Rare Disease State Policy Priorities

The EveryLife Foundation for Rare Diseases is powered by the rare disease community to improve health outcomes by driving change through evidence-based policy, leading science-driven policy and regulatory research, activating the community to advocate for their rights and needs, and strengthening the rare disease community.

Over 30 million Americans are living with one or more rare diseases. According to the National Economic Burden of Rare Disease Study, navigating a rare disease diagnosis can require more than 6 years and 17 medical interventions, on average, after symptoms begin.[i] This study also uncovered the nearly $1 trillion annual economic impact associated with rare diseases in the U.S. Of the 10,000+ known rare diseases, 95% do not have an FDA-approved therapy.[ii]

We must act with urgency to ensure all rare disease patients and families receive timely and accurate diagnoses so they can access expert care and available therapies that can improve lives and limit preventable economic impacts. Below are a few policy areas that are important to the rare disease community.

- **Newborn Screening**: The Recommended Uniform Screening Panel (RUSP) is a list of disorders recommended for states to screen as part of their universal newborn screening programs. The RUSP contains 38 conditions that without early detection and intervention can cause disabilities or even death. RUSP alignment legislation, as passed in 13 states, creates an automatic and streamlined process for the state to evaluate, at a minimum, all new RUSP conditions, taking advantage of established medical standards and decreasing burdens for state officials and patient advocates.

- **Patient and Expert Representation in Payer Decision Making**: Rare disease patient and expert engagement with committees that determine coverage policy such as Medicaid and Commercial Drug Utilization Review (DUR) Boards and Pharmacy and Therapeutics (P&T) Committees is essential to ensuring timely and appropriate coverage policy. For the rare disease community, where 95% of conditions do not have an FDA-approved therapy[ii], making these processes transparent, timely, and accessible for public participation is critical for incorporating appropriate rare disease expertise.

- **Access to Genetic Testing Services:** Navigating a rare disease diagnosis can require more than 6 years, on average, after symptoms begin.[i] A recent study revealed that for some rare diseases, the economic impact of a delayed diagnosis can be over $500,000 per patient.[iii] Genetic testing can play a crucial role in shortening a patient's diagnostic journey, decreasing financial and other burdens on families and the health care system. Unfortunately, although an estimated 70- 80%[iv] of rare diseases are genetic, many rare disease patients still lack access to appropriate genetic and genomic tests. States can play an important role in ending the diagnostic odyssey for rare disease patients by supporting innovative pediatric genomic screening initiatives, ensuring privacy of genetic data, and providing simple and robust Medicaid coverage for a wide range of DNA sequencing services as recommended by a physician.

- **Copay Adjustment Policies:** In recent years, health plans have started using copay adjustment policies—also called copay accumulators or copay maximizers—to pocket charitable copay assistance without counting those payments toward the enrollee's deductible and cost-sharing requirements. While copay adjustment programs can reduce costs for health plans, they leave patients with unexpected and unaffordable costs once their copay assistance is exhausted. These harmful policies eat into the already tight budget patients have, restricting access to rare disease patients' high-cost, lifesaving medications. Many states have banned copay adjustment practices for state-regulated health plans.

- **Genetic Data Non-Discrimination:** With approximately 70-80% percent of rare diseases being genetic in origin,[iv] genetic testing that rapidly informs appropriate patient care and treatment is fundamental to rare disease patients' health and well-being. However, concerns regarding privacy and discrimination have deterred many from seeking genetic testing. Legislation that prohibits life, long-term care, and disability insurers from accessing genetic test results and from making coverage decisions based on such information, provides much-needed protection against discrimination and financial repercussions for individuals with genetic predispositions.

- **Access to Expert Care via Telehealth:** Rare disease patients, much like the rest of the nation, received greater access to care under expanded telehealth provisions during the Covid-19 public health emergency. By identifying solutions that enable rare disease patients to see experts via telehealth across state lines, patients can maintain already established provider relationships and begin seeing expert providers, often unavailable in their home states due to the highly specialized nature of rare disease care.

- **Prior Authorization Reform:** Health insurers and pharmacy benefit managers increasingly require healthcare professionals to obtain approval, called prior authorization, before covering pharmaceuticals and medical services. While prior authorization requirements can help health plans keep their costs low, these requirements interrupt care, divert resources from patients, and complicate medical decision-making. The EveryLife Foundation supports efforts to increase transparency, timeliness, and the use of appropriate experts in the prior authorization approval process.

To inform the EveryLife Foundation's policy priorities, we convene the Community Congress, a forum for collaboration across stakeholders, representing over 200 rare disease patient advocacy organizations in addition to over 90 other healthcare and biotechnology organizations.

You can improve the lives of millions of patients and families in your state by supporting policies that facilitate early, accurate and affordable access to diagnostic services and access to the best care and available therapies.

To learn more, please visit www.everylifefoundation.org.

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[i] EveryLife Foundation for Rare Diseases. *The National Economic Burden of Rare Disease Study*, everylifefoundation.org/burden-landing/
[ii] Fermaglich, Lewis J, and Kathleen L Miller. *A Comprehensive Study of the Rare Diseases and Conditions Targeted By Orphan Drug Designations and Approvals Over the Forty Years of the Orphan Drug Act."* Orphanet journal of rare diseases vol. 18,1 163.
[iii] EveryLife Foundation for Rare Diseases, *The Cost of Delayed Diagnosis in Rare Disease: A Health Economic Study,* https://everylifefoundation.org/delayed-diagnosis-study/
[iv] National Human Genome Research Institute, Accessed April 2024. *Rare Genetic Diseases*, www.genome.gov/dna-day/15-ways/rare-genetic-disease.