Rare Disease Advisory Council
2024 Summation of Activities

Health-General Article §13-5004

Maryland Department of Health

July 2025

Table of Contents

Executive Summary

The Maryland Rare Disease Advisory Council (the Council) is required to report annually on its activities, funding, accomplishments, and recommendations in accordance with Senate Bill 188/House Bill 302 (2023). This report outlines the accomplishments of the Council over the past year, notably after the Council meetings officially started in June of 2024.

The Council prioritized establishing its operational procedures, developing plans for council workflow, and identifying action items for future research. Currently, eight of twenty-one council seats remain vacant, including the positions of chair and vice chair.[1] Existing council members, which include individuals with rare diseases, those engaged in rare disease research, members of the Senate, and other rare disease stakeholders, share a common goal to improve the lives and health of those impacted by rare diseases. As the Council is newly formed, vacancies must be filled by governor appointment and are expected to be filled within the year.

Since June of 2024, the Council has accomplished the following:

- Researched best practices and created a planning document to develop the Maryland Rare Disease Needs Assessment;
- Identified rare disease stakeholders in Maryland, including individuals with rare diseases, parents of individuals with rare diseases, primary care providers, professional organizations, teaching hospitals, nursing associations, geneticists, and more;
- Developed potential dissemination strategies for the needs assessment, including but not limited to channels through the surrounding teaching hospitals and professional medical societies;
- Prioritized legislative categories of future research; and
- Collaborated with peer contacts in other state rare disease advisory councils (RDACs) and the National Organization for Rare Disorders (NORD).

The Council also prioritized future work with plans to create and disseminate the needs assessment survey within the next year, to understand baseline rare disease data and needs, as well as perform functional analysis on rare disease prevalence in the state. Additionally, as the Council continues to grow its membership and body of work through 2025, it will continue to advocate for and serve Maryland’s rare disease community through council activities.

Background

Rare diseases are defined as conditions that impact less than 200,000 individuals, and are typically genetic, hereditary, and progressive in nature (NIH, n.d.). Despite rare diseases’ individuality, together, rare diseases affect an estimated 30 million people in the United States,

---

[1] RDAC vacancies as of October 1, 2024.

and over 350 million people worldwide (Sequeira et al., 2021). Rare diseases place stress on a varied group of stakeholders, including individuals, organizations, and the healthcare system at large. Individuals with rare diseases face challenges including, but not limited to, delays in diagnosis, insurance issues, high out-of-pocket costs, limited access to treatment, and dissatisfaction with providers (Bogart et al., 2022). On a health care systems level, rare diseases are often associated with high costs that increase the economic impact of rare diseases (Delaye et al, 2022; Sequeira et al., 2021). Nationally, it is estimated that the economic burden of rare disease is $997 billion in the United States, with inpatient care and prescriptions contributing the highest percentage of costs (Yang et al., 2022).

In Maryland, rare disease is a significant public health issue. The prevalence of rare disease in Maryland is not known, however, the advocacy coalition, Maryland Rare, estimates there to be 750,000 individuals in Maryland living with a rare disease (Maryland Rare, 2024). Diagnostic delays are common experiences for individuals with rare disease; research reports an average of four to five years before individuals with a rare disease obtain an accurate diagnosis (Marwaha et al., 2022). Even when diagnosed, only five percent of rare diseases have Food and Drug Administration (FDA) approved treatments (NIH, 2023). Further, cost reduction strategies and regulations such as Prescription Drug Affordability Boards (PDABs) do not account for rare diseases, making it even more difficult for individuals with a rare disease to obtain timely, safe, and affordable care (Gronde et al., 2017).

A recent worldwide review of the literature suggests that increasing knowledge of rare disease through education, advocacy for individuals with rare disease, standardizing healthcare guidelines on rare disease, and emphasizing research and development are some of the best ways to lower the burden of rare disease on individuals and communities at large (Adachi et al., 2023). Therefore, initiatives like the Council are critical in reducing the burden of rare disease in the state.

