Rare Disease Advisory Council

Annual Report 2025

Table of Contents

Letter from the Rare Disease Advisory Council (RDAC) Chairperson

Dear Governor Braun and Indiana Legislative Council,

It is with great pride that I present the annual report on the activities of the Indiana Rare Disease Advisory Council (RDAC) as required in HEA 1201, which was enacted in 2023 by the Indiana General Assembly.

The RDAC is pleased to be seeking solutions to treat and mitigate the effects of rare diseases in Indiana and beyond with the hope of someday eliminating them so all Hoosiers can live long, productive and healthy lives. This committee is proud to be part of the solution and is looking forward to continuing this important work for many more years.

Within this report is a summary of the impressive accomplishments of the council which began meeting monthly on Sept. 26, 2023, and the RDAC's recommendations for consideration and adoption in the coming general assembly. These recommendations are bulleted below:

- Establish a working group of pediatric oncologists whose goal is to meet with and provide recommendations to the state and insurers on issues related to treatments and diagnostic modalities that affect pediatric oncology. This working group should be placed under the RDAC.
- For those cases that are subject to prior authorization and peer review, establish a requirement of the insurance provider to have a pediatric subspecialist in disease expertise as the peer reviewer
- Provide a state subsidized position for a learning specialist/school and community liaison at every pediatric hospital in the state of Indiana to provide enhanced educational support for pediatric cancer and rare disease patients/survivors
- Require insurance coverage for fertility preservation of rare disease and pediatric cancer patients, when medically appropriate
- RDAC granted waiver to the Indiana Open Door Law

The expertise of the RDAC is immense with representation from industry, research, pharmacy, advocacy organizations, medical providers, minority health groups and state agencies. These members bring their diverse perspective to assessing the landscape of care in Indiana, gathering and sharing resources and developing actionable recommendations for the Indiana General Assembly to enact.

Sincerely,

Wade Clapp, MD
Rare Disease Advisory Council Chairperson; Chair, Department of Pediatrics; Richard L. Schreiner Professor of Pediatrics; Indiana University Distinguished Professor

Introduction

Established by the Indiana General Assembly in 2023, the Rare Disease Advisory Council is charged with the following tasks:

Establish a council whose duties include the following:

- Conduct public meetings to survey the needs of patients in Indiana with rare diseases and their caregivers and providers

- Provide testimony, comments and recommendations concerning legislation and rules that impact patients in Indiana with rare diseases

- After consulting with experts on rare diseases, develop policy recommendations to improve patient access to and the quality of rare disease specialists; affordable and comprehensive health care coverage; relevant diagnostics; timely treatment; and other needed services for patients with rare disease

- Research and make recommendations to state agencies and insurers that provide services to people with rare diseases on the impact of prior authorization, cost sharing, tiering or other utilization management procedures on the provision of treatment and care for patients

- Evaluate and make recommendations to improve Medicaid coverage of drugs for patients with rare diseases to improve coverage of diagnostics and facilitate access to necessary health care providers with expertise in the treatment of rare disease

- Publish a list of existing, publicly accessible resources on research, diagnosis, treatment and education relating to rare diseases on the state department's website

- Evaluate the current state and funding of pediatric cancer research taking place in Indiana and how the research interacts with the landscape of pediatric cancer research done nationally

- Study other issues and provide grants that impact patients with rare diseases

This report fulfills the requirements of the HEA 1201 (2023) to "submit an annual report to the governor and the legislative council not later than June 15. The report submitted to the legislative council must be in an electronic format under IC 5-14-6." The required report must include the following: "Summary of the council's activities and progress" and "Recommendations of the council to the governor and general assembly on ways to address the needs of people living with rare diseases."

