# Investigating African ancestry in Puerto Rican individuals via testing of mitochondrial and autosomal DNA to generate a personal phylogenetic family tree

**Honors Thesis** 

March 2015

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Ft. Lauderdale, Florida

## **Undergraduate Honors Thesis**

Of

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#### **PREFACE**

My research was first inspired by the work of Dr. Spencer Wells who documents human ancestry by combining genetic testing with genealogy research to understand better the origins of human populations and their movements across the globe over time. I remember the first time I saw Dr. Wells' film documentary The Journey of Man in my honors seminar, Genetics & Genealogy, and becoming immediately fascinated by the subject of early human migrations out of Africa. I recall thinking to myself that the idea that all humans in existence today arose from one common man and woman was simply unbelievable. The more I thought about it, the more my curiosity for the subject grew. At the end of the course, I received a genetic test on my own DNA sample from the National Genographic Project and was shocked by the results. According to the Genographic Project, my maternal lineage could be traced back to relatively recent and direct African ancestry. For me, these results were completely surprising and they ignited an insatiable curiosity to find out more. I had to learn more about my ancestors - to discover who they were, where they lived, and what they did. No matter how long ago, their simple presence on this earth played an incontrovertible role in my current existence. With a deep sense of combined determination and child-like curiosity, I embarked on the beginning of a lifelong journey to uncover my genetic and ancestral roots - a journey to uncover my past, present, and future.

Some time ago, I stumbled upon this quotation in my research from Marcus Tullius Cicero (106-43 B.C.): "To be ignorant of what occurred before you were born is

to remain perpetually a child. For what is the worth of a human life unless it is woven into the life of our ancestors by the records of history?" I believe that these words perfectly exemplify the journey that I have taken throughout this entire process. I have unearthed the stories of my ancestors and the history of my origins, and now it has become interwoven with my life. My past has melded with my present and, as a result, has undoubtedly shaped my future. This research and the knowledge that I have gained about my ancestral history has become a part of me.

Of course, my research and this thesis project would not have been made possible without the support and encouragement of two very special individuals, Dr. Emily Schmitt and Dr. James Doan. Their brilliance and the depth of their knowledge inspired me and motivated me every day. From the bottom of my heart, I thank you both for everything.

#### ABSTRACT

People have often been interested in their ancestral roots, and the ability to document ancestry via the use of genetic tools has never been greater and more economical. Inspired by the Human Genographic Project, the purpose of this study was to similarly employ the usage of genetic testing (via mitochondrial DNA, Y-chromosomal DNA, and autosomal DNA) as well as genealogical records to uncover details and information on a specific individual's personal ancestry. For this study, the principal investigator delved into the examination of her personal genetic ancestry. Previous research had shown that the investigator's mitochondrial DNA was linked to Haplogroup L1 Subclade L1c1, which is one of the human subpopulations to have more recently migrated from Africa. By focusing on specific molecular markers that represent human ancestral migration patterns, the researcher gained a better understanding of the connections that reveal more direct and recent African ancestry along her maternal lineage. The researcher also performed further investigations on her paternal lineage. The results of her brother's Y-DNA had originally shown that the paternal lineage belonged to Haplogroup R1b, M343, with origins in Western Europe. Additional testing confirmed this haplogroup and added to the classification. DNA test kits were obtained commercially through Family Tree DNA and 23andMe. Completion of this study resulted in the generation of a personal phylogenetic tree with further classification of the haplogroups on both maternal and paternal lineages, as well as a fictional family history narrative using both genetic and genealogical data obtained.

#### **ACKNOWLEDGEMENTS**

To the Honors Thesis Program of the Farquhar College of Arts and Sciences (NSU), as well as the Beta Beta Beta (TriBeta) National Biological Honor Society Research Foundation, thank you for helping to fund my research project and for making this entire journey possible.

To Dr. Emily Schmitt and Dr. James Doan, I extend my greatest thanks for your unwavering support and encouragement throughout this entire process. It was such a great honor to have you both as my faculty advisors. I have learned so much from the two of you over the past few years and you have both helped to shape the student and person that I have become today. Words cannot convey my appreciation and gratitude for you both.

To Dean Don Rosenblum, Dr. Matthew He, and Dr. Marlisa Santos, thank you so much for approving my project, funding my research, and supporting me every step of the way.

To Dr. Diego Castaño, thank you for sharing your genealogy story with me and for all your advice on the best technology for analyzing my results.

To Nora Quinlan and the Genealogy Collection at the NSU Library, thank you for the extensive resources and databases available for me to complete my genealogical research.

To my family, thank you for your undying love and support always. And a special thank you for supplying all the coffee that was consumed throughout the writing of this thesis.

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The Family Finder test examined my autosomal DNA in order to estimate the ancestral populations that make up by background. Note the low margin of error percentages indicating greater accuracy for all ancestral estimations.

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The results from the autosomal DNA test conducted on my maternal grandmother. These results are more indicative of African ancestry within my maternal lineage.

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The percentages of each region that makes up my ethnic background.

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Investigating African ancestry in Puerto Rican individuals via testing of mitochondrial and autosomal DNA to generate a personal phylogenetic family tree

#### I. Introduction

Humans have been in existence for hundreds of thousands of years. All human migration patterns originated in Africa and spread out as groups of people began to inhabit other regions. These migration patterns can be studied by analyzing the DNA (mitochondrial DNA and Y-DNA) of individuals for specific molecular markers that are indicative of particular types of ancestry (Wells, 2006). Mitochondrial DNA (mtDNA), which is passed down to offspring from the mother, can be traced back to three central lineages of ancestry (Pierce, 2012; Rosa and Brehm, 2011). These lineages are known as the L1, L2, and L3 Haplogroups (Wells, 2006). The L1 and L2 lineages gave rise to peoples who stayed in Africa while the L3 lineage is thought to have given rise to the group of people who left Africa to populate the rest of the world (Cavalli-Sforza, Lurquin, and Stone, 2007; Campbell and Tishkoff, 2008; Mitomap, 2013; MitoWheel, 2013).

#### a. Migrating Out of Africa

The "Out-of-Africa" theory is the most commonly acknowledged and established idea explaining the geographic origin of all modern humans. This theory states that all modern humans originated in Africa between 200,000 and 60,000 years ago, and then began migrating to other parts of the world over time (Wells, 2006). The "Out-of-Africa" theory is heavily supported by genetic studies involving mitochondrial DNA and fossil evidence. Upon examining a map of human migration routes out of Africa, it is clear that there

is one common point of origination in central Africa (Figure 1). This common origin signifies the common ancestor for all people.



**Figure 1: Human Migration Routes Out of Africa.** Note the point of origin for all routes of migration just to the east of central Africa. Image taken from *The National Genographic Project*.

If this map of human migrations is further compared to a map of haplogroup (populations with similar genetic markers) migrations, it becomes evident that the two figures are very similar. All the different haplogroups originate from the very same central origin in Africa despite each being characterized by different sets of mutations (Figure 2).

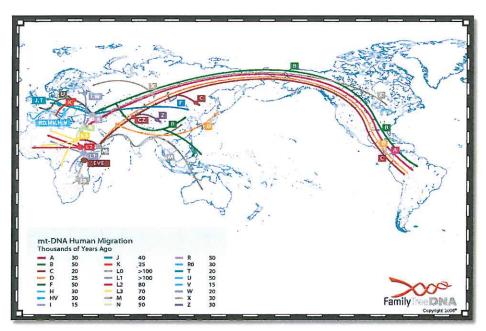
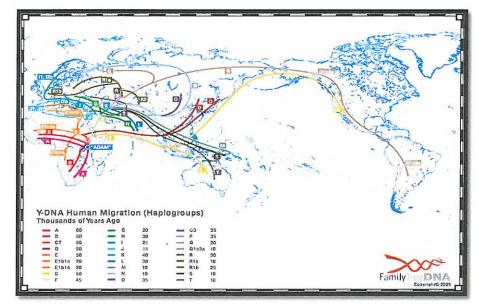


Figure 2: Mitochondrial DNA Haplogroup Migration Map. The migration patterns of each of the mtDNA haplogroups. Note that the origin in central Africa is composed of Haplogroups L1, L2, and L3, and all other haplogroups branch out from these three initial groups. Image taken from *Family Tree DNA*.

Similar migration patterns can be documented using Y-DNA (Figure 3).



**Figure 3: Y-DNA Haplogroup Migration Map.** The migration routes of the Y-DNA haplogroups as they spread out of Africa. Image taken from *Family Tree DNA*.

#### b. Documenting my Maternal Lineage (my mitochondrial DNA)

Scientists have concluded that members of Haplogroup L1 are the direct descendants of "Mitochondrial Eve" from East Africa. Individuals of Haplogroup L1 possess markers that can be traced back approximately 150,000 to 170,000 years into the past (Wells, 2006). The name "Mitochondrial Eve" does not represent the first female human, but rather the oldest female human that has descendants living today. Because of any number of reasons all other human lineages went extinct and her genetic information has been able to survive until today.

As the years progressed, the descendants of "Mitochondrial Eve" moved throughout much of sub-Saharan Africa and later divided into two major haplogroups, L1 and L0. Individuals of the L1 and L0 haplogroups are characterized by the special set of mutations that they possess. It has been said that these individuals "represent the deepest branches of the mitochondrial tree" (National Geographic, 2013). Today, the descendants of Haplogroup L0 primarily inhabit parts of Africa. Preliminary research of my maternal lineage (mtDNA at a few key markers) was conducted via genetic testing as part of The Human Genographic Project in January 2012 (National Geographic, 2013). These tests revealed the unique haplogroups that characterize both my maternal and paternal lineages. Genetic testing of my mtDNA through a sample of my own buccal cheek cells revealed that my maternal ancestral lineage belonged to Haplogroup L1. When I began my genetic research, I originally hypothesized that my DNA samples would be linked to either

Haplogroup L3, which gave rise to every currently non-African haplogroup in the world, or another haplogroup with greater ties to Hispanic origins (Wells, 2006).

Recent evidence has shown that some individuals of Haplogroup L1 made the Nile River Valley their home. A small subsection of this haplogroup broke off and began to populate the rest of the world. One of the West African tribes that is also a part of Haplogroup L1 is the San tribe (*National Geographic*, 2013). A physical comparison of my grandmother to a photo of a San woman reveals striking similarities in skin color, cheekbone structure, rounded face and nose, and small eyes (Figure 4).

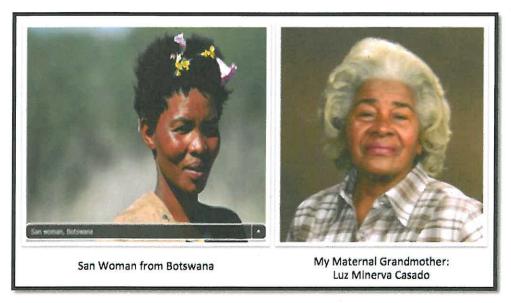


Figure 4: Comparing my Grandmother to a San Woman. Visual physical similarities can be seen between my grandmother and this San woman. Image on the left was taken from *The National Genographic Project*, and image on the right was taken from the collection of Rodriguez family photos.

By further examining our mtDNA and genetic similarities to African peoples such as those of the San tribes, I hope to provide better resolution to our maternal family ancestry within the last 300-1,000 years.

Research focusing on Amerindian ancestry in the Puerto Rican population has shown that, although the native Taíno culture is no longer extant within the population, the influence of the culture is still very much evident (Martinez-Cruzado, 2001). The physical traits of people from the Taíno culture are still existent within the population. By analyzing the haplogroup frequencies of the population, it is possible to draw linkages between the Taíno culture and their South American ancestors. This has been useful for better understanding the genetic influences of other ancestries on the Puerto Rican population. 91.5% of the mitochondrial DNA within the Puerto Rican population belongs to Haplogroups A and C, which are Amerindian haplogroups (Martinez-Cruzado, 2001). Genetic testing places my lineage in Haplogroup L, which is interesting in that it differs from typical Amerindian ancestry.

By delving further into the maternal aspect of my genetic history, molecular markers were investigated to confirm my linkage to the L1C1 haplogroup and further specify the subdivision to which my ancestry belongs. As a result of this study, I was able to trace my African ancestry further down the branches of the phylogenetic tree. Additionally, I continued assembling my family tree from information gained through genealogical records and family interviews.

#### c. Uncovering African roots in Puerto Rico

Since the arrival of the European explorer Christopher Columbus in 1493, Puerto Rico has undergone hundreds of years of Spanish rule followed by political, economic, and social influence from the United States. As a result, both the history and culture of Puerto Rico reflect a number of different international and indigenous roots. The people of the island are a physical representation of the diversity of cultures present among the population – most notably, Taíno (Amerindian), Spanish, African, American, and Latino/Caribbean.

#### i. Pre-Colonial Puerto Rico

The island of Puerto Rico was first settled nearly 4,000 years ago by the Ortoiroid culture from the Orinoco region in South America (Rouse, 1992). One of the subcultures of the Ortoiroid people is the Coroso culture. Rouse holds that the Coroso subculture of the Ortoiroid were the first settlers of the island. The earliest dated site in Puerto Rico is the Angostura site of the Coroso culture. Further archaeological research, however, suggests the influence of other early cultures like the Arawakan-speaking Taíno people.

The Taíno were one of the major native peoples of the Caribbean region. They were the dominating inhabitants of the island of Puerto Rico prior to Spanish rule. Archaeologists estimate that the island had approximately 30,000 Taíno inhabitants when Columbus arrived in the island in 1493 (Schimmer, 2010).

The Taíno had named the island Boriquen meaning "the land of the brave lord." They divided the island into several chiefdoms each governed by a cacique (chieftain) (Figure 5).



**Figure 5: Map of Pre-Colonial Boriquén.** At the time of European discovery, the island of Puerto Rico was divided into approximately 20 different villages each governed by a cacique. Image taken from the Genocide Studies Program at Yale University, 2015.

Historians approximate that the biggest Taíno tribes on the island were comprised of over 3,000 individuals each.

#### ii. Significance of Spanish Rule

When Columbus first arrived in Puerto Rico, the island was dominated by the native Taíno culture. Upon arrival, he renamed the island San Juan Bautista after Saint John the Baptist. The Spanish explorer Juan Ponce de Leon came to the island in the early 1500s and was openly

welcomed by the Taíno cacique Agüeybaná. Ponce de Leon governed the first settlement in 1508 (Schimmer, 2010).

The settlers soon began to conquer and enslave the native Taíno within a year of settlement. The indigenous Taíno people became decimated as a result of exposure to infectious diseases brought by the Spanish. The first smallpox outbreak recorded in the Hispaniola region took place around December of 1518 and January of 1519, killing off roughly 90% of the native population (Crosby, 2003). A large number of Taíno deaths can also be attributed to warfare and harsh bondage systems in place under Spanish rule. Some historians even view the treatment of the Taíno people as an example of early colonial genocide citing that the Taíno lived under oppressive conditions and strict enslavement that contributed significantly to their demise (Schimmer, 2010).

Despite the oppressive Spanish treatment of the natives, many Spaniards freely intermarried with the women of the Taíno tribes (Martinez, 2007). This intermarriage between the Spaniards and Taíno would help to ensure the continuation of the Taíno ancestral lines to some extent throughout history. Today, there is still a great genetic influence of the Taíno ancestry among the Puerto Rican population. According to Veran (2003), the Taíno genome project initiated in 1999 examining mitochondrial DNA found that 62% of Puerto Ricans have direct maternal ancestry consistent with Amerindian Taíno ancestors.

