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S9.3

Glucose-6-Phosphate Dehydrogenase (G6PD) Deficiency: Genotype And Phenotype Correlation

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G6PD is the first enzyme of the pentose phosphate pathway. The phenotype in affected individuals is highly variable. Some individuals with G6PD deficiency remain asymptomatic throughout most or all of their lives. Others, however, develop neonatal jaundice, chronic non-spherocytic hemolytic anemia (CNSHA) or acute hemolysis that is triggered off by Fava bean ingestion, infection, medications or unknown oxidative stress.

Different variants of the G6PD enzyme have been identified, on the basis of their amino acid change. G6PD variants are classified according to their residual enzyme activity levels as: class 1 (enzyme deficiency with CNSHA), class 2 (<10% activity or severe deficiency), class 3 (10-60% activity or moderate to mild deficiency), class 4 (>60% activity, very mild to no deficiency) and class 5 (increased enzyme activity).

The correlation between genotype, biochemical characteristics (electrophoretic mobility and enzymatic activity) and clinical phenotype among those with G6PD deficiency had been studied but appeared to be variable. Furthermore, residual enzymatic activity was not predictive of the clinical picture. More studies will be required to elucidate the interaction of environmental factors, oxidative stress and enzymatic activity on the clinical expression in individuals with G6PD deficiency.

S9.4

Quality Assurance Program For Neonatal Screening Of G6PD Deficiency

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Objective: The nationwide neonatal screening of Glucose-6-Phosphate Dehydrogenase (G6PD) deficiency in Taiwan was started on July 1, 1987. The effective collection rate has reached >99% of all newborns since 1996 and the overall incidence rate of G6PD deficiency is about 2%. The referral hospitals distributed all around Taiwan were organized for follow-up, confirmatory test, medical care and genetic counseling. To assess the reliability of the confirmatory and screening tests, an external quality assurance (QA) program for G6PD assay was developed.

Methods: For quantitative assay, lyophilized quality control (QC) materials were prepared from red blood cells. For screening test, the QC materials were prepared from whole blood by spotting on to Guthrie cards. Periodically (1-2 month), the QC materials were sent to referral and screening laboratories by speed post delivery. The external QA results were evaluated and compared to the reference value (and medium/mean for quantitative test). The test results were submitted through internet and the summary reports were published on the webpage within two weeks for each survey <http://g6pd.tw>.

Results: Twenty-one referral laboratories and 15 screening laboratories (3 in Taiwan, 5 in Mainland China, 2 in Philippines, and 1 each in German, Lebanon, Thailand, Turkey, and Vietnam) are participating in the QA program at the present time. From January 1988 to December 2006, 138 QA surveys (3 to 5 QC specimens for each survey) to referral laboratories were performed and 2,496 reports were received in reply to these QA surveys. Two hundred and eighty-nine (11.6%, 289/2,496) abnormal QA results were found. Interlaboratory C.V. for the quantitative test has reached below 10% in recent years. For the screening test, 10 blood spots were sent to each screening laboratory for each survey. From March 1999 to December 2006, 46 surveys were performed and 415 reports were received. Seventy-six (18.3%, 76/415) abnormal QA reports were found. Seventy-one false negative and 172 false positive results were reported from the 4,150 blood spots tested by all the screening laboratories.

Conclusion: The external quality assurance program has been useful for monitoring the performance of the referral hospitals and screening laboratories, and might be a guidance for the participating laboratories to correct the analytical errors.

SYMPOSIUM 10

Hearing Impairment 3

S10.1

Early Intervention: Appropriate, Accessible and Affordable

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A hearing impairment is said to be an "invisible disability," yet it is the most common major birth defect. Before the onset of universal newborn hearing screening, children with hearing loss are not identified until they fail to meet important speech and language milestones at two years old and beyond. But with the current widespread implementation of universal newborn hearing screening programs, more infants with hearing loss can now be identified in the first few weeks of life and be fitted with amplification within the first few months.