Membership

Chairperson: Dr. Wade Clapp (Chief Physician, Riley)
Term: Ends Sept. 30, 2025
Biopharmaceutical Representative: Vacant
Term: Ends Sept. 30, 2027
Health Carrier Representative: Vacant
Term: Ends Sept. 30, 2025
Hospital Administration: Dr. Jodi Skiles (Medical Director at Riley)
Term: Ends Sept. 30, 2025
Pharmacist with relevant experience: Dr. Tara Jellison (Ambulatory Services, Parkview)
Term: Ends Sept. 30, 2026
Physician with relevant experience: Vacant
Term: Ends Sept. 30, 2027
Rare Disease Caregiver: Laura McLinn (President of Best Day Ever Foundation and Caregiver of Son Living With a Rare Disease - Duchenne Muscular Dystrophy)
Term: Ends Sept. 30, 2027
Rare Disease Organization: Mindy Cameron (Muscular Dystrophy Family Foundation and Caregiver of Son Living With a Rare Disease - Duchenne Muscular Dystrophy)
Term: Ends Sept. 30, 2025
Rare Disease Patient: Dr. Doug Cipkala (Pediatric Oncology, Peyton Manning)
Term: Ends Sept. 30, 2026
Rare Disease Patient: Dr. Michael Busk (Physician, Ascension St. Vincent)
Term: Ends Sept. 30, 2026
Rare Disease Researcher: Dr. Santiago Schnell (Director, Notre Dame Center for Rare Disease)
Term: Ends Sept. 30, 2026
Registered Nurse with relevant experience: Lucy Paskus (CPNP at Peyton Manning)
Term: Ends Sept. 30, 2026
Director of Office of Medicaid Policy and Planning Designee: Dr. (Jeremy) Ty Sullivan (OMPP)
Term: Appointment does not expire
Commissioner of the Department of Insurance Designee: Cory Best (IDOI)
Term: Appointment does not expire
Indiana Minority Health Coalition chief operating officer: Carl Ellison (President of the Indiana Minority Health Coalition),
Term: Appointment does not expire

Summary of Activities and Progress

RDAC Fiscal Year 2025 Highlights

- Presentations were made to the RDAC by
    - Dr. Mary Ciccarelli, Medical Director of the Center for Youth and Adults with Conditions of Childhood
    - Amanda Moore representing the Angelman Syndrome Foundation
    - Holly Wheeler and Carrie Le representing Indiana Family to Family
- Conducted a pediatric cancer clinician survey. Responses were analyzed and results were shared in a March 17, 2025 letter to Indiana House Rep. Timothy O'Brien supporting House Bill 1453 which ultimately became law. See Appendix I.
- Pursuant to IC 16-46-17-3(6), RDAC assembled, evaluated and selected resources which are published on the RDAC website addressing research, diagnosis, treatment and education relating to rare diseases
- Supported the submission of the National Organization for Rare Diseases' (NORD) letter to Department of Health and Human Services Secretary Robert F. Kennedy encouraging reconsideration of his decision to eliminate the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). See Appendix II.
- Following on the success of the Pediatric Cancer Clinician Survey, the (Indiana Rare Disease Advisory) Council intends to launch the Indiana Rare Disease Survey in the summer of 2025. The purpose of this survey is to gather information about the needs of individuals and families living with rare diseases in the state, with the aim of improving quality of life and access to resources for affected Hoosiers.

  The Rare Disease Survey is designed to be confidential, with no identifying information collected. It is intended for individuals with rare diseases residing in Indiana, or their parents or guardians. Outreach about how to participate in the survey will be conducted through state-based patient advocacy groups, select medical facilities, and the Rare Disease Advisory Council website. Results of the survey are expected in early 2026 with publications to follow.
- A representative of the RDAC engaged with the Perinatal Genetics and Genomics Advisory Committee (PGG)

Recommendations

The RDAC proposed six recommendations as part of its 2024 report reflecting the widespread varied needs of patients living with rare diseases. The council is pleased to see much of its first recommendation, which was to fund a pediatric cancer research and treatment grant program with a priority on innovative research and treatments for pediatric cancer, addressed by the adoption of House Enrolled Act 1453 during the 2025 legislative session.

The RDAC reiterates three recommendations from the 2024 report which are hereby restated below. A fourth 2024 recommendation is also restated with the amendment adding rare disease patients in addition to pediatric cancer patients. Recommendation number five is new for 2025.

1. **Establish a working group of pediatric oncologists whose goal is to meet with and provide recommendations to the state and insurers on issues related to treatments and diagnostic modalities that affect pediatric oncology. This working group should be placed under the RDAC.**
2. **For cases that are subject to prior authorization and peer review, establish a requirement of the insurance provider to have a pediatric subspecialist in the area of disease expertise as the peer reviewer**

Recommendations one and two are closely linked. Therefore, the following justification supports both recommendations.

Historically, prior authorizations (PAs) were established to ensure that the utilization of health care resources (surgical, diagnostic, and pharmacy) was cost effective by maintaining consistency with published standard of care guidelines.

Barriers to an efficient and effective PA system, particularly in the pediatric oncology setting, include the burden of providing (often by phone and fax and not electronically) relevant clinical documentation, a lack of universally established clinical guidelines for such rare diseases, rapidly changing standards of care established by collaborative pediatric oncology groups such as the Children's Oncology Group, and a lack of reviewers with any pediatric oncology experience.

