

HAPLOGROUP DETERMINATION FOR MITOCHONDRIAL DNA IN THE TAIWANESE POPULATION

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Introduction:

To construct a Taiwanese mitochondrial DNA (mtDNA) phylogeny, we performed a large-scale association study on 5377 Taiwanese individuals.

Material and Method:

Genomic DNA was collected from peripheral blood cells of all subjects. We used a high-throughput method for detection of a single nucleotide polymorphism by combining polymerase chain reaction (PCR) and sequence-specific oligonucleotide probes (SSOPs) protocols with the Luminex 100 xMAP flow cytometry dual-laser system to quantify fluorescently labeled oligonucleotides attached to color-coded microbeads. We created 94 oligonucleotide probes to detect 45 haplogroups, 24 in macro-haplogroup M, 21 in macro-haplogroup N.

Result:

Specifically, macro-haplogroup M and N encompass almost all mtDNA lineages in Taiwan. From 5377 samples collected we identified 45 haplogroups: C, D4*, D4a, D4b1, D4b2, D4g, D4h, D4k, D4n, D5a, D5b, D6, E, G, M1, M7a, M7b, M7c, M8, M9, M10, M11, M12, M13, Z in macro-haplogroup M; F*, F1a, F1b, F1c, F2, F3, F4, B*, B4a, B4b, B4c, B4d, B4e, B4f, B4g, B5, B6, H2, A, N9a, N9b, N1b, Y, in macro-haplogroup N. After analyzing, 2708 samples belonged to M and 2669 samples belonged to N. Four major M-haplogroups (D4, M7, D5, M8) and four major N-haplogroups (F*, B*, B5, A) accounted for more than 78.6% of the variations observed in the Taiwanese population.

Discussion:

About 94.6% of the mtDNA is subsumed within 45 mtDNA haplogroups suggesting that the identified haplogroups could encompass virtually all Taiwanese mtDNA, whereas the 5.4% ambiguity are thought to have been mixed with outside genetic material. The percentage of the Taiwanese population is almost equal in macrohaplogroup-M and N, 50.4% and 49.6%, respectively. In comparing the mtDNA haplogroup composition in Taiwan with southeast Asia and Japan, haplogroup F, D4, M7 and B cover most of the lineages among Taiwanese, while haplogroup E, B4a1a, M7, B4c cover most of the lineages in southeast Asia and Japan. .