Maryland Rare Disease Advisory Council

The Council was established in May of 2023 through the enactment of Senate Bill 188/House Bill 302, making Maryland the 26th state to house a RDAC. The Council aims to serve, advocate, and make recommendations for Marylanders with rare diseases. Inherently, the burden of rare disease spans across many disciplines, and it is the responsibility of the Council to help alleviate this burden in the state.

To fulfill its purpose, the Council will accomplish the following tasks as outlined in Health-General Article §13-5001-§13-5004:

(1) Convene public hearings, make inquiries, and solicit comments from the public to assist the Council with a first-year survey of the needs of individuals with a rare disease, caregivers, and health care providers in the state;
(2) Consult with experts on rare diseases to 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;
(3) Research and make recommendations to state agencies and insurers that provide services to individuals with a rare disease on the impact of prior authorization, cost-sharing, tiering, or other utilization management procedures on the provision of treatment and care for patients;
(4) Establish best practices and protocols to include in state planning related to natural disasters and public health emergencies or other emergency declarations to enable continuity of care for rare disease patients and ensure that safeguards against discrimination for rare disease patients are in place;
(5) Evaluate and make recommendations regarding coverage of prescription drugs for rare disease patients, including patients with private health insurance coverage and patients enrolled in the Maryland Medical Assistance Program, to improve coverage of diagnostics, and to facilitate access to necessary health care providers with expertise in the treatment of rare diseases;
(6) Publish a list of existing and publicly accessible resources on research, diagnosis, treatment, and education relating to rare disease on the Council's webpage;
(7) Identify areas of unmet needs for research that can inform future studies and reports by the Council;
(8) Identify and distribute educational resources for health care providers to foster recognition and optimize treatment of rare diseases in the state; and
(9) Research and identify best practices to ensure continuity of care for rare disease patients transitioning from pediatric to adult care.

Council Membership

As of September 2024, the Council consists of 13 members. As the Council is still in its first year, eight seats remain vacant. Additionally, the Council does not yet have a chair or vice chair. The current members, as well as council vacancies, are listed in the Appendix.

Council Funding

The Council is currently unfunded. Initial Council meetings have included robust discussions regarding possible funding sources, such as the Global Genes RARE Impact Grant (Global Genes, 2024). Any funds acquired could be used to create and disseminate a needs assessment, which would include incentives for survey participants. In the future, funding may also be utilized to address gaps identified through the needs assessment.

Council Activities

As the Council began its work to serve those impacted by rare diseases, its activities were primarily research-driven and focused on planning for the future. Council members engaged in thoughtful and thorough discussions to determine important rare disease stakeholders and potential collaborators in conducting a needs assessment for those impacted by rare disease in the state.

Council Meetings

Between June and September 2024, the Council met four times and will continue to meet monthly through June 2025. After this date, meetings will shift to quarterly unless Council work necessitates more frequent meetings.

All meetings are open to the public in accordance with Maryland's Open Meetings Act, and meeting agendas, minutes, and meeting information are available on the council's website.[2] Only meetings for which a quorum was present are included on the website.

Meetings Timeline
During 2024, the Council met on the following dates:

- June 17
- July 15
- August 19
- September 16

Council Accomplishments

Through the first four meetings of the Council, members brainstormed how to accomplish the council duties and produce the required deliverables. Steps were taken to plan the needs assessment to be distributed to rare disease stakeholders in Maryland, gather resources for those impacted by rare disease to share on the council website, and learn from the best practices of RDAC peers.

Council members started planning for the needs assessment topics and survey that will be conducted in the next year. Rare disease stakeholders were identified, including individuals with rare diseases, parents of individuals with rare diseases, primary care providers, professional organizations, teaching hospitals, nursing associations, coordinating centers, rare disease

---

[2] Rare Disease Advisory Council Website.
https://health.maryland.gov/phpa/cyshcn/Pages/Rare-Disease-Advisory-Council.aspx

specialists, and geneticists. Potential dissemination strategies were also discussed, including through the surrounding teaching hospitals, professional medical societies, patient advocacy groups, and primary care providers who serve individuals with rare disease. The Council also prioritized needs of individuals with rare disease, patient access to care, and needs of rare disease providers as required by legislation.