Whereas the PA process is intended to ensure patients receive medically appropriate care and reduce costs by eliminating unnecessary care, there are studies suggesting that the process itself has significant costs, including patient suffering and stress when delays in or denial of therapy occur. Delays in therapy because of PAs and appeals, particularly in the pediatric oncology setting, can significantly affect the overall outcome and survival of the patient.[1] Lengthy PAs with appeals can add to the financial costs to the health care system. The added cost of physician and staff burnout leads to reductions in overall provider availability also impacts the care provided to the patient.

We appreciate the immense complexities related to the prior authorization process and its objective to minimize health care dollars spent on unnecessary or unproven, costly therapies

and diagnostic studies. With the goal of providing state-of-the-art, highest quality care for Hoosier children, recommendations 2 and 3 were adopted.

We believe this bill will benefit pediatric oncology patients with greater efficiencies in providing timely, up to date care for them. We expect, from a pediatric oncology standpoint, that streamlining the PA process will also lead to improved overall survival outcomes for the pediatric oncology patient with the potential for short and long term savings in expenditures.

3. Provide a state subsidized position for a learning specialist/school and community liaison at every pediatric hospital in the state of Indiana to provide enhanced educational support for pediatric cancer and rare disease patients/survivors

We propose a state-subsidized innovative position for learning specialist/school and community liaison at every state hospital location serving pediatric cancer patients to provide enhanced educational support for survivors and those affected by childhood cancer. Education is a key predictor of future employment, income, and integration into society therefore, educational attainment is considered a key measure of the quality of long-term survivorship. As well as the impact that treatment may have on cognition[2] the education of survivors may also be adversely affected by missing time in school due to treatment, thus falling behind on schoolwork[3].

The purpose of this role is to function at the expanding intersection of medical, educational, and community institutions. These specialists will promote collaboration among professionals serving pediatric cancer patients, navigate networks dedicated to ongoing medical, educational, advocacy, and research, act as a liaison between the patient's medical team, family, and school, inform educational plans to best support the patient's needs, determine homebound needs, provide education information and resources and ongoing assessment for school needs and support, dissect/summarize neuropsychological reports with patients, families, and schools, and offer transitional support by creating transition plans when medically appropriate to get students back to school in a safe and meaningful way.

Transitioning does not happen at one point in time but is a process composed of multiple transitions that occur over time, preparing the individual for the next phase of life[4].These specialists will create and implement plans to raise awareness and increase the utilization of services for people affected by cancer. They will build awareness and establish relationships with healthcare providers and other organizations that will increase their knowledge of and referrals to educational services.

We propose the creation of an Indiana statewide tutoring program consisting of retired teachers recruited from the Indiana Retired Teachers Association who support and encourage volunteerism. Children suffering from a chronic medical condition, such as cancer, may experience lifelong impacts on learning, achievement, and employment opportunities, hence hindering their potential economic, social, and human development. Seeking ways to promote educational support within the educational or home environment ensures the presence,

participation, and achievement of all children. Schools often do not have the resources to provide the level of support these children need.

4. Require insurance coverage for fertility preservation of rare disease and pediatric cancer patients, when medically appropriate

According to the American Cancer Society, 220,000 individuals aged 0-49 are diagnosed with cancer each year. While it varies greatly depending on type of cancer, due to increases in treatments an estimated 86% 5-year survival rate exists for those individuals.[5, 6] Some cancer treatments, however, can cause infertility. Chemotherapy, radiation, and surgery can damage reproductive cells (eggs and sperm), reproductive organs, and/or endocrine functioning; they can also impact the ability to carry a pregnancy. Because this damage is primarily treatment-based, it can affect patients with any type of cancer.

Patients with other conditions requiring similar therapies (e.g., sickle cell disease, lupus, and thalassemia, etc.) are also at risk. In this age group, concerns about family building are second only to mortality, and infertility after cancer can cause depression, anxiety, and a lower quality of life.

Fertility preservation is now considered part of the standard of care for age-eligible patients. Standard procedures available for preserving fertility include sperm, egg, embryo, and ovarian tissue banking; all these approaches are supported by all the relevant medical associations, including the American Society of Clinical Oncology (ASCO), the American Society for Reproductive Medicine (ASRM), and the American Medical Association (AMA).

According to the National Infertility Association, cost is the biggest barrier to fertility preservation.[7] Nationwide, costs can range from several hundred dollars for sperm banking, to approximately $15,000 for egg banking. Without insurance coverage, these treatments are unaffordable for many patients. The costs are exacerbated by the short window of opportunity that patients have before starting potentially sterilizing cancer treatment. While the costs faced by an individual patient are high, the cost when spread across a population of insured people is extremely low. Independent analyses in states where coverage has been enacted have estimated costs (per member per month) ranging from a low of $.01 (California); to a high of $.10-$.24 (Maryland).[7]

Over the past six years, 16 states and the District of Columbia have implemented some coverage for medically necessary fertility preservation: California, Connecticut, Colorado, Delaware, the District of Columbia, Illinois, Kentucky, Louisiana, Maine, Maryland, Montana, New Hampshire, New Jersey, New York, Rhode Island, Texas, and Utah.