#### iii. The Atlantic Slave Trade

As a result of the diminishing Taíno population and efforts put forth by Friar Bartolome de las Casas to protect Indian rights, the Spanish government decided to import enslaved Africans beginning in 1513 (Figure 6).



Figure 6: Transport of African Slaves. Slaves acquired through the Atlantic Slave Trade were the first Africans on the island of Puerto Rico. The 19th century drawing shows the transport of African slaves. Image taken from *Lehrbuch der Weltgeschichte oder Die Geschichte der Menschheit*, by William Rednbacher, 1890.

The Spanish royal government relied heavily on these slaves for mining and the building of forts (e.g., the construction of El Morro in the capital of San Juan, one of the most famous forts on the island of Puerto Rico). In 1517, Spanish subjects were allowed by the Spanish crown to import twelve African slaves each (Oregon State, 2003).

Historian Luis M. Diaz argues that the greatest number of slaves came from the present-day Gold Coast, Nigeria, Dahomey, and the Guineas (1985). Many of these slaves were from the Yoruba, Igbo, and Bantu tribes of the present-day regions of Nigeria and Guinea. It is also estimated that slaves were brought in from the Congo and Senegal. Within 25 years, the number of slaves in Puerto Rico increased from 1,500 in 1530 to over 15,000 in 1555 (Martinez, 2007). Upon entry to Puerto Rico, each slave was branded on the forehead with a hot iron stamp, and sent to work in either the gold mines or the ginger and sugar industries. The establishment of sugar plantations on the island further heightened and strengthened the demand for slave labor importation from Africa. Slaves were not permitted to earn or buy their own freedom until the Spanish Decree of 1789. Slavery was not abolished until nearly a hundred years later on March 22, 1873.

#### iv. Influence in Puerto Rican Culture

Unlike the Taíno population that was overtaken by the Spaniards, the African slave population survived through adaptation. Although they still faced great discrimination, African slaves learned the ways of the Spaniards and did their best to assimilate into the Spanish culture, in many cases adding their own cultural influences. African cultural influences played a pivotal role in shaping the culture and society present today in Puerto Rico.

The descendants of former African slaves contributed greatly to the island's language, art and literature, music and entertainment, sports, and scientific institutions. As assimilation in the Spanish culture occurred, African slaves added words from their own native language like cocolo (a person who likes to dance salsa), dengue (mosquito or virus), guineo (banana), and malanga (vegetable) to the Spanish spoken on the island (Lipski, 2005). Similarly, African slaves influenced music and entertainment through the introduction of such instruments like the barriles and African-rooted dance forms as the Bomba and Plena (Bailyn, 2014). The exact origins of the bomba dance form are unknown but it is believed that it is derived from West Africa. The plena dance, on the other hand, is thought to have been brought over to Puerto Rico from blacks that emigrated from other Caribbean islands. Puerto Rican cuisine also has a very strong influence from African culture. African women first created one of the most traditional holiday dishes, pasteles (small bundles of meat wrapped in plantain leaves).

#### v. Current Puerto Rican Demographics

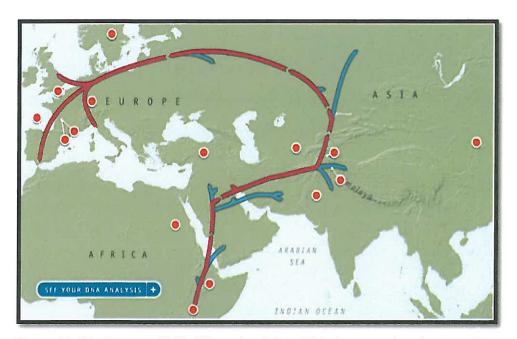
In 1802, the racial distribution of the Puerto Rican population was predominantly non-white (58%) with a remaining 42% characterized as white (Vincent, 2011). Classifications of white versus non-white were made based solely on visual observations of skin color (blacks and mulattos were considered non-whites) (Loveman & Muniz, 2006). Over time, the population grew significantly with the heightened importation of

African slaves onto the island for labor. Within two hundred years, the composition of the population changed drastically. In the 2010 census, it was found that 75% of the population was classified as white while 24.2% of the population was non-white.

A recent study examining mitochondrial DNA for maternal ancestry found that approximately 61.1% of those tested had Amerindian ancestry (Martinez-Cruzado, 2002). The study also concluded that 26.4% of individuals tested had African ancestry while another 12.5% had European ancestry. These findings further support the historical record that male Spaniards intermarried with women from the native Taíno and black slave populations.

#### d. Documenting my Paternal Lineage (my brother's Y-DNA)

My younger brother's buccal cell sample was sent in to the National Genographic Project for analysis. The results of my brother's DNA sample revealed that my paternal lineage belonged to Haplogroup R1b, M343, and, more specifically, to Subclade R1b1a2, M269 (Figure 7). The molecular markers that characterize this haplogroup are representative of the very first common ancestor of all non-African men. High numbers of individuals of Haplogroup R1b reside largely in the countries of Spain and Ireland, as well as the southern portion of England. The notion that my paternal lineage belongs to Haplogroup R1b coincided perfectly with my hypothesis that my father's ancestors possessed Spanish origins.



**Figure 7: Haplogroup R1b Migration Map.** This human migration map is based on Y-DNA and dates back roughly 60,000 years. Image taken from *The National Genographic Project*.

Researchers have found that the earliest ancestor of Haplogroup R1b originated roughly 50,000 years ago in Africa. The descendants of this ancient ancestor, who has been dubbed M168 after his unique molecular marker, formed the only lineage to survive outside of Africa (*National Geographic*, 2013). He is considered to be the shared common ancestor of all non-African men. Significant climatic environmental changes have been attributed as the cause of my ancestor's migration out of Africa. Neurobiologist William Calvin believes that "the climatological downturn after 50,000 years" in which conditions in the Saharan desert turned so dry that the land was not fit for living is the reason for the migration out of Northern Africa (Wells, 2006). The route of exodus then followed the path of movement through North Africa to Central Asia to Europe. Each of these new settlements denotes a new

fundamental lineage on the family tree of our human race. Each new lineage is connected to a distinct marker characteristic of a specific haplogroup. The last marker that scientists have traced in Haplogroup R1b is the marker M343. My brother's Y-DNA displays this very marker. Thus, all the members along my paternal lineage can be categorized under this specific subsection of Haplogroup R1b. Individuals that possess M343 are considered to be the direct descendants of Cro-Magnon (*National Geographic*, 2013). Originating in Western Europe approximately 30,000 years ago, Cro-Magnon dominated the human expansion and gave rise to all non-African men.

The results that my paternal lineage belongs to Haplogroup R1b and possesses Spanish origins are supported by the idea that before the 19th century, a great number of European men inhabited Puerto Rico and fathered the children of non-European women (Martinez-Cruzado, 2001). Because of this, it is appropriate to expect a higher percentage of European origins and ancestry especially among the population using male genetic markers.

I examined this part of my lineage further using new tests that include more specific ancestral markers to trace paternal lineage. Although my paternal lineage yielded the expected results, I wanted to delve further and see if I could uncover more information about my paternal ancestors. I wanted to create a very detailed understanding of both sides of my ancestry. I also researched genealogy databases to search for ancestral historical documents.

#### II. Methods

To obtain a more accurate understanding of my ancestral lineages, I employed various genealogical databases and commercial genetic testing. Both my maternal and paternal lineages were researched thoroughly through ancestral documents and also genetically tested. After familial DNA samples were tested, I analyzed them using different programs available through the genealogical databases. In order to better understand the historical movements tying Puerto Rican individuals to African ancestry, I researched the historic literature available. The tests that I conducted and the ensuing data analysis extended my understanding of my relationship and that of my ancestors within various human sub-populations.

#### a. Examining Historical Events

A complete understanding of the historical ties between Puerto Rican individuals and African ancestry was absolutely imperative for this research study. Because of this, historic literature was surveyed on prominent Puerto Rican historical events and human migrations. This research further added to my understanding of African roots along my maternal lineage.

I conducted this research through careful analysis of various primary sources related to Puerto Rican history and culture (original documents, creative works, photos of artifacts, etc). I also used available secondary sources such as publications, articles, and textbooks.

#### b. Researching Genealogical Databases

utilized numerous genealogical databases including www.MyHeritage.com, www.Ancestry.com, and www.familysearch.org to investigate historical and family records. These databases house countless archives, historical records, and family trees. The majority of my genealogical research came from Ancestry.com because of the extensive anthology of U.S. census, birth, and death records that is constantly being updated. Additionally, Ancestry.com is especially useful for studying Puerto Rican genealogy because it holds the largest online collection of Puerto Rican records spanning nearly 170 years. Because U.S. birth and census records in Puerto Rico have only been documented starting in the late 1880s, I also searched for other databases where such necessary information for the Puerto Rican population was available and reexamined the databases that I had already searched (as additional data are constantly being found, digitized, and added). Detailed records and copies of each document pertaining to a family member were kept in both electronic and print copies, and archived electronically.

I conducted extensive interviews with available family members, where I took notes on relatives based on stories and family memories. However, because of the elderly age of many relatives and their diminishing memories, the accuracy of certain interviews was sometimes uncertain, and that uncertainty was noted in the records.

#### c. Commercial Genetic Testing

Prior to embarking on this study, my understanding of my ancestral background was limited to the idea that the vast majority of my family originated in Puerto Rico. However, it is becoming established that the Puerto Rican population is largely the result of a great variety of more recent and distant human ancestral lines (Baran et al., 2012; Price et al., 2009; Via et al., 2011; Tang et al., 2007; Galanter et al., 2012). In the nearly two years since my original mtDNA testing, the ability to have the entire mtDNA sequence has become much more economical. As a result, I conducted many different genetic tests on familial DNA samples from cheek (buccal) cells. Buccal cheek cell samples were obtained by brushing the inside of the cheek followed by a mouth rinse. These samples were then sent to commercially available testing services for further in-depth analysis.

Table 1: Genetic Tests Conducted. A total of five commercially available genetic tests were performed from three different testing centers: Genographic Project, Family Tree DNA, and 23andMe. Three of the tests focused on examining mitochondrial DNA to study my maternal lineage. One of the tests analyzed Y-DNA in order to look at my paternal lineage. The final other test examined the DNA of both my maternal and paternal lineages by using autosomal DNA to narrow down regions of ancestral origin.

Test #	Name of Test	Type of DNA Tested	Person(s) Tested	Testing Center	Date of Test
1	1 19-Marker mtDNA Student		Student	Genographic Project	January 2012
2 12-Marker Y DNA Test		Y-DNA	Brother	Genographic Project	January 2012
3	Full mtDNA Sequencing Test	mtDNA	Student	Family Tree DNA	October 2013
4*	4* Family Finder Autosomal Grandmo		Student Grandmother Brother	Family Tree DNA	October 2013
5	Basic DNA Test Kit	Autosomal DNA and mtDNA	Student	23andMe	February 2014

In chronological order of when the tests were conducted, the first two were the 19-marker mtDNA test on my own mtDNA and the 12-marker Y-DNA test on my brother's DNA sample through the Genographic Project. Afterwards, three more tests were run through Family Tree DNA (FTDNA), each on a different DNA sample. The first of these FTDNA tests was a full mitochondrial-sequencing test on my personal DNA sample. Family Finder tests were also run on three individuals (my maternal grandmother, my brother, and myself). I only tested for mitochondrial DNA using my own DNA sample, rather than test both my grandmother and myself. I did not test

my maternal grandmother's mtDNA because it should be the same as mine since it is maternally inherited (Campbell et al., 2011).

The FTDNA Family Finder tests that I used examined the autosomal DNA to identify the geographical region of the world where my ancestors most likely originated from and to find some of my relatives within 5 generations (based on comparisons to the DNA of other users in their database). This test was run on my grandmother, my brother, and me.

The final type of test that was performed was a basic DNA test kit on my own autosomal DNA and mtDNA from the company, 23andMe. This test was especially helpful because of its ability to generate a detailed ancestral composition, determine the percentage of Neanderthal ancestry, and identify top possible countries of ancestry.

Genetic testing of these samples in my own university laboratory was not proposed because it has become much more economical and efficient to pay for this type of testing in a commercial lab setting where the entire mitochondrial genome can be sequenced at high quality for approximately \$200 and results automatically become part of a worldwide database to aid in the analysis of ancestry.

I was particularly interested in a few genetic markers linked to more recent African ancestry that were prominent in the literature as being ancestrally informative (van Oven and Kayser, 2009; and Mitowheel, 2011). After carefully surveying the literature, I selected 23 markers found to be indicative of African ancestry (Table 2). Upon receiving the results of my full

mitochondrial sequence, I looked for the presence/absence of these specific mitochondrial markers. These markers were chosen since they are beneficial as indicators of either the presence or absence of more recent African ancestry.

Table 2: Genetic markers linked to L1 Haplogroup proposed for in-depth analysis. A list of molecular markers compiled from various sources. All 23 markers are associated with African ancestry or Haplogroup L1 specifically.

Name of genetic marker	Background Information on the Marker
	Protein ND1 (gene MT-ND1)
G3666A	Subunit of NADH dehydrogenase (complex 1)
G3000A	Amino acid G120 (glycine)
	Third nucleotide in codon GGG
	Protein CO1 (gene MT-CO1)
A7055G	Subunit of cytochrome oxidase (complex IV)
A7055G	Amino acid G384 (glycine)
	Third nucleotide in codon GGA
	Protein CO1 (gene MT-CO1)
T7389C	Subunit of cytochrome oxidase (complex IV)
173690	Amino acid Y496 (tyrosine)
	First nucleotide in codon TAT
	Protein NDS (gene MT-ND5)
T13789C	Subunit of NADH dehydrogenase (complex I)
1137870	Amino acid Y485 (tyrosine)
	First nucleotide in codon TAC
	• Protein ND6 (gene MT-ND6, reverse)
T14178C	Subunit of NADH dehydrogenase (complex I)
1141760	Amino acid I166 (isoleucine)
	First nucleotide in codon ATT
	Protein ND6 (gene MT-ND6, reverse)
G14560A	• Subunit of NADH dehydrogenase (complex I)
G14300A	Amino acid V38 (valine)
-	Third nucleotide in codon GTC
	Protein ND1 (gene MT-ND1)
A3796t	Subunit of NADH dehydrogenase (complex I)
A37701	Amino acid T164 (threonine)
	First nucleotide in codon ACC

Part William	
	<ul> <li>Protein ND1 (gene MT-ND1)</li> <li>Subunit of NADH dehydrogenase (complex I)</li> </ul>
A3843G	Amino acid W179 (tryptophan)
	Third nucleotide in codon TGA
	Protein ND5 (gene MT-ND5)
A14148G	• Subunit of NADH dehydrogenase (complex I)
717 17 100	Third nucleotide in the stop codon TAA
	• Located in the hypervariable segment 1 (locus MT-HV1,
A16293G	16024-16383)
	Protein ND4 (gene MT-ND4)
	• Subunit of NADH dehydrogenase (complex I)
T11899C	Amino acid S380 (serine)
	Third nucleotide in codon TCT
	Located in the hypervariable segment 2 (locus MT-HV2, 57-
T198c1	372)
W-12-12-12-12-12-12-12-12-12-12-12-12-12-	• Located in the mtTF1 binding site X (locus MT-TFX, 233-260)
A249d	• Located in the hypervariable segment 2 (locus MT-HV2, 57-
712470	372)
	Protein CO1 (gene MT-CO1)
	Subunit of cytochrome oxidase (complex IV)
G6267A	Amino acid A122 (alanine)
	First nucleotide in codon GCA
3,00	Protein ATP8 (gene MT-ATP8)
	• Subunit of ATP synthase (complex V.)
G8387A	Amino acid V8 (valine)
	First nucleotide in codon GTA
112-121	Protein ATP8 (gene MT-ATP8)
	• Subunit of ATP synthase (complex V)
G8389G	Amino acidV8 (valine)
	Third nucleotide in codon GTA
	Protein COIII (gene MT-CO3)
	• Subunit of cytochrome c oxidase (complex IV)
T9233C	Amino acid H9 (histidine)
	Third nucleotide in codon CAT
2000 - 11 2000 - 1000 -	Protein ND4 (gene MT-ND4)
C11335T	basame of the bit doily drogonass (complex t)
	<ul> <li>Amino acid N192 (asparagine)</li> <li>Third nucleotide in codon AAC</li> </ul>
	Protein ND5 (gene MT-ND5)     Subunit of NADII debutes company (complex I)
T12879C	• Subunit of NADH dehydrogenase (complex I)
	Amino acid G181 (glycine)  Third avalantida in and are CCT.
	Third nucleotide in codon GGT

T16172C	• Located in the termination associated sequence (locus MT-TAS, 16157-16172)
1101/2C	• Located in the hypervariable segment 1 (locus MT-HV1, 16024-16383)
T16187C	<ul> <li>Located in the hypervariable segment 1 (locus MT-HV1, 16024-16383)</li> </ul>
T16294C	• Located in the hypervariable segment 1 (locus MT-HV1, 16024-16383)

#### d. Data Analysis Methods

Once the results from the various genetic tests were obtained, I compared these data to the large worldwide database of mitochondrial and autosomal genealogical marker results of all other participants from FamilyTreeDNA. I did the same with the Y-DNA results from my brother's test for my paternal lineage.

