

**Genelex Laboratory #:**2XXXX-14 **Participant:** Jane Doe

## **Summary of Findings:**

The results of the mtDNA sequencing test for Jane Doe found differences with the Cambridge Reference Sequence at positions 16314 and 16356. The mtDNA sequence of Jane Doe. does not match sequences from the Native American database.

### **Results:**

The mtDNA signature may be used to identify an individual and their direct maternal lineage. The mtDNA signature may also be helpful in identifying ethnic origins. A portion of the hypervariable region I (HVS1) from the DNA in the mitochondrial genomome were amplified and sequenced from base pair positions 16100-16383. This sequence was then systematically compared to a database of HVSI sequences that match exactly or are one mutation step away from your HVS1 sequence. This mtDNA sequence data was also compared to the Cambridge Reference Sequence (CRS). By comparing your sequence to the CRS we can identify the name of the lineage to which you belong. These lineages are called haplogroups. Any differences from the Cambridge Reference Sequence outside of the stated region will not be detected by using this test. This lineage information was then referenced with current mtDNA haplogroups and archaeological information sources to provide a description of the origins of the maternal line.

MtDNA sequence for Jane Doe (nucleotide positions 16001-16383):

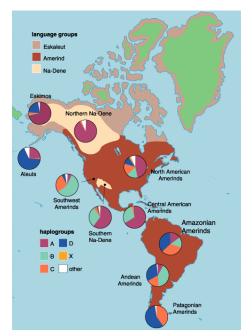
**Haplogroup Assignment**: You most probably belong to Haplogroup U4, a lineage most frequent in Central and South East Asia (see attached report). This genotyping would have to be confirmed through typing the hypervariable region II from the mtDNA. Current mtDNA haplogroup and archaeological information sources to provide a description of the origins of the maternal line are attached in a separate report.

# Native American Haplogroup Description:

Nearly all Native Americans belong to one of five mtDNA haplogroups: A, B, C, D or X which are broadly distributed throughout the Americas. All five haplogroups are observed in populations throughout the Americas and are also found in the three proposed Native American linguistic groups (Amerind, Na-Dene, Eskaleut). However, the original Na-Dene Indians and Eskimo-Aleuts appear to have lacked haplogroup B. Although mtDNAs from haplogroups A, B, C, D or X are often found together in a single population, many tribes lack at least one of these



haplogroups. Tribal isolation and founder effects may have led to the divergence of tribal gene pools in different regions.



Additional other mtDNAs have been detected in Amerindian tribes, all of which appear to have different genetic affinities. Because many Native populations exhibit European admixture in nuclear genetic studies, it seems likely that some of their "other" mtDNAs were acquired through gene flow with modern Europeans. Indeed, this does appear to be the case, as European haplogroup H and T mtDNAs are found in the Ojibwa, and European haplogroup H and J mtDNAs are seen in the Cherokee. There has also been some African gene flow into certain Amerindian populations, because African slaves and their descendents intermixed with Native Americans in historical times. Evidence of African-Amerindian admixture has been detected in Seminoles of Florida and the Narragansett of Massachusetts, in the form of African haplogroup L mtDNAs. Similar observations have been made for the Mixatec and Zapotec Indians of Southern Mexico. African mtDNA haplotypes may also be

present among other Native American groups. The remaining "other" mtDNAs exhibit features similar to those seen in the majority of Asian haplogroups. The great majority of these "Asian" haplogroups have been observed in South American tribes from the Brazilian Amazon, such as the Makiritare and Yanomami, but a few have also been detected at very low frequencies in a handful of North American groups.

# Prehistorical History of Native American Haplogroups:

According to genetic theory, all humans descend from a woman nicknamed "African Eve." This unknown woman probably lived in Africa, perhaps in Kenya, about 145,000 years ago (~5,700 generations). She was a very distant cousin of the Neanderthals, but unlike them, she became an ancestor of all modern humans. She wasn't the only human female in her generation -- some scientists think that she belonged to a human population of about 2,000 people. The other women of her time certainly left daughters and granddaughters, but African Eve was the only woman in her generation whose descendants in the female line are still living today.

