

January xx, 2025

President-Elect Donald Trump  
1401 Constitution Ave., NW  
Washington, DC 20230

Dear President-Elect Trump,

On behalf of the EveryLife Foundation for Rare Diseases and the XX undersigned organizations representing leading rare disease patient advocacy organizations serving the estimated 30 million Americans living with a rare disease or disorder, we are pleased to provide you with a set of priority policies to support the rare disease community as you begin your term as our nation's 47<sup>th</sup> President.

The policy priorities articulated below are informed by the needs of our community and our shared missions of ensuring all Americans affected by one or more of the estimated 10,000 rare diseases can access a timely diagnosis, expert clinical care, and optimal treatment options. We are a community who feels an intense sense of urgency on behalf of the millions of Americans living with rare diseases and their families, a disproportionate percentage of whom are children. Having worked with this administration before to help advance the use of patient experience data in research and therapy development, we know that you prioritize the health of the children of this nation, especially those living with rare diseases. As you may be aware, over two thirds of rare diseases are genetic in origin and about 70 percent of rare diseases begin in childhood, a statistic that becomes more sobering when acknowledging that fewer than 5% of the estimated 10,000 rare diseases have an FDA-approved therapy<sup>1</sup>.

Over 1,120 orphan designated approvals are changing lives of the patients and families affected thanks in large part to remarkable advances in science and technology, investments in rare disease research, significant efforts by patient communities, and the leadership of both the National Institute of Health (NIH) and the Food and Drug Administration (FDA) in embracing regulatory science innovation and the application of appropriate flexibility in rare disease product evaluation<sup>2</sup>.

Our optimism is balanced by the staggering extent of unmet need that remains and the recognition that in some cases, it is process and policy hurdles preventing scientific possibility from reaching patients. Rare disease patients and their families face significant challenges at every stage of diagnosis, treatment, and disease management. These barriers often lead to avoidable diagnostic and treatment delays, irreversible disease progression, and devastating social and economic consequences.

The National Economic Burden of Rare Disease Study estimated that in 2019 the overall annual economic impact of rare diseases in the United States exceeded \$966 billion<sup>3</sup>. Of the total health economic assessment, indirect costs from productivity losses comprised the most significant economic

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<sup>1</sup> [https://ncats.nih.gov/sites/default/files/NCATS\\_RareDiseasesFactSheet.pdf](https://ncats.nih.gov/sites/default/files/NCATS_RareDiseasesFactSheet.pdf)

<sup>2</sup> <https://www.accessdata.fda.gov/scripts/opdlisting/oopd/listResult.cfm>

<sup>3</sup> Yang, G., Cintina, I., Pariser, A. et al. The national economic burden of rare disease in the United States in 2019. *Orphanet J Rare Dis* 17, 163 (2022). <https://doi.org/10.1186/s13023-022-02299-5>

impact at \$437 billion, followed by direct medical costs at \$418 billion, and non-medical and uncovered healthcare costs of \$111 billion absorbed directly by families living with rare diseases<sup>4</sup>. These findings highlight the necessity of focusing on policies that address rare disease needs spanning the overall healthcare system<sup>5</sup>.

Our nation's rare disease community deserves access to comprehensive, affordable health insurance that enables us to receive timely diagnoses, recommended clinical care, and access to prescribed therapies. While rare disease patients are included among every public and private insurer's membership, Medicaid and CHIP play a critical role in ensuring kids and adults with rare diseases and disabilities can access the care they need. Covering almost half of all children with special healthcare needs, Medicaid coverage is an essential component to diagnosis, care, and treatment for the rare disease community<sup>6</sup>.

Alongside patients, families, researchers, clinicians, the diagnostic and biopharmaceutical industry, and other committed rare disease community stakeholders, our organizations drive evidence-based policy solutions and partner with regulatory agency leaders to ensure our public health infrastructure is optimized to work for all Americans, regardless of the rarity of one's condition.

