TYPES OF ERYTHROCYTIC GLUCOSE-6-PHOSPHATE DEHYDROGENASE IN NORMAL AND IN DEFICIENT TURKISH SUBJECTS*

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A deficiency of glucose-6-phosphate dehydrogenase (G-6-PD) of the erythrocytes is responsible for a group of hemolytic disorders such as primaquine-induced hemolytic anemia, favism, neonatal hyperbilirubinemia and some types of congenital non-spherocytic anemias.[1] Quantitative and qualitative variations of erythrocytic G-6-PD have been demonstrated in recent years.[2-5] Two commonly occurring forms of G-6-PD, namely A and B, have been identified in the erythrocytes of American Negroes by starch gel electrophoresis.[6,7] Negro males with G-6-PD deficiency usually have the A phenotype. The B phenotype, which is identical in electrophoretic migration to G-6-PD from erythrocytes of most Causasians, moves more slowly than type A on starch gel electrophoresis. About 35 per cent of Negro males are reported to possess the A form. Negro females may have any of the forms A, B or AB. Males may have either A or B, but not AB. The appearance of the AB, or hybrid form only in females suggests that the type of electrophoretic variation of G-6-PD is also X-linked. Family studies by various investigators have confirmed this hypothesis.[4,5]

Further investigations have revealed that the genes for G-6-PD deficiency and the type A variation are usually carried on the same X chromosome. On the other hand, the gene for normal G-6-PD activity may be either on the X chromosome bearing the A gene or on the one bearing the B gene. Thus, the following phenotypes may

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be designated: type A and normal enzyme activity (A+); type A and enzyme deficiency (A—); type B and normal enzyme activity (B+); and type B and enzyme deficiency (B—). Negroes rarely have this last phenotype (B—), and Caucasians posses phenotypes (B+) or (B—). Continued work on the subject has shown that some uncommon electrophoretic variants of the enzymes exist in different ethnic groups among American and Nigerian Negroes, Sardinians and Papuans. Porter et al reported that 13 G-6-PD variants (types A—, A+, B+, B—, Baltimore, Eyssen, Barbieri, Ibadan, Sardinia-1, Sardinia-2, Seattle, and two types unnamed by Kirkman et al), had been distinguished by 1964.[8] However, additional variants (Tel-Hashomer,[9] Canton,[10] Chicago,[11] Chinese[12]) in other population groups have been reported in recent publications.

Erythrocytic G-6-PD deficiency has been reported to be the cause of various hemolytic disorders in Turkey,[13, 14] but the types of electrophoretic variants of G-6-PD in the Turkish population have not been reported. In this paper, the electrophoretic pattern of the enzyme in Turkish subjects determined by starch gel electrophoresis will be presented.

Material and Methods

Five apparently unrelated mothers whose sons exhibited neonatal jaundice caused by G-6-PD deficiency were selected, along with their six sons and two daughters. One husband and two healthy laboratory technicians were also included in the study (Table 1). Blood from these subjects was drawn into sterile tubes containing ACD solution, and the specimens were kept at 4 C for 24 hours, until they could be shipped to the United States by air under refrigeration. Starch gel electrophoresis was done according to the method of Bowman et al,[15] with the exception that glucose-6-phosphate was substituted for 6-phoshogluconate. Controls were run with hemolysates from B(+) and B(—) Caucasian subjects. Enzyme determinations for erythrocytic G-6-PD were done according to the method of Narahara and Özand[16] as described in a previous publication.[14]

Results and Conclusions

This study indicated that the electrophoretic pattern of erythrocytes from G-6-PD deficient Turks is type B, the same as that seen in other Caucasians. It is well known that certain ethnic

TABLE 1

ELECTROPHORETIC PHENOTYPES OF G-6-PD IN RED CELLS FROM DEFICIENT AND NORMAL TURKISH SUBJECTS

Family No.	Case No.	Sex	G-6-PD assay phenotypes	G-6-PD units/10 ml RBC	G-6-PD phenotype on starch gel electrophoresis	Miscellaneous
1	1	F	unaffected	15.00 units	B	mother
	2	F	? intermediate		B	daughter
	3	M	affected	0.23 units		son, (16 months old, jaundiced 2 days old and had 5 exchange transfusions)
2	4	F	intermediate	5.70 units	B	mother
	5	M	affected	4.10 units	B	son (2.5 years old, jaundiced at 1 day of age. Had 5 exchange transfusions. Has mild sequalae of kernicterus)
	6	M	affected	3.50 units	B	son (had a mild jaundice when 3 days old)
3	7	M	unaffected	18.30 units	B	father
	8	M	affected	0.25 units	B	son (2.5 years old, came with hemoglobinuria after ingestion of fava beans)
	9	F	? unaffected	15.60 units	B	mother
4	10	F	? unaffected	9.00 units	B	mother
	11	F	intermediate		B	daughter
5	12	F	intermediate	5.90 units	B	mother
	13	M	affected	0.17 units	B	son
	14	M	unaffected	8.00 units	B	son
	15	M	unaffected		B	lab. technician (healthy)
	16	M	unaffected		B	lab. technician (healthy)

groups have relatively prevalent deficiencies of G-6-PD activity which are associated with specific clinical problems such as drug-induced hemolysis encountered in Negroes with the enzyme form A—. Individuals carrying the Mediterranean type of the enzyme also exhibit unexplained neonatal jaundice,[14, 17] and susceptibility

to favism, in addition to drug-induced hemolysis. Further studies may help to determine whether or not other possible variants exist in the Turkish population.

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