

Understanding and Improving Newborn Screening

Alex R. Kemper, MD, MPH, MS,¹ Jeffrey P. Brosco, MD, PhD²

In Greek mythology, the centaur Chiron had the ability to see hidden ailments that were often not obvious.^{1,2} He is credited as being the developer of the science of medicine and was considered to be the teacher of all physicians. Achilles and Asclepius were among his pupils; he also helped them when they were young children and most vulnerable. Given his medical acumen and dedication to children, Chiron could represent newborn screening. Even his appearance, combining horse and human features, suggests the complex interplay between the public health systems and traditional health care services necessary to make newborn screening function.

Of course, state- and territorial-based newborn screening in the United States is only around 60 years old. It remains unique in American medicine in that it promises universal access to screening and, when needed, diagnostic follow-up and linkage with subsequent care. Over the decades, newborn screening has expanded from testing just for phenylketonuria to panels including several dozen conditions. The innovations in screening, diagnosis, and treatment driving this expansion now threaten to disrupt the universal nature of this extraordinary public health program. With the advent of rapid genomic sequencing and gene therapy, it will be possible to detect and perhaps treat hundreds if not thousands of conditions in the newborn. This raises issues of informed consent, evidence review, public health laboratory capacity, health care resources, and

even the public health rationale for state-based newborn screening.

In 2025, a consensus report from the National Academies of Sciences, Engineering, and Medicine addressed many of these challenges by recommending that the Department of Health and Human Services establish a 10-year strategic plan to review how conditions are added to newborn screening, strengthen state programs, and coordinate public and private efforts to improve newborn screening.³ However, in the same year, the federal Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) was terminated, which for 2 decades had provided guidance to states and the broader newborn screening community on these and similar issues. The ACHDNC also made recommendations to the Secretary of Health and Human Services about the conditions that should be on the Recommended Uniform Screening Panel (RUSP), a list of conditions recommended to the state- and territorial-based newborn screening programs for screening. Fortunately, the RUSP still exists and much work continues to be done to advance newborn screening in the United States.

This supplement highlights some of the exciting ongoing work and thoughts related to newborn screening. It begins with a description of the current challenges in newborn screening noted in this introduction, as well as opportunities to strengthen it.⁴ The next article considers consequences of recent expansion of the RUSP on the overall benefit of early

¹Division of Primary Care Pediatrics, Nationwide Children's Hospital, Columbus, Ohio; and ²Department of Pediatrics and Institute for Bioethics and Health Policy, University of Miami Miller School of Medicine, Miami, Florida

Address correspondence to: Alex R. Kemper, 700 Children's Drive, LAC5411, Columbus, OH 43205-2664. alex.kemper@nationwidechildrens.org

AR Kemper drafted the initial manuscript. JP Brosco critically revised the manuscript. Both authors approved the final manuscript as submitted and agree to be accountable for all aspects of the work.

CONFLICT OF INTEREST DISCLOSURES: Neither of the authors has a commercial association that might pose or create a conflict of interest with the information presented in this manuscript.

FUNDING: No funding was secured for this study.

Accepted for Publication Date: June 11, 2026

<https://doi.org/10.1542/peds.2026-077357A>

Copyright © 2026 by the American Academy of Pediatrics

To cite: Kemper AR and Brosco JP. Understanding and Improving Newborn Screening. *Pediatrics*. 2026;158(S2):e2026077357A

detection and describes potential lessons as states consider further expansion, including through genomic sequencing-based screening.⁵ Clinical and public health system approaches are needed to assess decisions to expand to conditions that might benefit from newborn screening beyond traditional public health criteria. The supplement describes the experience of Early Check, a research platform to do just this.⁶ Next are 2 evidence reviews that ultimately helped support the addition of metachromatic leukodystrophy and Duchenne muscular dystrophy to the RUSP in December 2025.^{7,8} Whereas these articles represent the traditional approach to evidence review for the RUSP, the next article uses metachromatic leukodystrophy newborn screening to present a generalizable decision-analytic tool that could help evaluate the impact of candidate conditions for newborn screening in the future.⁹ The following 2 articles describe opportunities and challenges related to newborn hearing screening.^{10,11} Newborn hearing screening is a point-of-care test done at the bedside of the newborn, so public health newborn screening programs have had less direct involvement than they do with screening conducted using dried blood spot testing. There is only 1 other point-of-care screening test on the RUSP, critical congenital heart disease newborn screening, but it is likely that requests to add such point-of-care approaches to newborn screening will grow. Critical to ensuring the success of newborn screening is long-term follow-up after identification through newborn screening. Effective screening relies on comprehensive follow-up. However, long-term follow-up has been a major challenge. The next article describes specific options and provides a blueprint for state newborn screening programs to help ensure the full benefits of screening.¹² The supplement concludes with a historical evaluation to underscore the current and future challenges of newborn screening.¹³ This article asks whether newborn screening is facing just the latest examples of issues that have surfaced for decades or if this is the end of an era.

