

Some Aspects of the Genetic Composition  
of the Japanese

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**ABSTRACT**

This paper deals with the main characteristics of the Japanese as viewed from the distribution of genetic markers. Firstly, it is shown by the genetic distance and cluster analyses that three ethnic groups of Japan, namely, the Wajin, the Ainu and the Ryukyuan, are clearly distinguished to each other. Among them, particularly the Wajin has the genes supposedly introduced from the continental north-east Asia through Korea in high frequencies. Secondly, it is demonstrated that the Ainu belongs to the Mongoloid race, rather than to the Caucasoid. Their phenotypic features are explained as generalized Proto-Mongoloid characters. Thirdly, the genetic link between the Ainu and the Ryukyuan is suggested genetically, and they both are considered to be descendants of Proto-Japanese populations. Fourthly, several cases of gene frequency cline are observed, suggesting that relatively large number of immigrants came through Korean peninsula to north-eastern Kyushu and then spread eastwardly and southwardly since the Yayoi period.

In this paper I shall review the following four topics which I have been concerned in the studies of past 25 years: (1) Genetic nature of the major Japanese population, the Wajin; (2) Genetic affinities of the Ainu; (3) The Ainu and the Ryukyuan; (4) Gene frequency cline in Japan.

I will start with a dendrogram (Fig. 1) based on the gene frequencies of 8 polymorphic blood proteins. The dendrogram shows clearly that there are three genetically distinguishable populations in Japan [1]. Incidentally, they correspond to three ethnic groups of Japanese Islands, namely, the Ainu of Hokkaido, the Ryukyuan (or Okinawan) of south-western Islands, and the majority of the Japanese inhabiting all of the Japanese Islands and called Wajin or Yamato, or Hondo-Jin. There is no consensus about how we may call ourselves. Because of the large standard error, further clustering of Wajin into several local groups is probably meaningless so far as this dendrogram is concerned.

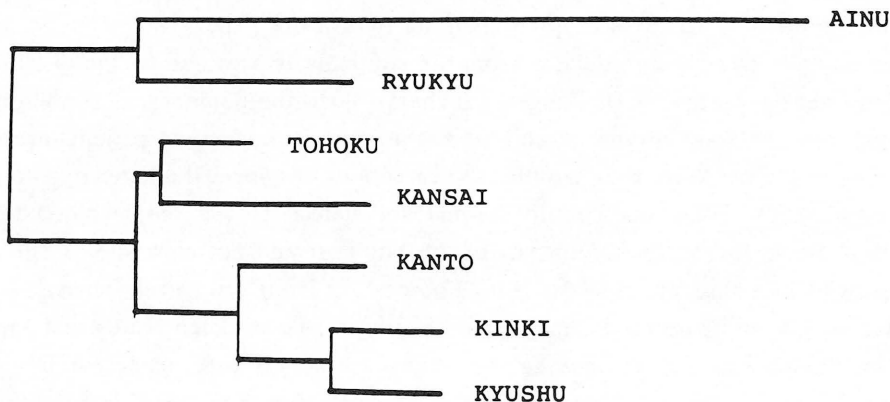


Fig.1. Dendrogram among 7 local groups of Japan based on gene frequencies of 8 polymorphic blood protein loci. The method of Cavalli-Sforza and Edwards (1967) was used.