**To further your policy goals to create efficient, affordable, and high-quality healthcare, our organizations recommend the inclusion of the following policy areas among those of your administration.**

- 1. Preserve and enhance therapeutic development incentives**
- 2. Support predictable and consistent approaches to the evaluation of rare disease therapies**
- 3. Modernize the nation's newborn screening program to ensure all babies can benefit from lifesaving health interventions at the earliest moment possible**
- 4. Support policies that facilitate timely and affordable access to approved therapies**

### **Preserve and enhance therapeutic development incentives**

Rare disease research and development is often more complex, time-intensive, and costly compared to non-orphan products. A condition qualifies as a rare disease if it affects fewer than 200,000 people in the U.S.<sup>7</sup>, but many innovative therapies target conditions with far smaller patient populations. Developing treatments for such small populations requires significant investment to address challenges like understanding the natural history of diseases, resolving diagnostic uncertainties, and validating

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<sup>4</sup> Ibid

<sup>5</sup> Ibid

<sup>6</sup> Musumeci, M., Chidambaram, P., & O'Malley Watts, M. (2019, June 14). Medicaid financial eligibility for seniors and people with disabilities: Findings from a 50-state survey - issue brief - 9318. KFF. <https://www.kff.org/report-section/medicaid-financial-eligibility-for-seniors-and-people-with-disabilities-findings-from-a-50-state-survey-issue-brief/>

<sup>7</sup> 21 U.S.C. § 316 (1983)

outcome measures. Clinical trials for rare disease products take twice as long as those for non-orphan products<sup>8</sup>, with higher risks of failure.

Since the Orphan Drug Act (ODA) was enacted in 1983<sup>9</sup>, policymakers have acknowledged the complexities of rare disease research and the critical need for policies that affirm no disease is too rare to warrant treatment. The ODA, along with various policies introducing targeted incentive mechanisms in the years since, has successfully spurred investment in the development of therapies for rare diseases. However, recent regulatory changes, potential threats to the sustainability of these incentives, and shifts in the scientific and economic landscape have created an uncertain future—particularly for the development of treatments targeting extremely small patient populations.

### ***Reauthorize the Rare Pediatric Disease Priority Review Voucher Program***

The Rare Pediatric Disease Priority Review Voucher (PRV) Program is a market-based incentive that has led to new treatments for 40 rare diseases at no cost to taxpayers. Unfortunately, this life-saving program lapsed on December 20, 2024, despite being extended for six years in the House-passed Give Kids a Chance Act and having broad bipartisan support. Over 200 patient organizations have advocated for the PRV Program to be extended<sup>10</sup>. Immediate action is needed from Congress to rectify this oversight early in 2025. We urge your administration to work with Congress to prioritize an extension to the PRV Program in the early days of the 119<sup>th</sup> Congress. Children’s lives are at stake.

### ***Preserve and Restore the Orphan Drug Tax Credit***

The Orphan Drug Tax Credit (ODTC), established as part of the ODA, allows companies to claim a tax credit for a portion of their qualified research expenses. Originally set at 50% of qualified expenses, the credit was reduced to 25% in 2017. Over the years, the ODTC has played a vital role in the approval of orphan drugs, reducing the financial burden of clinical trials that can be more complex, longer, and pricier than non-orphan products. Rare disease patients and families facing life-changing diagnoses need more tools in the drug development toolchest, not fewer. A robust ODTC, as intended in the bipartisan ODA, means more companies can take a chance on a novel therapy prospect. It also enables companies to conduct clinical studies to find new uses in orphan diseases after a treatment is already on the market.

### ***Enable federal research agencies to drive innovation and fill critical gaps in resources for rare disease therapy development***

With over 10,000 rare diseases, each presenting differently across patients, orphan drug investments remain high risk. Federal programs like the Orphan Products Grant Program (OPGP) at the FDA, the

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<sup>8</sup> Jayasundara, K., Hollis, A., Krahn, M. *et al.* Estimating the clinical cost of drug development for orphan versus non-orphan drugs. *Orphanet J Rare Dis* **14**, 12 (2019). <https://doi.org/10.1186/s13023-018-0990-4>

<sup>9</sup> 21 U.S.C. § 316 (1983)

<sup>10</sup> EveryLife Foundation for Rare Diseases. (2024, December 6). EveryLife Foundation Joins Forces with NORD and Leading Patient Organizations to Call for Congress to Pass Long-Term Extension of the Rare Pediatric Disease PRV Program. <https://everylifefoundation.org/wp-content/uploads/2024/12/Rare-Pediatric-Disease-Priority-Review-Voucher-Program-Reauthorization-Community-Letter.pdf>

