Genetic History of Polynesians and New Zealand Maori

Out of Africa: Genetic studies are increasingly expanding our knowledge of human population movements and variation around the globe. One technique that allows us to do this is complete mitochondrial genome sequencing. The mitochondrial genome can be used to trace maternal ancestors and assess population variation[1,2]. A haplogroup is a set of mitochondrial DNA (mtDNA) sequences that have particular shared, derived mutations in common. As humans migrated out of Africa, mtDNA sequences diverged and this is reflected in the geographic distribution of haplogroups.

Asian Ancestry and Polynesian Variation: As people moved throughout the Pacific and into Polynesia, genetic interactions took place. The movement of mitochondrial haplogroups represent the migration of people from South East Asia through Near Oceania into Polynesia. B and Q are two such haplogroups which made it through to Polynesia. . The B4 subclade arose about 44,000 years ago in mainland Southeast Asia[3]. From there it diverged into many more subclades including B4a1a which is restricted to Taiwan, Island Southeast Asia and the Pacific[4]. The lineage B4a1a1is prevalent in Near Oceania and has become almost fixed in Polynesia, making up more than 90% of all Polynesian mtDNA haplotypes[5]. Haplotypes from the Q1 lineage have also been reported in Polynesia, in particular Gambier and the Cook Islands[5-7].

New Zealand Studies and Variation: The Africa to Aotearoa project is part of the National Geographic Genographic Project looking at the migration of humans across the world. Preliminary data from this project shows that variation in Polynesian mitochondrial sequences is much higher than previously thought[9]. It also shows that all of the New Zealand Māori sequences identified to date have origins in East Polynesia. Māori-specific haplotypes

have already been described, all which are also found in East Polynesia[1,2,8]. These haplotypes have been described in both modern and ancient DNA studies[1,2].

At current, there is a lack of East Polynesian, including New Zealand Maori, data in Genbank. Also lacking, is the sequencing and analysis of complete mitochondrial genomes. Until recently, most mitochondrial genome studies have focussed only on the hypervariable region of the genome. However, this is not enough to fully understand the scope of variation within these sequences.

Aims

The aim of this project is to investigate the level of haplotype variation present in populations in West and East Polynesia, including New Zealand, using complete mitochondrial genome sequencing.

Methods

Cheek swab samples were collected from populations in Tokelau and French Polynesia by Professor Matisoo-Smith as part of the Africa to Aotearoa Project. These samples (n=165) were extracted, purified and quantified following the protocol described in Clarke et al. [10]. Preparation for sequencing was then undertaken, also described in Clarke et al. [10]. At this stage, samples that did not have an adequate result from the quantification step were dropped out. Sequencing of the complete mitochondrial genome on the Illumina MiSeq platform was carried out for 46 samples.

Sequence analysis and identification of haplotypes was performed using the in-house pipeline, adapted from Clarke et al. [10]. Data from analysis of Gisborne samples (collected and analysed by the Matisoo-Smith Lab) has been used for comparison.

Results

Population	Haplotypes (Frequency)	Where is this haplotype commonly seen?
Tokelau (West Polynesia)	B4a1a (2)	Derivative of B4 clade seen commonly in Pacific Islanders
	B4a1a1 (7)	"Polynesian Motif" – Polynesia, also: Micronesia, Coastal Papuans, Solomon Islands, Philippines
	B4a1a1a (4)	Tokelau, New Guinea, Solomon Islands
French Polynesia (East Polynesia)	B4a1a1 (9)	See Above
	B4a1a1a (1)	See Above
	B4a1a1c (14)	East Polynesia, including NZ Maori, Hawaii, Marquesas
	B4a1a1m (7)	Solomon Islands, Cook Islands, NZ Maori, French Polynesia
	B4b1a2a (1)	Derivative from haplogroups seen in Southern China, South East Asia, Philippines, Taiwan, Japan, Siberia.
	H2a2b1a1 (1)	Russia, Sweden, England, Scotland, Germany
Gisborne, NZ (comparative data)	B4a1a1 (4)	See above
	B4a1a1a (1)	See above
	B4a1a1m (5)	See above
	H1b1h (1)	Derivative from haplogroups seen in Eastern Europe, Central Asia, North Asia

Table 1: Haplotype identification, frequency and places were the haplotypes have been identified for samples from populations in Tokelau, French Polynesia and Gisborne (NZ).

Discussion

Haplotypes: From the haplotype data, we can see that there is variation within each population. This is to be expected, and expands on our ever increasing knowledge of population variation. The majority of the haplotypes can be found within the B4 clade, which is well represented in South East Asian and Pacific regions. Two haplotypes were found to be European, falling within in the H clade. No new haplotypes were identified.

Within each of the three populations, haplotypes derived from the B4a1a1 haplogroup was the most common. This haplogroup is seen in high frequency throughout Polynesia, Micronesia, Coastal Papua New Guinea, Solomon Islands and Philippines. As this is common to all of the populations investigated here, it shows that they have a shared ancestry. One haplotype common to all three populations is B4a1a1a (see table 1), which is thought to have arisen in the Bismarck Archipelago[11].

Populations: The haplotypes can be separated into those from West Polynesia (Tokelau) and those from East Polynesia (French Polynesia and New Zealand). The haplotypes seen in West Polynesia are found deeper in the B4 clade than those from East Polynesia. This can be used as further evidence of population migration from West to East[8].

There are also haplotypes present in East Polynesia that are not seen throughout West Polynesia, such as the B4a1a1c haplotype (see table 1). This can represent novel mutations in the expanding Polynesian populations or possibly genetic interactions with other groups of people in the Pacific, such as Micronesians[12]. Finding evidence of interaction between Polynesia and Micronesia can further enhance the story of the migration history of Polynesians and their ancestors[12].

The haplotypes present in the New Zealand population are most similar to those from French Polynesia, for example haplotype B4a1a1m (see table 1). This haplotype is restricted to the

French Polynesian and New Zealand Maori populations. This contributes further to the hypothesis that New Zealand Maori are descended from Eastern Polynesians[2,12].

Conclusions

From this project, we can see that complete mitochondrial genome data is necessary to understand population origins from a genetic perspective. More data from throughout the Pacific Region, and South East Asia is still required to answer further questions of population origins and migration histories.

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