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Inevitably, wherever your pediatric practice is, you are familiar with the call that comes late Friday late afternoon from the state health department, notifying you of an abnormal or critical newborn screen. This call is certainly difficult from the pediatrician's standpoint, as it requires not only knowing what to do with the screen but oftentimes trying to get the baby in to be seen within a 24–48 hour time frame, depending on the abnormality. States certainly don't time their calls this way on purpose, but the calls often seem to come at a very inconvenient moment.

Pediatricians are well-equipped as the first line providers to manage newborn screen results, but it may feel at times that every one or two years, more and more disorders are being added. That begs the question, how exactly do diseases become added to the newborn screening panels?

Despite the common misconception by the public that the addition of a disease to the newborn screen is a simple process, it involves a series of well-thought-out decisions by several national groups that then must be accepted by each individual state to move forward. The Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) is a federal panel that makes recommendations to the Secretary for Health and Human services as to whether or not a condition should be added to the Recommended Uniform Screening Panel (RUSP), which began in 2002. The RUSP is a list of core conditions that are not required to be added to newborn screens but are strongly encouraged based on a rigorous review by technical committees. Currently there are 35 core conditions and 26 secondary conditions that exist on the RUSP. Prior to the development of the RUSP, it was up to each individual state to determine which conditions would be screened for.

In general, diseases that are nominated for inclusion on the RUSP must undergo a complex decision-making matrix that includes the existing evidence for the disease and typically follows the key principles for newborn screening developed by Wilson and Jungner in 1968. These principles include, but are not limited to, having an available treatment for the disease, the existence of available diagnostic tools for the condition, understanding the natural history of the disease, and the existence of an early symptomatic or latent stage. However, these principles have been adapted over the years to better serve the needs of the population and allocation of potentially scarce resources.[1]

There are instances in which well-meaning support groups or disease foundations will lobby their local representatives to have a certain disease included on their state's newborn screen. Certainly all diseases that cause significant morbidity and mortality would be ideally picked up before a child is symptomatic, but unfortunately, not all of them will meet the necessary criteria that make the disease a) detectable by replicable and feasible means, b) treatable, and c) have that treatment be equably accessible for all babies across all socioeconomic statuses. Having these nominations bypass the usual mode puts states at risk of screening disorders which may not have ordinarily been put through the robust decision-making of the ACHDNC. A well-known example of this was the addition of Krabbe disease to New York's newborn screen in 2006.

Even after a disease is added to the RUSP, it may be years before an individual state adopts it for its own newborn screening panel[2] (which also undergoes a thorough review by state stakeholders) and actual implementation may take several years as well. Understanding this complex process doesn't necessarily change the day-to-day practice of a general pediatrician, particularly at 4:00 pm on a Friday afternoon, but does enable all first-line clinicians to know that the result they have in their hand is the end of a long road of science, evidence, and thorough review to ensure the best outcomes for all newborns in the U.S.

For more information on the new, updated neonatal metabolic screen and the important questions raised by an expanded screen, visit *Pediatrics in Review's* November article, Updated Neonatal Metabolic Screen.

References

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