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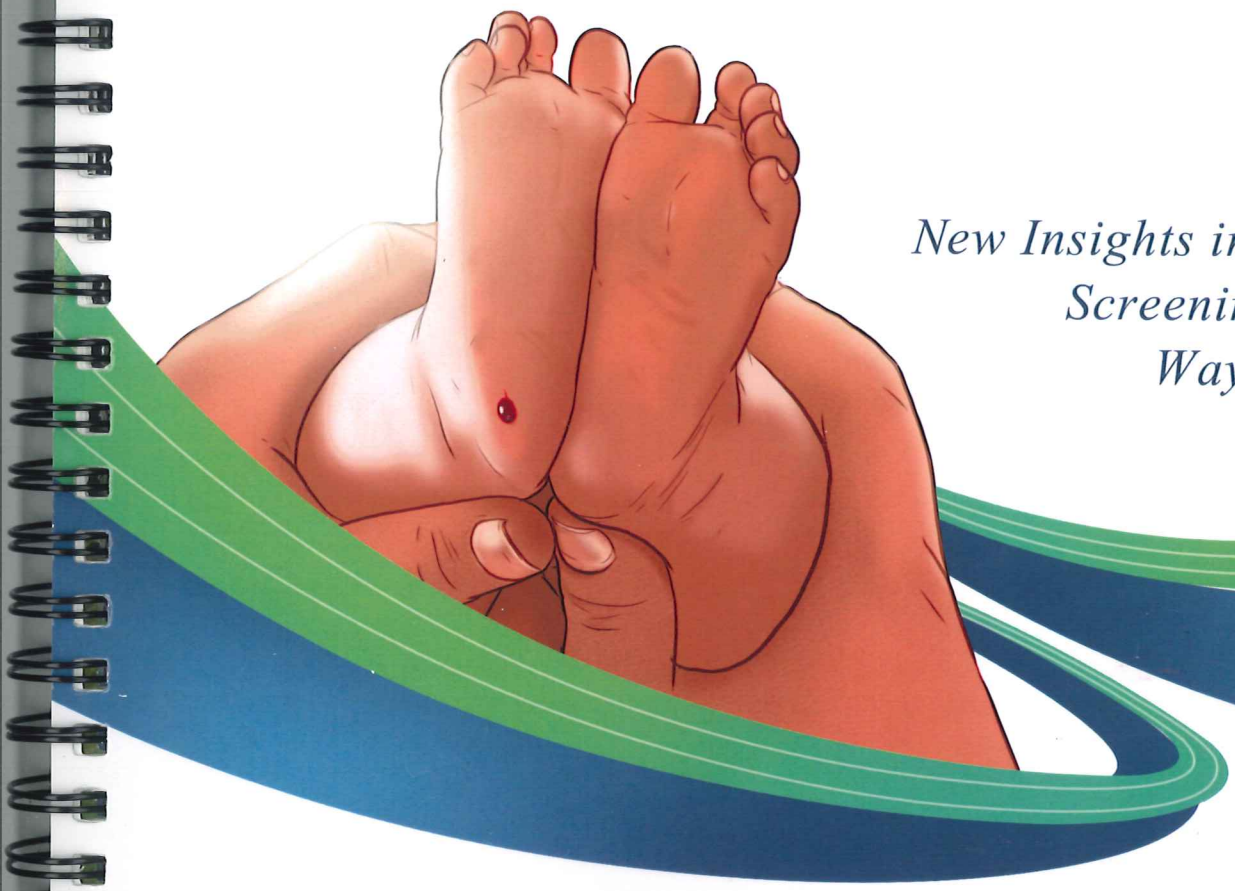

New Insights in Neonatal Screening and the Way Forward

GALT is also called classical galactosemia. Affected baby presented with jaundice and prone to septicaemia and later developed liver dysfunction, bleeding tendency and death within few weeks of life. GALE deficiency showed similar symptoms with less severity. Newborn screening laboratories around the world used either total galactose (TG) or GALT assay in dried blood spots (DBS) to screen for galactosemia. We present our experience in high-risk screening of galactosemia for the past 5 years.

Methodology: A total of 5200 DBS samples were received from physicians all over Malaysia, taken from patients with prolonged jaundice and liver dysfunctions for exclusion of galactosemia. Determination of TG were done using validated in-house method and GALT activity were performed using kit from Perkin-Elmer. Further testing were done to support our diagnosis like amino acids analysis and/or molecular findings for conformation.

