The glucose-6-phosfate dehydrogenase (G6PD) deficiency: clinical, biochemical and molecular features

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The aim of this work was to evaluate the presence of G6PD deficiency in healthy blood donor population and to study G6PDpatients as regards clinical, biochemical and molecular features.

Methods: 1900 blood donors of Policlinico Umberto I were screened for G6PD deficiency by differential pH-metry (CL-10 PLUS, Biocontrol). 1032 patients from S. Eugenio Hospital were studied for diagnosis of G6PD deficiency. The definitive diagnosis of G6PD deficiency was achieved by a diagnostic protocol including family study and both enzymatic and molecular test. G6PD and PK activities and GSH concentration were determined by spectrophotometric methods; hematological parameters were determined by ADVIA 120 (Siemens). The molecular characterization of G6PD variants was performed by ARMS, RFLP and DNA sequence analysis.

Results: 21/1900 blood donors were G6PD deficient and the main Italian G6PD variants G6PD Seattle 844C, Mediterranean 563T and A- 376G,202A were found. 507/1032 patients were G6PD deficient and the DNA analysis confirmed the frequency distribution of G6PD variants in Central Italy: 63% G6PD Mediterranean, 15% G6PD Seattle, 5% G6PD A-, 3% G6PD Chatam 1003G,and 1% for G6PD Union 1360T, Cassano 1347C, and Sibari 634G. Moreover single cases of G6PD Ube 241T and G6PD Radlowo 679T were detected and three novel mutations were characterized: nt 130 GCC-ACC; nt 1021 GTC-TTC, and nt 751 GAT-AAT. The results of biochemical and hematological analyses showed significant differences between normal and Mediterranean patients. No difference between Seattle subjects and normal ones was found. Nevertheless in hemizigous A- subjects a significantly lower GSH content than normal one was observed. The acute hemolytic anemia occurred in 63/328 patients with Mediterranean variant, in 2/80 patients with Seattle variant, and in 13/25 A- patients. The hemolytic attack was triggered mainly by drugs.

Conclusions: Though the A- variant is considered milder than Mediterranean variant, the A- group shows lowered GSH concentration comparable to the Mediterranean group and many A- people showed hemolytic attack. The evaluation of blood donors population evidenced that G6PD deficiency is nota rare event among periodic donors, so the performance of the G6PD screening on all blood donors might be suggested