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**DNA Tribes® Digest Spring, 2015**  
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## Exploring the Genetic Relationships of Greenlandic Populations with DNA Tribes® World Regions and Individual Populations

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### Introduction

It is our pleasure to welcome you to the Spring, 2015 issue of DNA Tribes® Digest. This is the first issue of our Digest since September, 2014 as due to the unexpected passing away of the company’s co-founder, Lucas Martin, we had temporarily suspended the publishing of new Digests. We appreciate the outpouring of support and encouragement from our customers following Mr. Martin’s death and are committed to building upon his legacy of Digest articles that, in a scientifically rigorous way, explore world genetic structure and complex genetic relationships among various populations.

The first of our quarterly Digests presents the results of our analyses of newly published STR marker data for native Greenlandic Inuit populations and explores the relationships of these populations with DNA Tribes® World Regions and approximately 1,300 populations in our STR population reference database.

Sincerely,  
**Dr. Eddie Valaitis**  
Scientific Adviser and Co-Founder of DNA Tribes

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## Background

Greenland (Kalaallit Nunaat in Greenlandic) is an autonomous territory within Denmark that is located between the Arctic and Atlantic Oceans, east of the Canadian Arctic Archipelago. Greenland has been inhabited for the last 4,000 – 5,000 years by 1) various Arctic populations that are understood to have migrated to Greenland from the present day Alaska and Canada and 2) Norse populations from Europe. Table 1 summarizes the settlement of Greenland by various cultures.

Period	Settlement Summary
<b>Saqqaq &amp; Independence I</b> <b>2,500 – 800 BC</b>	Southern and western Greenland is inhabited by populations associated with the <b>Saqqaq</b> culture; the culture exploits both marine and terrestrial resources using small tools made out of stone and bone. <sup>1</sup> Genetic evidence suggests that the earliest migrants into Greenland were descended from populations in the Bering Sea area and not directly related to later inhabitants that replaced them. <sup>2</sup> Starting in 1,300 BC, the <b>Independence I</b> culture is attested in northern Greenland with appearance of small towns such as Deltaterrasserne. <sup>3</sup>
<b>Dorset</b> <b>800 BC –</b> <b>1,500 AD</b>	The Saqqaq culture is replaced with the <b>Dorset</b> culture – which is the first culture to eventually spread across both western and eastern Greenlandic coasts. <sup>4</sup> These people live primarily on hunting of caribou and whales.
<b>Norse (European)</b> <b>986 AD –</b> <b>1,400/1,500 AD</b>	<b>Icelanders</b> and <b>Norwegians (Norse)</b> reach Greenland in 986 AD in 14 boats led by Erik the Red during a period of relatively mild climate in the Arctic. <sup>5</sup> They establish three settlements near the southwestern-most area of the island; these settlements disappear sometime in the 15th century, perhaps at the onset of the Little Ice Age which made the practice of agriculture for the Norse settlers nearly impossible. <sup>6</sup>
<b>Thule (Inuit)</b> <b>1,300 AD -</b> <b>Present</b>	The <b>Thule (Inuit)</b> culture, that originated in Alaska, reaches Greenland and introduces various technological innovations in transportation (dog sleds), hunting techniques and tools (harpoon points made out of whale bone). <sup>7</sup> Recent genetic analyses suggest that the expanding Thule groups encountered and interbred with the existing Dorset populations in Canada and Greenland. <sup>8</sup>

**Table 1:** Summary of the various human migrations and settlements in Greenland.

<sup>1</sup> Gotfredsen, Anne Birgitte, and Tinna Møbjerg. Nipisat-a Saqqaq Culture Site in Sisimiut, Central West Greenland. Museum Tusulanum Press, 2004.

<sup>2</sup> Gilbert, M. Thomas P., et al. "Paleo-Eskimo mtDNA genome reveals matrilineal discontinuity in Greenland." Science 320.5884 (2008): 1787-1789.

<sup>3</sup> Rasch, M.; Jensen, J. F. (1997). "Ancient Eskimo dwelling sites and Holocene relative sea-level changes in southern Disko Bugt, central West Greenland". Polar Research 16 (2): 101–115.

<sup>4</sup> Ramsden, P.; Tuck, J. A. (2001). "A Comment on the Pre-Dorset/Dorset Transition in the Eastern Arctic". Anthropological Papers of the University of Alaska New Series 1: 7–11.

<sup>5</sup> Ogilvie, Astrid EJ, Lisa K. Barlow, and A. E. Jennings. "North Atlantic climate c. AD 1000: Millennial reflections on the Viking discoveries of Iceland, Greenland and North America." Weather 55.2 (2000): 34-45.

<sup>6</sup> Buckland, Paul C., et al. "Bioarchaeological and climatological evidence for the fate of Norse farmers in medieval Greenland." Antiquity 70.267 (1996): 88-96.

<sup>7</sup> Fagan, Brian M. Ancient North America: The Archaeology of a Continent. 2nd Ed. New York: Thames and Hudson, 1991. P. 196

<sup>8</sup> Helgason, Agnar, et al. "mtDNA variation in Inuit populations of Greenland and Canada: migration history and population structure." American Journal of Physical Anthropology 130.1 (2006): 123-134.

## Data

STR marker data for indigenous Greenlandic Inuit individuals from two academic articles were pooled resulting in a sample size of 700 people.<sup>9,10</sup> The published data overlapped with the DNA Tribes® population reference database on 21 STR markers.<sup>11</sup> Figure 1 summarizes the geographical distribution of Greenlandic samples and shows that the majority of samples are from the western and southern coasts of Greenland with only one sample from eastern Greenland (Ammassalik Island).