Results: 37 (0.71%) of DBS samples were found to have TG more than the cut-off of 528 µmol/L. Out of that, 18 patients were diagnosed to have citrin deficiency (Citrullinemia Type II) where citrulline was also elevated. Four patients have elevated TG and low GALT suggestive of Classical galactosemia. 15 patients were noted to have elevated total galactose with normal GALT and normal citrulline. Further confirmatory testing is currently ongoing to exclude GALE deficiency in this group.

Conclusion: Combination of TG and GALT measurement are effective screening method for screening of galactosemia. Classical galactosemia was found to be less common than citrin deficiency in causing elevated total galactose and jaundice in Malaysian children.

08 DEVELOPMENT OF NEWBORN SCREENING FOR CRITICAL CONGENITAL HEART DISEASES IN TAIPEI

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Introduction: Critical congenital heart diseases (CCHDs) are severe and life-threatening diseases requiring surgical or catheter intervention within first year of life. The incidence of CCHDs has been reported about 1 to 2 of 1000 live births. Recently, pulse oximetry is adopted for newborn screening to detect mild and differential hypoxemia, which is characteristic for most CCHDs and may not be recognized by clinical examination. Early detection of CCHDs can significantly reduce morbidity and mortality in infants.

Objectives: The object of this study is to investigate the feasibility of implementation a community-based newborn CCHD screening program in Taipei city.

Methods: Twelve birthing facilities were participated in this program between October, 2013, and March, 2014. The screening protocol had a shorter interval (30 min) between measurements and screening once more for cases with first measurement <90% oxygen saturation. All these units submitted data on-line timely to the CCHD database, and the data was compared to nationwide newborn screening databases (metabolic and hearing) and birth certificate registry system to identify missing cases. Public health nurses followed up all missed or refused cases.

Results: Of 6,387 live births in these twelve birthing facilities during this period of time, 98.9% (n = 6,296) underwent pulse oximetry. Sixteen (0.25%) newborns had a final failed result. Five neonates were confirmed as CCHDs, two of them had diagnoses solely attributable to the CCHD screening. All the CCHD cases were referred and confirmed before 3 days after birth. Through repeated measurements for those with first measurement <90% saturation, only eleven of seventeen cases had a final failed result. False-positive rate was 0.17%.

Conclusion: The result indicates this is an efficient screening system with high screening rate. The community-based newborn CCHD screening program in Taipei successfully integrated screening, referral and public health systems which provided a scheme for nation-wide implementation.

09 INTERNAL QUALITY CONTROL (IQC) PROGRAM FOR NEONATAL SCREENING OF GLUCOSE-6-PHOSPHATE DEHYDROGENASE DEFICIENCY

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Introduction: Glucose-6-phosphate dehydrogenase (G6PD) deficiency is the most common human enzymopathic disease. Many Southeast Asia countries have included G6PD as a routine newborn screening item.

Objective: IQC program has been developed to monitor the quality of screening and quantitative G6PD tests between the EQA surveys.

Methods: For screening test, the QC materials were prepared from human whole blood by spotting on to Guthrie cards. For quantitative test, lyophilized QC materials were prepared from human red blood cells. The homogeneity and stability of the QC materials were checked according to ISO 13528. Two levels of QC materials were provided. The results were reported via an online system. The real time statistics and control chart were available as soon as the results had been input. The summary statistic reports of all the participants were published online monthly for comparison. <http://iqc.g6pd>< http://nsiqc.qap.tw>.

Results: Fifteen screening laboratories (Taiwan, Mainland China, Philippines, and Turkey) and 26 referral laboratories (Taiwan and Philippines) participated in the IQC program in 2014. Total 16,515 pair screening test results were reported. In high level, the range of CV, TE, and σ were 5.8%~27.0%, 11.8%~681%, and 0.5~3.4, respectively. In low level, the CV, TE, and σ were 4.6%~17.0%, 9.3%~766%, and 0.7~4.6, respectively. Total 2,095 pair quantitative test results were reported. In high level, the CV, TE, and σ were 0.7%~17.8%, 2.0%~52.8%, and -0.8~>6, respectively. In low level, the CV, TE, and σ were 2.4%~65.2%, 8.5%~172.6%, and -1.3~>6, respectively.

Conclusions: The data indicated that the quality of daily routine G6PD tests was varied widely in different laboratories. These G6PD IQC programs are useful for monitoring the intra-laboratory daily performance of the laboratories and provide good third party QC samples for the laboratory to compile the requirements for the quality and competence required by international standards (ISO 15189:2012, CLSI C24-A3:2006).