I analyzed the data received from the genetic tests using different programs available through their respective databases. The company 23 and Me had four ancestry tools that were especially advantageous for analysis of DNA (23 and Me, 2015). These four tools allowed me to analyze DNA for possible countries of ancestral origin, Neanderthal ancestry, global similarity comparisons, and haplogroup tree mutations. Similarly, the FTDNA site also allowed me to analyze the resulting genetic data through the Population Finder and My Origins programs. I examined the autosomal DNA in order to identify possible regions of ancestral origin along with margins of error for each approximation based on comparing the tested DNA to all samples in the FTDNA database.

#### III. Results

Each test allowed me to gain a more in-depth understanding of my genetic ancestry and origins for both my maternal and paternal lineages.

#### a. Maternal Lineage

I found the following genealogical and genetic results for my direct maternal ancestry.

#### i. Historical Documents

Historical records such as marriage, census, birth and death records, and other genealogical documents were acquired predominantly through searches done in genealogical databases such as Ancestry.com. Some historical records pertaining to my recent ancestry were obtained from interviews conducted with family members. I found 11 historical documents ranging from items such as census records to diplomas for my maternal lineage (Appendix II, a-m).

#### ii. Genetic Results

Commercial testing by the National Genographic Project in January 2012 revealed that my maternal ancestors belonged to Haplogroup L1, Subclade L1c1. The L1c group is a subclade of the L1 haplogroup. L1c1 is s further subgroup of the L1c subclade. The results from this initial National Genographic test allowed me to classify my maternal haplogroup by two subgroupings.

Two important tests were then conducted on autosomal and mitochondrial DNA samples from both my grandmother and me through

Family Tree DNA. The first test was an autosomal Family Finder test plus a full mtDNA sequencing test for myself. The results of this test included genetic matches and maternal ancestral origins, as well as a haplogroup migration map and frequency map. My haplogroup results from Family Tree DNA further confirmed Haplogroup L1, but were also able to take it a step further and isolate the marker that was associated with my particular subclade (T198C) (Appendix I-c).

The tests from Family Tree DNA were also able to give me a glimpse into the populations that make up my ancestral roots. According to the test, my ancestry was predominantly 49.95% European, but also comprised of 11.48% Native American, 20.30% African (West African), and 18.28% Middle Eastern (Table 3).

Table 3: Population Finder Ancestral Composition Results for Student from Family Tree DNA. The Family Finder test examined my autosomal DNA in order to estimate the ancestral populations that make up by background. Note the low margin of error percentages indicating greater accuracy for all ancestral estimations.

Continent (Subcontinent)	Population	Percentage	Margin of Error
Europe	Finnish, Russian, Spanish	49.95%	±7.28%
Africa (West African)	Mandenka, Yoruba	20.30%	±3.28%
Middle East	Mozabite	18.28%	±12.87%
Native American	Surui, Maya, Columbian	11.48%	±2.42%

The Mandenka and Yoruba tribes are the most likely origins for my maternal lineage. For my paternal lineage, it is most likely that my ancestors have Finnish, Russian, or Spanish roots.

The results of my grandmother's Population Finder test are more supportive of the notion that our maternal lineage belongs to Haplogroup L1. Her ancestry is comprised of 49.26% African (West African), 11.07% Native American (Central American), and 37.84% European (Table 4). The margin of error is very minimal (ranging from 0.9-12.9%) which further supports our haplogroup classification.

Table 4: Population Finder Ancestral Composition Results for Maternal Grandmother from Family Tree DNA. The results from the autosomal DNA test conducted on my maternal grandmother. These results are more indicative of African ancestry (specifically, the Mandenka tribe) within my maternal lineage.

Continent (Subcontinent)	Population	Percentage	Margin of Error
Africa (West African)	Mandenka	49.26%	±0.91%
Europe	Romanian, Spanish	37.84%	±2.38%
Central American	Maya, Pima	11.07%	±1.32%

One of the most recent updates, My Origins, in Family Tree DNA allows individuals to approximate exact regions of ancestral origins using autosomal DNA. My African Ancestry makes up about 23% of my ancestry (Table 6). It is most probable that my ancestors came from the Niger-Congo Genesis region (21% probability) or were part of the East African Pastoralists (2% probability).

Table 5: My Origins Test Results for Student from Family Tree DNA. The percentages of each region that makes up my ethnic background.

Continent (Subcontinent)	Region	Percentage
Г	Southern Europe	43%
Europe	British Isles	15%
Africa	West Africa	21%
	East Central Africa	2%
New World	Native American	10%
Middle East	North Africa	10%
East Asia	Northeast Africa	4%

When I ran the My Origins test on my grandmother's DNA sample, the probability of African origins doubled in all the subcategories. Her ethnic makeup has the strongest possibility of being from the Niger-Congo Genesis region (43% probability) (Table 6). It is also possible that our ancestors are from the East African Pastoralists (4%) or the Kalahari Basin (2%).

Table 6: My Origins Test Results for my Maternal Grandmother from Family Tree DNA. The percentages of each region that makes up my grandmother's ethnic background.

Continent (Subcontinent)	Region	Percentage
Africa	West Africa	43%
	East Central Africa	4%
	South-Central Africa	2%
Europe	Southern Europe	35%
	Finland and Northern Siberia	3%
New World	Native America	9%
East Asia	Northeast Africa	2%
Middle East	North Africa	2%

The results for both my grandmother and me suggest that our ancestors were part of the Mandenka or Yoruba tribe in West Africa

(Table 3 and Table 4). According to Historian Luis M. Diaz, the Yoruba tribe was one of the groups imported as slaves from Africa through the Atlantic Slave Trade (1985).

The 23andMe autosomal DNA and mtDNA test sequenced all my chromosomal and mitochondrial DNA and allowed me to examine ancestral composition, maternal lineage, and even percentage of Neanderthal ancestry. My results were in concordance with the results from Family Tree DNA showing a considerable portion of my African ancestry from Sub-Saharan Africa (20.8%), and, specifically, West Africa (14.4%) (Table 7). 5.8% of my autosomal DNA was associated nonspecifically with Sub-Saharan Africa and 0.6% was linked to Central and South Africa.

Because these data are generated from all autosomal DNA, the results reflect both sides of an individual's familial ancestry. My results indicate 50.4% connection to European ancestry, primary Southern European (14.7% nonspecific Southern European and 10.0% Iberian region).

**Table 7: Ancestral Composition for Student based on Autosomal DNA from 23andMe.** Data acquired from analysis of my own autosomal DNA. The test conducted through 23andMe analyzed my DNA for the percentage of ancestral composition for each of the 31 tested locations around the world.

General Region	Specific Region	Location	Percentage of Ancestral Composition (%)
European (57.3%)	Southern European	Iberian	29.1
		Sardinian	0
		Italian	0
		Balkan	0
		Broadly Southern European	18.8
	Northern European	British & Irish	0.1
		Scandinavian	0
		Finnish	0
		French & German	0
		Broadly Northern European	3.9
	Eastern European	-	0
	Ashkenazi	Ashkenazi	0
		Broadly European	5.3
Sub-Saharan	West African	-	19.7
African	Central & South African	):=	1.5
(22.8%)	East African	.=	0
		Broadly Sub-Saharan African	1.6
East African	Native American	-	10
& Native	East Asian	Japanese	0
American		Korean	0
(12.3%)		Yakut	0
		Mongolian	0
		Chinese	0
		Broadly East Asian	0.1
	Southeast Asian	-	<0.1
		Broadly East Asian & Native American	2.2
Middle	North African	-	3.2
Eastern &	Middle Eastern	-	0
North African	The second of the second secon	Broadly Middle	0.4
(3.6%)	g.	Eastern & North African	14000000
South Asian	_	-	<0.1
Oceanian			0
Unassigned	-	-	4

#### b. Paternal Lineage

By examining my brother's Y-DNA and autosomal DNA, I found the following genealogical and genetic results for my direct paternal ancestry.

#### i. Historical Documents

Through my research of historical documents, I uncovered 6 documents ranging from vocational certificates to marriage records (Appendix II, n-r).

#### ii. Genetic Results

Two tests were conducted on samples of my brother's Y-DNA in order to uncover the ancestral roots of my paternal lineage. Genetic tests were acquired through the National Genographic Project and Family Tree DNA.

Preliminary testing results through the National Genographic Project revealed that a sample of my brother's Y-DNA was linked to Haplogroup R1b (by the marker M343). The genetic markers that define ancestral history consistent with Haplogroup R1b reach back roughly 60,000 years to the first common marker of all non-African men, M168, and follow my lineage to the present day ending with M343, the defining marker of Haplogroup R1b. Many of the men in this haplogroup are commonly located in England, Spain, and Ireland (Figure 8).

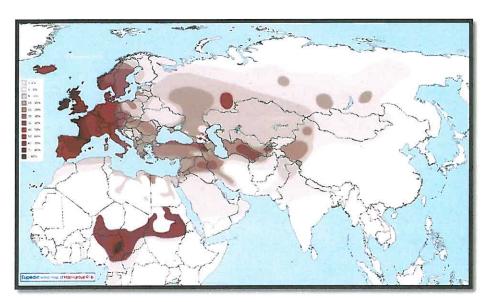


Figure 8: Frequency Map for Haplogroup R1b based on Y-DNA. The highest frequencies of individuals belonging to Haplogroup R1b are found in Europe and even some parts of Africa. Image taken from *Eupedia*.

The frequency of occurrence in Europe for Haplogroup R is 45.43%, the highest occurring haplogroup in Western Europe. Haplogroup R is also found in South Asia (40.51%), Central Asia (31.44%), the Middle East (20.56%), North Asia (7.66%), and Northern Africa (6.11%).

The results for my brother's Y-DNA sample through Family Tree DNA predicted that my paternal lineage belongs to Haplogroup R1b at the Marker M269. According to the Western Atlantic Modal Haplotype, this is the most commonly occurring Y-DNA marker for this haplogroup. Compared to the results from the National Genographic Project, this particular marker subcategorizes the paternal haplogroup by three subclades (R1b-M343  $\rightarrow$  R1b1-P25  $\rightarrow$  R1b1a-P297  $\rightarrow$  R1b1a2-M269).

### IV. Discussion

Through my analysis of historical movements and documents, and the results of my genetic testing, I have obtained a greater understanding of my ancestral roots for both my maternal and paternal lineages.

## a. Major Events in Puerto Rican History

Using the historical documentation that I uncovered, I came to comprehend the roles that major events played on the diverse population and history of Puerto Rico. Many events over the course of Puerto Rico's history have heavily influenced the ethnic makeup of the population. The first major event to drastically impact the island's population was the downfall of the Taínos. The downfall of the Taíno culture after the arrival of the Spaniards led to the increased need for additional slave labor. As a result, the Spaniards began importing African slaves through the Atlantic Slave Trade to work in the gold mines and the sugar and ginger industries. The establishment of sugar plantations in Puerto Rico further strengthened this demand for slave labor and brought about the importation of even more slaves from Africa. The population of the island thus increased significantly and, as a result, the racial demographic of this population diversified greatly.

Although African slaves faced a great deal of discrimination, many Spaniards intermarried with the African women. These intermarriages further helped to propagate the spread of African ancestry on the island, which greatly contributed to the genetic makeup of the population. Today,

African ancestry can be found in many Puerto Rican individuals.

## b. Piecing Together the Genealogy

Through my analysis of genealogical records, I was able to piece together the lives of my more recent ancestors.

# i. My Maternal Lineage: Meeting the Rodriguez Family

Using data obtained from family records and interviews, I compiled the following narrative information for my maternal lineage.

# My Great-Great-Grandparents, Pio Rivera and Fermina Lopez

My great-great-grandparents were Pio Rivera y Dias and Fermina Lopez. Pio and Fermina are my maternal grandfather's grandparents. My great-great-grandfather Pio was born around 1880. It is most probable that "Pio" was his nickname and that his birth name was Pedro. The majority of the records I gathered, however, documented his name as Pio. Fermina, on the other hand, was born approximately around 1883. Both individuals were born on the island of Puerto Rico. Once married, the two had seven children who were named Ramona, Monserrate, Dominga, Felix, Hipolito, Monica, and Julia (listed in order of birth) (Appendix II-a). It is also curious to note that another name appears as living at the same household at the time that this census was taken. Further information on this individual has not been found.

# 2. My Great-Grandparents, Serafin Rodriguez and Ramona Rivera

My great-grandparents, Serafin Rodriguez and Ramona Rivera, are my maternal grandfather's parents. Serafin was born in 1905, while Ramona was born in 1900. The exact dates of birth are unknown because various records display conflicting years and information. The six children of the Rodriguez family were Luis, Sara, Daniela, Lydia, Pedro, and Angel Manuel (my grandfather) (Figure 9).



**Figure 9: Rodriguez Family Photo from 1989.** Pictured from left to right: Angel, Ramona, Sara, Daniela, and Luis Rodriguez. Photo taken from Rodriguez Family Records.

Together, Serafin and Ramona owned several acres of land in Cayey where they grew an assortment of fruits and vegetables such as coffee, lemons, guava, and grapefruits. As she recalls her young childhood, my mother remembers sitting by the river and picking coffee beans with her cousins on Abuela (grandma) Ramona's farm. While they would talk and share stories, the children would grind up the coffee beans for their grandmother. On their breaks, Abuela Ramona would reward them with deliciously juicy mangos.

Although I never got to meet Abuelo (grandpa) Serafin, I was fortunate enough to meet Abuela (grandma) Ramona as a child. Despite struggling with diabetes, Ramona led a long life. She followed a very strict diet regimen enriched with healthy foods. Abuela Ramona died just three months before her 100th birthday in 1999 (Figure 10).



