Kasuistik / Casuistic

## Gc Subtypes Determined by Ultrathin-Layer Isoelectric Focusing. Distribution in the Veneto Population (Italy)\*

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**Summary.** The distribution of Gc phenotypes in the population of Veneto was investigated by ultrathin-layer isoelectric focusing. In our sample (n = 732) the six common phenotypes, Gc1S, 1F, 1S1F, 2, 2-1S, 2-1F and a further phenotype, Gc1S1C3, were observed and the following frequencies calculated: Gc1S = 0.560792; Gc1F = 0.159153; Gc2 = 0.277323; Gc1C3 = 0.002732. Our gene frequencies have been compared with those found in other populations.

**Key words:** Group-specific component, polymorphism – Veneto population, Gc subtypes

**Zusammenfassung.** Die Verteilung der Gc-Phänotypen wurde bei der venetischen Bevölkerung mittels Isoelektrofokussierung auf sehr dünnem Gel untersucht. In unseren Stichproben wurden sechs gemeine Gc-Phänotypen: 1S, 1F, 1S1F, 2, 2-1S, 2-1F, und ein weiterer Phänotyp Gc1S1C3 beobachtet und folgende Frequenzen berechnet: Gc1S = 0.560792; Gc1F = 159153; Gc2 = 0.277323; Gc1C3 = 0.002732. Die von entdeckte Genfrequenz wurde mit den bei anderen Bevölkerungen gefundenen Frequenzen verglichen.

Schlüsselwörter: Gc-System, Polymorphismus – venetische Bevölkerung, Gc-System – Blutgruppen, Gc-System

The group-specific component (Gc) polymorphism was originally detected by Hirschfeld in 1959 using immunoelectrophoresis on agar gel [1, 2]. By means of gel electrophoresis on starch or polyacrylamide gels [3–5] it was possible to identify three common phenotypes (Gc1-1, Gc2-2, Gc2-1) determined by two autosomal codominant alleles Gc<sup>1</sup> and Gc<sup>2</sup>. In 1975, Daiger et al. [6] clarified the biologic function of Gc, i.e., that it is a vitamin D-binding protein.

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By means of the new technique of isoelectric focusing, Constans and Viau [7] observed in 1977 a further heterogeneity of the Gc<sup>1</sup> allele, which is the sum of two alleles, Gc<sup>1S</sup> and Gc<sup>1F</sup>, and distinguished a three-allele model and six common phenotypes (Gc1S, Gc1F, Gc2, Gc2-1S, Gc2-1F, Gc1S-1F). Subsequently, Constans et al. [8] demonstrated the application of this classification method to population studies. At an International Workshop held in 1978 [9] it was demonstrated that the Gc phenotypes were determined by 30 different alleles, 21 double-band variants and nine single-band variants. Double-band variants were designated A (anodal) or C (cathodal) to indicate the anodal or cathodal positions relative to the common Gc1S band. Lately, several new variants have been observed [10–13]. In our study we observed six common Gc phenotypes and a variant phenotype, 1S1C3, which was classified by Dr. Weidinger of Munich (FRG). In this paper we report the distribution of Gc subtypes and their frequencies in the Veneto population.

#### **Materials and Methods**

Sera from 732 unrelated, apparently healthy blood donors were provided by the Transfusion Center of the Civil Hospital of Padua (Italy). Gc typing was performed by ultrathin-layer isoelectric focusing on polyacrylamide gels ( $250 \times 120 \times 0.3$  mm), on a LKB 2117 Multiphor apparatus connected to a LKB 2103 Power Supply. Each gel was brought to a final concentration of 4.85% acrylamide (w/v), with 3% ampholine in the 4–6 pH range and 15% glycerol (v/v). After 15 min of degassing, polymerization was achieved with 0.045% ammonium persulfate (w/v). The electrodic solutions used were 0.1 *M* glutamic acid in 0.5 *M* H<sub>3</sub>PO<sub>4</sub> (anolyte) and 0.1 *M*  $\beta$ -alanine (catolyte). After 30 min of prefocusing, undiluted sera were applied at 2 cm from the cathodal end by means of small papers (Whatman 3MM, 7 × 4 mm), and focusing was carried out for 4 h (the papers were removed after 45 min) with the following maximal conditions: 2000 V, 35 mA, 20 W; cooling temperature was kept at + 5°C. Evaluation of Gc bands was carried out as described by Kühnl et al. [14].

