A Price Tag of $38.50 Saved My Son

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The National Perinatal Association (NPA) is an interdisciplinary organization that strives to be a leading voice for perinatal care in the United States. Our diverse membership is comprised of healthcare providers, parents & caregivers, educators, and service providers, all driven by their desire to give voice to and support babies and families at risk across the country.

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Congenital hypothyroidism (CH) is the most common neonatal endocrine disorder affecting 1 out of every 2000 newborns. (1) Prior to 1976, when the American Thyroid Association recommended the inclusion of CH as part of the newborn screen (NBS). (2) CH was the leading cause of severe intellectual disabilities in children. With neonatal screening, CH can be diagnosed and treated in the newborn period, which significantly decreases the damaging effects of this disorder. Because of the continued support for NBS programs by the American Thyroid Association and the American Academy of Pediatrics, CH is now known as the most preventable cause of intellectual disabilities in children.

Best practices and guidelines have evolved as science has advanced. Unfortunately, national guidelines and standardization of best practices based on empirical evidence are lacking, and current NBS programs vary among states. Kilberg and colleagues(3) reviewed current practices for all NBS programs in the United States (n=51). The authors reported that regardless of the initial screening approach of measuring thyroxine (T-4) or thyroid-stimulating hormone (TSH), or both, all 51 programs did have TSH level, initiating immediate follow-up and further testing. However, the authors highlighted current variations among the states that result in potential limitations for CH screening protocols in some NBS programs. Specifically, if the TSH is borderline or the NBS is late, states vary on follow-up procedures. The concerning finding the authors presented was the lack of age-adjusted TSH thresholds for late initial screens and follow-up tests. Keeping the TSH threshold at higher levels consistent with the initial NBS (>40 mU/L) may result in false-negative reports. In 2006, the American Academy of Pediatrics reported that 10% of CH cases have TSH values <40mU/L and emphasized the need for age-adjusted values(4). According to the 2018 publication by Kilberg and colleagues(3), most programs do not adjust their TSH thresholds based on age. The authors provide a poignant example of an NBS collected at day of life 4 with a TSH of 24mU/L. Under their current practices, 28 out of 51 state programs (55%) would report this case as a false negative and miss the identification of CH.

Another strategy to be considered for reducing false negatives is the implementation of a second metabolic screening at 2-6 weeks of age. Few NBS programs in the United States have reported on the benefits of this additional neonatal screen. An eight-year review in Colorado revealed a false negative rate for CH on the initial NBS of 15.6%, and an additional 46 CH cases were identified using the second metabolic screening method. (5) Forty-six additional newborns received therapeutic medication to reduce the effects of hypothyroidism and decrease the risk and severity of intellectual disabilities. A more recent eight-year review in Alabama followed 146 newborns who were identified on the first or second NBS as having abnormal thyroid levels. Permanent CH was confirmed in 92 of the 121 newborns identified on the initial NBS (75%) and 5 of the 25 newborns identified from the second NBS (20%)(6). Five newborns would not have been able to receive medication for the prevention and reduction of severe intellectual disabilities if there was not a second NBS. In Utah, a four-year retrospective review revealed 20% of CH cases were missed on the initial NBS resulting in 25 additional cases detected because of the second NBS. (7)

There is the science. Now here is the reality. $38.50 saved my son's life. He was 6 days old. I finished a feeding, rocked him to sleep, and checked my phone. I noticed three missed phone calls from my pediatrician. My heart sank. Something was wrong. I quickly called the pediatrician. "Your son has an abnormal newborn screen." My mind raced through the list of congenital diseases included on the NBS that I had collected many times as a newborn intensive care nurse. "Your son's thyroid levels are abnormal. Can you come to the hospital to run some more tests today?"

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In 2018, 1 out of 1300 newborns in Nebraska were diagnosed with a form of hypothyroidism because of abnormal results identified on their NBS. (8) Seventeen babies that year were potentially saved from a known developmentally devastating disease. The cost of a newborn screen in Nebraska for 2018 was $38.50. (9) A price tag of $38.50 saved my son's life.

What is the total cost to society and to taxpayers? As a nurse scientist, I have searched the literature for numbers and facts. But, of course, each state varies because every program is funded differently. We can assume that lowering the TSH threshold will increase false positives. As identified in the literature, a study in Greece found lowering the TSH threshold did increase the follow-up rate from 0.12% to 1.20%. (10) The additional follow-up costs accounted for 1.8% of the NBS budget. The authors also discussed the adverse emotional effects on parents for false-positive cases. As a mother, I would be more devasted if my son's case were missed only to learn that my son's condition could have been prevented if the cut-off threshold was adjusted appropriately. The implications of a false positive significantly outweigh the negative consequences from a false negative CH diagnosis, including cost. The increased overall cost is another assumed barrier and the implication from decreasing the TSH threshold or adding a second metabolic screen. The study in Colorado did report a cost-per-case increase from $6108 to $9730 after adding the additional screening. (5) But as a mother, I cannot put a price tag on my son's health, and I cannot justify any rationale to miss a CH case when CH is a known preventable and treatable disorder. Can you?

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Reviewing the literature identifies potential major gaps in NBS programs related to CH. The risks of parental distress from a false positive and the minimal additional funding required for a second NBS or lowering the TSH threshold seem insignificant compared to the potential developmental consequences of delayed diagnosis and treatment for CH. In a world with so many unknowns and uncertainties, very few diseases can be identified early and, with simple treatment, drastically alter outcomes. A few things to consider as a neonatal provider:

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- Does your state use age-appropriate thresholds for TSH levels? Why not?
- What is the process for follow-up on NBS abnormal values? Is there someone dedicated to ensuring 100% follow-up rates?
- Has your state considered a second metabolic screen? Why not?
- Does your program have sufficient lab support and adequate funding? If not, advocate for change. Lawmakers listen to you.

For more information and resources, please visit the Centers for Disease Control and Prevention https://www.cdc.gov/newborn-screening/index.html or the National Newborn Screening and Global Resource Center http://genes-r-us.uthscsa.edu/

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Disclosure: The National Perinatal Association www.nationalperinatal.org is a 501c3 organization that provides education and advocacy around issues affecting the health of mothers, babies, and families.

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