Screening Tests (Available at Hospitals or Medical Centers) for Heritable Traits

G-6-PD deficiency

Numerous screening tests have been designed to identify the glucose-6 -phosphate-dehydrogenase (G-6-PD) deficient erythrocytes. The most simple, reliable, and specific screening procedure is the fluorescent spot test (2), This procedure has been widely employed (4,9,13, 15) and has been shown to be highly reliable in detecting the deficiency. It has also been automated and made available for widespread screening (3,14). Detection of G-6-PD levels lower than about 50 percent normal is achieved.

Sickle-cell trait

Several screening methodologies for sickle-cell abnormalities have been developed. The procedure consisting of cellulose-acetate electrophoresis (CAE) followed by solubility testing has been favorably regarded because of speed, cost, simplicity, accuracy, and ability to differentiate the various types of hemoglobin (1). Quantification of hemoglobin types is easily performed. In order to verify the electrophoresis procedure for HbS, any blood found to have HbS is subsequently evaluated via the solubility test as HbS displays abnormal solubility. CAE followed by a solubility test to confirm the presence of HbS has been the procedure recommended by the National Sickle Cell Disease Program and the National Hemoglobinopath) Standardization Laboratory at the Centers for Disease Control (1 1,1 2).

Thalassemias

Pearson, et al, (10) have developect an electronic measurement of mean corpuscular volume (MCV) which meets the requirements for a screening test for alpha and beta thalassemic heterozygotes. The procedure is rapid, automated, and inexpensive. It yielded no false negatives out of a study population of *300. However,* it is possible that false positives may occur for persons with an iron-deficiency condition. Further, persons who are so-called "silent carriers" (exhibiting no clinical symptoms) of alpha or beta thalassemia cannot be detected by this screening test. The frequency of the silent carrier is thought to be uncommon for beta thalassemia.

NADH dehydrogenase deficiency

The definitive diagnosis of hereditary methemoglobinemia requires the demonstration of deficient NADH dehydrogenase activity in red cells. The Hegesh, et al, *(6)* assay *is* considered preferable because of its specificity, accuracy at low enzyme activity levels, and ease of operation.

Serum alpha$_1$-antitrypsin (SAT) deficiency

Several reliable, easily administered, and inexpen - sive tests have been developed for the screening of large populations for SAT deficiency. All of these tests are sensitive for the recessive homozygous condition, but only one of them *(8)* can reliably detect the intermediate heterozygous Ievels. The authors claim that this test is a practical screening procedure which could be applied in large scale.

Slow v. fast acetylation

Urine tests for detecting slow and fast acetylators have been developed in order to deal with the potential medical problem of slow acetylators being at enhanced risk of developing adverse reactions to isoni - azid, an antitubercular treatment. The procedure is straightforward and simple, displaying an excellent capability to distinguish fast from slow acetylators (7).

HLA typing

The methodology for determining human leukocyte antigen types is considered simple and is frequently conducted in numerous medical centers in the United States. The typical cost is now less than $100 for a complete analysis (5).

Appendix F references

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