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Taiwanese aborigines: genetic heterogeneity and paternal contribution to Oceania

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ABSTRACT

In the present study, for the first time, 293 Taiwanese aboriginal males from all nine major tribes (Ami, Atayal, Bunun, Rukai, Paiwan, Saisat, Puyuma, Tsou, Yami) were genotyped with 17 YSTR loci in a attend to reveal migrational patterns connected with the Austronesian expansion. We investigate the paternal genetic relationships of these Taiwanese aborigines to 42 Asia-Pacific reference populations, geographically selected to reflect various locations within the Austronesian domain. The Tsou and Puyuma tribes exhibit the lowest (0.1851) and the highest (0.5453) average total genetic diversity, respectively. Further, the fraction of unique haplotypes is also relatively high in the Puyuma (86.7%) and low in Tsou (33.3%) suggesting different demographic histories. Multidimensional scaling (MDS) and analysis of molecular variance (AMOVA) revealed several notable findings: 1) the Taiwan indigenous populations are highly diverse. In fact, the level of inter-population heterogeneity displayed by the Taiwanese aboriginal populations is close to that exhibited among all 51 Asia-Pacific populations examined; 2) the asymmetrical contribution of the Taiwanese aborigines to the Oceanic groups. Ami, Bunun and Saisiyat tribes exhibit the strongest paternal links to the Solomon and Polynesian island communities, whereas most of the remaining Taiwanese aboriginal groups are more genetically distant to these Oceanic inhabitants; 3) the present YSTR analyses does not reveal a strong paternal affinity of the nine Taiwanese tribes to their continental Asian neighbors. Overall, our current findings suggest that, perhaps, only a few of the tribes were involved in the migration out of Taiwan.

1. Introduction

Located in East Asia, insular Taiwan has a population of about 23 million largely composed of Han Chinese (of the Min and Hakka subgroups) who migrated to the island from southeast China during the last half millennium (Chiung, 2001). The Taiwanese aboriginals comprise about 1.5% of the populace and speak Austronesian languages considered indigenous to the island (Trejaut et al., 2005). More specifically, the aboriginal population currently encompasses nine major tribes: Ami, Atayal, Paiwan, Bunun, Puyuma, Rukai, Tsou, Saisiyat and Yami (http://www.apc.gov.tw/). The Hans mostly reside in the west plain, whereas the indigenous populations live mainly in the central and east mountainous regions.

Linguistic and archeological evidence suggest that Austronesian speakers migrated into Taiwan from Southeast China (Gray and Jordan, 2000; Li et al., 2008) and, according to theory, embarked on a subsequent expansion into the vast geographical domain of Oceania. Various hypotheses concerning the Austonesian diaspora have been presented. Three of the most popular models, namely the "express train," "entangled bank," and "slow boat", are supported by various lines of genetic, linguistic and archeological evidence (Li et al., 2008). In the last several decades, genetic data encompassing both autosomal and uniparental markers have been employed to ascertain a more detailed view of the routes and times of migrations as well as the various genetic sources of the Pacific Islanders.

Mitochondrial DNA profiles of Taiwanese aborigines and Oceanic populations offer one perspective on the genetic affinities within this region. A distinct "Polynesian motif", B4a1a1, established a genetic

Abbreviations: MDS, multidimensional scaling; AMOVA, Analysis of Molecular Variance; mtDNA, mitochondrial DNA; YSTR, Y short tandem repeats; YSNP, Y single nucleotide polymorphism; PCR, polymerase chain reaction; DC, discriminatory capacity; HD, haplotype diversity; GD, gene diversity; NDH, number of different haplotypes; FUH, fraction of unique haplotypes; NUH, number of unique haplotypes.

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link between Taiwan aborigines and the Polynesian Islanders (Trejaut et al., 2005). Phylogenetic analysis of the B4a1a1 sub-types suggest parallel linguistic and maternal relationships (Bellwood, 1990; Donohue and Denham, 2010) among inhabitants of this region and delineates a dispersal initiating in Taiwan, transversing the Philippines and Indonesia en-route to the Polynesian Islands (Hill et al., 2007; Melton et al., 1995; Razafindrazaka et al., 2010; Tabbada et al., 2010). However, according to a more recent study by Soares et al. (2011), whole mitochondrial genome sequencing of Taiwanese aborigines and individuals from Island Southeast Asia and Oceania reveals that the mitochondrial sequence closest to that of the Oceanic groups is detected in the Bismarck Archipelago (Indonesia) and the number of accumulated mutations suggest a much ancient arrival of the haplogroup into Polynesia (Soares et al., 2011). Together, these findings indicate a non-Austronesian source for the original mtDNA lineages of remote Oceania.

Autosomal genetic data yields a different view from that provided by mtDNA. A previous analysis by Mirabal et al. (2013) analyzing autosomal STRs for the nine major Taiwanese aboriginal tribes (a total of 451 individuals) indicates that the aboriginal populations from Taiwan, as opposed to those from Indonesia, Mainland or Southeast Asia, exert a greater autosomal impact on the peripheral Polynesian populations of the Pacific. This finding lends support to the "Out of Taiwan" hypothesis espousing the view of minimal admixture with inhabitants of the Melanesian and Micronesian archipelagos en-route to distant Oceania. Further, the authors reveal that of the nine major Taiwanese tribes studied, three, Paiwan, Puyuma and Saisiyat, levied the greatest impact on the autosomal genome of Polynesian and Madagascar Austronesian populations.

