DISTRIBUTION CHARACTERISTICS OF POLYMORPHIC ALLELES AMONG THE POPULATIONS OF THE NANSEI ISLANDS, JAPAN

Kazutoshi AGO

Abstract

The characteristics of the distribution of blood group polymorphism alleles among the populations of the Nansei Islands, Japan, were examined using currently available blood group data, such as red cell types, enzyme types and serum protein types. 1. A comparison of genetic diversity was made between the Amami-Oshima and Tanegashima populations, and the neighboring large populations such as the mainland of Kagoshima and the main island of Okinawa. Gene diversity in the island populations is similar to that of Kagoshima and Okinawa. 2. Geographical clines of allele frequency of blood groups in Japanese populations are discussed. Clines constructed using the Nansei Islands data support the 'dual structure model hypothesis for the origin of the Japanese more strongly than previously described clines. 3. Two rare alleles, TF Damami and AHSG 5, which are uniquely distributed in the Nansei Islands, are described.

Key words: Nansei Islands, Island population, Blood groups, Geographical cline

Introduction

The Nansei Islands form an archipelago stretching between the mainland of Kagoshima and Taiwan, and are potentially important in tracing the origin of the Japanese people. I investigated the distribution of blood group alleles with the purpose of revealing the genetic characteristics of this area and to gather basic data for forensic practices. In this paper, on the strength of the data accumulated so far, I discuss the genetic homogeneity of the Tanegashima and Amami-Oshima populations, geographical clines of allele frequencies in Japanese populations and rare variants distributed specifically in the Nansei Islands.

The Genetic Homogeneity of Tanegashima and Amami-Oshima Populations

Gene diversity, meaning the degree of genetic homogeneity in each population, was calculated by the following equation (ROYCHOUDHURY & NEI, 1988) using the available data (AKAISHI & KUDO, 1975; ISHIMOTO, 1975; OMOTO, 1975; ROYCHOUDHURY, & NEI, 1988, AGO et al., 1998a, b).

$$H = 1 - \sum_{i=1}^{k} xi2$$

where xi is the frequency of the i-th allele in the population and k is the number of alleles. Comparison of gene diversity among populations from Amami-Oshima, the mainland of Kagoshima and the main island of Okinawa was carried out using eight blood group loci. As shown in Table 1, the average value of the Amami-Oshima population (0.415) laid between those of the Kagoshima and Okinawa populations (0.4064 and 0.4189 respectively) and statistically significant differences were not observed. The average value of the Tanegashima population (0.5044) was also similar to those of populations

from Kagoshima, Amami-Oshima and Okinawa (0.4950, 0.5157 and 0.5054 respectively), as shown in Table 2, and there are also no statistically significant differences. These results indicate that Amami-Oshima and Tanegashima have sufficiently large populations to maintain allele heterogeneity and avoid island effects usually associated with isolated populations. Similar investigations on islands with small populations, such as Yakushima, Okinoerabu and Yoron, are necessary, but data from these areas are not currently sufficient to allow detailed analysis.

	Amami-oshima	Kagoshima*	Okinawa
GC	0.6574 ± 0.0075	0.6506 ± 0.0084	0.6578 ± 0.0080
TF	0.3998 ± 0.0084	0.3693 ± 0.0145	0.3942 ± 0.0119
AHSG	0.4213 ± 0.0156	0.3754 ± 0.0147	0.3913 ± 0.0120
HP	0.3565 ± 0.0091	0.4009 ± 0.0119	0.3752 ± 0.0094
AP	0.3482 ± 0.0176	0.3523 ± 0.0135	0.3960 ± 0.0114
PGD	0.1130 ± 0.0160	0.1440 ± 0.0134	0.1456 ± 0.0126
PGM1	0.4234 ± 0.0141	0.3743 ± 0.0130	0.4125 ± 0.0120
ABO	0.6003 ± 0.0081	0.5846 ± 0.0012	0.5782 ± 0.0014
Average	0.4150 ± 0.0586	0.4064 ± 0.0545	0.4189 ± 0.0536

Table 1. Comparison of gene deversity between Amami-oshima and neighboring populations.

*:the mainland of Kagoshima

Table 2. Comparison of gene deversity between Tanegashima and neighboring populations.

	Tanegashima	Amami-oshima	Kagoshima*	Okinawa Honto
GC	0.6359 ± 0.0102	0.6574±0.0075	0.6506 ± 0.0084	0.6578 ± 0.0080
TF	0.4013 ± 0.0124	0.3998 ± 0.0084	0.3693 ± 0.0145	0.3942 ± 0.0119
AHSG	0.3959 ± 0.0120	0.4213 ± 0.0156	0.3754 ± 0.0147	0.3913 ± 0.0120
ABO	0.5845 ± 0.0074	0.6003 ± 0.0081	0.5846 ± 0.0012	0.5782 ± 0.0014
Average	0.5044 ± 0.0620	0.5197 ± 0.0642	0.4950 ± 0.0721	0.5054 ± 0.0670