**Figure 1:** Geographical distribution of Greenlandic samples. Satellite image courtesy NASA's Earth Observatory

## Relationship of Pooled Greenlandic Population with DNA Tribes® World Regions

### MDS Analysis

When summarizing world genetic variation using 21 STR markers and applying multidimensional scaling (MDS) technique, the first two most informative dimensions (Dimensions 1 and 2) separate all DNA Tribes® World Regions into three clusters that can be described as Sub-Saharan African, Native American and Other (see Figure 2).<sup>12</sup> The pooled Greenlandic population (GR) is genetically closest to the Arctic populations (NR in the plot; the Arctic region consists of native Inuit and Yupik populations of Alaska), which confirms the well-established theory of Thule (Inuit) migration into Greenland dating back to 1,300 AD. In addition, the Greenlandic populations are farther from Other World Regions

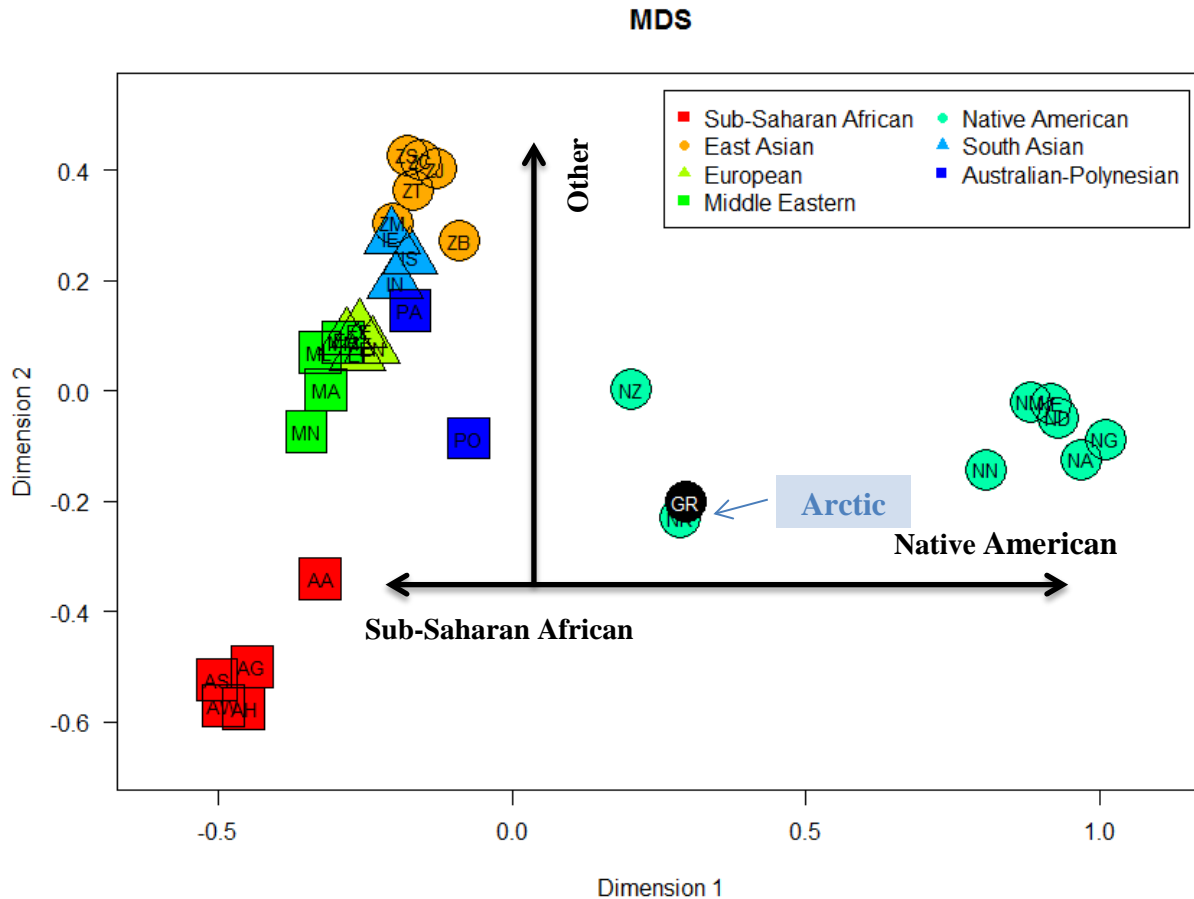
<sup>9</sup> Pereira, Vania, et al. "The peopling of Greenland: further insights from the analysis of genetic diversity using autosomal and X-chromosomal markers." *European Journal of Human Genetics* 23.2 (2015): 245-251.

<sup>10</sup> Tomas, C., et al. "Concordance study and population frequencies for 16 autosomal STRs analyzed with PowerPlex® ESI 17 and AmpFℓSTR® NGM Select™ in Somalis, Danes and Greenlanders." *Forensic Science International: Genetics* 11 (2014): e18-e21.

<sup>11</sup> The overlapping STR markers are: D3S1358, VWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, D7S820, D16S539, TH01, TPOX, CSF1PO, D2S1338, D19S433, D1S1656 and D2S441 [18] D10S1248 D12S391 D22S1045 SE33

<sup>12</sup> MDS is a dimensionality reduction technique often-used to visualize high-dimensional data in fewer dimensions.

compared to the Mestizo (NZ) World Region, which is comprised of mixed Native American and mostly European populations, yet they are closer to these Other World Regions than all other Native American World Regions. This indicates that the Inuit populations of Greenland retain potentially archaic proto-Asian genetic signatures linking them to East Asian World Regions as well as traces of European admixture.



**Figure 2:** Representation of world genetic structure using 21 STR markers for DNA Tribes® World Regions and the first two most informative dimensions from MDS analyses. Refer to Table 2 for abbreviations.

When DNA Tribes World Regions are plotted against Dimensions 1 and 3 (in Figure 3), which correspond roughly to Other and Native American division (Dimension 1) and East Asian and European division (Dimension 2), the Greenlandic populations:

- are still very close to the Arctic populations (NR); and
- are much closer to the East Asian populations than other Native American World Regions; this implies that the Saqqaq and/or Dorset cultures could have migrated into Greenland from Siberia via the Bering Sea and Alaska.

