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GENETIC POLYMORPHISM OF THE HUMAN COMPLEMENT COMPONENT C3 IN THE POPULATION OF NORTHERN POLAND[1]

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Summary. Studies on the human complement component C3 were carried out. An object of the studies were blood samples taken from adult, unrelated persons. The sera were separated by agarose gel electrophoresis. Agarose manufactured by Windsor Berkshire Co. was applied to veronale buffer at pH 8.6. The C3 components were detected in Coomassie Blue R 250 solution and in decolorizer. The analysed material was found to have three phenotypes: two homozygous C3F and C3S and one heterozygous C3FS. In one case the phenotypic variant $C3FS_{0.4}$ was observed.

The applied method of electrophoretic separation and a high degree of the component C3 genetic polymorphism permits to use that trait in studies on serological differentiation of human population.

Genetically determined polymorphism of the human complement component C3 was displayed by Wieme and Demeulenaere (1967) and Alper and Propp (1968). It has been found that two codominant alleles $C3^F$ and $C3^S$ determine the occurrence of three frequent phenotypes defined as C3S, C3F and C3FS. Their frequencies were the subject of population studies performed by Teisberg (1971), Dissing et al. (1971), Turowska et al. (1976) and Scacchi et al. (1979). The presence of rare phenotypic varients of C3 was also revealed Agarwal et al. (1972), Farhud and Walter (1972), Schlesinger et al. (1979).

This paper presents results of the studies on C3 distribution in the population of northern Poland.

MATERIAL AND METHODS

Phenotypes of the C3 components were determined in the blood serum of 420 normal, unrelated adults, applying to the Forensic Department of the Medical Academy in Gdańsk to establish paternity and in the blood serum of 120 mothers and their children.

The studied serum was separated from blood immediately after its coagulation

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and stored at −20°C till the moment of use. The C3 phenotypes were determined on the first or second day after blood sampling and also in the serum stored at −20°C during several days. A high voltage agarose gel electrophoresis according to the method of Teisberg (1970) with our own modifications was used. For the studies were used veronalo-lactate buffer at pH 8.6 and 1% agarose, manufactured by Windsor Berkshire, England. To the agarose gel on a glass 20×10 cm plate 1.5 cm apart from the cathode margin was introduced each time 1 l of the studied serum. The electrophoresis was conducted for 2.5 hours at 490 V and 45 mA using cooling. The preparation was then fixed for 30 min at 4% acetic acid, rinsed in distilled water and dried under filter paper at a room temperature. The C3 component was detected after 30 min of staining in 1% solution of Coomassie Blue R 250 and decolorizing in the solution of: methanol, distilled water, acetic acid (in the ratio 1 : 4 : 1).

RESULTS AND DISCUSSION

The activity zones of the C3 component were detected in the serum samples subjected to studies during 1 - 4 days after taking blood. After a longer time of serum storage at −20°C the spectral lines became broadened and after 6 days — the pattern was unreadable.

The analysed material was found to have 3 frequent phenotypes of C3: a slow-migrating homozygous type S, fast-migrating F and heterozygous type FS (Fig. 1). In one case a rare phenotypic variant $C3FS_{0.4}$ was detected.

The pictures of the observed C3 phenotypes are presented in Fig. 1. The phenotype and gene frequencies in the analysed population are summarized in Table 1.

Table 1. C3 phenotype and gene frequencies in the North Polish human population

Phenotype	Phenotype frequencies				χ^2	Gene	Gene frequencies
	observed		expected				
	n	%	n	%			
C3S	284	0.6762	283	0.6719	0.0035	$C3^S$	0.8195
C3FS	120	0.2857	123	0.2926	0.0731	$C3^F$	0.1785
C3F	15	0.0357	13	0.0317	0.0225	$C3^W$	0.0020
C3W	1	0.0024	1	0.0038	1.0000		
Total	420	1.0000	420	1.0000	1.0991 ($0.70 > p > 0.50$)		1.0000

No significant differences were found between the observed and expected phenotype and gene frequences. Table 2 presents a comparison of the gene frequency in the studied population with the frequencies in other populations.

As follows from Table 2 the gene frequencies of the studied population are very similar to those observed in other regions of Poland, but show no significant differences in relation to other European populations. A significantly smaller polymorphism

