January 3rd, 2023

Re: Support Rare Disease Community Legislative Priorities – establish a Rare Disease Advisory Council (SB 5097) and expand coverage for rapid whole genome sequencing (HB 1079)

As patients, clinicians, caregivers, researchers, and community leaders who advocate for policy changes that benefit individuals managing rare diseases, we're writing to urge your support of legislation that

will benefit rare disease patients and their families in the 2023 Washington state legislative session. A rare disease is defined as a condition that impacts fewer than 200,000 patients across the US[1]. While individual diseases may be rare, recent estimates indicate there are over 10,000 rare diseases[2], and they cumulatively impact over 750,000 Washingtonians. 80% of these conditions are genetic in origin, and about half of all rare disease patients are children[3]. On average, rare disease patients see 8 physicians[4] over 5 – 7 years[5] and are misdiagnosed 2 – 3 times[6] before receiving a correct diagnosis. This "diagnostic odyssey" causes significant hardship and expense for patients, caregivers, families, and our shared healthcare delivery system. We hope you'll join us in supporting policy changes that will address systemic gaps that rare disease patients and their families face every day.

SB 5097 – to establish a Rare Disease Advisory Council in Washington State

A rare disease advisory council (RDAC) has the potential to bridge gaps in healthcare access, facilitate earlier diagnoses, and provide resources for families managing rare diseases across Washington state. Previously, meaningful policy changes to help rare disease patients in Washington have been thwarted by a lack of actionable information about the incidence and prevalence of rare diseases, or the challenges that rare disease families navigate. SB 5097 would remedy these issues. Washington should join the 24 other states across the US that have implemented RDACs to assure greater connectivity between rare patients and the healthcare systems that serve them.

HB 1079 – to expand coverage for rapid whole genome sequencing and end the diagnostic odyssey

For rare disease patients, time is of the essence in achieving diagnosis. During the diagnostic odyssey, rare patients often miss key development milestones where earlier diagnosis and intervention could

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[1] Valdez R, Ouyang L, Bolen J. Public Health and Rare Diseases: Oxymoron No More. [Erratum appears in Prev Chronic Dis 2015;12. http://www.cdc.gov/pcd/issues/2015/15_0491e.htm.] Prev Chronic Dis 2016;13:150491. DOI: http://dx.doi.org/10.5888/pcd13.150491

[2] Haendel M, Vasilevsky N, Unni D, Bologa C, Harris N, Rehm H, Hamosh A, Baynam G, Groza T, McMurry J, Dawkins H, Rath A, Thaxon C, Bocci G, Joachimiak MP, Köhler S, Robinson PN, Mungall C, Oprea TI. How many rare diseases are there? Nat Rev Drug Discov. 2020 Feb;19(2):77-78. doi: 10.1038/d41573-019-00180-y. PMID: 32020066; PMCID: PMC7771654.

[3] Dicken, John. Rare Diseases: Although limited, available evidence suggests medical and other costs can be substantial. Government Accountability Office. October 2021. https://www.gao.gov/assets/gao-22-104235.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] Global Commission. Ending the diagnostic odyssey for children with a rare disease. 2019. globalrarediseasecommission.com;
Posada de la Paz M, Taruscio D, Groft SC. Rare diseases epidemiology: Update and overview. 2nd edition. Chapter Two. Springer 2017. Cham, Switzerland.

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

have averted costly and invasive cycles of care that ultimately produce worse outcomes over a person's whole lifetime. Rapid whole genome sequencing has a higher diagnostic success rate than sequential standard tests[7] and has been demonstrated to reduce costs for patients and the healthcare system[8]. HB 1079 would broaden access to rapid whole genome diagnostics and bring an end to the long, frustrating diagnostic odyssey for many Washingtonians urgently in need of care.

Rare disease patients and their families experience the inequities of our healthcare system in unique and profound ways. Both SB 5097 and HB 1079 would create systemic changes that would end the odyssey for chronically undiagnosed patients, provide rare families with more resources to navigate the healthcare system, and importantly, give rare disease advocates a formal voice to guide Washington state's approach to caring for their community. We would urge your support of these legislative priorities in the 2023 session.

Thank you very much for your time and consideration,

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[7] 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.
[8] 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.