Fertility preservation services are medically necessary. Fertility preservation for iatrogenic infertility is not "elective" or "experimental," but rather a needed intervention to prevent potential sterility and/or reproductive damage. Patients cannot rationally defer or forego life-saving treatments to spare their fertility. Independent clinicians uniformly find fertility

preservation medically necessary in the context of gonadotoxic threat. These services address a side effect of cancer treatment. Remedies for other side effects, such as breast reconstruction, chemo-induced anemia, wigs and prostheses, etc., are typically covered by insurance.

Studies show that significant numbers of patients make sub-optimal treatment decisions (e.g., stopping tamoxifen or choosing less gonadotoxic treatment) to minimize reproductive impact.[7] These decisions may adversely affect both medical outcomes and treatment costs. Infertility causes distress, depression, anxiety; these have financial and medical consequences, and result in overall lower quality of life for survivors. Loss of fertility is not merely a medical complication; it permanently affects reproduction and parenthood – basic human activities worthy of the highest levels of protection.

5. RDAC granted waiver to the Indiana Open Door Law

The Rare Disease Advisory Committee (RDAC) respectfully request your consideration in granting RDAC an exemption from the in-person attendance requirements under the Indiana Open Door Law, similar to the exemptions currently provided to boards regulating healthcare professions. This flexibility would allow our committee to better accommodate the unique needs of our members and the populations we serve.

RDAC is composed of patients, caregivers, clinicians, researchers, and other members of the health care community, many of whom are living with or caring for individuals affected by complex and often debilitating rare diseases. These individuals often face significant challenges related to travel, accessibility, and immunocompromised health statuses. About half of our members are busy health care professionals whose input is invaluable to our committee. Committee members also reside across Indiana, complicating consistent in-person participation in Indianapolis.

We believe that our mission to advise on and improve the diagnosis, treatment, and quality of life for those affected by rare diseases can be carried out through electronic participation without compromising transparency or public access, especially with the livestream requirement.

References

1. Trapani D, Kraemer L, Rugo HS, Lin NU. Impact of Prior Authorization on Patient Access to Cancer Care. Am Soc Clin Oncol Educ Book. 2023 May;43:e100036
2. Askins, M. A., & Moore, B. D., 3rd (2008). Preventing neurocognitive late effects in childhood cancer survivors. *Journal of child neurology*, *23*(10), 1160–1171. https://doi.org/10.1177/0883073808321065
3. Vance, Y. H., & Eiser, C. (2002). The school experience of the child with cancer. *Child: care, health and development*, *28*(1), 5–19. https://doi.org/10.1046/j.1365-2214.2002.00227.x
4. Sabet, R. F. (2019). *Health disparities among sickle cell disease patients: A grounded theory model* [Doctoral dissertation, University of Miami].
5. American Cancer Society Cancer Facts and Figures 2025 https://www.cancer.org/content/dam/cancer-org/research/cancer-facts-and-statistics/annual-cancer-facts-and-figures/2025/2025-cancer-facts-and-figures-acs.pdf
6. CDC United States Cancer Statistics: Data Visualizations https://gis.cdc.gov/Cancer/USCS/#/explore/survival?cancer=1&datatype=1&dataset=standard&indicator=value&area=1&timeperiod=1&sexes=1&races=1&ages=23&tab=1&view=map&xaxis=sexes
7. Resolve, The National Infertility Association, Fertility Preservation Coverage – Key Points

Appendices

Appendix I: Letter in support of HB1453

Riley Hospital for Children
Indiana University Health

SCHOOL OF MEDICINE
DEPARTMENT OF PEDIATRICS

March 17, 2025

Representative Timothy O'Brien

Re: House Bill 1453

Dear Representative O'Brien,

The Rare Disease Advisory Council appreciates the consideration of House Bill 1453. We would like to provide the following evidence in support of this bill.

Briefly, a survey was conducted by the Rare Disease Advisory Council with approximately 80% of all Indiana clinical pediatric oncologists responding. The survey covered a wide range of topics, including:

- Access to clinicians/services
- Access to testing and medications
- Access to information and support services
- Variation in getting needed services
- Travel for services
- Research and research access
- Biggest problems in providing care and its
    - Impact on quality of care
    - Impact on outcomes
- Opportunities and challenges

A need for funding was a theme. This includes funding for basic research as well as clinical trials and health services research to measure long-term clinical outcomes. Funding was reported to attract pediatric-specific basic science and translational researchers and availability of seed funding to start projects while trying to get larger grants or other types of financial support.