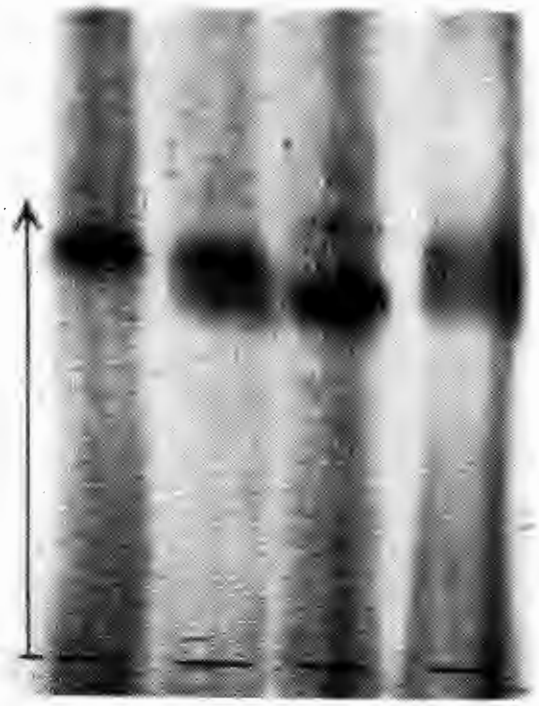

Fig. 1. C3 phenotypes: from left C3F, FS, S, FS

Table 2. Frequency of C3 genes in human populations from some countries

Country	Number	C3S	C3F	C3 rare	References
Poland	420	0.8195	0.1785	0.0020	Own studies
Poland	307	0.8127	0.1873	—	Turowska et al. (1976)
Norway	400	0.8000	0.1900	0.0070	Teisberg (1970)
Denmark	406	0.8160	0.1820	0.0020	Dissing et al. (1971)
Italy	252	0.8170	0.1790	0.0040	Scacchi et al. (1979)
Angola	268	0.9500	0.0480	0.0010	Kühnl et al. (1972)

of the C3 component was found in the negroid population. The study of that protein fraction phenotypes in 120 mothers and their children was used for verification of the inheritance model of C3 types. The obtained results are given in Table 3. The

Table 3. Distribution of C3 phenotypes of mothers and their children

Phenotype of mother	Child						Total	χ^2
	S		FS		F			
	Number							
	observed	expected	observed	expected	observed	expected		
S	68	67.4	14	14.1	—	—	82	0.006
FS	14	14.3	18	17.1	2	2.9	34	0.332
F	—	—	3	3.2	1	1	4	0.013
Total	82		35		3		120	0.351
							$(0.90>p>0.80)$	

numerical phenotype distribution found in children in individual groups of mothers with a definite phenotype does not deviate from the expected one. In no case a homozygous child (C3S or C3F) was found to have a phenotype opposite to homozygous mother.

CONCLUSIONS

1. The methodics of type determination of the C3 component applied in this paper permits to obtain readable and repeatable results.

2. The study of C3 phenotypes of mothers and their children confirms a codominant inheritance model of that trait.

3. A relatively high polymorphism of the C3 distribution permits to use it in population genetics.

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POLIMORFIZM GENETYCZNY KOMPONENTU C3 KOMPLEMENTU SUROWICY LUDZKIEJ W POPULACJI POLSKI PÓŁNOCNEJ

Streszczenie

Przeprowadzono badania polimorfizmu komponentu C3 komplementu surowicy ludzkiej w populacji Polski Północnej. Przedmiotem badań były próbki krwi pobrane od dorosłych, niespokrewnionych osób. Surowice poddano rozdziałowi elektroforetycznemu w żelu agarozowym. Stosowano agarozę firmy Windsor Berkshire w buforze weronalowym o pH 8.6. Komponenty C3 ujawniano w roztworze Coomassie Blue R 250 i w odbarwiaczu. W anamateriale stwierdzono występowanie dwóch fenotypów homozygotycznych C3F i C3S oraz fenotypu heterozygotycznego C3FS. W jednym przypadku obserwowano wariant fenotypowy $C3FS_{0,4}$.

Zastosowana metoda rozdziału elektroforetycznego i wysoki stopień polimorfizmu genetycznego komponentu C3 pozwala na wykorzystanie tej cechy w badaniach nad zróżnicowaniem serologicznym populacji ludzkiej.

ГЕНЕТИЧЕСКИЙ ПОЛИМОРФИЗМ КОМПОНЕНТА С3 КОМПЛЕМЕНТА ЧЕЛОВЕЧЕСКОЙ СЫВОРОТКИ В ПОПУЛЯЦИИ СЕВЕРНОЙ ПОЛЬШИ

Резюме

Проведены исследования компонента С3 комплемента человеческой сыворотки. Предметом исследований были пробки крови, взятые от взрослых, неродственных особ. Сыворотки были подвергнуты электрофоретическому разделу в агарозовом геле. Использовался агароз фирмы Windsor Berkshire в вероналовом буфере с pH 8,6. Компонент С3 был обнаружен в растворе Coomassie Blue R 250 и в обесцвечивателе. В анализируемом материале было обнаружено три фенотипа: два гомозиготных C3F и C3S, а также гетерозиготный C3FS. В одном случае наблюдался фенотипический вариант $C3FS_{0,4}$.

Применённый метод электрофоретического раздела и высокая степень полиморфизма компонента С3 позволяют использовать этот признак в исследованиях серологической дифференциации популяции человека.