NW Rare Disease Coalition (NWRDC) Policy Priorities Overview 2025

Objective: though there are a wide range of policy outcomes that the NWRDC has supported to advance the health and well being of rare disease families since its inception in 2022, this memo seeks to outline the priorities where NW Rare is serving as a lead advocacy organization – engaging legislators directly, soliciting cosponsor support, and advocating for committee hearing and floor time in the process.

Overview / Challenges Faced by the Rare Disease Community: some fast facts that can inform any conversation about the importance of advancing policy solutions in support of the rare disease community:

- Rare diseases are defined as conditions that affect fewer than 200,000 people in the United States.

- There are over 7,000 known rare diseases affecting approximately 30,000,000 Americans and about 50 percent of rare disease patients are children. It's estimated that rare diseases impact about 750,000 Washingtonians.

- Sadly, about a third of children impacted by rare disease will not live to see their fifth birthday. Rare diseases are responsible for 35% of all deaths (of any cause) during the first year of life for all children[1].

- Though they are individually rare, as a category, rare diseases impact more Americans than HIV and Cancer combined.

- 95% of rare diseases **do not** have an FDA approved therapy.

- On average, Rare Disease patients spend more than six years searching for diagnosis[2]. This process is referred to patients as the "diagnostic odyssey"

- During the diagnostic odyssey, rare disease patients typically see upwards of 8 physicians[3] and are misdiagnosed 2 – 3 times[4] before receiving a correct diagnosis.

- According to the EveryLife Foundation, the avoidable costs attributable to delayed diagnosis, in terms of medical costs and productivity loss in the pre-diagnosis years is between $86,000 and $517,000 per patient cumulatively for the years of delay.[5] Those are expenses borne directly by patients in out-of-pocket spending, by family members who are supporting them, and by the healthcare institutions responsible for their care.

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[1] Adachi T, El-Hattab AW, Jain R, Nogales Crespo KA, Quirland Lazo CI, Scarpa M, Summar M, Wattanasirichaigoon D. Enhancing Equitable Access to Rare Disease Diagnosis and Treatment around the World: A Review of Evidence, Policies, and Challenges. Int J Environ Res Public Health. 2023 Mar 8;20(6):4732. doi: 10.3390/ijerph20064732. PMID: 36981643; PMCID: PMC10049067.

[2] Hendriksz, 2013; Benito-Lozano, Lopez-Villalba, Arias-Merino, Posada de le Paz, & Alonso-Ferreira, 2022; Yang, Cintina & Pariser, 2022)

[3] Rare Disease Impact Report: Insights from patients and the medical community. https://globalgenes.org/wp-content/uploads/2013/04/ShireReport-1.pdf

[4] Rare Disease Impact Report: Insights from patients and the medical community. https://globalgenes.org/wp-content/uploads/2013/04/ShireReport-1.pdf

[5] Everylife Foundation "The Cost of Delayed Diagnosis in Rare Disease: A Health Economic Study", accessed 11/27/2024: https://everylifefoundation.org/wp-content/uploads/2023/09/EveryLife-Cost-of-Delayed-Diagnosis-in-Rare-Disease_Final-Full-Study-Report_0914223.pdf

I. Forming a Rare Disease Advisory Council in Washington state (SB 5097 from 2023 – 2024 biennium)

Twenty-nine other states have already established a Rare Disease Advisory Council to formalize a process through which rare disease patients, families, clinicians, and researchers can collaborate and make recommendations to state governmental leaders on priority issues impacting the health and well-being of rare disease families.

WA's RDAC would be tasked specifically with the following outputs:

- Make a recommendation on the creation of an incidence and prevalence report for rare disease in Washington state. Legislative progress on rare disease community issues has stalled in WA due to an absence of data around how many rare patients there are in the state. An incidence and prevalence report would be a foundational tool that the rare disease community could leverage to help advocate for structural healthcare reforms to the benefit of the community in Washington state.

- Make a recommendation on the establishment of a WA-specific online resource for patients and families still seeking diagnosis, to assist newly diagnosed families trying to figure out "what's next" after achieving diagnosis, or for those seeking navigation assistance to understand what social service benefits that they might qualify for after achieving diagnosis.

- Make recommendations on how to better educate and inform medical providers, schools, and public health districts around the diagnosis, treatment, and access to care for persons living with a rare disease, including support for their families and caregivers.

A rare disease advisory council (RDAC) has the potential to bridge gaps in healthcare access, facilitate earlier diagnoses, enhance information sharing amongst clinicians, and provide resources for families managing rare diseases across Washington state. If passed, Washington would join the twenty nine other states across the US that have already implemented RDACs to assure greater connectivity between rare patients and the healthcare systems that serve them.

II. Providing coverage for rapid whole genome sequencing (rWGS) to help end the diagnostic odyssey (HB 1079 in the 2023 – 2024 biennium)

For rare disease patients, time is of the essence in achieving diagnosis. During the multi-year diagnostic odyssey, patients (half of whom are children) often miss key development milestones where earlier diagnosis and intervention could have averted costly and invasive cycles of care that ultimately produce worse outcomes over an individual's lifetime.

Instead, we propose to broaden access to rapid whole genome sequencing, which is an investigation of the entire human genome to quickly identify disease-causing genetic changes, for children in inpatient settings under the age of one who aren't able to be diagnosed through other means.

rWGS has a higher diagnostic success rate than sequential standard tests[6] and has been demonstrated to consistently reduce costs for patients and the healthcare system[7] in other states where Medicaid coverage is already provided.

III. Conclusion

Rare disease patients and their families experience the inequities of our healthcare system in unique and profound ways. Establishing a Rare Disease Advisory Council and expanding access to rWGS would create systemic changes that would help end the diagnostic odyssey for many chronically undiagnosed patients, provide rare families with more resources to navigate the healthcare system, and importantly, give rare disease advocates a larger institutional voice to guide Washington state's approach to caring for their community moving forward.

[6] Levenson D. Benefits of genomic sequencing evident in pediatric diagnoses: recent study finds testing method less costly, more effective than other medical, genetic tests. Am J Med Genet A. 2015 Mar;167A(3):vii-viii. doi: 10.1002/ajmg.a.37019. PMID: 25691429.

[7] Dimmock D, Caylor S, Waldman B, Benson W, et al.. Project Baby Bear: Rapid precision care incorporating rWGS in 5 California children's hospitals demonstrates improved clinical outcomes and reduced costs of care. Am J Hum Genet. 2021 Jul 1;108(7):1231-1238. doi: 10.1016/j.ajhg.2021.05.008. Epub 2021 Jun 4. PMID: 34089648; PMCID: PMC8322922.