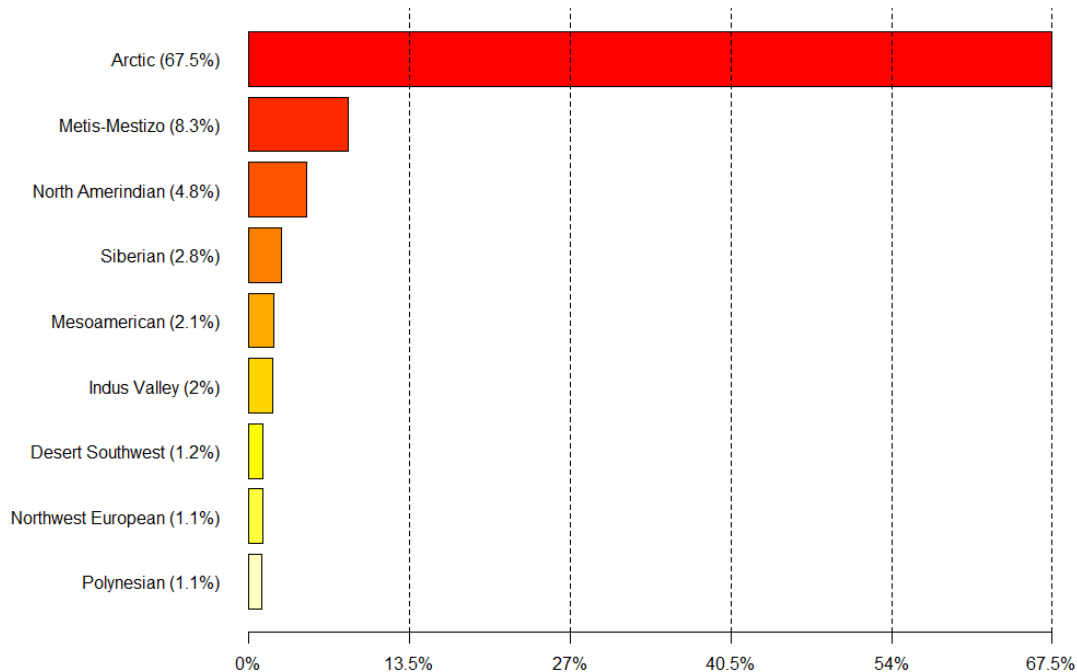
It is worth noting that while the Mestizo World Region (NZ) is close to the European World Regions, the pooled Greenlandic populations and the Arctic World Region (NR) are quite far from European World Regions indicating little mixture with Norse settlers.



### Top World Region Matches (including Arctic Region)

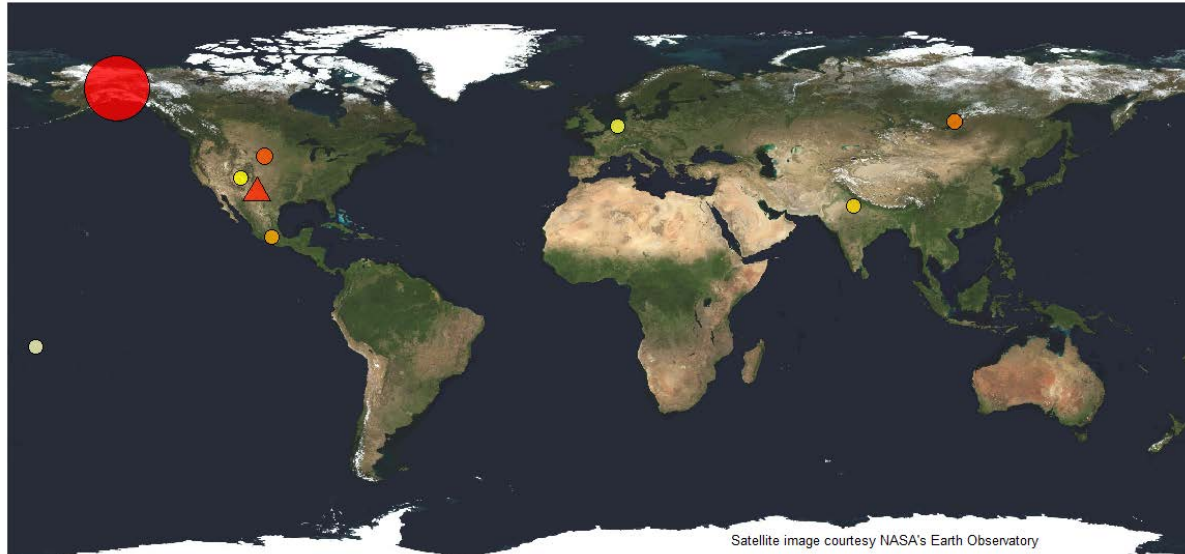
A statistical simulation was used to generate 5,000 synthetic Greenlandic profiles from the pooled Greenlandic population. Then, for each generated profile, a top DNA Tribes® World Region match was computed and retained (based on the highest MLI score for each profile). Figure 4.a summarizes the results of this analysis and shows that:

- 67.5% of simulated profiles have their top match with the Arctic World Region (consisting of Alaskan Inuit and Yupik individuals); this finding attests the expansion of the Thule culture from Alaska into Greenland;
- for 8.3% of Greenlandic profiles the top World Region match is with the Metis-Mestizo World Region (consisting of mostly mixed Native American and European populations). These matches may indicate that some modern Greenland Inuits have traces of European ancestry; however it is not possible to determine whether this European ancestry dates back to the Norse settlers (Middle Ages) or it is the product of admixture occurring in modern times;
- for 4.8% of the simulated profiles the top World Region match is with the North Amerindian World Region (containing Salishan, Athabaskan, and Algonquian speaking cultures of North America); the somewhat low top match incidence shows that the native Greenlandic populations are substantially different from the modern North Amerindian populations;
- for 2.8% of Greenlandic profiles the top World Region match is Siberian, which suggests that some of the modern Greenland inhabitants retain genetic signatures from ancient populations that used to inhabit the Asian continent in the Bering Sea area; and that older Greenlandic cultures (Dorset and even maybe Saqqaq) that were likely genetically more closely related to the ancient Asian populations may not had been replaced by later migrating cultures (Thule), but rather interbred with them to a certain degree.



**Figure 4.a:** Top World Region matches for 5,000 simulated Greenlandic individuals.

Figure 4.b depicts the geographical distribution of top matches for the simulated Greenlandic profiles and illustrates that the majority of top matches are with the Native American World Regions that are relatively geographically close to Greenland with some more distant World Regions (e.g., Polynesian and Northwest European) among the top matches as well.



**Figure 4.b:** Geographical distribution of top World Region matches for 5,000 simulated Greenlandic individuals. Metis-Mestizo World Region is denoted with a triangle.

