Two Cases of Duplication of the 1F and 1A2 Genes in the Vitamin D-Binding Protein (GC) System

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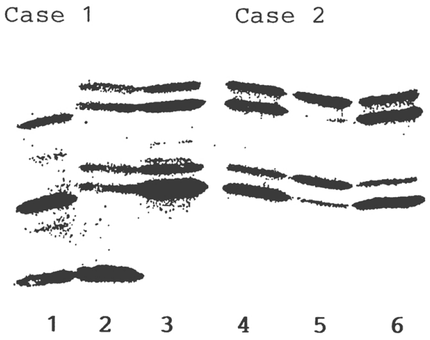
GC polymorphism in Japan is a bit unique in distribution of the phenotypes, because a variant allele(1A2) also retains in a polymorphic frequency (around 0.02) in addition to the three common alleles(GC 1F,1S & 2). Recently Yasuda et al. (1989)[1] found a new GC variant, GC 2-1F1A2, which is characterized by having double double-banded components with a common GC 2 band by PAGIF, and presumed it had arisen from duplication of the GC 1F and 1A2 genes. Later the same variant type was observed by others[2-4] in Japanese populations, and it is estimated such duplication is occurring with a frequency of 0.001 [2]. However, the variant has sofar been identified only in the samples with GC 2.

The samples described here were obtained from two cases of paternity test and their accused men were proven their fatherhood. GC types were determined by PAGIF(pH4.5-5.4, gel 0.2mm thick) followed by immunofixation. Densitometry was performed on stained membranes employing a Shimadsu double



1. Mother GC 2-1S
2. Child GC 2-1F1A2
3. Father GC 1F-1F1A2
4. Mother GC 1F-1F1A2
5. Child GC 1F1A2
6. Father GC 1F-1F1A2

Fig. 1. Photograph showing the GC variants.

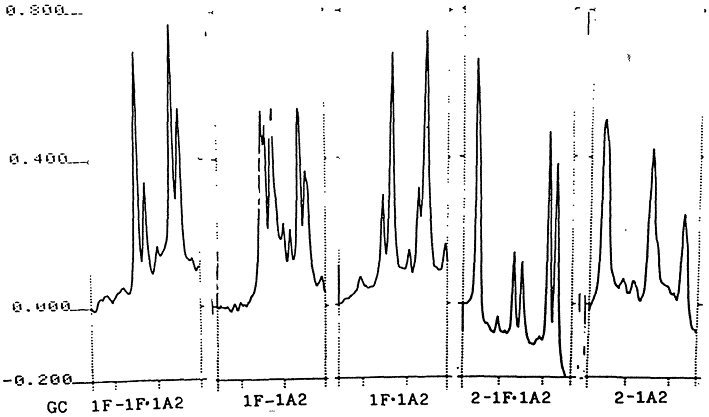


Fig. 2. Densitometric traces of the GC variants.

beam electroscanner(CS-930).

The GC patterns are shown in Fig. 1. In case 1, mother and the proven father had GC phenotype 2-1S and 1F-1A2, respectively, while their baby revealed a unique 5-band type, sharing only one band(GC 2) with the mother. The remaining four bands were obviously inherited from the father. The pattern is very similar to the socalled duplicated$^{1)}$ or recombined 3-allele$^{4)}$ variant in the previous reports. Therefore, the genotypes of the child and father were determined to be 2-1F1A2 and 1F-1F1A2,respectively.

In case 2, the mother and alleged father showed both GC 1F-1A2 phenotype, and their child had four GC bands in a position identical to those of 1F and 1A2, with the former bands very weak in intensity. Since the type is clearly different from that of GC 1A2 homozygote, we assumed it also arose from the same variant allele, and tried to examine it by scanning approach.

The densitometric traces in the samples with GC variants are presented in Fig. 2. It is evident that the scanning profile for GC 1F-1F1A2 is different from that of GC 1F-1A2. From the area calculation the ratio of 1F:1A2 in GC 1F-1F1A2 (father of case 1) is about 67:33, whereas the average ratio in GC 1F-1A2 is calculated to be 55:45. Using these figures the genotypes of the parents in case 2 were identified both to be 1F-1F1A2, which resulted the child should be GC 1F1A2. A position effect which was already pointed out in the original observation[1)] may be the most plausible explanation for the weak 1F factors in the homozygote.

1) Yasuda T, Ikehara Y, Takagi S, Mizuta K and Kishi K(1989) Hum Genet 82:89-91
2) Sawazaki K, Yamada T, Yasuda T, Mizuta K and Kishi K (1990) Hum Hered 40:187-189
3) Kishida T, Fukuda M and Tamaki Y(1991) Jpn J Legal Med 45:411-415
4) Kubo S, Kitamura O, Tsuda R, Hirose W, Matsumoto H and Nakasono I(1993) Hum Genet 91:71-72