C_3 Polymorphism in Korean Population***

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韓國人集團에서 C_3 多形 現象에 관한 硏究

吳 文 儒

Summary

The distribution of C_3 in Korean population has been investigated. In a total 1,115 individuals, 1,102 SS and 13 FS C_3 phenotypes were observed. The allele frequencies were; $C_3F = 0.00583$, $C3S = 0.99327$, respectively. This population was quite fit to the Hardy-Weinberg equilibrium.

Introduction

Since genetic polymorphisms of the third component of human complement were described, a lot of data have accumulated concerning their distribution in different populations. A polymorphism in C_3 was found by means of prolonged agarose gel electrophoresis by Alper and Propp(1968) and by Azen and Smithie (1968) using high-voltage starch gel electrophoresis. Both groups made observations consistent with the theory that the phenotype of C_3 is governed by autosomal co-dominant inheritance and presented evidence for the existence of two relatively frequently occurring genes as well as of rarer genes controlling this polymorphism.

High voltage (20V/cm) agarose gel electrophoresis in the study of C_3 polymorphism was used by Teisberg (1970), and he found that the method was quick, simple and gave a good separation of C_3 proteins. After that, many authors have been used the high voltage (20V/cm) agarose gel electrophoresis in the study of C_3 polymorphism.

Some authors used the immunofixation method on cellulose acetate strips (A. Germenis et al, 1982). According to the report of Z. Tongmao (1983) using high voltage agarose gel electrophorsis, in a total 388 individuals of Chinese population, 385 SS and 3 FS C_3 phenotypes were observed.

There is no information about the distribution of C_3 phenotypes in Korean population. In this paper, the phenotypes and gene frequencies of C_3 in Korean population, will be reported and compared with those in other populations.

Materials and Methods

Serum samples were obtained from unrelated, healthy adult individuals in Cheju-do population. The samples were stored at below -20°C. High voltage (20V/cm) agarose gel electrophoresis was used for the phenotyping of C_3, with barbital buffer

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*** This study was supported by the Korean Ministry of Education in 1983.

(pH 8.6), according to the method of Teisberg (1970).

To identify the C_3 bands, immunofixation on cellulose acetate strips was used, according to the methd of Germenis *et al* (1982).

Results and Discussions

Fig. 1 shows the relative mobility of the common C_3 phenotypes (C_3 SS, C_3 FS) obtained by agarose gel (1.25% agarose, 1mm thick) electrophoresis.

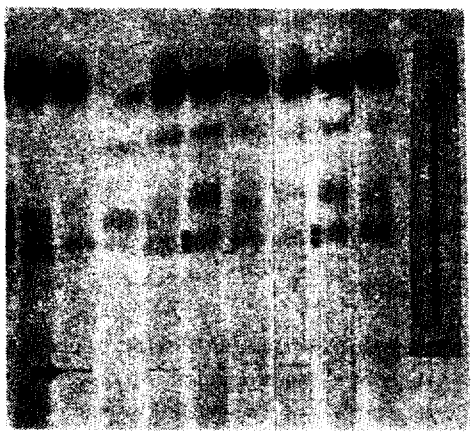

Fig 1. The common C_3 phenotypes after high voltage agarose gel electrophoresis. Arrow indicates the sample applying position and the dotted bands indicate FS types.

Fig. 2 shows the bands obtained by immunofixation on cellulose acetate membrane.

A total of 1,115 serum samples has been examined to determine the C_3 phenotypes by high voltage agrose gel electrophoresis and by immunofixation. Among 1,115 serum samples, 1,102 (98.83%) were SS, 13 (1.17%) were FS, but no FF samples were found (Table 1).

Gene frequences were; $C_3F = 0.0058$, $C_3S = 0.9933$ (Table 1). This population was shown in a good agreement with that assuming a Hardy-Weinberg equilibrium.

In Chinese population by Z. Tongmao (1983), the gene frequencies were; $C_3F = 0.0039$, $C_3S = 0.9961$. In these two Monoloid populations, the gene frequencies of C_3F were shown quite low (0.0058 and 0.0039).

On the other hand, in the Caucasoid populations, the gene frequencies of C_3F were higher than those of the Mongoloid populations; 0.25 (C.A. Alper *et al*, 1968), 0.21 (E.A. Azen and O. Smithies, 1968), 0.21 (E.A. Azen and O. Smithies, 1969), 0.19 in Norwegian population (P. Teisberg, 1970), 0.23 in Swedish population(R. Broennestam, 1971),0.1944 in Swedish population (R. Broennestam, 1973), 0.1702 in Finnish population (H. Arvilommi, 1973 in First Internationl Symposium and Workshop on

