Rare Disease Advisory Council

2025 Report

Health-General Article §13-5004

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Executive Summary

The Maryland Rare Disease Advisory Council (the Council) is required to annually report to the Governor and the General Assembly on its activities, funding, accomplishments, and recommendations in accordance with Health-General Article §13-5004. This report details the accomplishments of the Council in the past year, including the last three months of 2024.

The Council prioritized a legislatively mandated needs assessment this year, in addition to streamlining operational procedures, researching best practices, discussing legislation, and identifying Council priorities. Three appointments were made this year, including the appointment of a Council Chair. Currently, five out of the twenty-one council seats remain vacant. Sitting Council members continually strive toward the goal of improving the health of those in Maryland's rare disease community. Council vacancies must be filled by Governor appointment, and are expected to be filled within the year.

Since October 2024, the Council has accomplished the following:

- Engaged in research on best practices for rare disease advisory council (RDAC) needs assessments;
- Collaborated with peer contacts in other RDACs, including Pennsylvania and Florida, as well as the National Organization for Rare Disorders;
- Formed a subcommittee to focus on needs assessment efforts;
- Held discussions on current legislation and potential advocacy efforts;
- Wrote a letter to the Secretary of Health regarding the Rare and Expensive Case Management Program; and
- Finalized the Maryland RDAC Needs Assessment (the Assessment), which was approved by the Maryland Department of Health's Institutional Review Board.

The Council identified and prioritized future work with plans to disseminate the Assessment in Fall 2025, analyzed baseline rare disease data gathered from the Assessment, and involved the rare disease community through information sharing and education. As the Council continues to grow in membership and goals, it will continue to serve and advocate for Maryland's rare disease community.

Background

Rare diseases are defined as conditions that impact fewer than 200,000 individuals, and are typically genetic, hereditary, and progressive in nature.[1] Despite the individuality of rare diseases, collectively, they affect more than 30 million people in the United States and over 350 million people worldwide.[2, 3]

[1] U.S. Department of Health and Human Services, National Institutes of Health. (2025). *About GARD*. Genetic and Rare Diseases Information Center. https://rarediseases.info.nih.gov/about

[2] Fermaglich, L. J., & Miller, K. L. (2023). 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, 18(1), 163.

[3] 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.

In 2022, a global group of rare disease stakeholders, including patients, policy makers, and clinicians gathered to create an agreed-upon Operational Description of Rare Diseases that includes a core definition and three frameworks.[4] This definition highlights the complexity of rare diseases, including the distinct burdens faced by those living with a rare disease, how rarity creates challenges in care, and the variety of diseases and conditions included in rare diseases.[4]

Rare diseases, therefore, place a significant burden on a diverse group of stakeholders, including individuals, organizations, and the broader healthcare system. Individuals with rare diseases face numerous challenges, including but not limited to: delays in diagnosis, insurance issues, high out-of-pocket costs, limited access to treatment, and dissatisfaction with providers.[5] At the healthcare system level, rare diseases are often associated with high costs, which in turn increase their economic impact.[3, 6] Nationally, the economic burden of rare diseases is estimated at $997 billion, with inpatient care and prescriptions contributing the highest percentage of costs.[7]

In Maryland, rare disease is a significant public health issue. While the most common rare diseases in Maryland are not currently known, a recent literature review noted that the state of Maryland has the second most reported rare variant types for alpha 1 antitrypsin deficiency in the United States.[8]

Diagnostic delays are common for individuals with rare diseases- research reports an average of four to five years before an individual with a rare disease obtains an accurate diagnosis.[9] Even when diagnosed, only 5% of rare diseases have Food and Drug Administration (FDA) approved treatments.[10] Furthermore, 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 rare diseases to obtain timely, safe, and affordable care.[11]

A recent worldwide review of the literature suggests that increasing knowledge of rare diseases 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 diseases on individuals and communities at large.[12] Further, rare disease is becoming a global health priority

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[4] Wang, C. M., Whiting, A. H., Rath, A., Anido, R., Ardigò, D., Baynam, G., ... & Macchia, F. (2024). Operational description of rare diseases: a reference to improve the recognition and visibility of rare diseases. Orphanet journal of rare diseases, 19(1), 334.