There is also a lack of funding for new Children's Oncology Group protocols and National Cancer Institute-sponsored clinical research. We also need to test the establishment of statewide networking and partnerships to expand mental health and benefits of home health services.

Finally, there is a need to study the socio-economic gap in receiving the best care, i.e., those with financial resources have access to the best care while those with limited financial and social resources may not as easily access or demand the best care.

The following summarizes detailed responses related to research – it is important to note that this survey included physician scientists and clinicians.

- Eighty-seven percent of respondents reported being involved in clinical trials, 53% in other clinical research, and 6.7% in bench (basic science) research. (Numbers add to more than 100% because respondents could be involved in multiple types of research.)

- Twenty percent of clinicians stated that it was not a problem (not difficult) for their patients to participate in new research studies. While 67% said it was a slight problem. Of note, 14% either didn't answer the question or said they did not know.

- Respondents were asked where families go to participate in research studies: 100% reported at their own institution; 20% reported another institution within Indiana, and 53% reported an out-of-state institution. (Numbers add to more than 100% because respondents can have some patients who participate in different studies in different places.)

- Sixty percent of clinicians reported having at least five patients participate in a clinical trial network study within the past three years. Reasons patients have not participated included patients not meeting study criteria, no open study for the patients they see, and difficulty finding study criteria.

- Ninety-three percent of clinicians (with at least five patients participating in a clinical trial network study) participated in the Children's Oncology Group. Others include BMT CTN (13.%), PBTCTC (13%), PNOC (20%), CureWorks (7%), PTCTC (7%) and other pharma studies (7%).

- Respondents reported that families are educated on the availability of research studies at the time of diagnosis and during the course of treatment when possibly eligible by the clinician or the research team

- The most common reasons clinicians reported for patients choosing NOT to participate in recommended clinical trials were concern about the risks/side effects and not being interested in research, followed by distrust of research, requirement for extra tests or procedures, and time constraints

Opportunities and Challenges

There were *two questions* about opportunities and challenges:

The first question was: *Now, thinking about pediatric cancer care in all of Indiana, what do you see as the **greatest possibility** for developing new or improved childhood cancer therapies using the talent and resources available in Indiana?*

Two themes emerged from the responses. The first was regarding current research work and strengths. Riley has a large number of clinical faculty and strong basic scientists, which could lead to further bench-to-bedside collaboration. For example, studies coming out of the research labs inform how to utilize existing drugs to treat new neoplasms in pediatric and young adult patients. There are a growing number of clinical trials available at Riley, including CART-T cell therapy. Opportunities reported regarding clinical research were centered around increased pooling of resources and collaboration, further ensuring that clinical talent is connected to resources and made aware of what is available.

The second opportunity for developing new or improved childhood cancer therapies was related to health services research. The first theme was the need to explore and test better means of providing mental/behavioral health care that would lead to better short and long-term outcomes. The second theme was the need to discover and test better ways of providing a transition to adult care for cancer patients – many of whom are not aware of the potential late effects of cancer therapies.

The second question was: *What do you see as the **greatest challenge** for developing new or improved childhood cancer therapies using the talent and resources available in Indiana?*

Themes mirrored the opportunities question, but also several people felt that we need to think further, i.e., not just to be the best in Indiana but to be a national model for innovation, improved translation of bench-to-bedside to community care, more collaborative care, and more comprehensive, holistic care.

New potential therapies are coming out of the research labs, but getting those new therapies into patients can be a daunting task. Respondents reported a lack of time, resources, and investments. There is a need for support staff to support these trials and more protected time for physicians to lead them. One physician said, "There are many of us who are interested in pursuing research opportunities, but the support system is not there to step away from the bedside. We need additional resources to carry out the care on the clinical side for us to be effective. You do not need to recruit 100% of researchers, but you do need to protect the time of those who want to do research."

Another said, "If we want new therapies, we need advanced practice providers to help cover patients to allow for time to develop concepts."

A third reported, "[We] need protected time for faculty… to be able to create and lead translational and clinical research studies, which requires creating more non-physician support systems for help with administrative and other tasks that don't require a physician to do them. Clinical research teams are currently swamped with opening studies from the major consortia so that new research will require more clinical research associates.

Finally, a respondent reported concern about the decline in the number of medical students and residents choosing pediatric hematology-oncology as a career. Published research reports a shortage of pediatric oncologists and that trainees are choosing careers with better work/life balance and better pay.