*:the mainland of Kagoshima

Geographical Clines of Allele Frequencies in Japanese Populations

The 'dual structure model ' of HANIHARA (1991), describing the origin of the Japanese people, is currently thought to offer a reasonable explanation. It is thought that the modern Japanese consist of the Ainu and Ryukyus peoples descended from the Jomon people and the Hondo Japanese, who mixed with immigrants from the Korean Peninsula during the Yayoi era. Therefore, differences in genetic traits exist in the eastern and western parts of the Japanese Islands. Geographical clines in allele frequencies have so far been reported in the ABO system (FUJITA et al., 1978), the GPT type (a red cell enzyme type; ISHIMOTO & KUWATA, 1974) and the GC type (a serum protein type; YUASA et al., 1983). It is thought that these clines provide evidence in support of the differences in genetic traits mentioned above. In previous studies, clines starting from northern regions, such as Aomori, were examined. However, if genetic traits were influenced by immigrants from the

Korean Peninsula, clines starting from western areas, such as the northern part of Kyusyu, Yamaguchi and Kinki, should be examined. As shown in figure 1 for the clines of the ITI alleles, two types of cline originating from Aomori and Yamaguchi were examined. Linear clines originating from Aomori similar to the ABO, GPT and GC clines described in previous reports were observed (figure 1A). Clines from Yamaguchi, shown in figure 1B, demonstrate that different clines exist in the eastern and western parts of the Japanese Archipelago (AGO et al., 2000). V-shaped and inverted V-shaped clines were similarly observed in the GC and TF types (AGO et al., 1998a, b), adding further support to the ' dual structure model ' hypothesis.



Fig.1. Geographical clines of ITI gene frequencies in Japanese populations. Distances were measured from Aomori in (A) and from Yamaguchi in (B).

Rare Variants Specifically Distributed in the Nansei Islands

I discovered that two rare alleles, TF Damami (AGO et al., 1998a) and AHSG 5 (TAMAKI et al., 1996) appear to be distributed exclusively within the Nansei Islands. As for the above mentioned TF type, three phenotypes, TF C1, TF C1C2, TF C2, which are controlled by the TF C1 and TF C2 alleles, were commonly observed in Japanese populations. Rare variants controlled by the TF B and TF D alleles were also observed. These phenotypes are shown in figure 2. Most of the TF D variants in Japanese populations are TF Dchina. In Amami-Oshima, a TF D variant, tentatively named TF Damami, was frequently observed. TF Damami was also observed in each sample from Yakushima and Okinoerabu. TF Damami moved slightly faster than TF Dchina on the electrophoretogram shown in figure 2. In the AHSG type (a serum protein type) AHSG 1 and AHSG 2 are common alleles. The AHSG 5 allele is extremely rare throughout most of Japan, but not in Amami-Oshima, Tokunoshima, Okinoerabu, Okinawa, Miyako and Ishigaki. The frequency of the AHSG 5 allele in these areas was greater than 1% that can be called polymorphic frequency. The distribution of the AHSG 5 allele indicates that it is specific to the Nansei Islands.

Kazutoshi Ago



Fig. 2. Three common phenotypes controlled by TF C1 and TF C2 and rare variants by TF B and TF D alleles (Dchi: Dchina, Dama: Damami).

Conclusion

Our study of geographical clines of polymorphic alleles, and of rare alleles that are uniquely distributed within the Nansei Islands, suggests that the genetic makeup of people from this area can help us to understand the origin of the modern Japanese people. Additional data should provide greater insight in the future.

References

- AGO, K., YUASA, I. & TSUGANEZAWA, O. 1998a. Anthropological Science, 106: 209-219.
- AGO, K., YUASA, I. & TSUGANEZAWA, O. 1998b. Anthropological Science, 106: 385-395.
- AGO, K., YUASA, I., UMETSU, K. & TSUGANEZAWA, O. 2000. "Inter-alpha-trypsin inhibitor polymorphism in southwestern Japan: geographical clines of allele frequencies in Japanese populations." Progress in Forensic Genetics 8 (ed. SENSABAUGH, G.F., LINCOLN, P.J. & OLAISEN, B.), 356-358. Elsevier, Amsterdam.

AKAISHI, S. & KUDO, T. 1975. "Blood groups." Anthropological and genetic studies on the Japanese (ed. WATANABE, S., KONDO, S. & MATSUNAGA, E.), 77-107, University of Tokyo press, Tokyo.
FUJITA, Y., TANIMURA, M. & TANAKA, K. 1978. Jap. J. Human Genet., 23: 63-109.

HANIHARA, K. 1991.Japan Review, 2: 1-33.

ISHIMOTO, G. & KUWATA M. 1974. Jap. J. Human Genet, 18: 373-377.

- ISHIMOTO, G. 1975. "Red cell enzymes." Anthropological and genetic studies on the Japanese (ed. WATANABE, S., KONDO, S. & MATSUNAGA, E.), 109-139, University of Tokyo press, Tokyo.
- OMOTO, K. 1975. "Serum protein groups." Anthropological and genetic studies on the Japanese (ed. WATANABE, S., KONDO, S. & MATSUNAGA, E.), 141-162, University of Tokyo press, Tokyo.
- TAMAKI, N., YUASA, I., IRIZAWA, Y., AGO K., TSUGANEZAWA, O. & UMETSU, K. 1996. Jpn. J. Legal Med., 50: 143.

ROYCHOUDHURY, A.K. & NEI, M. 1988. Human polymorphic genes world distribution, 3-244, Oxford University Press, Oxford.

YUASA, I., SANESHIGE, Y. & OKADA, K. 1983. Jap. J. Human Genet. 28: 255-261.