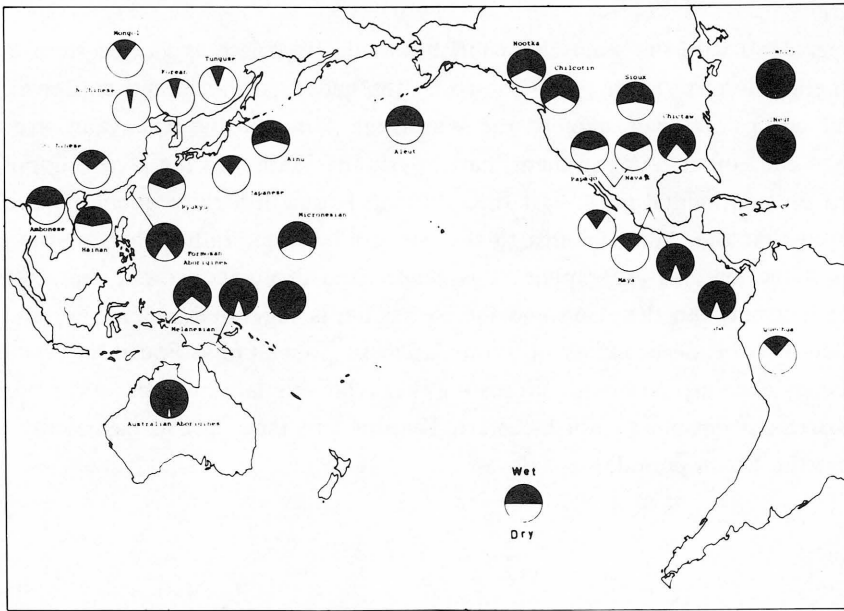


Fig. 2. The distribution of ear-wax types. Black portion indicates the frequency of wet type.

It is obvious that the Wajin is genetically greatly influenced by the continental Mongoloid groups. A good example to show this is the distribution of ear-wax type which is one of the genetic polymorphism of human being [2]. There are two phenotypes, wet and dry, and the dry type is homozygous for a recessive, autosomal allele. The peculiar fact is that the dry type is characteristic to the Mongoloid populations and has its center of distribution in northeast Asia (Fig. 2). In Japan, more than 80 percent of the Wajin have dry type, while in the Ainu the frequency of dry type is less than 40 percent, and in the Ryukyuan the value is less than 60 percent. So, it is quite likely that the ancestors of the Wajin came probably through Korean peninsula to Japan and mixed to a large extent with the native Japanese. I will come back to the problem of the earwax type in the later part of this paper.

Further example of gene introduction from the continent is provided by the distribution of acatalasemia gene reported by Dr. Shigeo Takahara [3]. In the homozygous condition of this gene, usual form of blood enzyme catalase is not produced, causing the patient severe symptoms leading to the necrosis in the mouth. The carriers of this gene, the heterozygote and called hypocatalasemia, are phenotypically normal and detected by a simple blood test. This gene occurs also in the north-eastern part of the Far East and not elsewhere. The average frequency of hypocatalasemia in Wajin is 0.23 percent. It is high in Korea, namely, 0.81 percent. Interestingly, in Tsushima Island situated in the middle between Korea and Japan, the value is intermediate at 0.40 percent. In the Ryukyus it is very rare, namely, 0.007 percent. Unfortunately no data of the Ainu is available. Nevertheless, this gene distribution clearly suggests migration from the continent to Japan.

Also, interesting data are provided recently by Dr. Hideo Matsumoto [4]. He showed that a gammaglobulin (Gm) type called  $ab^3st$  is a marker of northern Mongoloid populations and it occurs in Japanese in uniquely homogeneous frequencies. However, the different point of this

marker from the above mentioned markers is that no difference in gene frequency among the Ainu, the Wajin and the Ryukyuan are observed. Dr. Matsumoto considers that this gene has been introduced into Japan in very old time, perhaps prior to the Jomon period.

Now, I turn to the problem of the genetic affinities of the Ainu. In classical anthropology up to about 1960, the Ainu was often considered to belong to the Caucasoid race, mainly based on the facial appearance and hairiness. Our genetic study contradicts to this theory and put the Ainu clearly to the Mongoloid race as is shown by the dendrogram using 16 genetic markers (Fig. 3) [5]. Blood genetic marker studies also showed that the Ainu lack the Caucasoid marker such as blood group K and red cell enzyme AK\*2 and instead they have the Mongoloid marker such as blood group Diego (a) and serum protein transferrin Tf\*Dchi. [5, 6].