**Figure 10: Rodriguez Family Photo from 1999.** Pictured from Left to Right: Gloria, Luis, Daniela, Angel, Pedro, Sara, and Pedro Rodriguez at Ramona's funeral. Photo taken from Rodriguez Family Records.

#### 3. My Maternal Grandfather, Angel Manuel Rodriguez

My maternal grandfather, Angel Manuel Rodriguez, was born on July 5, 1941, in Cayey, Puerto Rico (Appendix II-b). He was the youngest one of the seven children of Serafin Rodriguez and Ramona Rivera. As a child, Angel Manuel was nicknamed "Nolly" by his older brothers and sisters. In his early twenties, Angel tried enrolling in the military but because of mental health and anxiety issues, he was honorably discharged after a few weeks (Appendix II-c).

Later, when he moved to New York with his wife Luz, Angel found work as a taxi driver (Appendix II-d). One night he received a phone call from a customer needing to be picked up at a certain location. Upon arrival, he was assaulted by a masked group who beat him and left him unconscious and without clothes. Fortunately, he was able to survive. Yet, the psychological side effects of such a trauma still remained. For the rest of his life, he suffered from anxiety issues. As a result of this trauma, Angel and his family moved back to Puerto Rico.

#### 4. My Maternal Grandmother, Luz Minerva Casado

My maternal grandmother, Luz Minerva Casado, was born on June 3, 1936, in Canovanas, Puerto Rico (Appendix II-e). She was the daughter of Amalio Toledo, a painter, and Bernardina Casado, a maid. The couple had another younger son Manolo after

the birth of the first child Luz, but he died at a very young age. Growing up in Canovanas, Luz's family was very poor and struggled greatly to make ends meet. She was forced to withdraw from her schooling once she graduated from the 6th grade. Because her father did not play a strong role in her upbringing, Luz was raised primarily by her mother Bernardina and her grandmother Ramona Canales (Appendix II-f). As a result, she took on her mother's maiden name.

She also found that she possessed a great passion for her faith and religion. Luz was an avid churchgoer and community member. On several occasions, she spoke as a leader at her church and was recognized for her undying dedication to her faith (Appendix II-g). It was also here at her local church that she met the handsome young Angel Manuel Rodriguez. After some period of dating, the two were married on November 28, 1964 (Appendix II-h). Shortly afterwards, my grandparents moved to New York to start a new life. Luz found work as a hat maker in a clothing store to help support her family. Several years later, Luz, Angel, and their nine-year old daughter Marilu moved back to Puerto Rico. They bought a home in the high mountains of Cayey, Puerto Rico, and Luz and Angel lived there up until three years ago when they moved to Florida (Appendix II-i). Despite all the struggles that she

has endured, Abuela Luz is one of the strongest and most resilient women that I know.

## 5. My Mother, Marilu Rodriguez

My mother, Marilu Rodriguez, was born on December 2, 1965, at Beth Israel Medical Center in Manhattan, New York (Appendix II-j). Marilu was the only child to my grandparents Angel and Luz. My grandparents tried several times for a baby but, unfortunately, my grandmother Luz suffered several spontaneous abortions. During one of her pregnancies, she lost a set of twins because of an unsafe fall from an elevator shaft in New York. Luz Minerva often says that she should have named my mother Milagros, which means "miracle" in Spanish. Marilu spent the first 9 years of her life growing up on the streets of Manhattan. As a child, Marilu was very quiet and loved going to school and learning.

At 9 years of age, my mother and her parents moved to my great-grandmother Bernardina's home in Canovanas, Puerto Rico, for a couple of weeks until they were able to purchase a house of their own. Within a number of weeks, Angel and Luz bought a house up in the mountains of Cayey, Puerto Rico. As an adult, Marilu attended the University of Puerto Rico and graduated with a Bachelor's Degree in Elementary Education (Appendix II-k). She was later hired as an elementary English schoolteacher for La

Escuela Julio Vizcardi. This school was located only five minutes away from my grandparent's home.

#### ii. My Paternal Lineage: Meeting the Negrón Family

The following personal narrative information for my paternal lineage was compiled from family interviews and data obtained from genealogical records.

#### 1. My Paternal Grandfather, Luis Negrón

My paternal grandfather, Luis Negrón was born on October 1, 1940, in Cayey, Puerto Rico. As a child, Luis was fascinated with music and art. Although he never learned how to formally read music, he taught himself how to play numerous instruments such as the saxophone, the guitar, and the piano. He has dedicated much of his life to music and spends many nights performing at various venues in Cayey. Before marrying my grandmother Ruthy in 1964, Luis had a previous marriage and fathered my aunt Carmen Yvette. Ruthy and Luis were later married and had four children: Nelly, Angel Luis, Eliesel, and Joanne (Figure 11). During the marriage, however, Luis was unfaithful and fathered three other children as well. Although the difficulty of infidelity loomed over their marriage, Luis and Ruthy were able to reconcile and are still together today.



**Figure 11: Negrón Family Photo in 1974.** Pictured from left to right: Mayra, Angel Luis, Joanne, Eliesel, and Nelly. Photo taken from Negrón Family Records.

### 2. My Paternal Grandmother, Ruthberta Maldonado

My paternal grandmother, Ruthberta ("Ruthy") Maldonado, was the daughter of Juan Maldonado and Victoria Cotto. Born on January 12, 1941, Ruthy always demonstrated a great passion for food and cooking. As an adult, she worked as a chef at a local restaurant. Similarly to my grandfather, Ruthy also had previous marriages before marrying Luis. She had two daughters, Yvonne and Mayra. Abuela Ruthy married Luis in 1964 after meeting him in New York.

### 3. My Father, Angel Luis Negrón, Jr.

My father, Angel Luis Negrón Jr., was born on December 13, 1966, in Manhattan, New York (Appendix II-l). Angel was one of many children to Luis Negrón and Ruthy Maldonado. As a

child, he demonstrated a great passion for education and his schooling. His favorite subjects were math and science. Some of his most cherished childhood memories were when he and his siblings would hold competitions with one another. Junior, as he was commonly referred to, would also spend countless hours in the garage with his father working on cars. By age 13, he already knew how to drive both automatic and stick-shift cars. After graduating high school, Junior attended Miguel Such Vocational School to study aviation (Appendix II-m,n). He left Puerto Rico a year later to start a new life in 1988 and work for American Airlines in Tulsa, Oklahoma, with only \$500 in his pocket.

### iii. Uniting the Negrón and Rodriguez Families

Both the Negrón and Rodriguez families had been good family friends since their children's youth. My parents, Angel Luis and Marilu, were reunited in 1989 when my father returned to Puerto Rico from working in Oklahoma. Marilu had acquired employment as an English schoolteacher at La Escuela Julio Vizcardi in Cayey, Puerto Rico. This elementary school was located right across the street from the Negrón home. One sunny day in May of 1988, Marilu took her students out to recess in the school's playground. As she watched her students play, Marilu took a look across the street at all the homes. She saw two figures standing outside the Negrón home and recognized them as Eliesel (my uncle) and Junior (my father). She waved a

cheerful hello to the brothers and received the same response back. At the time, she did not know that Junior didn't recognize her because of his poor eyesight. Later that afternoon, Junior and his parents decided to visit the Rodriguez family. As my mother recalls, Luz Minerva spent much of the time interrogating Junior and asking whether or not he was currently in a relationship. Before leaving, Junior made sure to ask Marilu out on a date. Their first official date was to the local Pizza Hut. The pair dated for the next year and a half before getting married on June 2, 1990, at the Baptist Church of Caguas in Puerto Rico (Appendix II-o).

Shortly after getting married, the couple moved to Dallas, Texas in search of new opportunities. Over the course of the following years, Marilu and Junior had two children: my brother and me. I was born on August 20, 1993, in Arlington, Texas (Appendix II-p). I am currently a senior in college studying biology, history, and behavioral neuroscience at Nova Southeastern University. My younger brother, Izak, was also born in Arlington on July 26, 1995 (Appendix II-q). He is currently a sophomore at Florida International University studying mechanical engineering and wishes to study aviation like our father.

#### c. Understanding the Genetic Data

#### i. Overview of My Maternal Lineage

Prior to beginning testing, I conducted a great deal of research surveying the literature available on African ancestry specifically connected to Puerto Rican individuals. From my research, I selected 23 markers to be looked at for more in-depth analysis (Table 2). The markers were chosen based on their linkage to either a specific haplogroup or subclade (Figure 12).

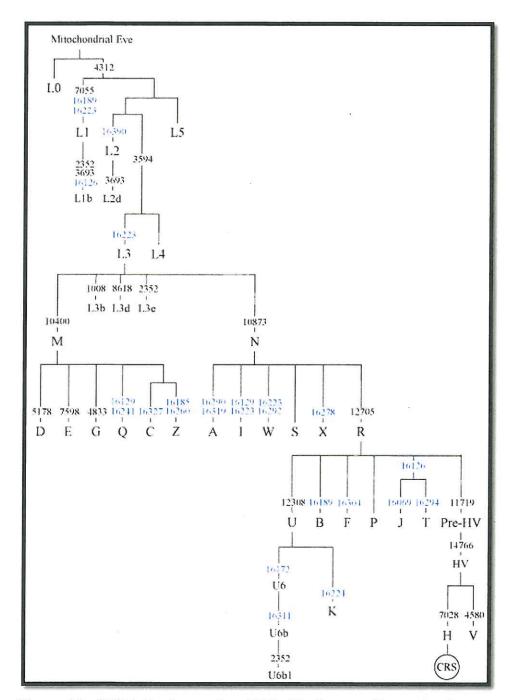


Figure 12: HVR1 Region and mtDNA Haplogroup Determination Map. There are certain molecular markers associated with each haplogroup. Some of the markers chosen for closer examination for my research are listed as linked to Haplogroup L1 in the top left corner of the figure. Image taken from *Genebase*, 2015.

Out of the 23 markers examined, my DNA results had the following 7 associated with African ancestry and the L1 haplogroup: G3666A, A7055G, T7389C, T13789C, T14178C, G14560A, and A3796T (Figure 13).

The genetic tests used in this study examined the HVR1 and HVR2 coding regions of mtDNA. These coding regions reveal the most information about a person's ancestry. The HVR1 and HVR2 regions contain an abundance of single nucleotide polymorphisms encompassing over 16,000 molecular markers (Wells, 2006). Therefore, studying these regions is a good way to obtain a detailed understanding of a person's ancestry because each individual's set of mtDNA SNP markers is unique to that person and their maternal line. For this reason, my SNP markers are identical to the markers of my grandmother and everyone else along my maternal line.

The studies that I conducted through the National Genographic Project, Family Tree DNA, and 23andMe allowed me to confirm that my maternal lineage belongs to Haplogroup L1, and helped me to further specify this characterization by two subclades. I found that my mitochondrial DNA possesses markers consistent with the L1c1 subclade (A3796t, A3843G, A14148G, and A16293G) and the L1c1a subgrouping (T198C) (Figure 13).

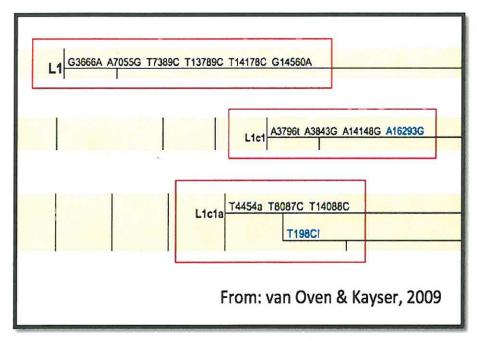


Figure 13: African Ancestral Informative Markers. Some of the genetic markers associated with Haplogroup L1, Subclade L1c1, and the subgrouping L1c1a are outlined above. Image taken from van Oven & Kayser, 2009.

Genetic data from 23andMe also allowed me to approximate specific regions of ancestry within Africa for my maternal lineage. Both my grandmother and I possess mtDNA that is consistent with ancestry from the Yoruba tribe in West Africa. The Yoruba tribe was one of the African groups brought to the Caribbean through the Atlantic Slave Trade, and so it is very plausible that our maternal ancestor was part of this group. Unfortunately, there are very few records documenting the individuals who were brought to the Caribbean in this manner. However, perhaps one day I might find a record of who my grandmother's grandmother was and it is possible that she may be a descendant of a Yoruba tribeswoman brought from Africa between 1513 and 1873 when

the Atlantic Slave Trade was active. If the ancestor came in the later 1800s, perhaps I will be able to find documentation of her import into Puerto Rico and having a child named Bernardina. Currently, there are only slave registry records from Puerto Rico for the year 1872 and my maternal grandmother's last name does not show up there at all (*Registro Central de Esclavos*, 1872). If additional records become available, I plan to search them for any such evidence of my maternal ancestry and their connection to the importation of slaves.

### ii. Overview of My Paternal Lineage

For my paternal lineage, I was able to confirm Haplogroup R1b and further specify my subclade by three groupings (R1b-M343 → R1b1-P25 → R1b1a-P297 → R1b1a2-M269). The Y-DNA37 test from Family Tree DNA provided me with 30 distinct markers associated with this particular haplogroup and gave me a better idea of where my paternal ancestry originated.

# d. Implications for Puerto Rican Identity

The population of Puerto Rico represents a fusion of the three major genetic groups on the island (European, African, and Amerindian). Racial classifications on the island are based primarily on skin color and any observable physical characteristics like hair texture, nose shape, and lip form (Duany, 2000). Contrary to most other racial models, Puerto Ricans also may take into account socioeconomic factors in determining race.

According to Loveman and Muniz, the Puerto Rican perception of how whiteness was characterized changed significantly with social and cultural changes (2006). As the number of interracial unions rose, the percentages of individuals who were considered white in census records also increased greatly. Loveman and Muniz attribute this to spousal reclassification where the majority of spouses changed racial classifications to match the spouse of lighter color with a higher social status. Thus, intermarriage was commonly seen by many Puerto Ricans as a mechanism to "improve one's race" ("adelantar la raza") during the early 20th century.

However, this general whitening effect that is argued by Loveman and Muniz cannot adequately describe the racial identity of Puerto Ricans. Rather, the racial identity of this population is much more complex. Three different racial groups came together to shape the Puerto Rican culture and identity in many ways. As a result, Puerto Ricans today view themselves as a unified group that does not seem to differentiate between whites and blacks. This unified identity has led to a strong sense of nationalism and pride, as well as a general racial tolerance. European, African, and Taíno influences can be seen in art, language, music, and food. This atmosphere of interracial harmony is void of the more explicit forms of racism and racial violence. This is the multiracial atmosphere that I was exposed to every summer growing up when I visited Puerto Rico. As a whole, the people of Puerto Rico are very racially tolerant and there seems to be a strong belief that there is not racial prejudice on the island.

Nevertheless, after interviewing different family members about their experiences and views on race, I found that the overwhelming consensus was that there still exists some racial prejudice on the island, particularly among the older generations. My father believes that the more widespread absence of racism is due to the diverse ancestral genetic composition of the people. However, he also recognizes that some are prejudiced against those with darker skin pigmentations. My maternal grandmother even argues that the prejudice exists amongst blacks on the islands ("de Negro a Negro"), and that this prejudice is founded on differences in power, education, and money.

When I asked my family members (my father, mother, and maternal grandmother) how they identified themselves, they either answered Hispanic or Puerto Rican. They each embrace having a multiracial background. When I discussed my findings with them, they were not surprised at having relatively recent African ancestry (within the last 500 years). Actually, I was the only member of my family who was surprised by such recent African roots. After concluding my research, I have come to take great pride in my triracial origins.