Collaboration with Other State RDACs

To understand how to best move forward with the Council, Council members facilitated key informant interviews with RDAC members from other states, including North Carolina, Pennsylvania, and Georgia. Additional resources were compiled from the states of Massachusetts, Florida, and Nevada.

As the Council prepares for 2025, members plan to continue to reach out to their contacts in other RDACs and the NORD. These conversations will guide the development of Maryland's needs assessment survey, increase understanding of the Council's best practices, and allow for resource sharing between peers of the Council. The Council was not able to start meetings until the summer, which caused delays in the initial outreach to other RDACs around the country. Additional collaboration with fellow RDACs will occur during the October NORD Rare Disease Summit, as will continued outreach via phone and email.

Council Recommendations

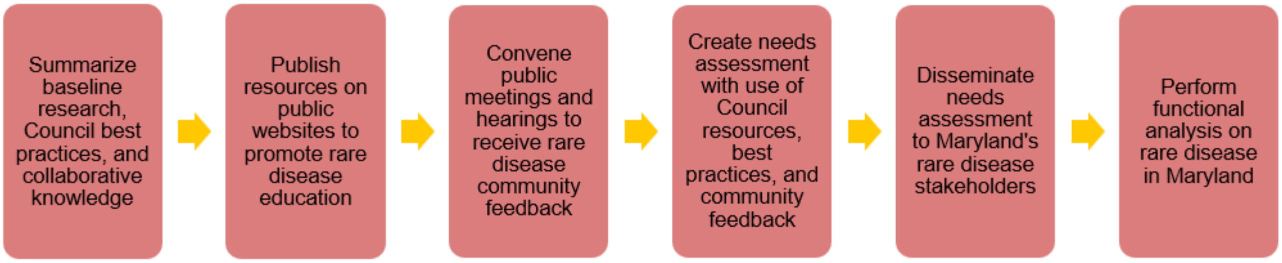
To make recommendations on behalf of stakeholders in the rare disease community, on matters related to rare disease in the state of Maryland, and for individuals with rare disease, the Council plans to first complete critical, foundational research to better understand rare disease in Maryland. Keeping this in mind, the Council has created the following recommendations to focus council efforts and resources:

1. Determine the overall prevalence of rare diseases in Maryland.
2. Determine which rare diseases are the most prevalent in Maryland.
3. Develop and conduct a needs assessment to pinpoint the needs of caregivers of, healthcare providers for, and individuals with rare disease.
4. Raise awareness of rare disease in Maryland to state constituents, policymakers, healthcare systems, and nonprofits.
5. Proliferate informational content on public websites to promote rare disease education to community stakeholders and the public.
6. Collaborate with other RDACs around the country and representatives from NORD, to identify best practices and research priorities for the Council.

Future Work of the Council

The Council was created to study, advocate for, raise awareness of, and make recommendations on behalf of the rare disease community in the state. The Council has made significant accomplishments in outlining processes, establishing collaborative contacts, and identifying research priorities. The below chart depicts a visual representation of the Council's future work to be completed in the next year. This framework will allow for the Council's continued success.

Figure 1: The Council Future Work Through 2025



The Council has initiated the process to develop a plan to complete several of the legislative tasks. These tasks include the needs assessment of rare disease stakeholders in Maryland, identifying best practices in care for individuals with rare disease, publishing resources on rare disease for the public to access, raising awareness of rare diseases in Maryland, and convening public meetings and hearings to receive community feedback. The next steps include creating and disseminating the needs assessment survey to understand baseline rare disease data and needs, and performing functional analysis on rare disease prevalence in the state. Once this baseline data is established, focused legislative recommendations can be made with the goal of improving the health of all Marylanders living with a rare disease.

