Newborn Screening, Inborn Errors of Metabolism

AB057. Inborn error of metabolism screening: timeliness and clinical service outcomes in Singapore

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Background: Inborn error of metabolism (IEM) Screening is an important healthcare program for the detection of metabolic and heritable disorders. Screening activity includes the collection of a blood specimen from the newborn, specimen transport to Hospital Screening laboratory, analysis and reporting. The impact of delayed screening at any stage of this process can lead to delays in treatment and potential harm to the newborn. In this report we aim to: (I) evaluate the screening laboratory efficiency and timeliness of presumptive positive results; (II) compare the outcomes with International recommended benchmark.

Methods: Data generated from the National Expanded Newborn Screening program from 2006 to 2017 were analyzed.

Results: We have screened 284,191 neonates since July 2006 of which 106 neonates have a screen positive result. Ninety-three [93] neonates were diagnosed with a metabolic disorder and another thirteen [13] with a maternal condition. The incidence rate is 1:2,700. Our recent data [2012–2016] showed that 97% (range: 95.9–98.6%) of all specimens' results were reported to service providers by day 7 of life. This met the benchmark of 95% recommended by the Department of Health and Human Services' (HHS) Advisory Committee on Heritable Disorders in Newborns and Children (GAO Highlights-17-196). For time critical conditions for which early recognition and treatment of acute symptoms can reduce the risk of illness and death, 94% (30/32) of presumptive positive cases have been reported to the provider and the neonates evaluated in a metabolic clinic by day 5 of life. Similarly, for presumptive positive results for non-critical conditions, 91% (67/74) of cases have been reported to the provider and the neonates evaluated clinically no later than 7 days after birth.

Conclusions: Our data showed good service delivery and timeliness in the screening program: from collection of sample to reporting of results and clinical evaluation by a specialist when indicated.

Keywords: Inborn error of metabolism (IEM) Screening; timeliness; clinical service

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