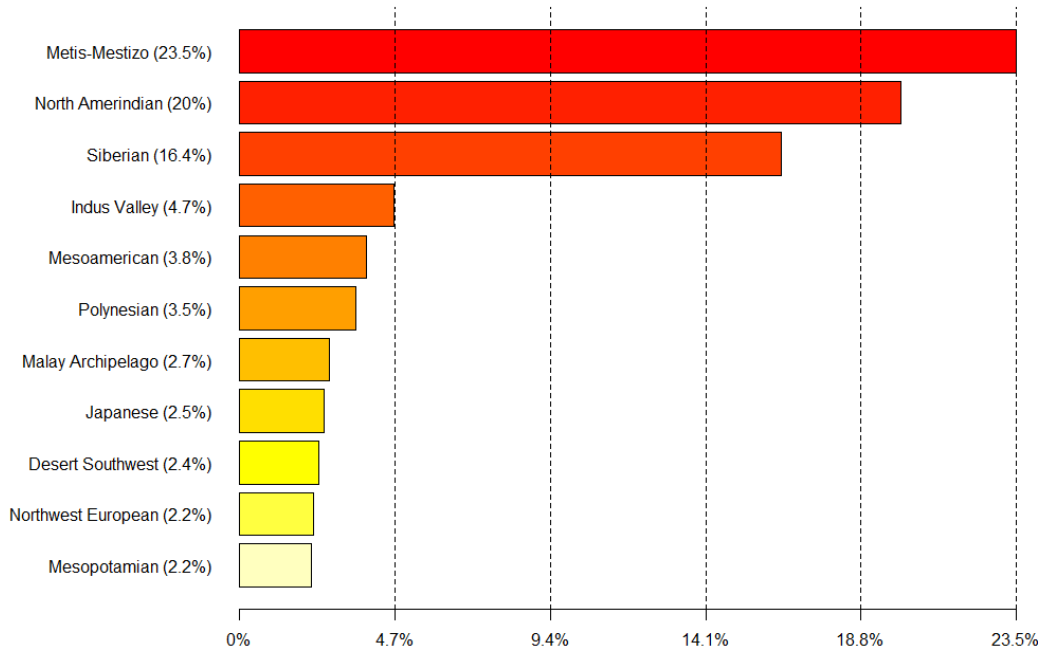
### *Top World Region Matches (excluding Arctic Region)*

To further assess the deeper genetic connections of Greenland Inuit populations to DNA Tribes® World Regions, the Arctic World Region was excluded when re-calculating the top matches for the 5,000 synthetic individuals. The resulting top matches are provided in Figure 5.a. and show that:

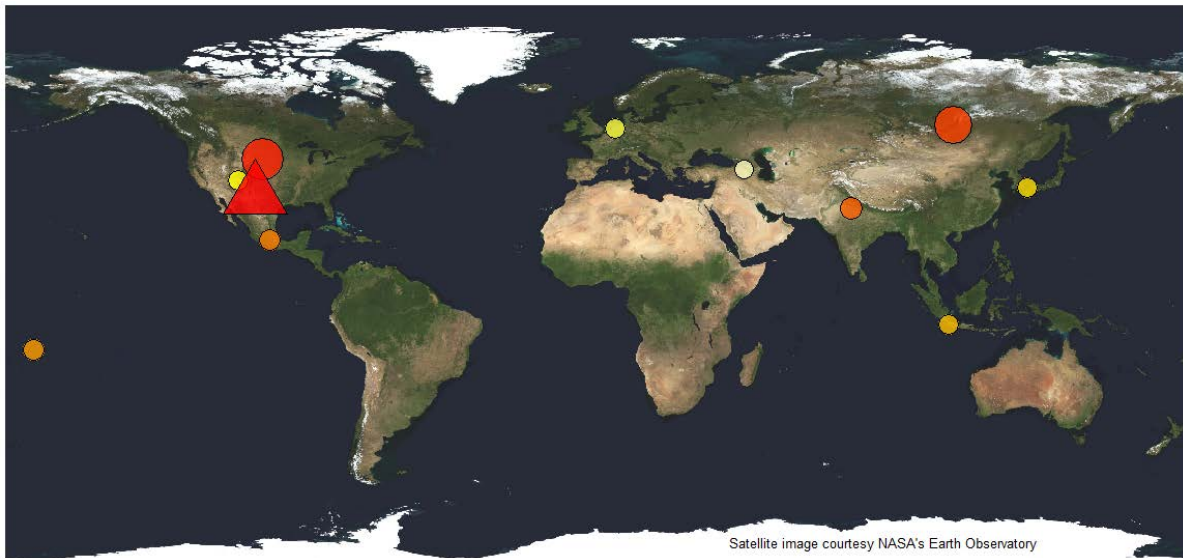
- the top three regions account for 60% of all individual's top matches and include Metis-Mestizo (23.5%), North Amerindian (20%), and Siberian (16.4%) World Regions and confirm the results obtained when the Arctic World Region is included;
- Indus Valley is the fourth most frequent top World Region match; this region consists of populations from the Northern Indian Subcontinent and Southern Central Asia (e.g., Tajikistan and Afghanistan). The matches of Greenlandic profiles with this World Region can be interpreted as an extension of the Siberian matches, as the latter region is genetically relatively close to the Indus Valley World Region;
- the next strongest matches with a non-Native American World Region are with the Polynesian World Region. This distant World Region is hypothesized to have been colonized around 1,000 BC by populations who left Taiwan around 3,000 BC.<sup>13</sup> These proto-Taiwanese are likely to have also been related to ancient Asian populations that migrated via the Bering Sea into Alaska/North America (at an earlier period). Therefore, these seemingly unlikely matches are sensible from the perspective of deep genetic signatures that, in highly endogamous (inter-

<sup>13</sup> Kayser, M.; Brauer, S.; Weiss, G.; Underhill, P.; Roewer, L.; Schiefenhövel, W.; Stoneking, M. (2000). "Melanesian origin of Polynesian Y chromosomes". *Current Biology* **10** (20): 1237.

breeding) populations such as Greenland, can be preserved over hundreds and thousands of years.



**Figure 5.a:** Top World Region matches for 5,000 simulated Greenlandic individuals when the Arctic World Region is excluded.



