

we used three primers ("3-primer PCR") to amplify in parallel wild-type and deleted mtDNA ("common deletion") and two characteristic fragments of wild-type and common deletion could be detected. Quantitation of these mutations is being done to see (possible) quantitative correlation between the amount of mutation and clinical severity. The present findings serve to emphasize the extent to which formalin-fixed paraffin sections represent a valuable repository of genetic material for a molecular genetic study.

SL804

Y Chromosomal DNA Variation in East Asian Populations and its Potential for Inferring the Peopling of Korea

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We have examined variation of five polymorphic loci (*DYS287*, *DXYS5Y*, *SRY465*, *DYS19* and *DXYS156Y*) on the Y chromosome in samples from a total of 1,260 males in eight ethnic groups of East Asia. We found four unique haplotypes constructed from three biallelic markers in these samples of East Asians. The Japanese population was characterized by a relatively high frequency of either the haplotype I-2b (-Y2/T) or II-1 (+Y1/C). These dual patterns of the distribution of Y chromosomes (I-2b/II-1) were also

found in Korea, although they were present at relatively low frequencies. The haplotype II-1 was present in Northeast Asian populations (Chinese, Japanese, Koreans and Mongolians), with the exception of a single male from the Thais in the Southeast Asian populations (Indonesians, Philippines, Thais and Vietnamese). The Japanese were revealed to have the highest frequency of this haplotype (27.5%), followed by Koreans (2.9%), Mongolians (2.6%) and mainland Chinese (2.2%). In contrast, the haplotype I-2b was found to be in the Japanese (17.1%), Indonesian (9.5%), Korean (6.3%), Vietnamese (3.8%) and Thai samples (2.7%). These findings suggested that the chromosomes of haplotype I-2b were likely derived from certain areas of Northeast Asia, the region closest to Southeast Asia. Phylogenetic analysis using the neighbor-joining tree also reflected a general distinction between Southeast and Northeast Asian populations. The phylogeny revealed a closer genetic relationship between Japanese and Koreans than to the other surveyed Asian populations. Based on the result of dual patterns of the haplotype distribution, it is more likely that the population structure of Koreans may not have evolved from a single ancient population derived from Northeast Asians, but through dual infusions of Y chromosomes entering Korea from two different waves of East Asians.