In addition to the autosomal and mtDNA findings, controversy surrounds incongruous Y-chromosomal results. Several early YSNP investigations suggest a paternal linkage between continental Asian and Polynesian groups (Su et al., 2000) and provide no genetic evidence connecting Polynesia to Taiwanese tribes (Kayser et al., 2000, 2003, 2006, 2008; Lum, 1998). However, more recent Y SNP analyses reported that the O3a2 (P201) lineage, widespread across Island Southeast Asia, Indonesia and Polynesia, has not been detected in mainland Asia (Karafet et al., 2010). Subsequently, Mirabal et al. (2012) established a link between the Ami and Polynesia via the shared polymorphic presence of a rare subhaplogroup, O3a2c* (P164), which is detected only at extremely low levels in some mainland East Asian groups (Yan et al., 2007).

In the previous decade, anthropological studies investigating the distribution of YSTR haplotypes among Taiwanese indigenous peoples were, for the most part, limited to seven YSTR loci — DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392 and DYS393 (Li et al., 2008) and the findings of these analyses lack consensus. In the present inquiry, for the first time, we explore the paternal genetic variation of all nine main aboriginal Taiwanese populations and their contribution to the genetic landscape of Oceania at a higher resolution by employing 17 YSTR loci. STR loci are hypervariable allowing for higher phylogenetic resolution and assessment of more recent demic events such as the Austronesian diaspora from Taiwan (Mirabal et al., 2013; Rowold and Herrera, 2003). By employing this sensitive uniparental marker system, we hope to gauge the paternal genetic contribution bestowed by the nine major Taiwanese tribes on inhabitants of Oceania and compare it to that imparted by Indonesia, Mainland and Island Southeast Asia. Further, we also evaluate the results provided by the high resolution YSTR diversity in the context of previous genetic studies and marker systems in an attempt to uncover migrational patterns among the Taiwanese aborigines, nearby mainland and Pacific island populations. In addition to the nine Taiwanese tribes, the three key geographically targeted populations of Mongolia, Han Chinese and Society Islands, French Polynesia were genotyped and analyzed to provide additional key points of reference. Forty geographically relevant reference populations from previous investigations including Northeast Asians, Southwest Chinese, Southeast Asians, Solomon/Papuan Islanders, Australian and eastern Polynesians are featured in the analyses as a basis for comparison (Table 1).

2. Materials and methods

2.1. Sampling and DNA extraction

Whole blood and buccal (swabs) samples were collected from a 293 unrelated male individuals from the major nine aboriginal tribes that inhabit Taiwan (Ami: 65; Atayal: 42; Bunun: 38; Rukai: 28; Paiwan: 29; Saisiyat: 24; Puyuma: 15; Tsou: 27; Yami: 25). Additionally, samples were procured from the Han Chinese (52) and Mongolian (67) populations of northeast Asia, as well as 36 unrelated Polynesian natives residing in the Society Islands of French Polynesia. Genealogical information was collected for a minimum of two generations to ascertain paternal descent.

DNA from blood was extracted using the standard phenol-chloroform method (Antunez de Mayolo et al., 2002; Novick et al., 1995) and the buccal swabs were processed with the Gentra Buccal Cell Kit (Puregene, Gentra Systems, Minneapolis, MN) according to the manufacturer's specifications. Samples were stored as stock solutions in 10 mM Tris–EDTA at −80 °C. All samples were procured from donors voluntarily with informed consent.

2.2. DNA amplification and STR genotyping

The DNA samples were genotyped with a panel of 17 YSTR loci (DYS19, DYS385a/b, DYS389I/II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635 and Y GATA H4) available using the AmpFlSTR® YFiler™ PCR Amplification Kit (Applied Biosystems of Life Technologies). Amplification reactions were performed in an Eppendorf Mastercycler® gradient (Eppendorf AG, Hamburg, Germany) according to the manufacturer's specifications. DNA fragments were separated by capillary electrophoresis on an ABI Prism 3130xl Genetic Analyzer using the ABI GeneScan 500 LIZ internal size standard as a basis for comparison. Fragment sizes were obtained using the GeneMapper® v3.1 software (Applied Biosystems of Life Technologies, Foster City, CA). Alleles were designated by comparison to an allelic ladder supplied by the manufacturer (Applied Biosystems). All microvariants, and duplicated alleles were confirmed by repeating the amplification process.

2.3. Reference populations used for comparison

A total of 39 previously published populations were used for comparison across the 17 Y-STR loci and are listed with corresponding references in Table 1. The geographical locations, sample size, abbreviations as well linguistic affiliations are provided. The geographic locations of the aboriginal populations within Taiwan as well as the genotyped Han, Mongolian and Society Islands Polynesian populations, and reference populations are illustrated in Fig. 1.

2.4. Analysis of data

All phylogenetic analyses were performed utilizing the 15 Y-STR loci (DYS19, DYS389I, DYS389II, DYS390, DYS391, DYS392, DYS393, DYS437, DYS438, DYS439, DYS448, DYS456, DYS458, DYS635, GATA H4) in common among the collections listed in Table 1. DYS385 was excluded from the phylogenetic analyses because is not possible to discriminate between the DYS385a and DYS385b loci with the Y-filer kit. In addition, the size of the DYS389I allele was subtracted from the DYS389II for all analyses. Allelic frequencies were calculated using PowerMarker V3.25 (Liu and Muse, 2005). Computation of both haplotype and gene diversity indices (HD and GD, respectively) was performed with the Arlequin V3.5 software package (Excoffier et al., 2005). HDs were estimated at the 17 YSTR loci level. Chromosomes