National Center for Advancing Translational Sciences (NCATS) at the NIH, and the Advanced Research Projects Agency for Health (ARPA-H) have spurred innovative collaborations, built infrastructure to attract private investment, and funded research targeting diseases that were once deemed too rare or complex to treat. These investments allow non-traditional drug development models, such as initiatives led out of academic centers, non-profit research institutes, and patient organizations to be viable when commercial interest will never be. The OPGP alone has funded clinical trials that led to over 80 FDA-approved therapies and in its 13-year history<sup>11</sup>; NCATS has contributed to 55 Investigational New Drugs and 14 approved therapies and encouraged innovation in areas such as cell and gene therapies and catalyzed industry interest in rare conditions that were previously overlooked<sup>12</sup>.

### ***Promote policies that will incentivize the development and repurposing of treatments and cures for rare diseases***

Despite the complex nature of rare diseases, advances in science, technology, and drug development platforms have increased the possibilities that one therapy might be effective across a variety of diseases with shared pathways. Increasingly, willingness to invest in rare disease therapy development is contingent on a target molecule's potential to treat multiple rare diseases. Decisions about investments, the order of trials and FDA submissions, and repurposing efforts often carry life-or-death consequences for patients. Policies should maximize a therapy's potential, letting science drive outcomes, and enabling the use of the established and broadly supported expedited pathways for FDA approval, that for some rare disease communities, may be the only viable option to get treatments to patients.

### **Support predictable and consistent approaches to the evaluation of rare disease therapies**

#### ***Support Successful Implementation of the Rare Disease Innovation Hub at the FDA***

Following years of advocacy by the rare disease community, in 2024 the FDA announced the establishment of the Rare Disease Innovation Hub to improve consistency and predictability of therapy approaches across and within the Centers and Divisions for rare disease patients and therapy manufacturers. We urge your Administration to commit to a clear set of priorities that align with those of the rare disease community, and that focus on addressing the most significant challenges, particularly building FDA regulatory science expertise in rare disease.

#### ***Developing Treatments for Ultra-Rare Diseases***

While all rare diseases pose challenges and additional costs to developing therapies and treatments, the needs are even more pronounced when it comes to ultra-rare or very small population diseases and conditions. Of the approximately 10,000 known rare diseases, more than 80 percent have a prevalence

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<sup>11</sup> U.S. Food and Drug Administration. (2024, October 1). *Clinical Trials Grants Program*. Orphan Products Grant Program. <https://www.fda.gov/industry/orphan-products-grants-program/clinical-trials-grants-program>

<sup>12</sup> U.S. Department of Health and Human Services. (2024, August 28). *Our Impact*. National Center for Advancing Translational Sciences. <https://ncats.nih.gov/research/our-impact>

of less than 1 in 1 million<sup>13</sup>, and advances in science are leading to identification of hundreds of newly identified rare genetic diseases every year. A particular challenge for ultra-rare diseases is the limited natural history data associated with any condition as well as very small patient populations that make clinical evaluation exceedingly difficult and more costly. To help address these concerns, we urge you to support policies and development incentives that enable pipelines for small populations to thrive.

### **Modernize the nation’s newborn screening program to ensure all babies can benefit from a timely and lifesaving diagnosis after birth for generations to come**

Obtaining a confirmed rare disease diagnosis takes an average of 6.3 years, and includes visits to 16.9 medical providers, and an average of 2.4 out-of-state trips<sup>14</sup>. This delay not only exacerbates physical and emotional suffering, but also leads to substantial financial burdens for patients, families, employers, and the healthcare system. Moreover, delayed diagnoses can result in irreversible disease progression and missed opportunities for timely interventions within optimal therapeutic windows. Newborn screening plays a critical role in identifying pediatric-onset conditions immediately after birth.

Each year, approximately four million newborns are screened in the U.S., and an estimated 12,000 infants are identified to have a treatable condition, benefiting from intervention<sup>15</sup>. In addition to the life-altering benefit of newborn screening, a timely diagnosis also provides an economic benefit for patients and the health care system more broadly. The Cost of Delayed Diagnosis in Rare Disease: A Health Economic Study calculated the avoidable costs associated with providing a timely diagnosis at birth as opposed to a delayed diagnosis for three newborn screening conditions<sup>16</sup>. The conditions studied – X-ALD, Pompe Disease, and SCID – showed savings of \$301,647, \$168,718, and \$517,638 in avoidable costs respectively when a timely diagnosis at birth was provided<sup>17</sup>. Newborn screening leads to better outcomes for families and the healthcare system, which is why it is often referred to as one of the most successful public health programs in the country.