#### e. Simply the Beginning

This undertaking has become one of my most treasured experiences. It represents the beginning of a lifelong endeavor to better understand both my genetic and ancestral roots. Although I have learned a great deal about my ancestors through this venture, there is still an entire world of knowledge waiting to be uncovered. I hope to continue this work throughout my life and

learn more about the ancestral origins that run through my blood making me who I am today. As more historical documents become available, I plan to continue expanding my collection of genealogical records to help me trace back my family tree as much as possible. It is also my hope that this thesis project will serve as a template for other individuals hoping to uncover their own personal ancestral roots and genealogy.

# V. Ancestral History Narrative: "Chione: Daughter of the Nile"

After learning of my deep ancestry, I became fascinated with this haplogroup and pondered over what my ancestors would have been like. The following passage is a fictional story that I created offering a small glance into the life of my ancient ancestor. The narrative tells the story of a possible ancestral relative, Chione of Maadi (present-day Cairo), Egypt, during the time period between 3600 and 3000 B.C. based on historical information consistent with my maternal haplogroup. Knowledge acquired from my ancestral studies of Haplogroup L1 serves as the foundation for my biologically and historically-based fictional family narrative.

## Borders of the Nile River, 3300 B.C.

As she sat quietly by the river, Chione filled the steatite (variation of mineral talc) pot she had crafted earlier that week with water. She couldn't help but become mesmerized by the intricate carvings she had etched into the water pot. It had taken her quite some time but she wanted to make sure they were absolutely perfect. Running down the center of the pot was the Eye of Horus and the hieroglyphics that signed out "I will always watch over you." Representing protection, the Eye of Horus was one of the most symbolic characters in Egyptian culture. Her father had always said those very words to her every night before bed. As she ran her fingers over each hieroglyphic taking care to truly feel the power of each one, she was transported back to a sea of memories from her childhood...

In the flicker of an instant, Chione was brought back to the present. She looked down the river at her friends and neighbors gathering water too. Above her, the sun burned brightly casting a powerful glimmer on the river. She quietly bowed her head and thanked Ra, the God of Sun, for blessing them once again with the presence of the bright sun.

Lifting up the now-filled pot, Chione set on her way back through the village to her home down at the end of the road. As she made her way through her small native village, Chione looked past the hustle and bustle of the traders to see the magnificent pyramids towering off in the distance. Her father had helped to build those very pyramids. Every time she saw them, she couldn't help but think of him.

The village of Maadi was especially busy today. Traders and merchants traveled great distances from all over to the town of Maadi for the chance to exchange goods. Maadi had become one of the foremost trade centers along the Nile River Valley. As Chione made her way through the city, she saw many merchants trading beautiful cloths, various foods, domesticated donkeys, and, of course, the coveted copper and gold. Her beloved city of Maadi was never quiet, and this was something for which Chione had always been thankful. As a young girl, she would always accompany her father into the village to trade the cloths and fabrics her mother had woven for an assortment of goods. It was a fast-paced world but she loved every minute of it. Sometimes she would even bring some of her pottery to town. Her natural gift for pottery had made art her passion. Chione

always dreamed that one day her sculptures and artwork would find a home in the great palace of the pharaoh.

After a long walk through the bustling village, Chione finally arrived at her home. As she made her way through the brick entrance, Chione saw her mother sitting by the bathing pool. Her golden skin glistened in the powerful light of the sun. She walked over to where her mother sat and placed the brown pot on the ground near her.

"Mut (Mother), what are you doing?" Chione asked.

"My daughter Chione, I am thanking the Gods for my beautiful children. They have blessed me with four wonderful children. One day, you too will sit here and thank them for your children." Mut replied in her soft velvety voice.

"I hope one day to be as blessed as you have been Mut." Chione uttered. For some time afterwards, the pair sat quietly by the bathing pool in simple silence. Although she was only 14 years old, Chione thought of her future children and what they would be like. Somehow she knew they would be children of the Nile - great and strong people. Unbeknownst to Chione, her children and her children's children would one day give rise to descendants that would come to populate the entire world.

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# **APPENDIX I: Genetic Testing Results**

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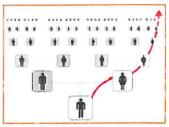


Thank you for testing at Family Tree DNA.

Your mtDNA (mitochondrial DNA) Full Sequence results are complete. These results are the first step in understanding your genetic ancestry through your direct maternal line.

This guide will explain how your results help with genealogy and distant ancestry as well as how to use your myFTDNA personal account.

#### The Direct Maternal Line



Your direct maternal lineage is the line that follows your mother's maternal ancestry. This line consists entirely of women, although both men and women have their mother's mtDNA. This means that fathers do not pass on their mtDNA to their children. Your mtDNA can trace your mother, her mother, her mother's mother, and so forth, and offers a clear path from you to a known, or likely, direct maternal ancestor.

Note that you and your matches may share ancestors on other parts of your family tree; however, those matches are coincidence.

## Matching for Genealogy

Your mtDNA may help you find genetic cousins along your direct maternal line. When we look at your mtDNA HVR1, HVR2 and Coding Region results, we look for differences in your mtDNA caused by small changes - copying errors - that we also call "mutations" or "polymorphisms." We compare those differences to the differences for other people in our database. The range of possible generations before you share a common ancestor with a match is wide. Your mtDNA HVR1, HVR2 and Coding Region exact matches may be recent, but they may also be hundreds or thousands of years in the past. We show this in the table below.

Note: The term "mutation" in this sense does not refer to anything medical.

Testing Level	AND THE REST	Generations to Common Ancestor				
	Matching Level	50% Confidence Interval	95% Confidence Interval			
mtDNA	HVR1	52 (about 1,300 years)	- NA*			
mtDNAPlus	HVR1 & HVR2	28 (about 700 years)	— NA*			
mtFullSequence	HVR1, HVR2, & Coding Region	5 (about 125 years)	22 (about \$50 years)			

<sup>\*</sup>The range of generations to a common ancestor at this level is too broad to calculate a 95% confidence period.

The wide range in the test results does not prevent those results from being useful. You can use this clear maternal line to provide evidence to support a relationship. You first trace two or more female lineage descendants of a single woman utilizing traditional genealogy research. The descendants then test their mtDNA. If they are exact matches, it is evidence that supports the relationship. Not matching usually disproves the relationship.

Family Tree DNA - Gene by Gene, Ltd. 1445 North Loop West, Suite 920 Houston, Texas 77009, USA Phone: (713) 868-1438 info@FamilyTreeDNA.com http://www.familytreedna.com

Appendix I-a: Family Tree DNA – Guide to Understanding Your Maternal Genetic Results



Planned comparisons are the best choice. However, you can still find your common ancestor with as little as an HVR1 or HVR2 match. To do so, use your known maternal genealogy. For each match, look for common geographic locations on the direct maternal line. Work through each of your ancestors on this line as well as their daughters, their daughters' daughters, and so forth.

Comparing genealogical records is vital when using mtDNA matching to help you in your research. You need to enter all that you know about your direct maternal line in your myFTDNA account. See the "Quick Steps" reference page for step-by-step instructions on how to set up your account and access your matching information.

## The Science of Your Direct Maternal Line

We can trace our direct maternal line with our mitochondrial DNA due to a special relationship between the power supply for human cells (the mitochondria) and the cell itself.

In every human cell, there are hundreds and sometimes thousands of mitochondria. Each mitochondrion has several copies of its genetic code (represented by the letters A, C, T, and G). This genetic code is mitochondrial DNA (mtDNA) and tells the mitochondria how to function. The code also tells the mtDNA how to copy itself. Over time, the copying process can create small changes - "polymorphisms" or "mutations." If these changes are in the mother's egg cell, the child produced from that egg inherits them. If female, the child may some day pass that same change on to her own children.

Slowly, these changes build up down maternal lineages. They define and mark branches on the maternal tree. We can look at your specific mtDNA code sequence to see which changes you have in your mtDNA.

There are two scientific baseline sequences to which scientists compare these changes in mtDNA: the Reconstructed Sapiens Reference Sequence (RSRS) and the revised Cambridge Reference Sequence (rCRS). By comparing your mtDNA changes to each sequence, we can distinguish the differences between the original values in the RSRS (or the comparative values in the rCRS) and your test results. Following scientific standards, Family Tree DNA compares all mtDNA results to the rCRS, and provides you with your comparisons to the RSRS.



The example above shows results for someone who has tested all three parts of their mtDNA: the HVR1, the HVR2, and the Coding Region.

In the example, each letter-number combination represents a difference between your results and the reference sequence. These individual changes do not indicate any particular ethnic or geographic origin. Their significance is in their ability to mark branches on the maternal tree, and those branches are geographically and sometimes ethnically significant.

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Appendix I-a: Family Tree DNA – Guide to Understanding Your Maternal Genetic Results



## Your Ancestral Origins

Your direct maternal ancestors have passed down their mtDNA generation after generation. The line began with our common maternal ancestor in Africa and it ultimately reached you. Our mtDNA marks the path from our ancestors in Africa to their locations in historic times. Your ancestors carried their mtDNA line on their historic migrations. Your line's present geography shows the path of this journey. Your mtDNA results are the most precise DNA tool for this line. Your mtDNA results use two strategies to explore your maternal origins.

The first method uses your main (backbone) branch on the maternal tree, known as an mtDNA haplogroup. Scientists study the history of populations across geography and time using mtDNA. They often use the frequencies of each branch in modern populations, as well as samples from ancient burial sites. With these, they are able to tell us much about the story for each haplogroup. This traces back hundreds, thousands, or even tens of thousands of years. Your branch on the tree tells you where your maternal ancestors are present today and about their likely path out of Africa. We summarize what scientists know about your mtDNA Haplogroup on the mtDNA - Results page of your myFTDNA account.

The second method uses your haplogroup information alongside your mtDNA Full Sequence differences. We use these to match you to the information of others in our database. These matches are more likely to share your lineage in closer genealogic time. Country frequencies thus reflect the more recent history of your ancestors. Both the mtDNA – Haplogroup Origins and the mtDNA – Ancestral Origins pages show this information. See the "Quick Steps" reference page for step-by-step instructions on how to access this information.

## Privacy and Courtesy Standards

#### Privacy

Family Tree DNA cares about your privacy. When you use our internal database, we can assure your privacy. However, please use caution if anyone asks you to export your results to other databases not endorsed by Family Tree DNA. Be sure you have confidence in the privacy protections of whoever is requesting your data. You should not ever share your kit number and password outside of Family Tree DNA.

#### **Courtesy Standards**

Family Tree DNA would like genetic genealogy and DNA testing to be an enjoyable experience for all. We ask that our customers treat each other, project administrators, and Family Tree DNA staff with courtesy and respect. When your matches reach out to you, please respond to them. Even an answer of "I don't know" is better than no answer. At all times, respect the privacy of others. Everyone has his or her own comfort level for sharing genetic information. The best policy is to ask before doing.

#### Resources

## Useful links:

myFTDNA 2.0 User Guide: mtDNA - http://www.familytreedna.com/fao/answers.aspx?id=49

Understanding Results: mtDNA - http://www.familytreedna.com/faq/answers.aspx?id=10

mtDNA Library of Scientific Papers - http://www.familytreedna.com/mtdna-papers.aspx

Glossary (complete) - http://www.familytreedna.com/faq/answers.aspx?id=21

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#### **Common Terms**

**Coding Region:** The coding region is the slow changing part of the mitochondrial genome. It contains genes, causing it to have a slower change rate than the rest of the genome.

Direct Maternal Line: The direct maternal line is the line that traces your mother, her mother, her mother's mother, and so forth. With the exception of yourself if you are male, this line consists entirely of women.

GEDCOM - An internationally recognized software file (.ged) that allows you to organize genealogical information. You may export this file type from a number of software programs and upload your GEDCOM to the Family Tree DNA site. Family Tree DNA does not host or provide this software for you to create your GEDCOM.

Genetic Cousins - Individuals whose DNA test results match one another, indicating shared genetic ancestry.

Genome: A genome is a full set of genetic code. Humans have two genomes, the nuclear genome and the mitochondrial genome. The human nuclear genome is composed of 46 chromosomes (23 pairs), which contain 3 billion base pairs. The human mitochondrial genome is composed of a single circular DNA sequence that contains 16569 base pairs.

Haplogroup - A haplogroup is a major branch on either the maternal or the paternal tree of humankind. Haplogroups are associated with early human migrations. Today these can be associated with a geographic region or regions.

HVR1 (Hypervariable Region 1): HVR1 is one of two faster changing parts of the mitochondrial genome. There are two human Hypervariable Regions, HVR1 and HVR2. They do not contain genes. Therefore, they have a faster change rate than the coding part of the mitochondrial genome.

HVR2 (Hypervariable Region 2): HVR2 is one of two faster changing parts of the mitochondrial genome. There are two human Hypervariable Regions, HVR1 and HVR2. They do not contain genes. Therefore, they have a faster change rate than the coding part of the mitochondrial genome.

mtDNA (Mitochondrial DNA): Mitochondria are parts (organelles) within human cells. Our mitochondria provide cell respiration and make energy for the cell. Mitochondria were once separate organisms. Early mitochondria evolved inside primitive nuclear cells. They formed a mutually beneficial relationship with the larger nuclear cell and have their own mitochondrial DNA (mtDNA) genome. Human mothers pass their mtDNA genome to their children.

MDKA (Most Distant Known Ancestor): Your most distant known ancestor (MDKA) is the furthest person who you have documented on a specific genealogical line. In genetic genealogy, it usually refers to someone on a direct maternal line (the mother, her mother, her mother's mother, etc.) or to a direct paternal line (the father, his father, his father, etc.).

Polymorphism: Polymorphisms are changes to your DNA code. They are natural copying errors. One analogy is to think of a copy machine that is making many copies of a page. Occasionally it will make a mistake; an e might look more like an o, for example. This is a "polymorphism." If you then take that page with the o and copy it, it will pass on its "polymorphism" to all of its descendant copies.

RSRS: The Reconstructed Sapiens Reference Sequence (RSRS) is the sequence against which scientists compare all tested results. Researchers created it using a global sampling of modern human samples and samples from ancient hominids. It is less likely to have bias toward one population or maternal lineage.

rCRS: The historic revised Cambridge Reference Sequence (rCRS) is the sequence based on the first complete mtDNA sequence completed.

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# **Genealogy Quick Steps**

#### **Entering Your Ancestry Information**

Your myFTDNA account has two places for you to enter ancestry information. They are the My Account - Most Distant Ancestors page and the My Account - GEDCOM/Family Tree page. After you enter information, you will be ready to access and contact your matches.

The My Account - Most Distant Ancestors page is where you enter basic information about your most distant known ancestor (MDKA) on your direct maternal line. To do so:

- Log in to your myFTDNA account. (https://www.familytreedna.com/login.aspx)
- 2. On the top menu bar, find the My Account menu.
- 3. From the My Account menu, select Most Distant Ancestors.
- On the My Account Most Distant Ancestors page, look for the Direct Maternal area in the Most Distant Ancestors section.
- In the Direct Maternal area, select the Country of Origin. If you are not sure of this, select Unknown.
- In the Direct Maternal area, enter the name, date of birth, and date of death of your most distantly known direct maternal ancestor in the Name field.
- Click the Save button to save your changes.
- On the My Account Most Distant Ancestors page, find the Direct Maternal area in the Ancestral Locations section.
- Click the Add Location button to launch the Ancestral Locations Map wizard.
- 10. In the Maternal Location section, click the blue Edit Location button.
- Follow the steps to add your direct maternal most distantly known ancestor's
  location. This is usually the place where she was born. If you do not know that, it can
  also be where she was married or where she died.