Furthermore, the Ainu have some interesting gene composition. They seem to lack red-green color blindness, and have extremely low frequency of PTC taste-blindness [7].

Today, most Ainu are more or less mixed. The average rate of admixture of our material, most of which we collected during the International Biology Program (IBP) from 1966 to 1972, was about 40 percent. So, I have corrected the gene frequencies before constructing the dendrogram. Now, if one hypothesizes that the mode of admixture is such that most genes introduced are from immigrant males rather than the females, it would be ideal if we had a maternally inherited genetic marker. Because male genes do not affect the maternal lineages, we could see genes from very old pure ancestral gene pool. In the last ten years, we have such a maternally inherited genetic marker, namely, mitochondrial DNA (mtDNA), which is the main topic of the next speaker, Dr. Satoshi Horai. We also have used mtDNA for our comparison of the Ainu and the Wajin [8]. The result was the same as in the above mentioned nuclear genetic markers, and the Ainu and the Wajin are genetically very close to each other.

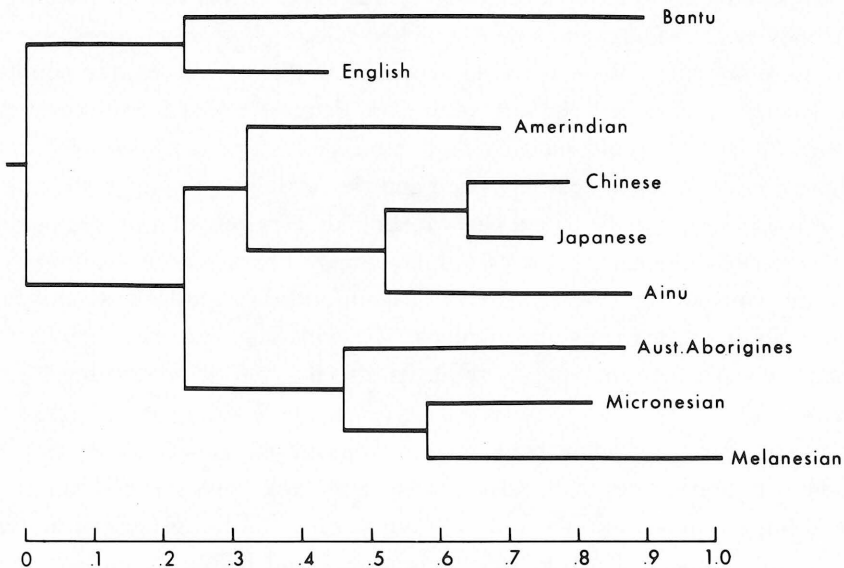


Fig.3. Dendrogram among 9 populations of the world showing the position of the Ainu. Gene frequency data of 16 polymorphisms (6 blood groups, 9 blood protein groups and ear-wax) were used by Cavalli-Sforza and Edwards' method.