Chiron, our metaphor for newborn screening, had a sad outcome that could still suggest a positive future for newborn screening. As an innocent bystander, Chiron was accidentally struck by a poisonous arrow from Hercules during a fight with other unruly centaurs. Because Chiron was immortal, he did not die but instead was destined without intervention to live in pain forever. Chiron instead chose to sacrifice himself, and with this he convinced Zeus to allow Hercules to release Prometheus to be unbound from the rock where he was imprisoned for giving fire to human beings. What is the future of newborn screening? Will this amazing benefit for children and their families, and society more broadly, continue in its current state? Or will we see an entirely new approach to public health newborn screening? Going back to our story, there is debate about what happened to Prometheus after he was free. We chose to believe that he brought fire to inspire scientists, clinicians,

public health practitioners, laboratory experts, advocates, the general public, drug and device makers, insurers, and policy makers to meet the promise of newborn screening. We hope that by reading this journal supplement you will have a greater understanding of newborn screening and what fire you can bring to help meet its goals.

ABBREVIATIONS

ACHDNC: Advisory Committee on Heritable Disorders
in Newborns and Children
RUSP: Recommended Uniform Screening Panel

REFERENCES

1. Apollodorus (Author), Frazer JG (Translator). *The Library*. G. P. Putnam's Sons; 1921.
2. Homer (Author), Fagles R (Translator). *The Iliad*. Penguin Books; 1990.
3. National Academies of Sciences, Engineering, and Medicine. *Newborn Screening in the United States: A Vision for Sustaining and Advancing Excellence*. National Academies Press; 2025.
4. Goldenberg AJ, Gaviglio A. Newborn screening at a crossroads: navigating the challenges reshaping a public health legacy. *Pediatrics*. 2026;158(S2):e2026077357B
5. Grosse SD, Wilfond BS. Policy implications of expanded newborn screening as a service to families. *Pediatrics*. 2026;158(S2):e2026077357C
6. Bailey DB, Gehtland LM, Peay H, et al. Early check: the first 5 years. *Pediatrics*. 2026;158(S2):e2026077357D
7. Kemper AR, Lam WKK, Ojodu J, et al. Evidence regarding Duchenne muscular dystrophy newborn screening. *Pediatrics*. 2026;158(S2):e2026077357F
8. Lam WKK, Ream MA, Grosse SD, et al. Evidence regarding metachromatic leukodystrophy newborn screening. *Pediatrics*. 2026;158(S2):e2026077357E
9. Smith HS, Stein S, Henry MW, et al. Decision analysis to inform screening of the young: an interactive newborn screening model. *Pediatrics*. 2026;158(S2):e2026077357G
10. Kemper AR, Hart LC, Redden KE, Findlen UM. Opportunities to improve outcomes for deaf and hard-of-hearing infants. *Pediatrics*. 2026;158(S2):e2026077357H
11. Findlen UM, Hart LC, Kovacs L, et al. A framework for illustrating the decisions for parents along the early hearing detection and intervention journey. *Pediatrics*. 2026;158(S2):e2026077357I
12. Sontag MK, Steyermark A, Shannon CM, Barringer L, Kellar-Guenther Y. Long-term follow-up for newborn screening: a road-map and guide with programmatic considerations. *Pediatrics*. 2026;158(S2):e2026077357J
13. Brosco JP. End of an era, or just another inflection point in the history of newborn screening in the United States? *Pediatrics*. 2026;158(S2):e2026077357K