The My Account – GEDCOM/Family Tree page is where you upload an electronic file of your pedigree. GEDCOM is the standard format for sharing electronic pedigree information, and almost all genealogy software packages are able to export to standard GEDCOM format (.ged). To upload your file:

- First, you must export your GEDCOM file from the software in which you developed
  your family tree (you should be able to do this by clicking on File in that program
  and then Export to save this file to your computer). Please note that Family Tree
  DNA does not host or offer customer support for any GEDCOM software.
- Log in to your myFTDNA account (https://www.familytreedna.com/login.aspx)
- 3. On the top menu bar, find the My Account menu.
- From the My Account menu, select GEDCOM/Family Tree.
- On the My Account GEDCOM/Family Tree page, look for the GEDCOM section and the Next button.
- Click on the Next button to start the GEDCOM Upload wizard.

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#### **Accessing Your Matches**

You may view your mtDNA Full Sequence matches on our website by following these steps:

- Log in to your myFTDNA account. (https://www.familytreedna.com/login.aspx)
- On the top menu bar, find the mtDNA menu.
- From the mtDNA menu, select Matches.

The mtDNA – Matches page has two sections. The top Filter Matches section is where you can change the testing level and filter for specific parameters. The bottom Matches section is where you can view your matches. To view your most recent mtDNA HVR1 and HVR2 matches:

- 1. In the Filter Matches section, change the For: field to HVR1, HVR2, Coding Region.
- 2. Click the orange Run Report button

By default, your most recent matches will be at the top of the report. You can read about additional settings for the mtDNA – Matches page by clicking on the blue page Help button at the top of the page.

# **Ancestral Origins Quick Steps**

#### mtDNA Haplogroup Information

To find information about your maternal branch (haplogroup):

- Log in to your myFTDNA account (https://www.familytreedna.com/login.aspx)
- 2. On the top menu bar, find the mtDNA menu.
- From the mtDNA menu, select Results.
- 4. On the mtDNA Results page, look for the Your Origin section.

The Your Origin section describes what we know about the origin and historic migrations of your direct maternal line.

#### Haplogroup and Ancestral Origins Information

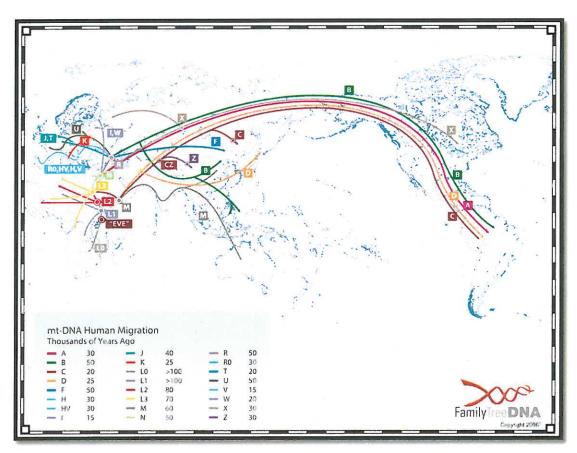
To find information about your more recent ancestral origins, begin with the mtDNA – Haplogroup Origins page:

- Log in to your myFTDNA account (https://www.familytreedna.com/login.aspx)
- 2. On the top menu bar, find the mtDNA menu, and then select Haplogroup Origins.
- On the mtDNA Haplogroup Origins page, look for blue Help button. The help section describes how to interpret the table of populations and frequencies on the page.

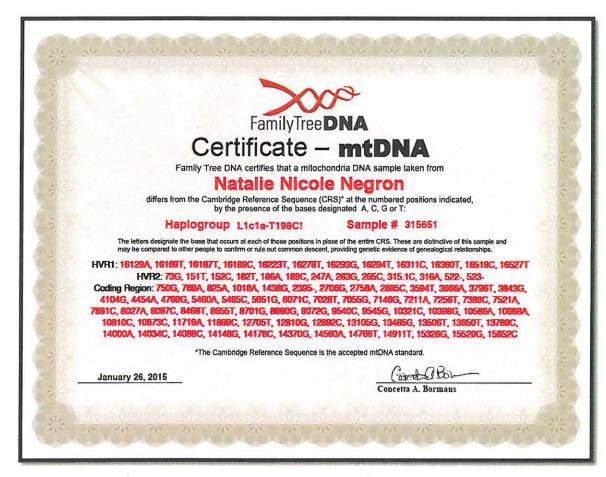
#### Next, check the mtDNA - Ancestral Origins page:

- Log in to your myFTDNA account. (https://www.familytreedna.com/login.aspx)
- On the top menu bar, find the mtDNA menu, and then select Ancestral Origins.
- On the mtDNA Ancestral Origins page, look for blue Help button. The help section describes how to interpret the table of populations and frequencies on the page.

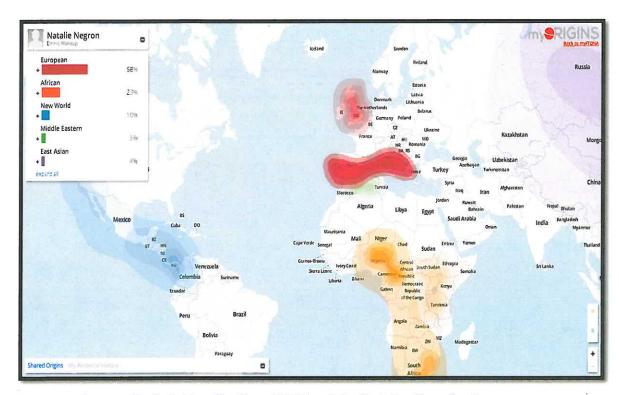
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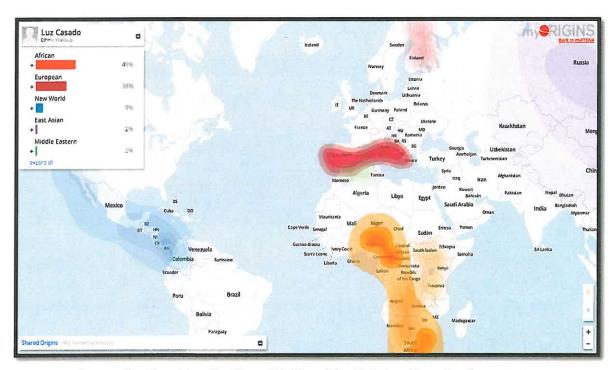
Appendix I-b: Family Tree DNA – mtDNA Migration Map (Enlarged Version)



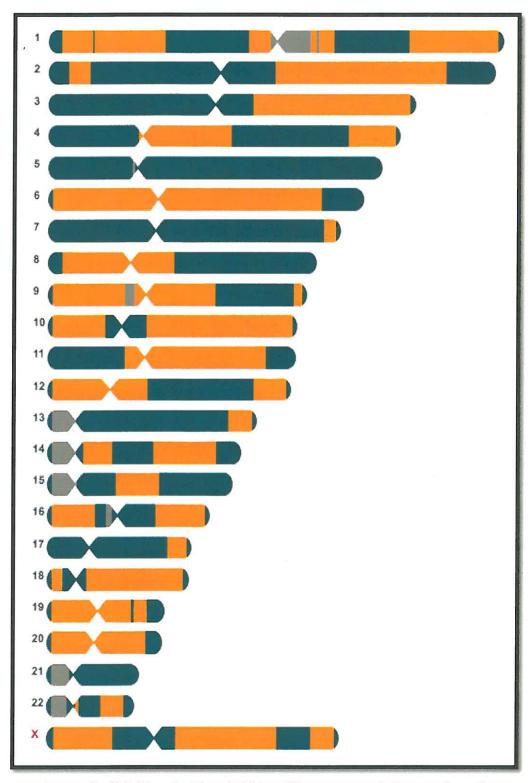
Appendix I-c: Family Tree DNA – mtDNA Certificate for Natalie Nicole Negrón



Appendix I-d: Family Tree DNA – My Origins Results for Natalie Nicole Negrón



Appendix I-e: Family Tree DNA – My Origins Results for Luz Minerva Casado



Appendix I-f: Family Tree DNA – Chromosomal Comparison for Natalie Nicole Negrón and Luz Minerva Casado

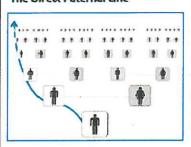


Thank you for testing at Family Tree DNA.

Your Y-DNA37 results are complete. These results are the first step in understanding your genetic ancestry through your direct paternal line.

This guide will explain how your results help with genealogy and distant ancestry as well as how to use your myFTDNA personal account.

#### The Direct Paternal Line



Your direct paternal lineage is the line that follows your father's paternal ancestry. This line consists entirely of men. Your Y-Chromosome DNA (Y-DNA) can trace your father, his father, his father's father, and so forth. It offers a clear path from you to a known, or likely, direct paternal ancestor.

Note that you and your matches may share ancestors on other parts of your family tree; however, those matches are a coincidence

#### Matching for Genealogy

Your Y-DNA may help you find genetic cousins along your direct paternal line. For Y-DNA37 results, we report your results for STR markers. STR marker values change slowly from one generation to

Steps	Probability that the MMCA lived no longer than this number of generations ago.						
	50%	904	95%				
0	2	5	7				
1	4	8	10				
2	б	12	14				

MRCA = Most Recent Common Ancestor

the next resulting in distinctive sets of results. We compare your set of results to those of other men in our database. The range of possible generations before you share a common ancestor with a match is wide. Your Y-DNA37 exact matches (0 Steps) may be recent, but they may also be hundreds of years in the past. Your matches that have one marker difference (1 Step) may be even more distantly

related. We show this in the table above.

The wide range in the test results does not prevent those results from being useful. You can use this clear paternal line to provide evidence to support a relationship. You first trace two or more male lineage descendants of a single man utilizing traditional genealogy research. The descendants then test their Y-DNA. If they are exact matches, it is evidence that supports the relationship. Not matching usually disproves the relationship.

Planned comparisons are the best choice. However, you can still find your common ancestor with matches. To do so, use your known paternal genealogy. For each match, look first for a shared

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surname if you come from a culture where surnames have followed paternal lines. Then look for common geographic locations on the direct paternal line. Work through each of your ancestors on this line as well as their sons, their sons' sons, and so forth.

Comparing genealogical records is vital when using Y-DNA matching to help you in your research. You need to enter all that you know about your direct paternal line in your myFTDNA account. See the "Quick Steps" reference page for step-by-step instructions on how to set up your account and access your matching information.

#### The Science of Your Direct Paternal Line

Your Y-Chromosome is a sex chromosome. Sex chromosomes carry the genetic code that makes each of us male or female. All people inherit two sex chromosomes. One comes from their mother and the other from their father. You and other men receive a Y-Chromosome from your father and an X-Chromosome from your mother. Men and only men inherit their father's Y-Chromosome. Thus, it follows the same path of inheritance as your direct paternal line.

STR markers are places where your genetic code has a variable number of repeated parts. We report your STR marker results as the measured number of repeats for each marker. In the example below, the marker DYS393 has 12 repeats.

Marker	DESSE	DY3390	DYS19**	DYSJ91	D18305	DYS 426	DIS447	DY8437	DYS 440	DY\$442	DY9430
Value	12	23	12	10	16-16	11	24	13	:9	13	21

Over many generations, the number of repeats in each STR marker changes. The number of repeats may go up or down. These changes create the patterns (haplotypes) of individual lines. This process is random. It is not possible to predict that any one marker will change between any set of generations. We do know though how often on average these random changes happen. Thus, we can estimate how closely related two men are by using the similarity of their results.

#### **Your Ancestral Origins**

Our Y-DNA marks the path from our direct paternal ancestors in Africa to their locations in historic times. Your ancestors carried their Y-DNA line on their travels. The current geography of your line shows the path of this journey. Your Y-DNA37 results use two ways to explore your paternal origins.

The first method uses your main (backbone) branch on the paternal tree. This is your Y-DNA haplogroup. Scientists study the history of populations across geography and time using Y-DNA. They use both the frequencies of each branch in modern populations and samples from ancient burial sites. With these, they are able to tell us much about the story for each branch. This traces back hundreds, thousands, or even tens of thousands of years. Your branch on the tree tells you where your paternal ancestors are present today and about their likely migration paths. We review what scientists know about your Y-DNA Haplogroup on the Y-DNA – Haplogroup & SNPs page of your myFTDNA account.

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The second method uses your haplogroup with your STR marker results. We use these to match you to others in our database. These matches are more likely to share your line in historic times. Country frequencies thus reflect your ancestors' recent history. Both the Y-DNA – Haplogroup Origins and the Y-DNA – Ancestral Origins pages show this information.

See the "Quick Steps" reference page for step-by-step instructions on how to access this information.

**Privacy and Courtesy Standards** 

#### Privacy

Family Tree DNA cares about your privacy. When you use our internal database, we can assure your privacy. Please use caution if anyone asks you to export your results to other databases not endorsed by Family Tree DNA. Be sure you have confidence in the privacy protections of whoever is requesting your data. You should not ever share your kit number and password outside of Family Tree DNA.

#### **Courtesy Standards**

Family Tree DNA would like genetic genealogy and DNA testing to be an enjoyable experience for all. We ask that our customers treat each other, project administrators, and Family Tree DNA staff with courtesy and respect. When your matches reach out to you, please respond to them. Even an answer of "I don't know" is better than no answer. At all times, respect the privacy of others. Everyone has his or his own comfort level for sharing genetic information. The best policy is to ask before doing.

#### Resources

#### Useful links:

myFTDNA 2.0 User Guide: Y-DNA - http://www.familytreedna.com/fag/answers.aspx?id=48

Understanding Results: Y-DNA STRs - http://www.familytreedna.com/faq/answers.aspx?id=9

Understanding Results: Y-DNA Haplogroups & SNPs http://www.familytreedna.com/faq/answers.aspx?id=26

Understanding Results: Y-DNA TiP - (Family Tree DNA Time Predictor) http://www.familytreedna.com/faq/answers.aspx?id=51

Understanding Results: Walk Through the Y (WTY) http://www.familytreedna.com/faq/answers.aspx?id=27

Y-DNA Library of Scientific Papers - http://www.familytreedna.com/y-dna-papers.aspx

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Glossary (complete) - http://www.familytreedna.com/faq/answers.aspx?id=21

#### Common Terms

Genetic Cousins - These are individuals whose DNA test results match one another indicating shared genetic ancestry.

Haplogroup - A haplogroup is a major branch on either the maternal or the paternal tree of humankind. Haplogroups are associated with early human migrations. Today, these designations can be associated with a geographic region or regions.

Haplotype - A Y-DNA haplotype is the set of values for a group of Y-DNA STR markers. Two individuals that match exactly on all markers have the same haplotype.

MRCA (Most Recent Common Ancestor) - The ancestor shared most recently between two individuals.

Mutation - Mutations are changes to your DNA code. They are natural copying errors. One analogy is to think of a copy machine that is making many copies of a page. Occasionally, it will make a mistake; an e might look more like an o, for example. This is a mutation. If you then take that page with the o and copy it, it will pass on its mutation to all of its descendant copies. Note, the term mutation in this sense does not refer to anything medical.

Mutation Rate - A mutation rate is how often on average DNA changes. This can be either for a specific STR marker or for a length of genetic code. It is how often small copying errors happen. Note, the term mutation in this sense does not refer to anything medical.