References

Adachi, T., El-Hattab, A. W., Jain, R., Nogales Crespo, K. A., Quirland Lazo, C. I., Scarpa, M., ... & Wattanasirichaigoon, D. (2023). Enhancing equitable access to rare disease diagnosis and treatment around the world: a review of evidence, policies, and challenges. *International journal of environmental research and public health*, *20*(6), 4732.

Bogart, K., Hemmesch, A., Barnes, E., Blissenbach, T., Beisang, A., & Engel, P. (2022). Healthcare access, satisfaction, and health-related quality of life among children and adults with rare diseases. *Orphanet journal of rare diseases*, *17*(1), 196.

Delaye, J., Cacciatore, P., & Kole, A. (2022). Valuing the "burden" and impact of rare diseases: a scoping review. *Frontiers in Pharmacology*, *13*, 914338.

Gronde, T. V. D., Uyl-de Groot, C. A., & Pieters, T. (2017). Addressing the challenge of high-priced prescription drugs in the era of precision medicine: A systematic review of drug life cycles, therapeutic drug markets and regulatory frameworks. *PloS one*, *12*(8), e0182613.

Rare patient impact grant opportunities. Global Genes. (2024). https://globalgenes.org/rare-impact-grant-opportunities/

Sequeira, A. R., Mentzakis, E., Archangelidi, O., & Paolucci, F. (2021). The economic and health impact of rare diseases: A meta-analysis. *Health Policy and Technology*, *10*(1), 32-44.

Tiger, G. (2024, January 8). *Opinion: Maryland policymakers must protect rare disease patients*. Maryland Matters. https://marylandmatters.org/2024/01/08/opinion-maryland-policymakers-must-protect-rare-disease-patients/

U.S. Department of Health and Human Services. (2023, March 21). *Rare disease day at NIH 2023: Putting hope into action*. National Center for Advancing Translational Sciences. https://ncats.nih.gov/news-events/news/rare-disease-day-at-NIH-2023-putting-hope-into-action

Yang, G., Cintina, I., Pariser, A., Oehrlein, E., Sullivan, J., & Kennedy, A. (2022). The national economic burden of rare disease in the United States in 2019. *Orphanet journal of rare diseases*, *17*(1), 163.

Appendix

Council Membership

Seat	**Member**
Hospital Administrator	Peter Hill, MD, MS, FACEP
Pharmacist licensed in the state with experience dispensing drugs used to treat rare disease	Kristopher Rusinko, Pharm D, PhD, MBA, M.Ed.
Caregiver of an individual with a rare disease	Jeneva Stone, PhD
Representative of the biopharmaceutical industry	Matthew Mehan, BA
Representative of a rare disease patient organization that operates in the state	Lauren Shillinger, BA
Member of the scientific community who is engaged in rare disease research	Constance Smith-Hicks, MD, PhD
Member of the Senate of Maryland	Clarence Lam, MD
Member of the Maryland House of Delegates	Jamila Woods, MDiv, MSW
Representative of the Office of Minority Health and Health Disparities	Camille Blake-Fall
Representative of the Maryland Medical Assistance Program	Mary Mussman, MD, MPH
Representative of the Maryland Insurance Administration	Jamie Sexton, JD
Individual who has been diagnosed with a rare disease	E. Felicia Brannon, MPA
Chair of the State Advisory Council on Hereditary and Congenital Disorders	Jamie Fraser, MD, PhD, FACMG
Secretary of Health or Designee	Vacant
Academic research institution that receives rare disease grant funding	Vacant
Geneticist practicing in Maryland	Vacant
Registered nurse or advanced practice nurse with experience treating rare disease	Vacant

Physician with experience treating rare diseases	Vacant
An individual who has been diagnosed with a rare disease	Vacant
Health insurance provider representative	Vacant
Organization representative that provides care management for individuals in Maryland’s Rare and Expensive Case Management Program	Vacant