Geneticists divide African Eve's descendants into haplogroups popularly called "clans" to make the subject easier for lay people to understand. Different types of mtDNA correspond to different haplogroups. Currently, there are only 33 major haplogroups. Again, there have been different maternal lines in existence in human history, but these are the only lines that can be found in existence today. Major haplogroups are continental or ethically specific. Three of them (L1, L2 and L3) group sub-Saharan African lineages; nine (H, I, J, K, N1b, T, U, V, W and X)



encompass almost all of mtDNAs from European, North African and Western Asian Caucasians. Finally, haplogroups A, B, C, D, E, F, G and M cover the majority of lineages described for Asia, Oceania and Native Americans. Central East Asian populations exhibit all five lineages common to Native American populations. The Altai of southern Siberia, near Lake Baikal, are the only known modern ethnic group whose membership represents all five Native American founding haplogroups.



MtDNA analysis of aboriginal Siberian populations has revealed that haplogroups C and D are widely distributed throughout Siberia, whereas haplogroup A reaches its highest frequency in the Chukchi of the Chukotoka peninsula adjacent to Alaska. As humans moved northwards, only A, C and D predominated. These three mtDNAs crossed the Bering land bridge to give rise to the Native Americans. The presence of Haplogroup B throughout coastal Asia raises the possibility that it cam to the Americas via coastal migration. HaplogroupX also represent as ancient migration from Asia or Europe to the Americas.

The earliest inhabitants of Beringia possessed a limited number of founding mtDNA haplogroups. The "reindeer" Chukchi had haplogroups A, C and D while the Siberian Eskimos only showed haplogroups A and D. Beringia existed during a time when much of Canada was covered in massive sheets of ice. Because of its arid climate, Beringia remained untouched by glaciers, allowing people to migrate to the New World. After the ice age, about 10,000 years ago,



the Bering land bridge disappeared separateing Chukotka and Alaska. Eventually, a few distinct populations evolved in the northern Pacific Rim of America and presumably reflected the Paeleoasiatic-speaking Chukchi, Na-Dene-speaking Indians and Eskimo-Aleuts.



### Migration to the Americas:

A widely accepted theory about the arrival of humans in North America has proposed that a wide ice-free corridor east of the Rocky Mountains split the giant ice sheet that covered Canada towards the end of the last Ice Age. Human populations would have crossed the Beringia land bridge and moved down through Alaska into the rest of North America about 12,000 years ago, when the glaciers receded, probably following the movement of prey animals. New research involving genealogy and archaeology, has cast doubt on this idea as it left too little time for migration through the Americas. Since the Monte Verde site in Chile puts humans in South America thousands of years before the opening of the Canadian ice sheet, these early human populations must have found a different entryway into the Americas. Convincing evidence suggests that humans were using watercraft 40,000 years ago, and recently, scientists have suggested coastal-boat "leapfrogging" that bypasses the glacial barriers. Your ancestors would have exploited the rich food environment provided by marine mammals, fishing and the gathering of shellfish.



Nearly all Native Americans belong to one of five mtDNA haplogroups: A, B, C, D or X which are broadly distributed throughout the Americas. Based on studies of Native Americans DNA, scientists have arrived at many different and often contradictory conclusions about the number and sequence of migrations from Asia to the Americas. A broad range of dates has been estimated (43,000 - 11,000 years ago). Early studies suggested that the Americas were peopled multiple migrations. Torrini suggested that the initial wave of immigrants consisted only of individuals having haplogroups A, C and D (34,000-26,000), while a later wave brought haplogroup B (15,000-12,000) and a third wave brought the Na-Dene (9,000 - 7,200). More recently single migration models support a continuous flow of

small groups across Beringia to America in search of food and other resources (30,000 - 20,000) years ago). Male genealogical studies of the Y-chromosome, support this initial large single wave and also a later wave from eastern Siberia to form the Na-Dene and northern Amerindian groups. Although there is still no agreement on the number of founding migrations, the exact age of the migrations, or the combination of Native American groupings, there is unqualified support for an Asian origin to early Americans and an early migration to America of approximately 30,000 - 20,000 years ago.

This information is meant to give you a plausible snapshot of what life was like when and where your maternal line originated. It combines the results of ongoing archaeological, linguistic and genetic research. Because the study of human pre-history is not exact and must rely on assumptions, scientists may disagree about the best interpretation of existing knowledge. As additional research results become available our assumptions may be updated or change completely. Your maternal inheritance is a small part of your overall inheritance but provides you with one of the clearest earliest views of your ancestry. It's like finding an especially



beautiful and informative artifact in the remains of an ancient village or campsite. Genelex hopes that this information has been exciting and informative to you. We are honored to have played a role in your search for your genetic ancestors.

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