Respectfully submitted,

D. Wade Clapp, M.D.
Rare Disease Advisory Council Chairperson
Chairman, Department of Pediatrics
Physician-in-Chief, Riley Hospital for Children

Appendix II: National Organization of Rare Diseases' Letter

May 5, 2025

Robert F. Kennedy Jr.
Secretary, Department of Health and Human Services
200 Independence Avenue, SW
Washington, DC 20201

Dear Secretary Kennedy,

On behalf of the 272 undersigned organizations committed to the health of our nation's mothers, infants, children, and families, we express our deep concern over the Administration's recent decision to eliminate the Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC). This sudden termination, alongside the proposed elimination of other federal newborn screening infrastructure, will delay time-sensitive detection of serious medical conditions in newborns and will impede delivery of clinical care and intervention to babies with devastating, treatable conditions during the optimal therapeutic window. We urge you to immediately reinstate the work of this important federal advisory committee and preserve our nation's federal newborn screening infrastructure.

Newborn screening is one of our nation's most successful public health programs, serving nearly 4 million infants each year and saving thousands of babies' lives. Our nation's newborn screening system detects life-threatening diseases in newborn babies before they can cause irreversible damage or death. Through timely detection and treatment within the first few days of life, our national newborn screening program provides American children the best chance at a healthy life — a purpose that aligns with the Administration's vision for a healthier America.

The ACHDNC is a critical part of the U.S. newborn screening system, providing guidance to the Secretary of the Department of Health and Human Services (HHS) on the most appropriate application of universal newborn screening tests, technologies, policies, guidelines, and standards. The Health Resources and Services Administration (HRSA), the National Institutes of Health (NIH), the Food and Drug Administration (FDA), and the Centers for Disease Control and Prevention (CDC) all provide guidance to the ACHDNC from their specific expertise, with the ACHDNC serving as the convenor and the central point of contact for all federal agencies and the newborn screening community overall. The proposed elimination of newborn screening programs across the federal agencies would significantly limit the expertise available both within the federal government and to state newborn screening programs. These programs provide grant funding, make recommendations, and track the impact of newborn screening to help improve health outcomes.

Policymakers created the ACHDNC through a bipartisan effort to bring uniformity to the U.S. newborn screening system as part of the Newborn Screening Saves Lives Act, passed by Congress in 2007 and signed into law in 2008. Prior to the passage of the bill, only 10 states and

the District of Columbia required infants to be screened for all 29 disorders recommended for screening by the American College of Medical Genetics and Genomics. Today, all 50 states and the District of Columbia require screening for at least 32 treatable conditions. The ACHDNC has served as the nation's chief newborn screening advisory body under Democrat and Republican administrations alike, making newborn screening one of the most successful public health programs in the country.

The ACHDNC plays an instrumental role in the maintenance of the Recommended Uniform Screening Panel (RUSP), a list of disorders that the Secretary recommends states to screen for as part of their universal newborn screening program. The ACHDNC oversees the evaluation of conditions considered for addition to the RUSP, reviewing and assessing the clinical and health outcomes of early detection and treatment and the readiness of the public health system to expand newborn screening. While states determine which conditions are screened as part of their respective programs, many states have limited resources to review evidence, and it is not feasible for all 50 states to conduct their own evidence review for every condition. The addition of new conditions to the RUSP guides the expansion of newborn screening at the state level, enabling early detection and treatment of serious rare disorders and saving thousands of lives.

The ACHDNC supports individual states' decision-making processes for adding conditions to their newborn screening panel, providing an evidence review that can be evaluated and implemented in every single state. The Committee's work guides federal recommendations that protect our nation's newborns from preventable death, enabling timely clinical interventions and optimized health outcomes. There is no comparable body to carry out this function in its absence. Without a clear path forward, the Administration's elimination of this committee risks the preventable death and suffering of children with treatable rare disorders.

We strongly urge you to preserve our federal newborn screening system and reinstate the work of the ACHDNC immediately so dedicated experts can continue to guide the lifesaving work of our nation's newborn screening programs without any further delay.

Sincerely,
Achalasia Awareness Organization
Acid Maltase Deficiency Association (AMDA)
ADCY5.org
Adrenal Insufficiency United
Adult Polyglucosan Body Disease (APBD) Research Foundation
Advocate Health
Akari Foundation
Akron Children's Hospital
Alabama Rare Disease Advisory Council
Alaska Chapter, American Academy of Pediatrics
ALD Alliance/Newborn Screening Alliance
Alliance for Regenerative Medicine
Alpha-1 Foundation