The newborn screening system needs modernization to keep pace with advancements in medical science and emerging treatment options for rare diseases. In the last 15 years, only nine conditions have been added to the federal recommended panel. For the conditions that have been added, states often lag behind the recommendations, taking more than five years to add new conditions. Advances in genomic sequencing and the potential to intervene in more rare diseases as science advances present

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<sup>13</sup> Schaefer, J., Lehne, M., Schepers, J. et al. The use of machine learning in rare diseases: a scoping review. *Orphanet J Rare Dis* 15, 145 (2020). <https://doi.org/10.1186/s13023-020-01424-6>

<sup>14</sup> Yang, G., Cintina, I., Pariser, A. et al. The national economic burden of rare disease in the United States in 2019. *Orphanet J Rare Dis* 17, 163 (2022). <https://doi.org/10.1186/s13023-022-02299-5>

<sup>15</sup> Centers for Disease Control and Prevention. (2024, May 13). Emergency preparedness for newborn screening programs. Centers for Disease Control and Prevention. <https://www.cdc.gov/newborn-screening/php/about/emergency-response.html>

<sup>16</sup> The Cost of Delayed Diagnosis in Rare Disease: A Health Economic Study, EveryLife Foundation for Rare Diseases In Partnership With: The Lewin Group, part of Optum Serve Expert Stakeholders The Rare Disease Community, 2023. [https://everylifefoundation.org/wp-content/uploads/2023/09/EveryLife-Cost-of-Delayed-Diagnosis-in-Rare-Disease\\_Final-Full-Study-Report\\_0914223.pdf](https://everylifefoundation.org/wp-content/uploads/2023/09/EveryLife-Cost-of-Delayed-Diagnosis-in-Rare-Disease_Final-Full-Study-Report_0914223.pdf)

<sup>17</sup> Ibid

great potential for improving children's health, but policy gaps and resource constraints stand to limit the possibilities.

To address these growing challenges, the federal newborn screening system must be modernized through a reauthorization of federal programs that will make needed improvements to ensure that conditions are added in a timely manner and that meets the needs of 21<sup>st</sup> century diagnostics and therapies. New policies are needed that address how conditions are added, the role of genetic testing, increased investment in public health infrastructure, education, and data systems to support a more effective and equitable newborn screening program. These issues can create a forward-looking system that keeps pace with medical advancements and delivers optimal outcomes for all children and their families.

### **Support policies that facilitate timely and affordable access to approved therapies**

Many of the transformational therapies that are approved face access barriers that prohibit or delay these life-changing treatments from reaching patients. This counteracts the benefits of expedited pathways that speed the availability of drugs to treat serious diseases that are intended to ameliorate unmet patient needs and reduce long term costs by allowing for early access to innovative treatments. Further, utilization management techniques and programs to divert charitable assistance programs are employed by public and private payers to limit their expenses, but these policies have the effect of increasing costs for patients and creating delays that can result in irreversible disease progression and loss of life.

Policy solutions should aim to eliminate the patient access hurdles of time and cost, ensuring access to appropriate FDA-approved therapies without burdensome and medically inappropriate utilization management requirements. To achieve these goals, policies should also enable innovative payment models that recognize the full value of a rare disease treatment and incorporate meaningful input from patient and scientific perspectives.

Please know our organizations will be a resource to you in the coming weeks, months, and years. We welcome the opportunity to meet with you about the priorities of the rare disease community. To help facilitate the engagement of our community organization partners in administration policy priorities, please reach out to Annie Kennedy, Chief of Policy, Advocacy and Patient Engagement, with the EveryLife Foundation for Rare Diseases at [akennedy@everylifefoundation.org](mailto:akennedy@everylifefoundation.org).

Thank you for your consideration of the estimated 30 million Americans living with one or more rare diseases, and our collective policy priorities as you and your team begin to assess opportunities to advance the nation's health and well-being in the early days of your administration.