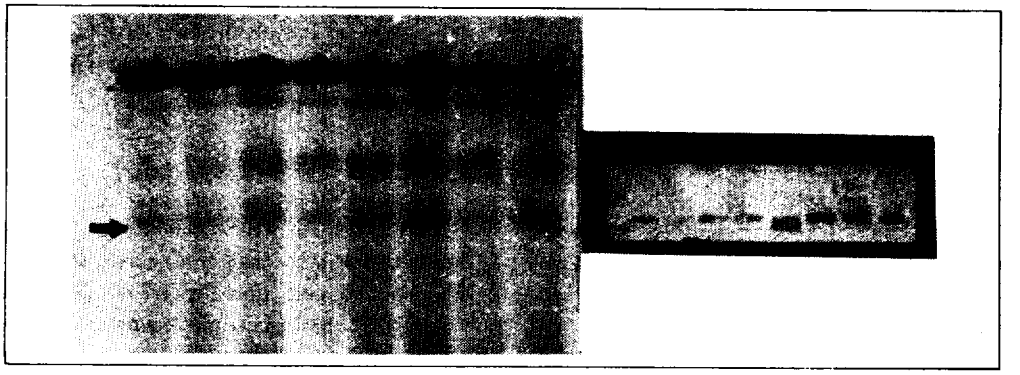

Fig 2. C_3 phenotypes seen by immunofixation on cellulose acetate membrane(right) and agarose gel electrophoresis patterns of C_3(left). Arrow indicates the sample applying position. Positive is at the top.

the polymorphism of Third Component of the Human Complement System), 0.2139 in Hessen (W. Spielmann, 1973), 0.221 in Swiss population (R. Pflugshaupt *et al*, 1973), 0.1990-0.2238 in four Spanish populations (H.W. Goedde *et al*, 1973), 0.193 in German of Cologne area (G. Mauff et al, 1974), 0.1939 in North German (S. Seth and H. Berndt, 1974), 0.14 in Norway (K. Berg and A. Heiberg, 1970), 0.207 in German and 0.218 in Spanish population (H.W. Gedde *et al*, 1970).

But, in Finnish Lapps and Greenland Eskimos populations, the C_3F gen frequencies were shown lower than those of the other Caucaoid populations, 0.05-0.056 and 0.0889-0.0316, respectively (K. Berg *et al*, 1972, E. Stoffersen *et al*, 1982, Arvilommi *et al*, 1973). These two populations (Finnish Lapps and Greenland Eskimos) are quite similar to Mongoloid populations in C_3F gene frequencies (Fig. 3).

On the other hand, the C_3F gene frequency was 0.04 in North American Blacks (E.A. Azen *et al*, 1969).

Table 1. Distribution of C_3 phenotypes and corresponding allele frequencies.

		Phenotypes		
No.	Total	FF	FS	SS
Obs.	1,115	0	13	1,102
Per cent	100.0	0	1.17	98.83
Exp.		0.0378	12.9	1,100.1

allele frequencies;

$S = 0.99327$

$F = 0.00583$

$X^2 = 0.0041$ $\quad 0.95 > P > 0.80$

Fig 3. Shows that Mongoloid populations are the lowest in gene frequencies of C_3F, Caucasoid populations the highest, and the Hlack seem to be situated between these two populations.

According to the reports of D.D.Farhud *et al*(1972), there was a significant excess of FF phenotypes in patients with rheumatic factor while in hepatitis the SS phenotype was significantly lower and a relatively high frequency of FF was stated. On the other hand, R.G. Holzhauer *et al* (1976) stated in their report that the C_3 levels were correlated with the cystic fibrosis

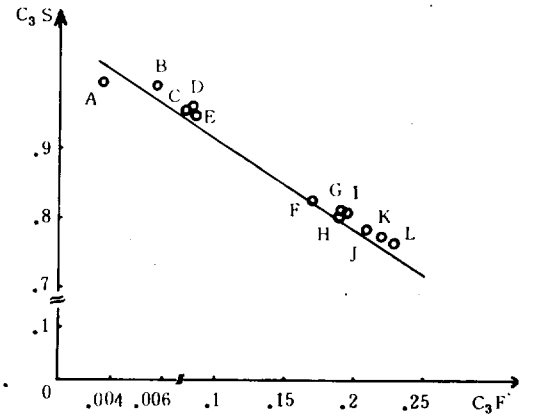

Fig 3. Distribution of the C_3 gene frequencies by the populations.

A ; Chienese B ; Korean
C ; American Blacks D ; Finn, Lapp
E ; Eskimos F ; Finn
G ; Icelanders H ; German
I ; Norwegian J ; Spanish
K ; Swiss L ; Swedish

satients' clinical status and the mean C_3 level was significantly higher (153 ± 16 vs. 136 ± 21; $p<0.001$), but no significant differences were found in the C_3 phenotypes or the S and F gene frequencies. We are sorry to say that we couldn't have a chance to study on the differences of C_3 gene frequencies between healthy population and the patient population, because of the difficulties of blood sampling from the patients.

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國 文 抄 錄

韓國人集團에서의 C_3 多形현상을 Agarose gel 電氣泳動法 및 Immunofixation 方法으로 총 1,115명을 대상으로 조사하여 다음과 같은 結論을 얻었다.

총 1,115명 중 FS type은 13명(1.17%)이었으며 SS type은 1,102명(98.83%)으로 나타났고 FF type은 한명도 발견치 못하였다.

Allele frequency는 $C_3F = 0.0053$, $C_3S = 0.9933$으로, C_3F 유전자빈도는 中國人集團(0.0039)보다는 조금 높게 나타났지만, 白人집단 보다는 아주 낮았다. 이러한 현상은 Finnish Lapp 및 Eskimo 집단에서도 아주 낮게 나타남으로써, Mongoloid 집단의 marker gene으로 이용할 수 있을 것으로 본다.