Newborn Screening for Congenital Hypothyroidism in India: Let’s Just Do It!

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Newborn screening (NBS) for congenital hypothyroidism (CH) has been ongoing in many developed countries since the early 1970s [1]. Interestingly, published studies on NBS for CH in India date to the early-1980s, only a few years after its implementation in Canada [2]; yet NBS for CH is still not widespread here. Over the years, a number of successful Indian NBS pilots for CH (and various other screening conditions) have supported the need for a national NBS initiative [3]. Researchers have repeatedly demonstrated the presence of CH in the Indian population along with the availability and value of early detection and treatment through NBS. Unfortunately, a national NBS policy aimed at early screening, detection and treatment for CH (or other screenable conditions) has been slow to evolve. The net result is thousands of individuals and families who must unnecessarily endure the adverse consequences of an easily and cheaply treated disease. Additionally, the resultant intellectual impairment results in a considerable negative societal impact and health care expense.

In this issue of Indian Pediatrics, the paper by Verma, et al. [4] adds valuable information to help define the preferred NBS laboratory process for identifying newborns at increased risk for CH. It is important that the NBS laboratory protocol minimizes patient recall for confirmatory testing while not missing true cases, if possible. This study helps to accomplish that goal by documenting a model screening laboratory protocol in support of CH screening protocols and data published recently by the Indian Council of Medical Research (ICMR) [5,6], and the Indian Society for Pediatric and Adolescent Endocrinology (ISPAE) [7,8]. It seems that at present there is adequate published laboratory data, screening and follow-up protocols, and treatment recommendations, to move forward with a national policy on NBS for CH (and/or other congenital conditions) and/or a national public health program for NBS in India.

In NBS, the laboratory screening test(s) performed must have demonstrated sensitivity and specificity such that as few individuals as possible are recalled for additional follow-up without missing true cases of the condition. The specimen matrix must also lend itself to both an effective and efficient laboratory testing protocol. In the case of NBS for CH, both thyrotropin (TSH) and thyroxine (T4) have been, and continue to be, used as productive laboratory screening tests in screening laboratories around the world. Strategies using either testing algorithm have long been shown to be effective as screening methods. As a result of physiologic surges in TSH soon after birth that might falsely elevate the test result, the use of TSH testing in newborns who must be screened in the first day of life (for various reasons, including routine early hospital discharge in some settings) is sometimes replaced by T4 testing despite its somewhat lower sensitivity [1,3,7]. Similarly, both liquid cord blood and heel stick specimens collected on special absorbent paper have been successfully used as the screening matrix [1,3,7,9]. The preferential use of heel stick blood in most screening programs results from its ability to provide a satisfactory specimen for many other metabolic screening tests that are not possible from cord blood, which can facilitate later program expansion. These testing and specimen issues and debates are not new in the NBS world and often the knowledge of the approach of others can provide a template to move the discussion forward. To this end, it may be useful to revisit the issues noted by the American Academy of Pediatrics in the early days of NBS in the US [9].

The apparent incidence of CH, which was thought to be about 1:3,000 newborns when screening began, now appears to be closer to 1:2,000 newborns, with even higher incidences reported in iodine-deficient areas, which include parts of India. This means that in India, over 13,000 babies with CH are either not diagnosed or diagnosed late each year (assuming 26 million births annually). While the direct benefit to newborns and their families are obvious, there are also substantial benefits to society and net cost savings to government health care
costs. While there are many variables to consider in determining cost benefit, and the value of these variables may differ markedly from country to country [10,11], I am unaware of a report of negative cost benefit for CH screening. Indeed, from a purely financial perspective, cost savings resulting from early diagnosis, treatment and normal integration into society can have extreme monetary benefit over time, especially for such a large patient cohort as exists in India. Of course, the benefits of successful screening extend well beyond finances.

As the Indian population continues to realize the benefits resulting from successful public health strategies that have steadily decreased the infant mortality rate, NBS has become increasingly important as a preventive public health strategy. The refinement of screening strategies, such as demonstrated by the study by Verma, et al. [4], continue to provide the basis for initiation of a sustainable and productive NBS program. While national implementation will take time, a national policy supporting initiation of screening seems attainable, and this will invariably lead to provincial programs, which can network over time to accomplish a national screening goal. Building on the recent increased government and professional interest illustrated here [4] and in other recent reports [5-8], the time to begin NBS is now. Let’s Just Do It!

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REFERENCES