Alport Syndrome Foundation
Ambry Genetics
American Academy of Allergy, Asthma & Immunology
American Academy of Neurology
American Academy of Ophthalmology
American Academy of Pediatrics
American Association for Pediatric Ophthalmology and Strabismus
American College of Allergy, Asthma and Immunology
American College of Medical Genetics and Genomics
American College of Obstetricians and Gynecologists
American Society for Clinical Pathology
American Society for Reproductive Medicine
American Society of Hematology
American Society of Human Genetics
Angelman Syndrome Foundation
Ann & Robert H. Lurie Children's Hospital of Chicago
Aplastic Anemia and MDS International Foundation
Arizona Chapter, American Academy of Pediatrics
Association for Creatine Deficiencies
Association for Diagnostics & Laboratory Medicine
Association of Public Health Laboratories
Autoimmune Association
Autoimmune Encephalitis Alliance, Inc.
Avery's Hope
Ayana's Hope Cells
BDSRA Foundation
Bionano Genomics, Inc.
Bionano Laboratories
Bloom Syndrome Association
Boomer Esiason Foundation
Bubba's Light, Inc.
CACNA1A Foundation
California Chapter 1, American Academy of Pediatrics
California Chapter 3, American Academy of Pediatrics
California Life Sciences
California Rare Disease Access Coalition Hemophilia Council of California Chiesi Global Rare Diseases
Child Neurology Foundation
Children's Craniofacial Association
Children's Hospital Colorado
Children's Hospital of Orange County (CHOC)
Children's Sickle Cell Foundation, Inc.
Chondrosarcoma CS Foundation, Inc.
Coalition to Cure Calpain 3

Coffin-Lowry Syndrome Foundation
Colorado Chapter, American Academy of Pediatrics
Colorado Rare Disease Advisory Council
COMBINEDBrain, Inc.
Congenital Adrenal Hyperplasia Research, Education & Support Foundation Connetics Consulting, LLC
CTNNB1 Connect & Cure
CTX Alliance
CureARS
Cure 4 The Kids Foundation
Cure CMD
Cure GM1 Foundation
Cure LGMD2i Foundation
Cure SMA
cureCADASIL
CureSHANK
Cyclic Vomiting Syndrome Association
Cystic Fibrosis Foundation
Cystic Fibrosis Research Institute
Cystinosis Research Network
Dana's Angels Research Trust
Danny's Dose Alliance
debra of America
District of Columbia Chapter, American Academy of Pediatrics DLG4 SHINE Foundation
Dravet Syndrome Foundation
Dup15q Alliance
Elpida Therapeutics
Eosinophilic & Rare Disease Cooperative
EveryLife Foundation for Rare Diseases
Fabry Support & Information Group
FACES: The National Craniofacial Association
Familial Dysautonomia Foundation
Firefly Fund
flok Health
Florida Chapter of the American Academy of Pediatrics, Inc.
Foundation for Angelman Syndrome Therapeutics
Foundation to Fight H-abc
Friedreich's Ataxia Research Alliance (FARA)
Galactosemia Foundation
Gaucher Community Alliance
Gene Giraffe Project
GeneDx
Genetic Alliance
Global Genes

Global Liver Institute
Grant's Giants Pompe Awareness Nonprofit
Greenwood Genetic Center
GRIN2B Foundation
Haystack Project
HCU Network America
Histiocytosis Association, Inc.
HNRNP Family Foundation
Hope in Focus
Hues for Hope
Hydrocephalus Association
Hypertrophic Cardiomyopathy Association
Idaho Chapter, American Academy of Pediatrics
Illinois Chapter, American Academy of Pediatrics
Immune Deficiency Foundation
Indiana Chapter, American Academy of Pediatrics
Indiana Rare Disease Advisory Council
Institute for Gene Therapies
International Foundation for CDKL5 Research
International Society for Mannosidosis & Related Diseases (ISMRD)
Iowa Chapter, American Academy of Pediatrics
Jett Foundation
Johns Hopkins All Children's Hospital
Kansas Chapter, American Academy of Pediatrics
Kentucky Chapter, American Academy of Pediatrics
Key Proteo
Kids Conquering Sickle Cell Disease Foundation
KIF1A.org
KrabbeConnect
Krishnan Family Foundation
Labcorp
Little Hercules Foundation
Little Miss Hannah Foundation
Louisiana Chapter, American Academy of Pediatrics
Louisiana Rare Disease Advisory Council
Lupus and Allied Diseases Association, Inc.
Maine Chapter, American Academy of Pediatrics
Malan Syndrome Foundation
Maple Syrup Urine Disease Family Support Group
March of Dimes
Marshall's Mountain, Inc.
Maryland Chapter, American Academy of Pediatrics
M-CM Network
MedGenyx, PLLC