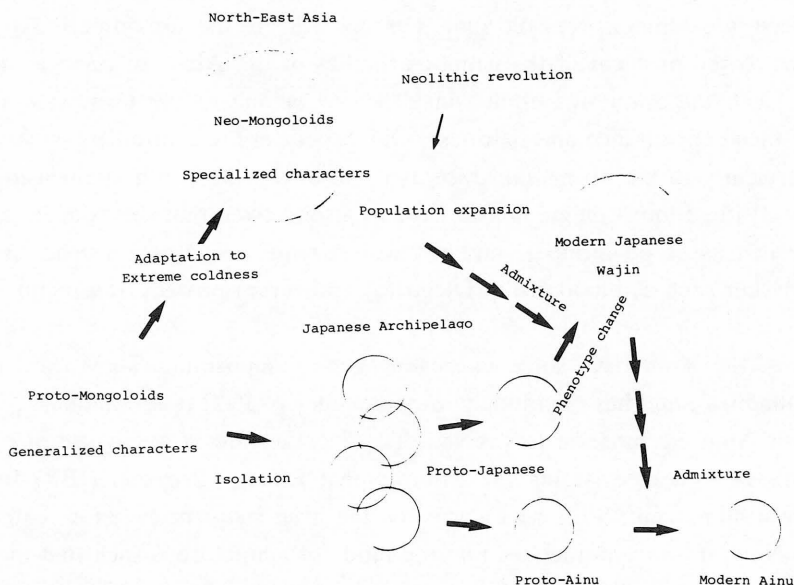


Fig. 4. Hypothetical diagram indicating the formation of modern Japanese and Ainu populations. Proto-Japanese may be understood as Jomonese.

If the Ainu is genetically close to east Asian peoples, how we explain the drastic difference in facial appearance and hairiness between them? One hypothesis may be as follows (Fig. 4). In the late paleolithic period there were populations with generalized morphological features in east Asia, which we may call Proto-Mongoloids. During the last glacial age with the peak at some 20,000 years ago, populations of north-eastern Asia suffering from extreme coldness phenotypically sustained a drastic change due to adaptation. This change includes flattened face, stocky body build, a flat eyelid with Mongoloid fold, and so on. Hairiness seems to have been disadvantageous among these northern peoples. On the other hand, the populations isolated in the Japanese archipelago did not suffer from extreme coldness and thus remained relatively non-specialized. I would imagine that some Proto-Japanese such as Jomon people were specifically hairy. After the glacial period and the neolithic revolution, the north-eastern population in Asia which may be called Neo-Mongoloids experienced a drastic population expansion, and some of them certainly migrated into Japan. Probably, the migration took place through Korean peninsula into western part of Japan, and then gradually to the eastern and northern parts, absorbing the native populations. The Ainu may have been the last of the native population who were phenotypically the least affected, but at present most of them are already mixed.

Now, let us consider the relation between the Ainu and the Ryukyuan. It may be argued that they both are descendants of Proto-Japanese, who have been relatively intact from the migrant Neo-Mongoloids giving rise to the Wajin because of the geographical remoteness. They share certain similarities in the facial appearance and hairiness, and as already mentioned they have relatively high frequency of wet ear-wax. However, the strongest evidence for their possible common origin comes from the frequency of a unique allele of Rh blood group, namely,  $r''$  or cdE. This allele is extremely rare all over the world, but as the excep-

tion it has appreciable frequency in the Japanese archipelago. The highest frequency is found among the Ainu, namely, 12.4 percent. It may be also relatively common in the Ryukyuan, namely, there is a record of 7 percent in Okinawa Island [9]. In Honshu, the frequency is about 3 percent. The distribution of this allele nicely fits the story of the origins of three Japanese populations mentioned above.

Finally, I will discuss about the geographical cline in Japan. It was well known that there is a cline in the frequency of the allele A of the ABO blood group system [10]. The allele frequency is about 30 percent in northern Kyushu, the westernmost Japan, and decreases as one goes east and north, until the frequency is 24 percent in Aomori, the northernmost part of Honshu. Until recently, no other example of cline was demonstrated. That is because the number of sample locality was not sufficient in the other genetic systems. Recently, I was able to show a cline similar to ABO blood group in the serum protein system called Gc, or vitamin D binding protein (DBP) [11]. A statistically significant cline was observed in the two alleles of Gc, Gc\*1F and Gc\*2, between Aomori, the northernmost Honshu, and Nagasaki, northwestern Kyushu. The interesting fact in this case is that this cline can be extended to the Ainu in the north and to the Koreans in Seoul in the west. Another serum protein, haptoglobin (Hp), may have also the cline of the similar kind.