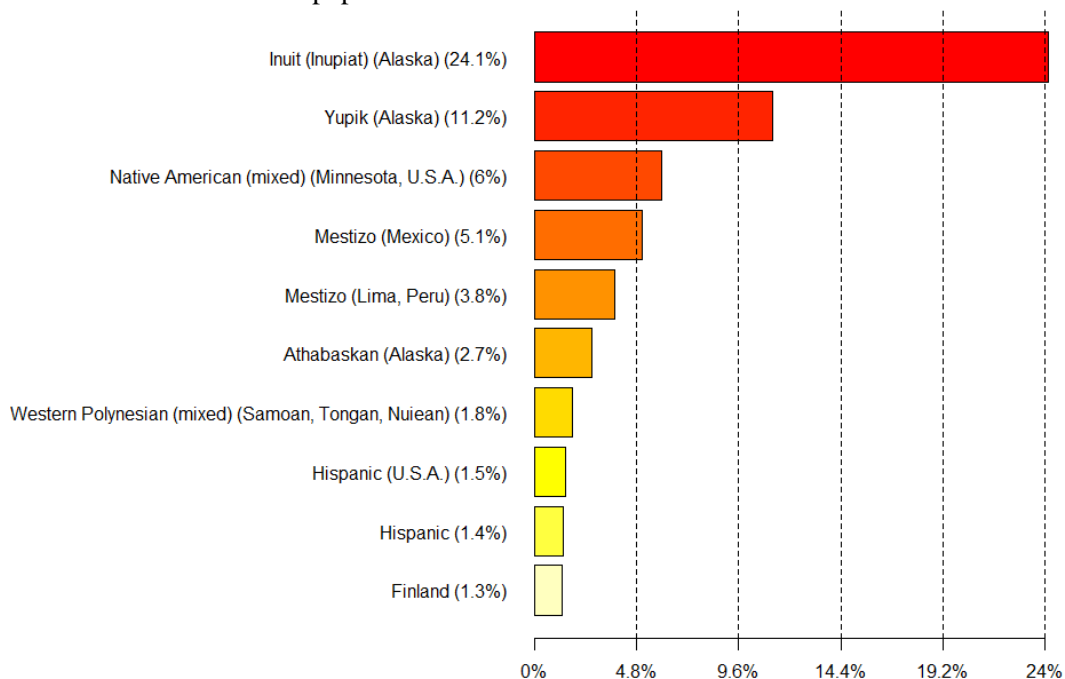
**Figure 5.b:** Geographical distribution of top World Region matches for 5,000 simulated Greenlandic individuals when the Arctic World Region is excluded. Metis-Mestizo World Region is denoted with a triangle.



## Relationship of Pooled Greenlandic Population with Individual Populations in DNA Tribes® Reference Database

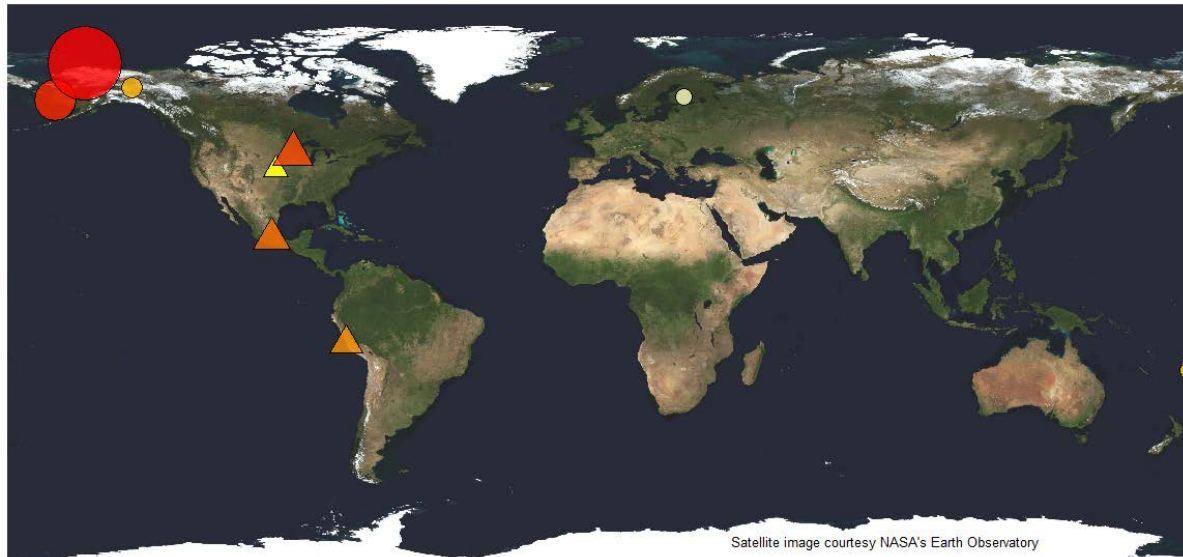
The 5,000 simulated Greenlandic samples were further compared to approximately 1,300 populations (both native and Diaspora) in the DNA Tribes® population reference database. Figure 6.a summarizes the results and shows that:

- some of the most frequent population matches for the simulated profiles are with the native Alaskan populations: Inuit (24.1%), Yupik (11.2%) and Athabaskan (2.7%); this is consistent with the historically attested Thule culture expansions from Alaska into Greenland;
- top matches also include a northern Native American population from Minnesota (exhibiting some admixture with Europeans) as well as Mestizo and Hispanic populations from Mexico, Peru and the U.S.A. (mixed Native American and European populations). This implies that there exist non-negligible European genetic signatures in modern Greenlandic populations; again, it is not possible to determine whether these signatures date back to the early (Middle Age) Norse settlers or reflect admixture from modern times;
- the Western Polynesian population (consisting of mixed sample of native Samoan, Tonga and Nuiean individuals) is the most frequent top match outside of the Americas (1.8% of top matches). This is consistent with the previously discussed connection of some Greenlandic individuals to the Polynesian World Region. Moreover, this attests to some individuals from Greenland having quite distinct genetic profiles from Native American populations (when Greenlandic populations are excluded from analyses);
- Finland is the top population match for 1.3% of Greenlandic individuals; this may imply that the native Finnish (Lapp) signatures in modern Finnish samples share some commonalities with the modern Greenlandic populations.



**Figure 6.a:** Top population matches for 5,000 simulated Greenlandic individuals when Greenlandic populations are excluded.

Figure 6.b exhibits the geographical distribution of top population matches for the simulated Greenlandic individuals; mixed Native American and European populations are denoted with a triangle. The only non-mixed Native American populations that Greenlandic profiles have top matches with are located in Alaska.