Michele Schoonmaker, LLC
Michigan Chapter, American Academy of Pediatrics
Michigan Medicine
Michigan Rare Coalition
Michigan Rare Disease Advisory Council
Minnesota Chapter, American Academy of Pediatrics
Minnesota Rare Disease Advisory Council
Mississippi Chapter, American Academy of Pediatrics
Mississippi Metabolics Foundation
Mississippi Rare Disease Advisory Council
MitoAction
MLD Foundation
MPS SuperHero Foundation
MTS Sickle Cell Foundation, Inc.
Muenzer MPS Research & Treatment Center
Muscular Dystrophy Association
Myasthenia Gravis Foundation of America
MyOme
Myositis Support and Understanding
Myotonic Dystrophy Foundation
National Adrenal Diseases Foundation
National Association of Pediatric Nurse Practitioners
National Ataxia Foundation
National CMV Foundation
National Health Council
National MPS Society
National Niemann Pick Disease Foundation
National Organization for Rare Disorders
National PKU Alliance
National Society of Genetic Counselors (NSGC)
National Tay-Sachs & Allied Diseases Association, Inc.
Nationwide Children's Hospital, Columbus, Ohio
Necrotizing Enterocolitis (NEC) Society
Nemours Children's Health
Nevada Chapter, American Academy of Pediatrics
Nevada Rare Disease Advisory Council
New Hampshire Chapter, American Academy of Pediatrics (NHAAP)
New Mexico Pediatric Society
New York State Department of Health
New York State Chapter 2, American Academy of Pediatrics (NYS AAP – Chapter 2)
New York State Chapter 3, American Academy of Pediatrics (NYS AAP – Chapter 3) Niemann-Pick type C Disease Group
Noah's Hope
NR2F1 Foundation

NTM Info & Research, Inc.
NW Rare Disease Coalition
Ohio Life Sciences Association
Ohio Rare Disease Advisory Council
Organic Acidemia Association
Parents Infant Children of Kernicterus
Pathways for Rare and Orphan Solutions
Patient Advocacy Strategies
Pennsylvania Chapter, American Academy of Pediatrics
Pennsylvania Rare Disease Advisory Council
Pharming Healthcare, Inc.
Phelan-McDermid Syndrome Foundation
Platelet Disorder Support Association
Pompe Alliance
Prader-Willi Syndrome Association | USA
PRISMS
Project Alive
Project GUARDIAN
Pyruvate Kinase Deficiency International Alliance
Quest Diagnostics
Rare Access Action Project
Rare and Black
Raregivers, Inc.
Rare New England
Rare STRIDES
Rare Wish
Revvity
SCAD Alliance
Sickle Cell Association of Kentuckiana
Sickle Cell Disease Association of America, Inc.
Sickle Cell Warriors Foundation, Inc.
SLC6A1 Connect
Smith-Kingsmore Syndrome Foundation
Society for Inherited Metabolic Disorders (SIMD.org)
South Carolina Rare Disease Advisory Council
Speak Foundation
Syngap Research Fund
TANGO2 Research Foundation
Tatton Brown Rahman Syndrome Community
Taylor's Tale
Team Telomere
Team Titin
TED Community Organization
Tennessee Chapter, American Academy of Pediatrics

Terumo Blood and Cell Technologies
The Bonnell Foundation: Living with cystic fibrosis
The Children's Medical Research Foundation, Inc.
The DDX3X Foundation
The Ehlers-Danlos Society
The E.WE Foundation
The Global Foundation for Peroxisomal Disorders
The Lambert-Eaton LEMS Family Association
The Louisa Adelynn Johnson Fund for Complex Disease
The MED13L Foundation
The Oxalosis and Hyperoxaluria Foundation
The Sudden Arrhythmia Death Syndromes (SADS) Foundation
The TBCK Foundation
Tourette Association of America
Travere Therapeutics
TrueNorth
TSC Alliance
Turner Syndrome Society of the United States
UDNF PEER
UH Rainbow Babies & Children's Hospital
United Mitochondrial Disease Foundation
United MSD Foundation
United Pompe Foundation
University of Washington
US Thrombotic Microangiopathy Alliance and Consortium
Usher Syndrome Coalition
Utah Chapter, American Academy of Pediatrics (UTAAP)
Utah Department of Health and Human Services Newborn Screening Program
Utah Rare Disease Advisory Council
Vasculitis Foundation
Virginia Chapter, American Academy of Pediatrics
Virginia Rare Disease Advisory Council
Wadsworth Center, New York State Department of Health
wAIHA Warriors
West Virginia Chapter, American Academy of Pediatrics
Wisconsin Chapter, American Academy of Pediatrics (WIAAP)
Wiskott-Aldrich Foundation
XLH Network, Inc.