Polymorphism - A Polymorphism is a mutation that has reached a greater than 1% frequency in a local or global population. In genetic genealogy, we most often use it to describe backbone branch defining mutations. These are related to backbone haplogroups. Note, the terms *polymorphism* and *mutation* in this sense do not refer to anything medical.

Polymorphism Rate - A polymorphism rate is how many polymorphisms a given length of DNA is likely to have. It and the mutation rate are related, but it is a measurement of the expected number or polymorphisms for a given length of DNA rather than how often they occur. Note, the terms polymorphism and mutation in this sense do not refer to anything medical.

STR (Short Tandem Repeat) - A short DNA motif (pattern) repeated in tandem. ATGC repeated eleven times would give the marker a value or allele of 11.

TMRCA (Time to Most Recent Common Ancestor) - The amount of time or number of generations since two or more individuals have shared a common ancestor. Since mutations occur at random, the estimate of the TMRCA is not an exact number (i.e., 7 generations), but rather a probability distribution. For Y-DNA, as more STR markers are compared the TMRCA estimate becomes more refined.

X-chromosome - One of the two sex chromosomes, X and Y. X is the sex chromosome that is present in both sexes, singly in males and doubly in females.

Y-Chromosome - One of the two sex chromosomes, X and Y. The Y-Chromosome passes down from father to son. Females do not receive it. As the Y-Chromosome is passed on through the paternal line, it is valuable for surname and male lineage based genealogy studies.

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#### Genealogy Quick Steps

#### **Entering Your Ancestry Information**

Your myFTDNA account has two places for you to enter ancestry information. They are the My Account – Most Distant Ancestors page and the My Account – GEDCOM/Family Tree page. After you enter information, you will be ready to access and contact your matches.

The My Account – Most Distant Ancestors page is where you enter basic information about your most distant known ancestor (MDKA) on your direct paternal line. To do so:

- Log in to your myFTDNA account. (https://www.familytreedna.com/login.aspx)
- 2. On the top menu bar, find the My Account menu.
- 3. From the My Account menu, select Most Distant Ancestors.
- On the My Account Most Distant Ancestors page, look for the Direct Paternal area in the Most Distant Ancestors section.
- In the Direct Paternal area, select the Country of Origin. If you are not sure of this, select Unknown.
- In the Direct Paternal area, enter the name, date of birth, and date of death of your most distantly known direct paternal ancestor in the Name field.
- Click the Save button to save your changes.
- On the My Account Most Distant Ancestors page, find the Direct Paternal area in the Ancestral Locations section.
- 9. Click the Add Location button to launch the Ancestral Locations Map wizard.
- 10. In the Paternal Location section, click the blue Edit Location button.
- Follow the steps to add your direct paternal most distantly known ancestor's location.
   This is usually the place where she was born. If you do not know that, it can also be where she was married or where she died.

The My Account – GEDCOM/Family Tree page is where you upload an electronic file of your pedigree. GEDCOM is the standard format for sharing electronic pedigree information, and almost all genealogy software packages are able to export to standard GEDCOM format (.ged). To upload your file:

- First, you must export your GEDCOM file from the software in which you developed your family tree (you should be able to do this by clicking on File in that program and then Export to save this file to your computer). Please note that Family Tree DNA does not host or offer customer support for any GEDCOM software.
- 2. Log in to your myFTDNA account. (https://www.familytreedna.com/login.aspx)
- 3. On the top menu bar, find the My Account menu.
- From the My Account menu, select GEDCOM/Family Tree.
- On the My Account GEDCOM/Family Tree page, look for the GEDCOM section and the Next button.
- 6. Click on the Next button to start the GEDCOM Upload wizard.

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Appendix I-g: Family Tree DNA – Guide to Understanding Your Paternal Genetic Results



#### **Accessing Your Matches**

You may view your Y-DNA Full Sequence matches on our website by following these steps:

- Log in to your myFTDNA account. (https://www.familytreedna.com/login.aspx)
- 2. On the top menu bar, find the Y-DNA menu.
- From the Y-DNA menu, select Matches.

The Y-DNA – Matches page has two sections. The top Filter Matches section is where you can change the testing level and filter for specific parameters. The bottom Matches section is where you can view your matches. To view your most recent Y-DNA37 matches:

- In the Filter Matches section, change the For: field to Y-DNA37.
- 2. Click the orange Run Report button

By default, your most recent matches will be at the top of the report. You can read about additional settings for the Y-DNA – Matches page by clicking on the blue page Help button at the top of the page.

#### **Ancestral Origins Quick Steps**

#### Y-DNA Haplogroup Information

To find information about your paternal branch (haplogroup):

- Log in to your myFTDNA account. (https://www.familytreedna.com/login.aspx)
- 2. On the top menu bar, find the Y-DNA menu.
- From the Y-DNA menu, select Results.
- On the Y-DNA Haplogroup & SNPs page, description of your haplogroup.

#### Haplogroup and Ancestral Origins Information

To find information about your more recent ancestral origins, begin with the Y-DNA – Haplogroup Origins page:

- Log in to your myFTDNA account. (https://www.familytreedna.com/login.aspx)
- On the top menu bar, find the Y-DNA menu, and then select Haplogroup Origins.
- On the Y-DNA Haplogroup Origins page, look for blue Help button. The help section describes how to interpret the table of populations and frequencies on the page.

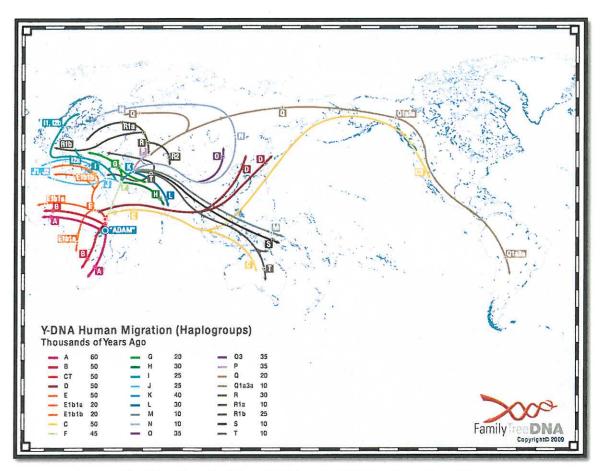
Next, check the Y-DNA - Ancestral Origins page:

- Log in to your myFTDNA account. (https://www.familytreedna.com/login.aspx)
- 2. On the top menu bar, find the Y-DNA menu, and then select Ancestral Origins.
- On the Y-DNA Ancestral Origins page, look for blue Help button. The help section describes how to interpret the table of populations and frequencies on the page.

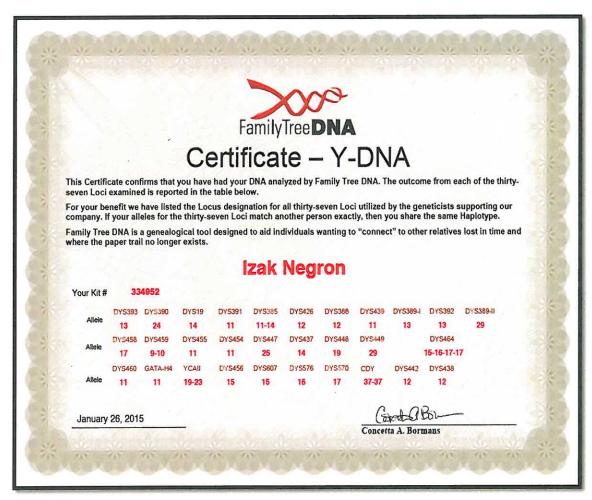
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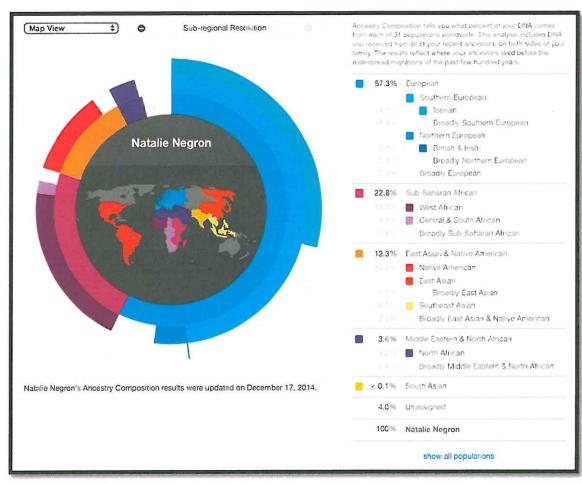
Appendix I-g: Family Tree DNA – Guide to Understanding Your Paternal Genetic Results



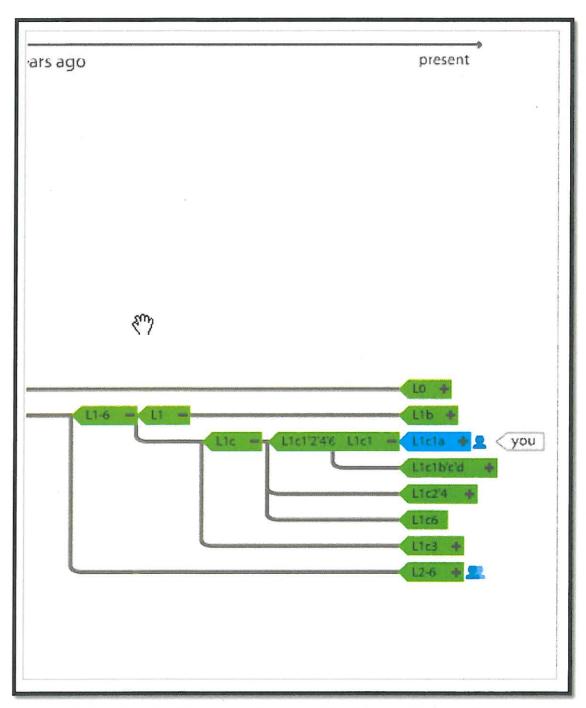
Appendix I-h: Family Tree DNA – Y-DNA Migration Map (Enlarged Version)



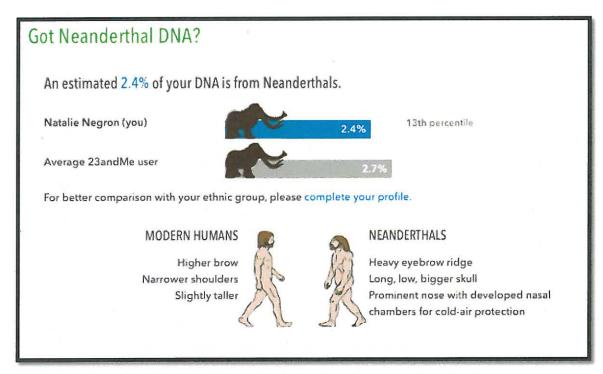
Appendix I-i: Family Tree DNA - Y-DNA Certificate for Izak Negrón



Appendix I-j: 23andMe - Ancestral Composition for Natalie Nicole Negrón



Appendix I-k: 23andMe – Maternal Haplogroup Results for Natalie Nicole Negrón



Appendix I-1: 23andMe – Neanderthal Ancestry Results for Natalie Nicole Negrón

## **APPENDIX II: Historical Documents**

Appendix II-a: 1920 U.S. Census Record (Ramona Rodriguez)

Appendix II-b: Certification of Birth for Angel Manuel Rodriguez (Part of Personal Family Collection, Not Included)

Appendix II-c: Certification of Military Service for Angel Manuel Rodriguez

Appendix II-d: Taxi and Limousine Licenses for Angel Manuel Rodriguez

Appendix II-e: Certification of Birth for Luz Minerva Casado (Part of Personal Family Collection, Not Included)

Appendix II-f: Funeral Booklet for Bernardina Casado Canales

Appendix II-g: Church Records for Luz Minerva Casado

Appendix II-h: Certificate of Marriage for Angel Manuel Rodriguez and Luz Minerva Casado

Appendix II-i: Contract of House Construction for Angel Manuel Rodriguez and Luz Minerva Casado

Appendix II-j: Certificate of Birth for Marilu Rodriguez (Part of Personal Family Collection, Not Included)

Appendix II-k: University Diploma in Elementary Education for Marilu Rodriguez

Appendix II-l: Certificate of Birth for Angel Luis Negrón (Part of Personal Family Collection, Not Included)

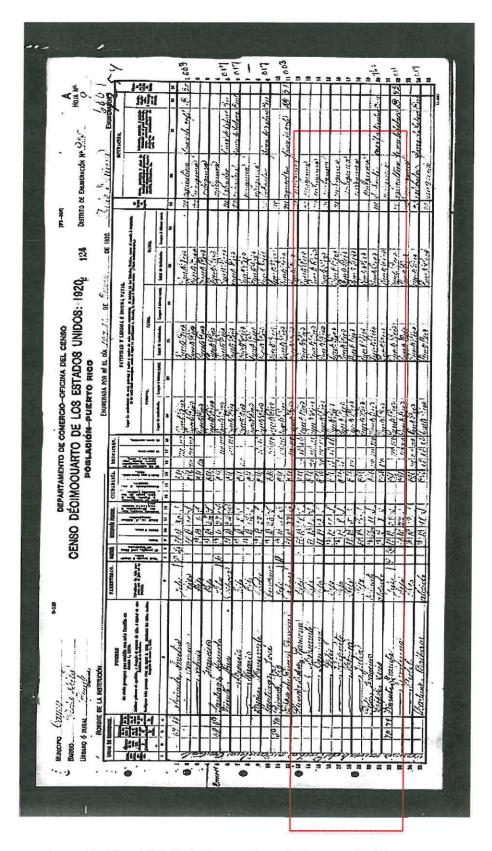
Appendix II-m: Vocational Certificate in Aviation Mechanics for Angel Luis Negrón

Appendix II-n: Vocational Certificate in Automotive Mechanics for Angel Luis Negrón

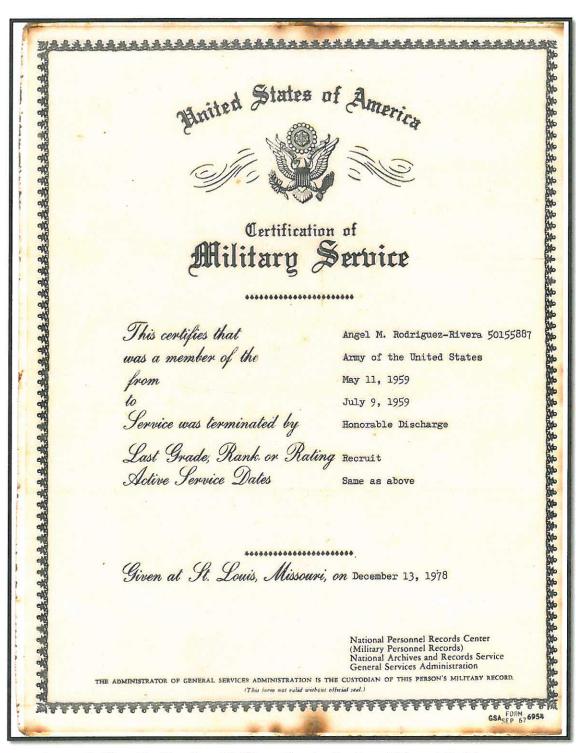
Appendix II-o: Marriage Certificate for Angel Luis Negrón and Marilu Rodriguez

Appendix II-p: Certification of Birth for Natalie Nicole Negrón (Part of Personal Family Collection, Not Included)