While the above mentioned cline is the east to west cline in the Japanese main islands, there are examples in the north to south cline in the south-western Islands or Ryukyu Islands. Ear-wax is the best example, but the allele of red cell enzyme glutamate-pyruvate transaminase (GPT) shows also a cline. The simplest way of explaining the cause of cline is migration. It is noteworthy that the both clines of Japan, that of east to west and that of north to south, may have been brought about by migration probably from Korea to westernmost Japan. It is difficult to estimate from the genetic data the time when the migration took place. However, a theoretical paper by Aoki and Omoto (1980) dealing with the ABO gene frequency cline suggested, under various assumptions, that the population mixture started about 2,000 years ago, roughly corresponding the middle of Yayoi period [12].

It is also difficult to estimate from the genetic data how many people came as migrants from the continent. Judging from the high frequency of dry ear-wax among the Wajin, namely on the average 84 percent, while the Korean has 92.4 percent, the admixture rate of the migrant should have been very high. Dr. Ei Matsunaga who showed that the ear-wax type is one of the genetic polymorphism of humans, made a preliminary calculation [2]. If we assume that the frequency of dry type of the migrant people be 90 percent and that of the native Japanese be 30 percent, a simple calculation leads to an average admixture rate of 92: 8. Of course the situation is more complex because the admixture was not a single, but must have been a continuing event. Nevertheless, the admixture rate may have been quite high.

I am inclined to think that Dr. Kazuro Hanihara's recent estimate of a very high number of immigrants from the continent to Japan, namely, one million per one thousand years [13], is not unrealistic. Future studies including simulation experiments may certainly shed lights on the problem relating the origins and microevolution of the Japanese peoples.

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日本人の遺伝子構成上の若干の様相  
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この論文では、著者が過去25年間に行った研究に関連する次の四つのトピックスについて概説したい。(1)日本人の主流派である和人の遺伝的性質、(2)アイヌの遺伝的近縁性、(3)アイヌと琉球人、(4)日本人における遺伝子頻度の地理的勾配。

まず、遺伝的多型を示す血液蛋白の遺伝子頻度にもとづく遺伝距離とクラスター分析により、日本人(日本列島に住むヒト)は三つの主要な集団に分かれることが示された。すなわち、アイヌ、琉球人および和人(本土人)である。このうち和人には明らかに大陸からの遺伝子拡散の影響が見られる。よい例は耳垢型で、東北アジアに最も多い乾型が和人では80パーセント以上の頻度で見られるのに、アイヌでは40パーセント以下で、琉球人でも60パーセント以下である。このことは、和人の先祖が東北アジアより多分朝鮮半島経由で日本に渡来し、原住民と混



血したことを示唆する。同様の例は血液酵素異常の一種ハイポカタラセミアの分布にも見られる。また最近、松本秀雄は免疫グロブリンの遺伝標識の一種がシベリアから日本にかけ類似の頻度で分布することを見出している。

次に、アイヌについては、遺伝学的調査の結果、過去にしばしば提唱されたコーカソイド説は完全に否定され、モンゴロイドに属することが明らかとなった。彫りの深い顔や多毛性は原モンゴロイドの非特殊化形態とみなされ、原日本人から原アイヌへとひきつがれたものであろう。一方、寒冷適応により表現型が特殊化した東北アジアの新モンゴロイドは新石器時代以降に人口増大により拡散して日本列島へも渡来し、和人となった。

アイヌと琉球人とはいずれも原日本人集団に由来する可能性がある。このことを強く示唆させるものは Rh 血液型の r<sup>+</sup> (cdE) 遺伝子の頻度で、アイヌで12パーセントと高く、沖縄では7パーセントで、本州の3パーセントよりいずれも高い。

日本における遺伝子頻度の地理勾配については、ABO 血液型の A 遺伝子が従来唯一の例であったが、最近になって血液蛋白の Gc 型と Hp 型とに青森から長崎までの勾配が検出された。これらとは別に、南西諸島には耳垢型と赤血球酵素 GPT 型に勾配が見られる。これら、東西および南北の勾配は、弥生時代以降主として朝鮮半島から西日本に大陸からの移住があったと考えると最もよく説明がつく。