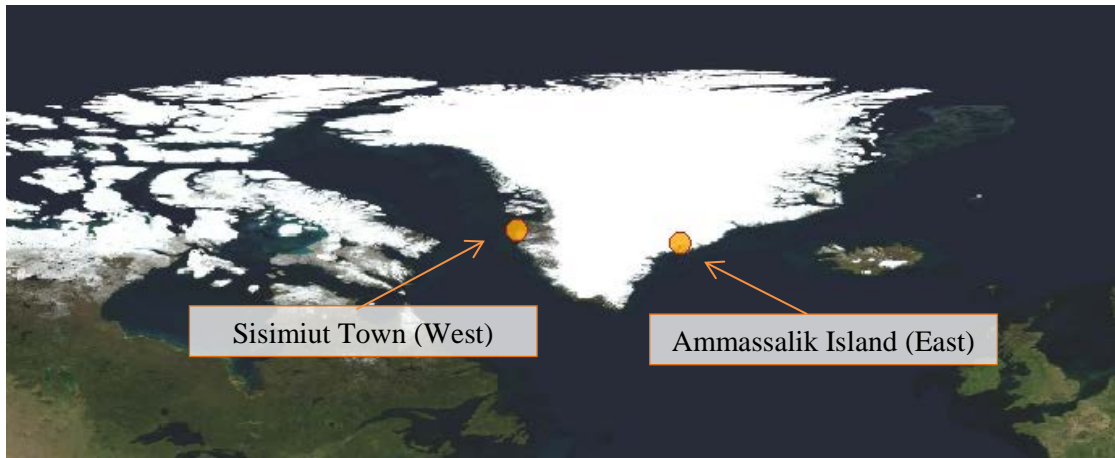


**Figure 6.b:** Geographical distribution of top population matches for 5,000 simulated Greenlandic individuals when Greenlandic populations are excluded. Mixed Native American and European (Mestizo) populations are denoted with a triangle.

## Comparing Connections of Eastern and Western Greenlandic Populations to World Regions and Individual Populations

In addition to assessing the genetic connections of the pooled Greenlandic population to the World Regions and individual populations in the DNA Tribes® population reference database, two individual Greenlandic Inuit populations were assessed by simulating 5,000 synthetic profiles from each population and calculating top World Region matches for each individual. The location of two individual Greenlandic populations is depicted in Figure 7 and they can be described as follows:

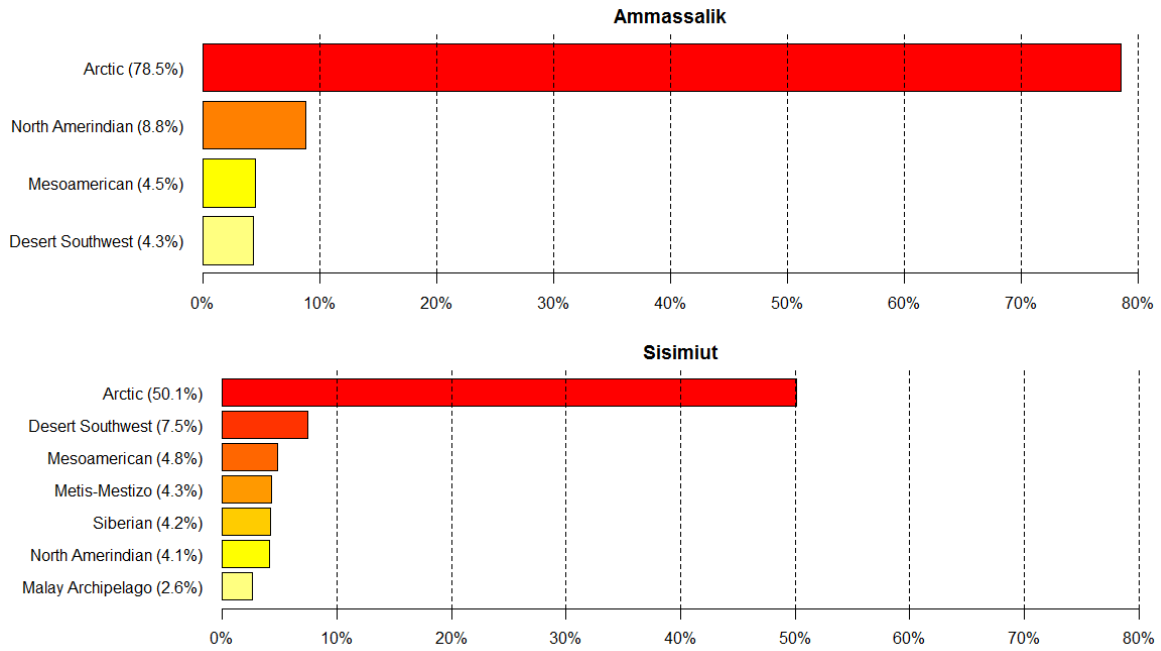
- **Ammassalik Island:** Historians assert that the Saqqaq culture reached eastern Greenland and was later replaced by the Dorset culture. The Thule culture migrated through the area in the 15<sup>th</sup> century; however, this area was not permanently settled until the 18<sup>th</sup> century.
- **Sisimiut (town):** This town has been first settled dating back to approximately 2,500 BC by the Saqqaq culture and then later occupied (after a few hundred of years of no inhabitation) by the Dorset and then Thule cultures. It is likely that the latter two cultures interbred to a certain degree, and unlike the Ammassalik Island, the location of the town has been continuously inhabited since the early Dorset culture arrival.