Appendix II-q: Certification for Birth for Izak Negrón (Part of Personal Family Collection, Not Included)

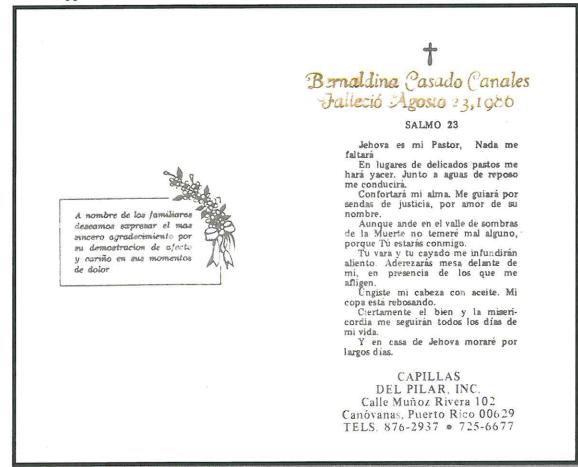


Appendix II-a: 1920 U.S. Census Record (Ramona Rodriguez)



Appendix II-c: Certification of Military Service for Angel Manuel Rodriguez





Appendix II-f: Funeral Booklet for Bernardina Casado Canales

PROGRAMA   10:30 a.m.   10:30	Muchas felicidades para esta familia que ha recibido tan precioso regalo de Navidad.  Nuestro marino Recordemos a nuestro marino en estas Navidades. Su nueva dirección es como sigue. P.F.C. Angel Lausell Jr. 2030739 D.Co. C. E. Bn. Class 15-66 M.C.R.D. San Diego, California	Cantata El viernes próximo celebraremos la cantata de Navidad. El coro ha preparado un repertorio de himnos aproplados para la ocasión. También exhibiremos la película "To Each a Gift"!	Programa de Navidad El domingo próximo invertiremos el programa de la forma siguiente: Servicio de adoración de 10:30 a 11:15 y luego bajamos a la planta baja para el intercamito y regalos a los niños.	No olvide traer su regalo y poneire en nomera a la persona a quien se dedica. Por la tarde los esposos Conde tienen un pequeño agasajo al coro. El viernes 31nos reuniremos desde las 7:30 p.m. para esperar el año con servicio de testimonios. oración, etc.	La hna. INocencia ha dado un buen donativo para la alfombra del pasillo. La hna. Ramonita Agosto donó el arbol de Navidad 7 otros hnos. sus adornos.
o i d	Lectura Antifonal: Exodo 35:20-29 y 36:2-7 Gración intercesoria Hno. José Bermudez Himno congregacional # 2 Adorando con diezmos y ofrendas Gfertorio Himno especial - coro Mensaje - Dr. S. Soto Fontanez	Promesas de mayordomía - Cte. de presupuesto Himno congregacional #32 Sendición - Doxología - Postludio	Nos complacemos en darle la bienvenida al Rev. Fontanez, Director del Trabajo Hispano en las Sociedados Bautistas de la Ciudad y amigo de nuestra iglesia.	La asistencia del domingo pasado: 101  " al Estudio Biblico: 28  Ofrenda regular 4217.61  " Fro-Templo 5.00	

Appendix II-g: Church Records for Luz Minerva Casado

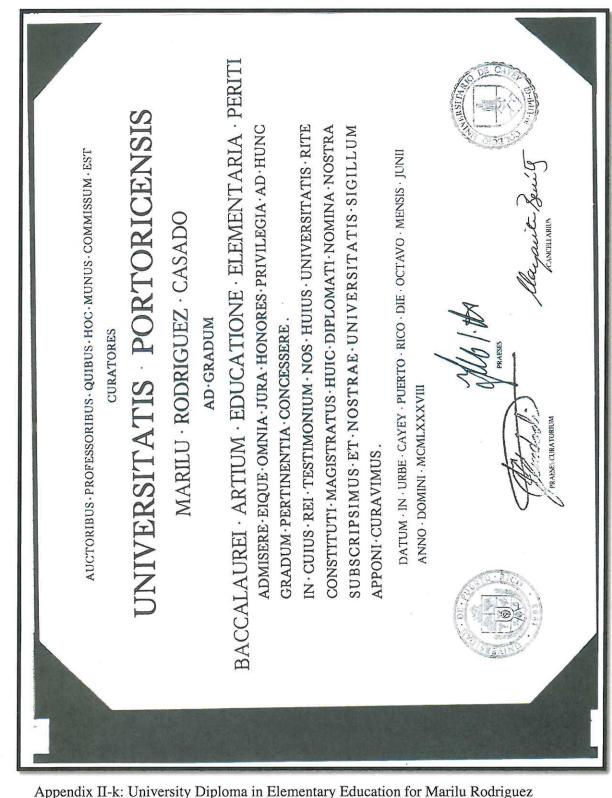
THE CITY OF NEW YORK Marriage License OFFICE OF CITY CLERK No. 30/93 19 64
enane Deputy City Clerk, duly designated by the
Herk of the City of New York to solemnize marriages, Do Hereby Certify that I did on this lay of hereby Certify that I
he Borough of Manhattan, City of New York, State of New York, solemnize the rites of matrimony between
in the Country of My Ostate of My
in the Country of My State of My
as witnesses.
Witness my hand at the office of the City Clerk, Borough of Manhattan, City of New York,  State of New York, this day of American A.D. 19
R.F. 76-8M Sets-701208 (63) Deputy City Clerk of the City of New York, Borough of Manhattan

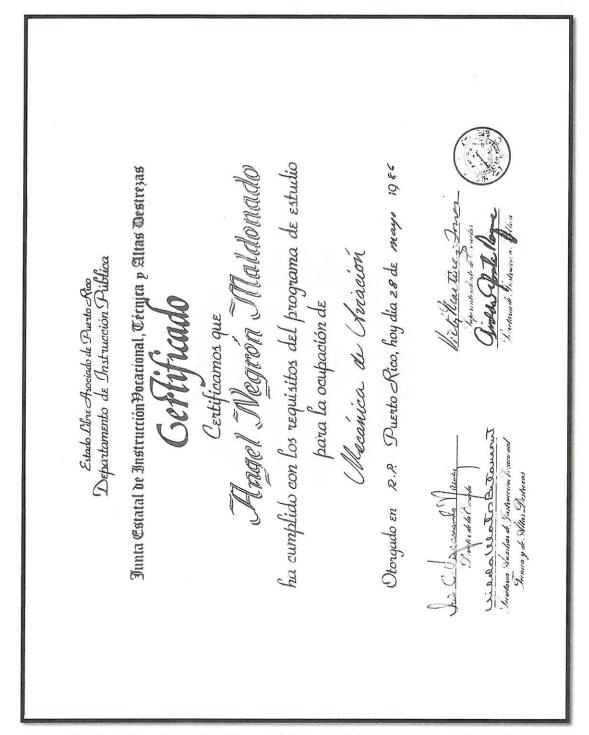
Appendix II-h: Certificate of Marriage for Angel Manuel Rodriguez and Luz Minerva Casado

	cayey, Puerto Rico, a los seis días del mes de sentiamina
	novectentos setenta y nueve,
	TOCK D THE MI
	de Puerto Pico
	de Puerto Rico, con residencia, vecindad y estudio abierto en cayey, Puerto Rico,,,
	Cayey, Puerto Rico.,
	COMPARECEN
	DE LA PRIMERA PARTE: DON ANGEL M. RODRIGUEZ RIVERA Y DONA LUZ
	M. CASADO DE RODRICUEZ, mayores de edad, casados entre sí, propie
	tarios y vecinos Cayey, Fuerto Rico,
	DE LA SECUNDA PARTE: DON LUIS RIVERA TORRES, mayor de edad, ca
	sado, contratista y vecino de Cayey, Fuerto Rico
	EXPONEN
-	PRIMERO. One las comparacientes de la primera parte son dueños
	de un solar con una estructura ubicada en el barrio Matón Abajo en
	el kilómetro cero de Cayey, Puerto Rico
	SEGUNDA: Que los comparecientes tienen convenido un contrato
	de contrucción bajo las siguientes
	PRIMERA: Que el compareciente de la segunda parte se compromete
	a realizar las siguientes terminaciones:
	A: Llenar las c smas,
	By Instalación de vigas
	C: Terminación del tiro de la torta
	SEGUNDA: Que el costo de dicha construcción es de TRES MIL NO
V	VECIENTOS DOLARES (\$3,900.00), los cuales han de ser pagados de la
9	iguiente forma:
-	A: La cantidad de DOS MIL DOLARES (\$2,000.00) en las próximas
	einticuatro horas a patir de la firma de este contrato
	B: La cantidad de MIL DOLARES (\$1,000.00) al terminar la insta
	ación de las vigas y el comienzo del tiro de la torta
	C: La cantidad de MOVECIENTOS DOLARES (SAMO DOL 2) ***********************************

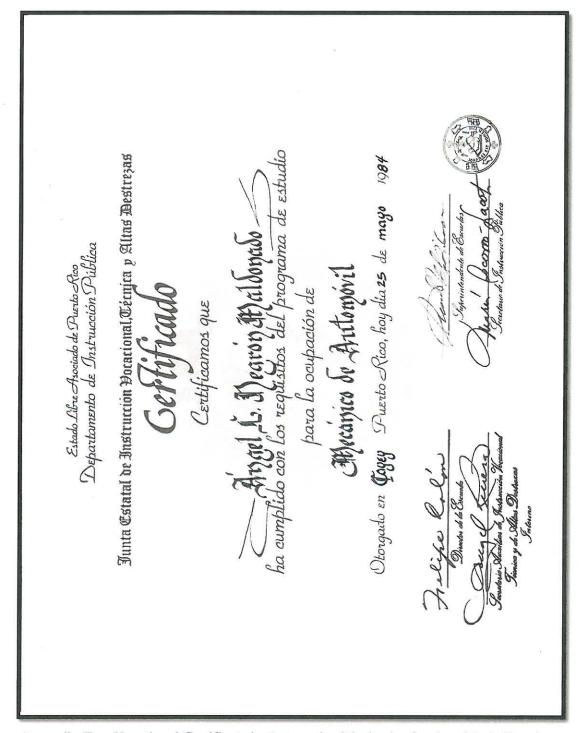
Appendix II-i: Contract of House Construction for Angel Manuel Rodriguez and Luz Minerva Casado

Appendix II-i: Contract of House Construction for Angel Manuel Rodriguez and Luz Minerva Casado





Appendix II-m: Vocational Certificate in Mechanical Aviation for Angel Luis Negrón



Appendix II-n: Vocational Certificate in Automotive Mechanics for Angel Luis Negrón

Mosela RD-to (Rev. 1745) ESTADO LIRRE ASOCIADO DE PUERTO RICO COMMONWEALTH OF PUERTO RICO DEPARTAMENTO DE SALUD - OFASS DEPARTMENT OF HEALTH - HESA Area of Demographic Registry Area de Registro Demográfica CERTIFICADO DE ACTA DE MATRIMONIO Certificate of Marriage Registration Certifica que en la Sección de Matrimonios del Registro a mi cargo aparece la siguiente inscripcion.

Libro Num.

Volume Vir.

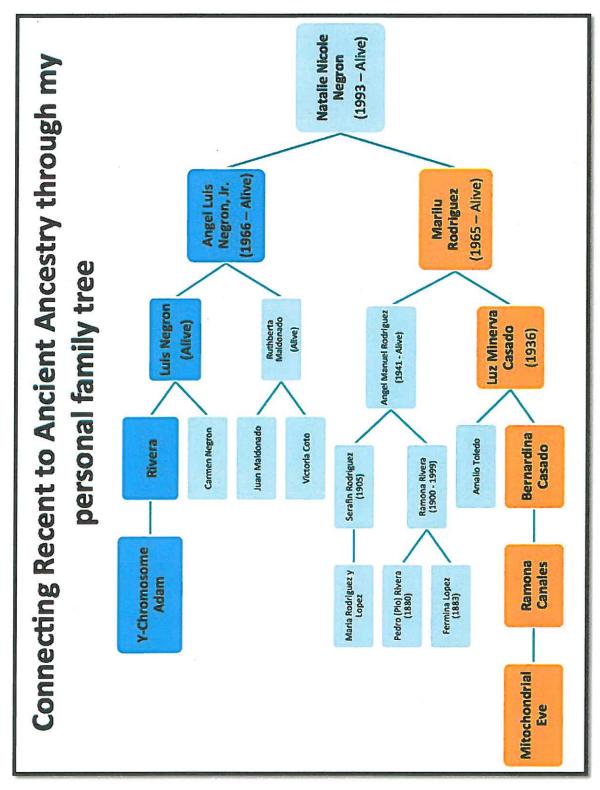
113 Folio Num.

Folio Num. I certify that in the Section of Marriages of the Registry under my custody the following marriage is registered. Municipio donde se cetebro el matrimonio: Municipality where merriage was performed Cagues, P.R. Fecha de Celebración Date of Celebration 2 junio ..5..... Nombre del Contravente Edad 23 años Angel Luis Negron Maldonedo Visido Divorcia Cayey, P.R. (State or Country) Estado o País Edad 24 años Marilu Rodriguez Casado Estado Civil Civil Status Cayey, P.R. Natural de Jirthplace Estado o Pais (State or Country) Nombre del Celebranie: Nume of Officialing Party Jose Angel Lopez Rios DATOS SOBRE EL SOLICITANTE Information on Applicant Si veterano, licencismiento honorable Num Il veteran, honorable discharge Number Iuz. M. Casado Rodriguez Direction Address Cayey, P.R. Si para uso oficial de una Agencia del Gobierno Estatal o Federal.

If for official use of an agency of the Commonwealth of Fuerto Rico or Federal Government Number de la Age Number of Agency ADVERTENCIA Este certificado, no será valido a est el mismo no se ha estampado el sello oficial del Hegisaro Demográfico correspondiente. Lembien se requiere la cancelación de un sello de remas internas de 22 segue expe la ley, excepto en aquellos casos en que el certificado se explita para uso exclusivo de un verezono o de ona seguencia de gobierna del Estado Inire Asociado de Puerto Rice o de los Estados Unidos de America. LA MERA POSESION DE ESTE DOCUMENTO, NO DEBE CONSTITUIR BASE PARA IDENTIFICAR AL PORTADOR COMO ENO DE LOS CONTRA ENTES EN ESTE MATULMONIO. This certificate is sold if the official seal of the corresponding Demographic Registry Office has not been stamped hereon. Also a \$2 internal receive stamp is required by law to be causeful of orces, unless this certificate is issued for the exclusion use of a velocity of the Commonwealth of Fuerto Rico or of the Federal Government of the United States THE WIRE FORSESSION OF THIS DOCUMENT SHOULD NOT CONSTITUTE ENOUGH EVIDENCE TO IDENTIFY THE PERMER AS ONE OF THE PARTIES IN THIS MARKILOR. ESTE CERTIFICADO NO SERA VALIDO SEEN EL MISMO APARECEN TACHADURAS, EORRADURAS O ALTERACIONES. par di Expedición 28 de junio 1990 Municipio de Municipality of Puerto Rico Caguas Fuerto Rico Encargado del Elegistos 22 al. graciosi) Aida Iuz Rivera Rodriguez Typed Name 3, 10-04-201 S0,000-29-1GPR

Appendix II-o: Marriage Certificate for Angel Luis Negrón and Marilu Rodriguez

# **APPENDIX III: Personal Family Tree**



Personal Family Tree