

# Microsatellite polymorphisms in two Taiwanese aboriginal groups

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**Abstract.** The aim of this study was the genetic characterization of two Taiwanese indigenous populations (*Ami* and *Atayal*) based on the short tandem repeat (STR) loci sanctioned by CODIS. Significant departures from genetic equilibrium were detected at the D8S1179 and TH01 loci in the *Ami*, which persisted even after applying Bonferroni-type corrections. Gene diversity (GD) values ranged from 0.5377 (TPOX) to 0.8674 (FGA) in the *Atayal*, whereas in the *Ami* GD oscillated between 0.6409 (TPOX) and 0.8764 (D21S11). The STR data were analyzed using distance-based methods to assess the genetic relationships of these two aboriginal groups with other Asian populations. © 2005 Elsevier B.V. All rights reserved.

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## 1. Introduction

In the island of Taiwan, formerly known as Formosa, nine indigenous groups have coexisted (*Tsou*, *Bunun*, *Paiwan*, *Rukai*, *Atayal*, *Saisiat*, *Ami*, *Puyuma* and *Yami*), which are highly homogeneous within each tribe, but diversified among the different tribes probably due to long-term genetic isolation. Two of these indigenous groups, the *Ami* and the *Atayal*, comprise about 60% of the total aboriginal population. The *Ami* live on the lowlands near the ocean, frequently intermixed with Han Chinese, whereas most of the

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*Atayal* live in remote mountain areas [1]. The aim of this work was to examine the genetic characteristics of these two Taiwanese populations based on 13 hypervariable autosomal STR loci. We then used distance-based methods to assess the phylogenetic relationships of these aboriginal groups with other Asian populations.

## 2. Material and methods

Whole blood samples were obtained from 108 unrelated healthy donors (40 *Atayal* and 68 *Ami*). Genomic DNA was extracted by standard phenol/chloroform procedure. DNA samples were amplified using the AmpF/STR® Profiler Plus™ and AmpF/STR® COfiler™ PCR Amplification kits (PE Applied Biosystems). PCR amplifications of the CODIS core STR loci (D3S1358, VWA, FGA, D8S1179, D21S11, D18S51, D5S818, D13S317, D16S539, TH01, TPOX, CSF1PO and D7S820) were carried out using 0.5–1.25 ng of DNA. Amplification products were run in a GeneAmp PCR System 9600 thermal cycler. Amplified STR fragments were separated in an ABI PRISM 377 DNA Sequencer using an internal size standard (GeneScan® 500 ROX). Genotyping of each sample was made with the Genotyper® 3.7 NT and GeneScan® 3.7 softwares by comparison with allelic ladders. Hardy–Weinberg equilibrium (HWE) was tested through

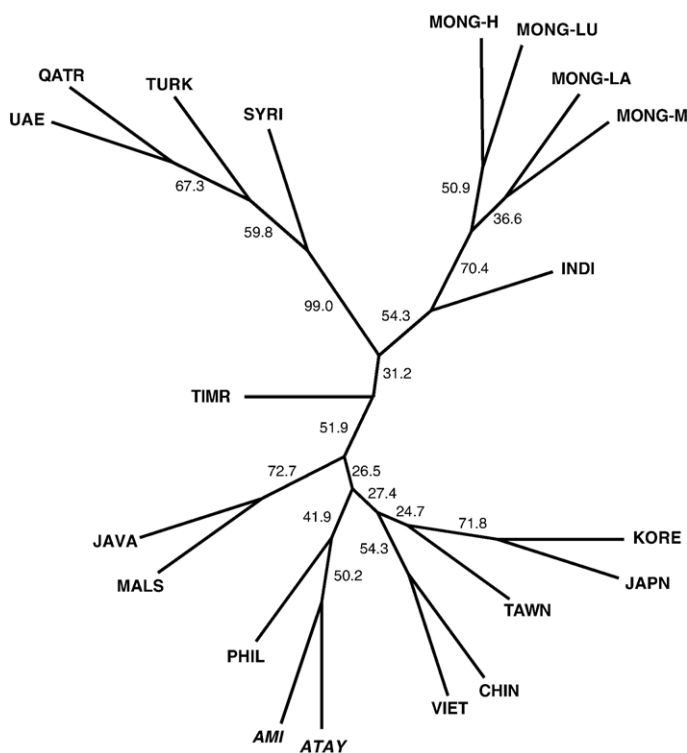


Fig. 1. Neighbor-joining (NJ) tree constructed from Reynold's  $F_{ST}$  genetic distances based on the allelic frequencies of 13 short tandem repeat (STR) loci in 20 Asian populations. Figures along the branches are percentage bootstrap values estimated from 1000 iterations.

the exact- $P$  method [2]. Some useful parameters were calculated including gene diversity [3] and power of discrimination [2]. To assess phylogenetic relationships based on the allelic frequencies of the STR markers, data compiled in previous studies were used to compute  $F_{ST}$  genetic distances [4] between pairs of populations. From the resultant  $F_{ST}$  distance matrix, a Neighbor-Joining tree [5] with bootstrap validation [6] was constructed.

### 3. Results and discussion

Significant departures from Hardy–Weinberg equilibrium were found at the D8S1179 and D18S51 loci in the *Ami*, which persisted after applying Bonferroni-type corrections. Deviations of HWE could associate to the limited genetic diversity of these populations [7], which in turn would be caused by the high endogamy levels typical of indigenous communities. The most polymorphic markers showed the highest values for the power of discrimination (PD). In the *Ami* these were D21S11 (PD: 0.969, 12 alleles), FGA (0.963, 10 alleles), D18S51 (0.948, 10 alleles) and VWA (0.941, 9 alleles), whereas in the *Atayal* the more variable loci were FGA (0.963, 9 alleles), D21S11 (0.929, 8 alleles) and D8S1179 (0.925, 7 alleles). Gene diversity (GD) values ranged from 0.5377 (TPOX) to 0.8674 (FGA) in *Atayal*, while in the *Ami* GD oscillated between 0.6409 (TPOX) and 0.8764 (D21S11). A notable heterozygosity was observed in both tribal groups, although it was slightly higher in *Ami* (average: 0.7867) than in *Atayal* (0.7036).

The Neighbor-Joining tree represented in Fig. 1 illustrates the genetic relationships of the Taiwanese indigenous groups in relation to other Asian populations. The phylogram generated from the STR data strongly correlated with geography. Three main subdivisions appeared in the analysis: Middle Eastern Arab populations (Qatar, United Arab Emirates and Syria) plus Turkey, Central Asian populations (India and the various samples from Mongolia), and a third cluster including both island and mainland populations from the Far East, where the Taiwanese aboriginal samples were grouped. Specifically, the study populations (*Ami* and *Atayal*) showed the strongest genetic affinity with the sample of the Philippines.

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