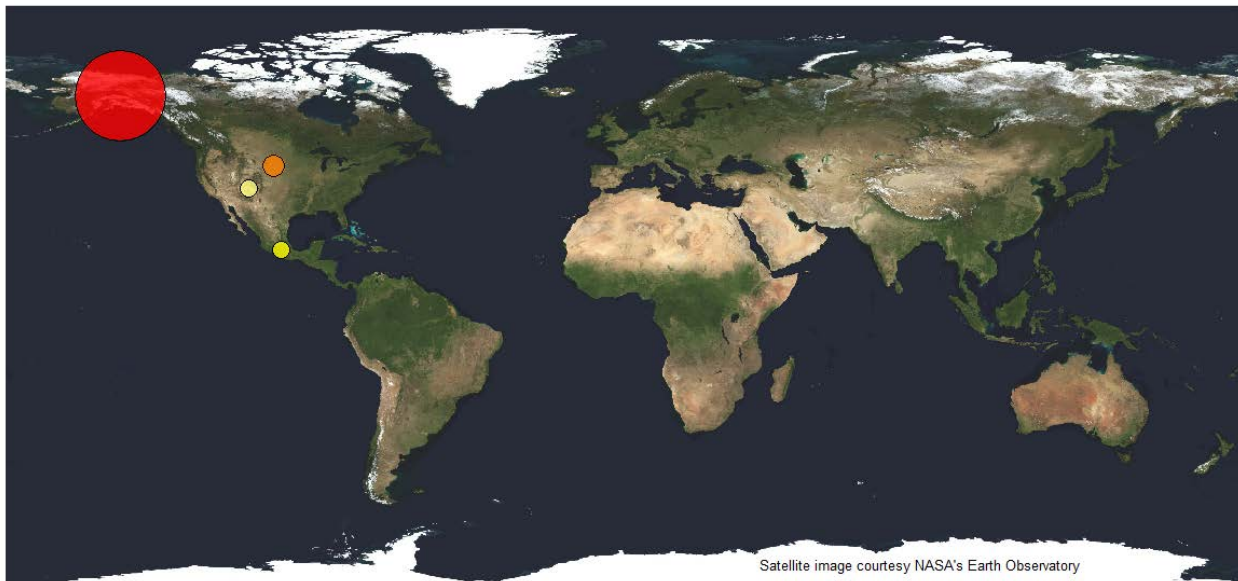
**Figure 7:** Location of two individual Greenlandic Inuit populations: Ammassalik Island on the east coast of Greenland and Sisimiut (town) on the west coast of Greenland.

Figures 8, 9.a and 9.b summarize the top World Region matches for the two Greenlandic Inuit populations (only the World Regions that are top matches for at least two percent of the simulated profiles have been included in the summary figures and discussion):

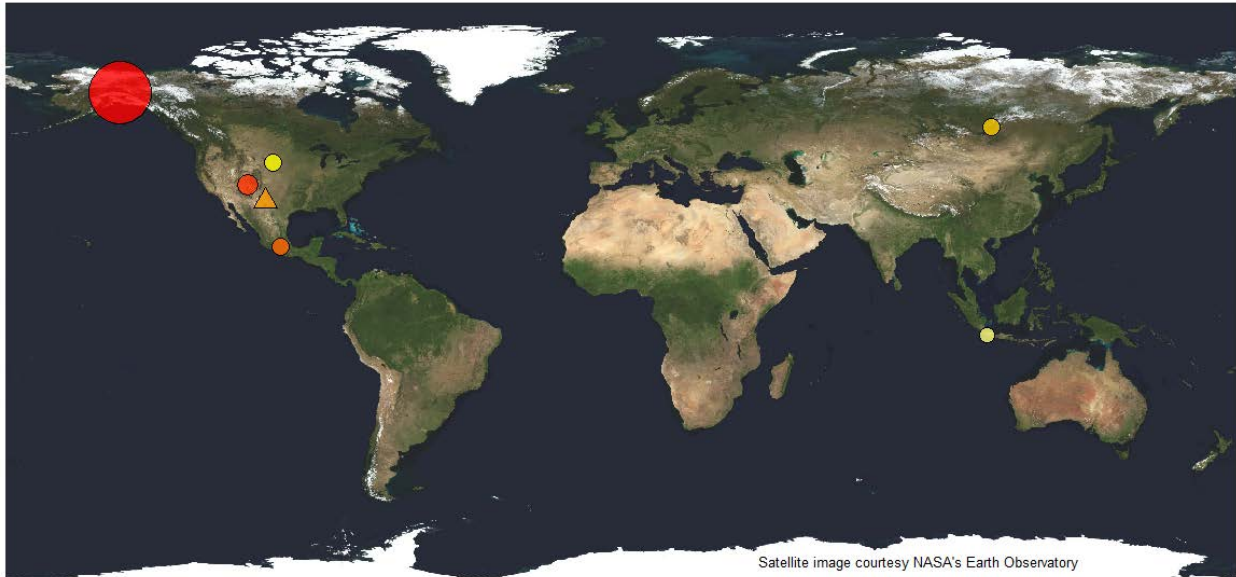
- The Ammassalik Island individuals overwhelmingly have top matches with the Arctic World Region (78.5%) followed by three other non-mixed Native American World Regions. These four regions represent the top matches for 96% of the simulated profiles indicating that the individuals inhabiting the Ammassalik Island do not exhibit European/Asian genetic signatures and are very closely linked to modern Alaskan Inuit and Yupik populations.
- To the contrary, the genetic profiles from the Sisimiut town are much more diverse; only 50% of them have a top match with the Arctic World Region with another 16.4% matching the three non-mixed Native American World Regions (Desert Southwest, Mesoamerican and North Amerindian) better. The remaining World Regions (Metis-Mestizo, Siberian and Malay, the latter being genetically more similar to the previously discussed Polynesian World Region) indicate that the Sisimiut Inuit population retains more archaic genetic signatures compared to the Ammassalik Island inhabitants:
  - The Siberian and Malay matches may indicate that the Dorset and Thule cultures may have interbred at this location as the poorly understood (from the origins perspective) Dorset culture was likely genetically more close to the proto-Asian populations from around the Bering Sea (Siberia).
  - The Metis-Mestizo matches may indicate that, in a larger (by Greenlandic standards) population center, some of the European admixture has happened either during the Norse Middle Age settlement or during modern times.



**Figure 8:** Top World Region matches for 5,000 simulated Greenlandic individuals from each native Greenlandic Inuit population.



**Figure 9.a:** Geographical distribution of top World Region matches for 5,000 simulated profiles from Ammassalik Island.



**Figure 9.b:** Geographical distribution of top World Region matches for 5,000 simulated profiles from Sisimiut (town). Metis-Mestizo World Region is denoted with a triangle.

## Conclusion

The newly available STR marker data for Greenlandic Inuit populations confirm the strong genetic links of these populations to the Inuit and Yupik populations of Alaska. In addition, non-negligible Siberian and European admixture is present in some Greenlandic Inuits potentially related to 1) the interbreeding of Dorset and Thule cultures on the western coast of Greenland and 2) later admixture with Europeans (either via the Middle Age Norse settlements or modern interbreeding). As more data become available for the Inuit populations across the Arctic Circle (e.g., from Canada), additional analyses may refine the conclusions presented in this Digest, especially as they relate to the genetic connections of the Inuits to the proto-Asian populations as well as the potential admixture with Norse settlers.

