Adding Conditions to State Newborn Screening Panels

Creating an Effective State Process

Newborn screening is a state-based public health service that identifies babies at risk for serious, rare, but treatable disorders so that they may be rapidly diagnosed and treated. As testing technology and clinical treatment advances, new disorders are being added to state newborn screening panels more frequently. In many states, legislators are being asked to formulate a process for adding new disorders to the state panel. Recently, legislative action has been substituted for other processes for adding new conditions to state authorized newborn screening panels. Such action, while often well-intended, has the potential to re-direct clinical, scientific and financial priorities away from time-tested strategies for adding new NBS conditions. This fact sheet presents guidelines for legislators and other decision makers charged with shaping this process.

Newborn screening is not new. It began in the early 1960's with screening for a single condition, phenylketonuria (PKU). The success of PKU screening, which prevented severe intellectual disability, paved the way for screening of other treatable disorders. Today most states screen for more than 30 disorders, and children who would have died or suffered life-long disability now lead healthy, normal lives. In the United States, newborn screening programs identify about 40 babies with a newborn screening disorder each day.

The Newborn Screening Process

Newborn screening is a complex process, not just a test. It begins with collection of a few drops of a baby's blood followed by laboratory testing and follow-up care for babies identified with a disorder. Parent education is essential to the process at all stages.

States rely on technical, clinical and community advisory groups to evaluate the addition of new newborn screening tests for particular disorders. These deliberations are public, with opportunity for input from health care providers, medical experts, parents, advocates, legislators and public health programs.

Most states have state advisory committees that review recommendations by stakeholder groups and compare them with recommendations by the Department of Health and Human Services Advisory Committee on Heritable Disorders in Newborn and Children. This federal advisory committee recommends new disorders added to the national Recommended Uniform Screening Panel (RUSP). Some states follow the RUSP in adding new disorders to state panels, some states consider the RUSP as one factor in decision making, while others take a more active and state-centered role in decision-making.

In 2015, the American Society of Human Genetics recommended that conditions be added to state newborn screening panels only after a state or federal review process evaluating the benefits and harms, impacts on systems of care, resources and capacity, and input from relevant stakeholders.[i]

[i] Botkin, J., Belmont, J., Berg, J., Berkman, B., Bombard, Y., Holm, I., Levy, H., Ormond, K., Saal, H., Spinner, N., Wilfond, B., & McInerney, J. (2015) ASHG Position Statement: Points to Consider: Ethical, Legal, and Psychosocial Implications of Genetic Testing in Children and Adolescents.The American Journal of Human Genetics 97, 6-21.

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Key Considerations

Both federal and state newborn screening advisory committees evaluate the addition of a new disorder based on the following key considerations:

1. Screening is needed to identify all newborns who may need treatment,
2. There is a significant risk of illness, disability, or death if babies are not treated promptly,
3. Effective treatment is available,
4. Treatment is more beneficial in the newborn period than later,
5. Resources and access to treatment and counseling are widely available,
6. The benefits to babies and to society outweigh the risks and burdens of screening and treatment.

Why a Defined Process?

All state stakeholders benefit from a clearly articulated review and approval process for the addition of new disorders to state newborn screening panels. A defined process ensures consistency, fairness and a full review of the many complex and highly technical factors involved in making a decision that may mean life or death for some infants.

The section below outlines the process for adding new disorders to Maine's newborn screening panel. It is vital to include key contacts who can correctly answer questions from state policy makers, parent advocacy groups and others about nomination, evaluation and selection of new disorders. These contacts should be able to explain the rationale for past state and federal decisions concerning the addition of new disorders to state and federal newborn screening panels.

Defining the State of Maine Process:

Maine adds newborn screening disorders to its newborn screening panel using the following collaborative process that relies on the expertise and experience of national and international newborn screening programs, genetic specialists, state health agencies, parents and families, health providers, state policy makers and other stakeholders. The state aims to select disorders that have the greatest potential to protect the health of Maine babies.

Role of Federal Advisory Committee: The Department of Health and Human Services Advisory Committee on Heritable Disorders in Newborn and Children (SDCHDNC) reviews nominated disorders. When there is sufficient evidence the SDCHDNC adds the disorder to the national Recommended Uniform Screening Panel (RUSP).

Role of Maine State Advisory Committee: The Joint Advisory Committee for Maine Newborn Screening (JAC) monitors and reviews SDCHDNC recommendations, data from medical experts and other newborn bloodspot screening programs. After discussion and deliberation the JAC makes a recommendation on whether to add a disorder to the Maine Newborn Bloodspot Screening Panel. The committee membership includes: Pediatric Specialists from the following disciplines: Endocrinology, Genetics, Hematology, Immunology, Neonatology and Pulmonology, Primary Care Providers, Parents and Family Advocates, Dieticians, Genetic Counselors, Laboratorians, representatives from Maine Birth Hospitals and national newborn screening advocacy groups.

Role of Maine Newborn Bloodspot Screening Program (MNBSP): Once a disorder has been recommended by the Joint Advisory Committee for Maine Newborn Screening Program, a workgroup is formed to plan for implementation. The workgroup is facilitated by MNBSP staff. Workgroup members include: a pediatric specialist for the new condition, MNBSP follow-up staff and laboratorians. The workgroup develops the system of care for the new disorder which includes: educating primary care providers and families, defining screening criteria, establishing follow-up recommendations for out-of-range results, identification of pediatric specialty care providers for follow-up care, and resources for follow-up testing and treatment. Once screening for the new condition is implemented, MNBSP uses a continuous quality improvement model which reviews and updates as needed: educational materials, reporting criteria and process, and resources for diagnosis and treatment. Identified cases and outcomes are also reviewed.

Contact for more information:

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