#### **Results and Discussion**

The distribution of Gc phenotypes and gene frequencies in the population of Veneto is shown in Table 1.

Besides the six common Gc phenotypes, a variant phenotype was observed in four samples. The distribution fits in well the Hardy-Weinberg equation, as shown by the  $\chi^2$  value.

Our results were compared to those obtained in other Italian regions and in other countries. All these gene frequencies are reported in Table 2. As regards the comparison with other Italian regions, we can see that in some cities, such as Arezzo, Roma, Lecce, Gc gene frequencies vary significantly from ours, but they concern a limited population. Instead, gene frequencies found in wider populations (Tuscany, Italy) are similar to ours. Besides Veneto, gene frequencies do not differ significantly from those found in most of the other European populations, except for regions in Germany (Hessen), France (Pyrenees), and Iceland. However, as regards the Pyrenees, we can think of an endogamic population, as the region itself shows. The same can be said for Iceland, which

Gc phenotypes	No. observed	Observed (%)	No. expected	
15	230	31.42	230.20	
1F	19	2.59	18.54	
1 <b>S</b> 1F	129	17.62	130.66	
2	56	7.65	56.30	
2-1S	228	31.15	227.68	
2-1F	66	9.02	64.62	
1S-1C3	4	0.55	2.24	

Gene frequencies: 1S = 0.560792; 1F = 0.159153; 2 = 0.277323; 1C3 = 0.002732 $\chi^2 = 0.064$ 

For 
$$3 df 0.9950 < P < 0.9990$$

Table 2. The distribution of Gc subtypes in	Veneto as compared to other popula	tions
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No. of cases		1 <b>S</b>	1F	2	V
	Veneto (this study)	0.560159	0.159153	0.277323	0.002732
244	Arezzo [15]	0.6455	0.2459	0.1086	
104	Roma [15]	0.6490	0.2260	0.1250	
162	Lecce [15]	0.6235	0.2469	0.1269	
965	Tuscany [16]	0.5934	0.1480	0.2570	0.0016
147	Italy [17]	0.5884	0.1497	0.2619	
440	Germany (Munich) [17]	0.5920	0.1443	0.2614	0.0023
146	Germany (Marburg) [17]	0.5514	0.1781	0.2705	
261	Germany (Hessen) [14]	0.603	0.125	0.272	
680	Germany (Hessen) [18]	0.5978	0.1412	0.2610	
1156	Western Germany [19] (Düsseldorf region)	0.5476	0.1561	0.2963	
744	Belgium [20]	0.5477	0.1727	0.2796	
290	France (Pyrenees) [8]	0.512	0.077	0.410	
195	Northern Israel [17]	0.5436	0.2231	0.2308	0.0025
253	Bolivia [8]	0.636	0.231	0.122	0.009
1674	Denmark [21]	0.5717	0.1589	0.2688	0.0006
243	North Indian population [22]	0.519	0.191	0.290	
385	Iceland [23]	0.631	0.107	0.262	
357	Senegal [24]	0.115	0.780	0.053	0.0052
510	Japan Mie [25]	0.241	0.465	0.263	0.032
531	Japan Tokyo [25]	0.241	0.487	0.246	0.027
110	USA, Pennsylvania [26] (Whites)	0.572	0.149	0.279	
273	USA, Philadelphia [26] (Negroes)	0.169	0.685	0.126	0.020
219	USA, Georgia [26] (Negroes)	0.121	0.790	0.080	0.009
267	Central African Empire [8]	0.191	0.584	0.064	0.161

 
 Table 1. The distribution of Gc
phenotypes and gene frequencies in the Veneto population is an island, and then the inhabitants, of course, constitute a segregated population.

At least, significant differences are found in the comparison between our gene frequencies and those of other ethnic groups. However, as far as the US population is concerned we can say that, while the Black people's frequencies obviously differ from ours, those of the Whites closely resemble those obtained in our study.

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