## Appendix: DNA Tribes® STR Update for Spring 2015

We are pleased to announce a Spring 2015 update to the DNA Tribes® STR analysis including:

- 83 native and local populations added to our reference database (*Figure A exhibits the geographical distribution of new populations*)
- Updated labeling for diaspora populations (*based on genetic distance to non-mixed World Regions*)
- Updated and refined world region memberships

### **New** Sub-Saharan African Populations:

- African (South Africa) (634)
- Ashanti (Ghana) (201)
- Madagascar (170)
- Somalia (198)

### **New** Diaspora Populations:

- Afro-Bolivians of Nor Yungas (57)
- Argentina (Bahia Blanca) (85)
- Argentina (Mar Del Plata) (180)
- Ashkenazi Jewish (DNA Tribes) (219)
- Colombian (Santander Province) (1,000)
- La Plata (Argentina) (100)
- Mestizo (Entre Rios, Argentina) (686)
- White (South Africa) (462)

### **New** East Asian Populations:

- Han (Guangzhou, China) (132)
- Han (Henan, China) (122)
- Han (Taiwan) (51)
- Han (Western China) (7,636)
- Japanese (1,501)
- Sichuan (China) (200)
- Uyghur (Yili) (128)
- Xibe (China) (226)

### **New** Middle Eastern Populations:

- Morocco (320)
- Morocco (86)
- Saudi Arabia (Riyadh) (190)
- Turkish (Bosnia and Herzegovina) (100)
- Turkish (Cyprus) (501)
- United Arab Emirates (109)

### **New** European Populations:

- Belgium (205)
- Calabria (Italy) (235)
- Denmark (362)
- Denmark (597)
- English (DNA Tribes) (300)
- Estonia (303)
- German (DNA Tribes) (331)
- Greece (737)
- Hungary (254)
- Netherlands (2,085)
- Northeast Spain (292)
- Northern Italy (303)
- Poland (2,041)
- Poland (Bialystok) (208)
- Poland (Gdansk) (293)
- Poland (Krakow) (285)
- Poland (Lublin) (204)
- Poland (Pomerania Kujawy) (306)
- Poland (Poznan) (301)
- Poland (Szczecin) (306)
- Poland (Warszawa) (236)
- Poland (Wroclaw) (208)
- Republic of Macedonia (205)
- Russia (1,118)
- Russia (358)
- Western Romania (336)

### **New** Central Asian Populations:

- Kirgiz (74)
- Mongolia (165)
- Mongolian (China) (291)

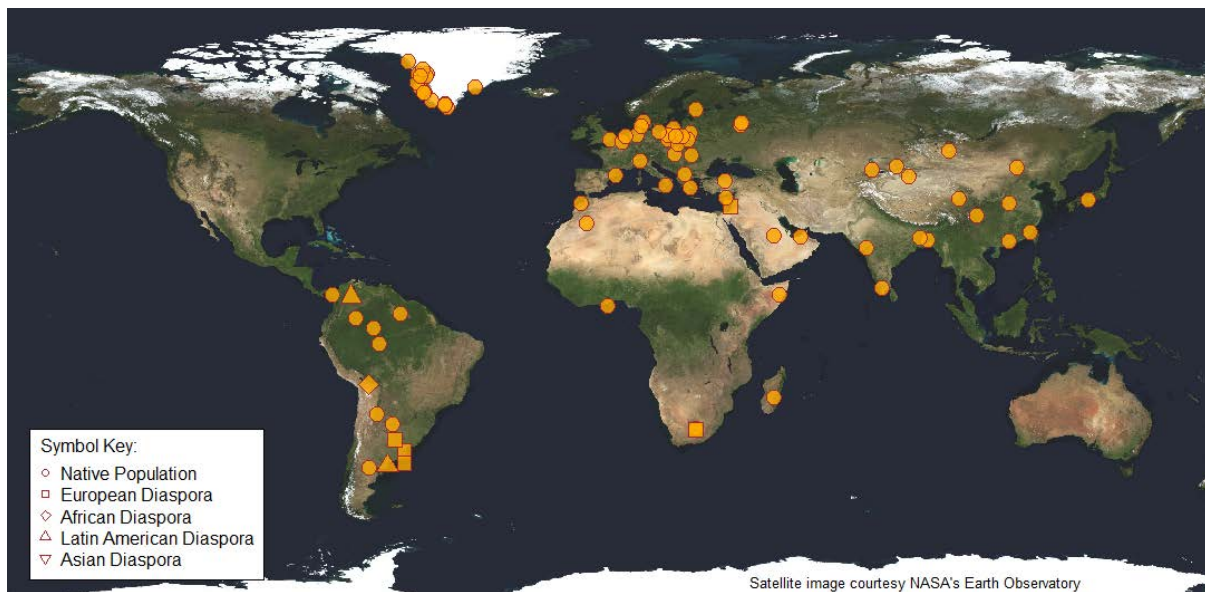
**New Native American Populations:**

- Desano (Colombia) (28)
- Embera (Colombia) (31)
- Greenland (194)
- Greenland (505)
- Greenland (Aasiaat) (26)
- Greenland (Ammassalik) (76)
- Greenland (Ilulissat) (54)
- Greenland (Maniitsoq) (51)
- Greenland (Nanortalik) (25)
- Greenland (Narsaq) (18)
- Greenland (Nuuk) (56)
- Greenland (Paamiut) (17)
- Greenland (Qaqortoq) (27)
- Greenland (Qasigiannuit) (18)
- Greenland (Qeqertarsuaq) (10)

- Greenland (Sisimiut) (81)
- Greenland (Upernavik) (18)
- Greenland (Uummannaq) (17)
- Mapuche (Rio Negro, Argentina) (50)
- Mocovi (Northern Argentina) (50)
- Nukak (Colombia) (29)
- Puinave (Colombia) (77)
- Wichi (Salta, Argentina) (50)
- Curripaco (Colombia) (32)

**New South Asian Populations:**

- East Bengal (India) (80)
- Badaga (Tamil Nadu, India) (65)
- Bengali (Bangladesh) (595)
- Bhil Tribe (Gujarat, India) (297)



**Figure A:** Geographical locations of new native and Diaspora populations added to the DNA Tribes® population reference database.