GROUP-SPECIFIC COMPONENT (Gc) POLYMORPHISM IN JAPANESE: AN INVESTIGATION ON THE PHENOTYPIC DISTRIBUTION WITH REGARD TO THE Gc\(^J\) ALLELE

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Summary Using the prolonged immunoelectrophoresis, Gc determination was performed in 1,347 plasma collected from 3 local populations in Japan. Six phenotypes determined by the codominant alleles, Gc\(^1\), Gc\(^2\) and Gc\(^3\), were observed, in which a new phenotype GcJ-J was included. This study revealed the presence of at least 3 polymorphic alleles at Gc locus in Japanese population.

INTRODUCTION

The group-specific component (Gc), now established to be the binding protein for vitamin D (Daiger et al., 1975), is one of the serum proteins which show genetic polymorphism in human populations. Since Hirschfeld (1959) first discovered three common phenotypes, Gc\(^1\)-1, Gc\(^2\)-1 and Gc\(^2\)-2, this Gc system has been utilized as a useful genetic marker for anthropological studies and in the field of forensic medicine.

In addition to the two common autosomal alleles, Gc\(^1\) and Gc\(^2\), at least 11 ‘rare’ electrophoretic variants have been detected in various populations, most of which occur sporadically in a few families (cf. Cleve, 1973; Johnson et al., 1975). Exceptions are Gc Ab (Aborigine) and Gc Chip (Chippewa), and they have been shown to be polymorphic (gene frequencies greater than 1%) in some population groups of Oceania (Gc Ab) and American Indians (Gc Chip) (Cleve et al., 1963).

Recently we encountered three family cases with Gc\(^J\) (Japan) variants in the course of paternity test carried out only in a year, and felt that the allele frequency for Gc\(^J\) is not extremely rare in this country (Nakajima et al., 1976; Ishimoto et al., 1976). However, there are no data on the incidence of this variant allele except for an initial report of a family with GcJ-2 (Omoto et al., 1972), although extensive population studies of Gc have been presented in Japanese.

Therefore, we conducted an examination of the Gc phenotypes among Japa-
nese with careful inspection, using the method of ordinary immunoelectrophoresis that was applied in most of earlier studies. Now, we can conclude from the result of 1,347 individuals that the variant allele, \( \text{Gc}^3 \), attains to be a polymorphic frequency in Japanese populations.

**MATERIALS AND METHODS**

In order to search the geographical variation of the phenotypic distribution within this country, the blood specimens were obtained from three different localities. They were from: 1) 337 blood donors in Fukushima Prefecture, Northeastern part of Japan. 2) 139 medical students at Tokyo Medical and Dental University and 295 healthy subjects living in Tokyo or its suburbs, which were mainly from the parents of family studies. 3) 346 medical students at Mie University and 230 blood donors in Mie Prefecture, Western Central part of Japan. To avoid any deterioration of the proteins, careful attention was paid for the storage of samples. About half of the plasma were examined within a week after bleeding in a fresh state. They were stored at 4°C prior to analysis. The other samples were once kept at -20°C for at most one month, but never thawed before use.

Agar gel immunoelectrophoresis was performed as described by Hirschfeld (1959) with a minor modification that electrophoresis allowed to proceed until the albumin front had migrated about 7 cm or more. This long run enhanced a good separation of Gc proteins and enabled us to determine the Gc variants. The antisera to develop the precipitation line were used both polyvalent anti-human rabbit sera prepared in our laboratories and commercially available anti-Gc sera (Behringwerke and DAKO). In some instances antigen-antibody crossed electrophoresis (Laurell, 1965) and immunofixation electrophoresis (Johnson *et al.*, 1975) were also carried out to ascertain the phenotype.

**RESULT AND DISCUSSION**

Six different phenotypes were classified as shown in Fig. 1. Among them 3 common phenotypes, Gc1-1, 2-1 and 2-2, were most popular and were observed in 94.9% of 1,347 sera examined. The less common phenotypes, GcJ-1 and J-2, showed peculiar patterns as described before (Nakajima *et al.*, 1976), and were observed in 3.5% and 1.4% of the samples, respectively. A new phenotype, proposed as GcJ-J, was also detected in 3 individuals. This type is characterized by having the fast moving variant with apparent lacking in any of the Gc 1 or Gc 2 component.

The two family materials with this homozygous phenotype were available, and the Gc determinations are presented in Fig. 2. As can be expected, the parents of the GcJ-J propositus had both the less common phenotypes with Gc J (Family Mat) and all children from the GcJ-J father showed the phenotype involving Gc J component (Family Om). Besides the pedigrees in Fig. 2, 15 families with either GcJ-1 or GcJ-2 have sofar been examined and the results are compatible with the