Proc. 15th Annual Conference of Screening of Inborn Errors of Metabolism, Osaka, Japan. pp.24-5 (1987)

教 育 講 演 (3)

Neonatal Screening of Glucose-6-Phosphate Dehydrogenase Deficiency

Kwang-Jen Hsiao, Ph. D.

Clinical Biochemistry Research Laboratory
Dept. of Medical Research
Veterans General Hospital
Taipei, Taiwan 11216, Republic of China

Glucose-6-phosphate dehydrogenase (G6PD) deficiency is an X-linked inherited disease and the most common disease-producing enzyme deficiency of human beings. Deficiency of G6PD in erythrocyte, in various forms, is the basis of favism, primaquine and some other drug-sensitive hemolytic anemias, neonatal jaundice, and chronic nonspherocytic hemolytic anemia. Different variants of the enzyme are found in high frequency (1-10%) in African, Mediterranean and South East Asian populations. The clinical presentations of the Mediterranean and Asian mutants are more severe than the African A^- type mutant. Severe neonatal hyperbilirubinemia may occur in Mediterranean and Asian newborns with G6PD deficiency when they are exposed to certain drugs or environmental factors (e.g. moth balls). Kernicterus frequently results from this clinical episode, which contributes to early neonatal death and mental retardation in those who survive. The clinical manifestation beyond the neonatal period is associated with hemolytic anemia which may not be so innocuous, as acute renal failure may result from the hemolysis. Early detection by neonatal screening and preventing affected newborns from contacting those risk factors may relieve the medical and social burdens caused by G6PD deficiency in Mediterranean and Asian populations.
In Singapore, a mass neonatal G6PD screening program was initiated in 1965 and it has been reported effective in preventing kernicterus. Recently, several mass neonatal G6PD screening programs have been carried out in Greece, Hong Kong and Taiwan.

The estimated average incidence of G6PD deficiency in the Chinese population in Taiwan is around 3%. Island-wide epidemioilogical study indicates that the male newborns with G6PD deficiency have a much higher risk to develop severe neonatal hyperbilirubinemia. A pilot project to establish an island-wide neonatal G6PD screening program was started in 1984 in Taiwan. Heel or cord blood was collected on the filter paper used for routine neonatal screening and sent to the screening laboratory. The G6PD activity on a 3 mm blood spot disc was than screened by a qualitative fluorescence method with 6.2

U/gHb (40 % of normal activity) as the cut-off standard. The G6PD activity in positive samples was then re-checked by a semi-quantitative fluorescence method which was developed in my laboratory. The final positive cases were referred to one of the six local centers for quantitative confirmatory tests for the whole family, medical care and education, and genetic counseling. From 1984. 11. 1 to 1986. 12. 31, 54,052 samples were collected from newborns delivered in 51 hospitals, 16 clinics, 8 midwifes, and 65 health stations all over Taiwan. 1853 cases (3.4 %) were found to be positive by the screening tests. 930 cases (50.2 %) were successfully recalled and 591 cases (male 458, female 127, unknown 6) were confirmed as G6PD deficiency. No kernicterus has been reported in the G6PD deficient newborns detected by this program. Based on these results and the conclusions of a recent consensus meeting in Taipei, mass neonatal screening for G6PD deficiency in Taiwan may become a national health policy in the near future. However, the comparison of efficacy, efficiency and practicability between the localized screening with a cord blood sample and centralized screening with routine heel blood spots plus maternal education remains to be established in Taiwan.