria, Apulia, Sardinia, Lazio, Campania, Calabria, Lombardy, Piedmont, Bacilicata and Liguria.

mtDNA Haplogroups in Italy

Most Italians belong to mtDNA haplogroup H (40%), however, other haplogroups found at moderate levels (more than 10%) in specific regions include:

- Haplogroup T2 in Veneto (19%), Friuli-Venezia-Giulia (12%), Marche (11%), and Sardinia (10%).
- Halogroup J in Calabria (14%), Sardinia (13%), Apulia (10%), and Veneto (10%)
- Haplogroup K in Lombarfy (11%)
- Haplogroup HV in Calabria (10%)
- Haplogroup HV0 and V in Liguria (10%)

Endogamy

Endogamy is the custom of marriage only within a limited population, clan, community or tribe over the course of many generations.[3] The reasons for this practice may be cultural or religious or they could be due to geographic limitations. In endogamous populations, their long-term isolation from other cultural groups and a lack of gene flow eventually leads to the development and accrual of unique genetic markers which distinguish members of the population from other surrounding populations.

Individuals with ancestry from endogamous populations frequently descend from the same ancestor(s) multiple times and they may share multiple ancestors in common with their genetic cousins. In these cases, shared DNA may be due to one of several possible relationships, and the closeness of an estimated relationship may be overstated due to multiple sources contributing to the overall shared DNA between two individuals.

One of the most effective strategies we have observed for overcoming the effects of endogamy is to prioritize, target test, and leverage the test results of individuals who are only distantly descended from the endogamous population of interest as descendants of genetic pioneers. Other strategies include:

- Focusing on the closest genetic cousins with the highest amounts of total shared DNA.
- Applying revised centimorgan thresholds to better prioritize DNA matches.
- Prioritizing DNA matches with the longest segments.
- Testing as many descendants of a research subject as possible.
- Target testing additional individuals to aid in filtering test results.
- Supplementing autosomal DNA test results with Y-DNA, mtDNA and X-DNA tests and analysis.
- Considering amounts of shared DNA between genetic cousins in the construction of In-Common-With (ICW), network and cluster groups.
- Focusing on shared geography and ancestral locations rather than shares surnames or common ancestors.
- Engaging in segment analysis.

Compiled family trees

[3] "Endogamy." *International Society of Genetic Genealogy* (ISOGG) *Wiki* (https://isogg.org/wiki/Endogamy: accessed 13 November 2018).

Common genetic genealogy methodologies rely on quick and speculative extension of family trees of groups or clusters of genetic cousins who are likely related to each other in order to identify patterns and clues of shared ancestral locations, shared ancestral surnames and even shared ancestors. Because many of the matches of Italian individuals are descended from recent migrants from Italy, it is our experience that few genetic cousins have extended their pedigrees beyond the first few generations, and typically not far past their earliest immigrant ancestors. As such, it is often necessary for genetic genealogy researchers to extend the family trees of genetic cousins using original documentary sources including civil registration and Catholic church records. Depending on the locality and the time period, this can sometimes be accomplished through online records, but other times may require contacting or visiting local civil registration offices, parish priests, or possibly contracting with onsite agents to perform research on an individual's behalf.

Foundlings

Genetic genealogy research in Italy sometimes centers around the identification of biological parents of a foundling child. For more information on practices of Italian Infant Abandonment, see:

- "Italian Infant Abandonment," *FamilySearch Research Wiki*, https://www.familysearch.org/en/wiki/Italian_Infant_Abandonment, accessed November 2022.
- Kim Running, "Finding Foundlings: Searching for Abandoned Children in Italy," 14 September 2017, https://www.legacytree.com/blog/finding-foundlings-italy, accessed November 2022.

In such cases, it is important to consider all potential clues offered by the church and civil registration records for the foundling child. We have experienced in several cases that the address of the midwife or individual reporting the birth may give important clues regarding the residence of the mother. We have even had instances where the individual reporting the birth was actually a close relative of the child.

Finding living people in Italy

In genetic genealogy research, it is sometimes necessary to locate contact information for living individuals, either to arrange targeted testing, or to reconstruct a non-responsive genetic cousin's pedigree. Some resources we have found helpful for these efforts in Italy include

- Community Facebook or social media groups.
- Family Status Certificates
    - For more information see: "Italy Census," *FamilySearch Research Wiki*, https://www.familysearch.org/en/wiki/Italy_Census, accessed November 2022.
- PagineBianche (https://www.paginebianche.it/)
- Working through contacts in the village, town, or city where your ancestors lived.
- Working with shared matches of an individual and where possible prioritizing descendants of a family in the United States given that it is often much easier to find contact information in the United States.