[5] 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.

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

[7] 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.

[8] Ferrarotti, I., Wencker, M., & Chorostowska-Wynimko, J. (2024). Rare variants in alpha 1 antitrypsin deficiency: a systematic literature review. Orphanet journal of rare diseases, 19(1), 82.

[9] Marwaha, S., Knowles, J. W., & Ashley, E. A. (2022). A guide for the diagnosis of rare and undiagnosed disease: beyond the exome. *Genome medicine*, *14*(1), 23.

[10] 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

[11] 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

[12] 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.

recognized by the United Nations, World Health Organization, and International Rare Disease Research Consortium, in which awareness, training, and education on rare disease are key recommended interventions.[13, 14] Therefore, initiatives like the Council are critical in reducing the burden of rare diseases in the state.

About the Maryland Rare Disease Advisory Council

The Council works to serve Marylanders affected by rare diseases by advocating and making recommendations on their behalf. As noted in the 2024 Council report, the Council works to accomplish the following tasks as outlined by Health-General Articles §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 2025, the Council consists of 16 members, with three new appointments made since the last report of this Council. Since the Council is still new, five seats remain vacant. The Council appointed

[13] Adams, D. R., van Karnebeek, C. D., Agullo, S. B., Faùndes, V., Jamuar, S. S., Lynch, S. A., ... & IRDiRC Diagnostic Scientific Committee. (2024). Addressing diagnostic gaps and priorities of the global rare diseases community: Recommendations from the IRDiRC diagnostics scientific committee. European Journal of Medical Genetics, 70, 104951

[14] Baynam, G., Hartman, A. L., Letinturier, M. C. V., Bolz-Johnson, M., Carrion, P., Grady, A. C., ... & Groft, S. (2024). Global health for rare diseases through primary care. The Lancet Global Health, 12(7), e1192-e1199.

Dr. Ada Hamosh as its Chair as of June 2025. The current members, as well as council vacancies, are listed in the Appendix.

Council Funding

The Council does not currently receive any funding. Council members have discussed that any funds awarded could be used to address gaps identified in the Assessment.

Council Activities

The Council's second year of work focused on thoughtful, research-driven discussions, dialogue on potential statutory changes, and planning for future work. The Council created a subcommittee that led the group in developing and finalizing the Assessment. The Assessment is a legislatively mandated Council activity and has been a high priority for the Council this year. Additionally, the Council wrote a letter to the Secretary of Health requesting a meeting with the Rare and Expensive Case Management Program in Maryland to discuss how it could better serve the rare disease community.

Council Meetings

Between October 2024 and September 2025, the Council met 12 times. The Council continues to meet monthly and plans to do so until its work no longer requires frequent meetings. The Council discussed a potential shift to bi-monthly or quarterly meetings after the Assessment is complete.

All meetings are open to the public in accordance with Maryland's Open Meetings Act. Council meeting agendas, minutes, and information about upcoming meetings are available on the Council's website.[15] Once adopted by a quorum of members, meeting minutes are available on the website.

Meeting Dates
During the reporting period, the Council met on the following dates:

2024
- October 30
- November 18
- December 16

2025
- January 27
- February 24
- March 17
- April 21
- May 19
- June 16
- July 8
- August 12

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

- September 9

Council Accomplishments

In the Council's second year, members focused efforts on creating the statutorily mandated Assessment. Council members engaged in research and informational interviews with peers to gather information on the needs assessment processes of other rare disease advisory councils. Members also sought expertise and advice from the National Organization for Rare Disorders (NORD) to guide the Assessment's questions. Council members, as well as Maryland Department of Health (MDH) staff, participated in NORD's annual convening, the 2024 NORD Breakthrough Summit. This convening brought together rare disease advisory council members from across the country, as well as rare disease experts, innovators in rare disease care, and other stakeholders.

Council members formed a subcommittee to focus on the development and creation of the Assessment. The subcommittee met five times and reported work to the larger council at subsequent Council meetings. Once the subcommittee completed a draft of the Assessment, the Council discussed survey questions, timing, audience, accessibility, dissemination, and participation. The council then voted on and approved the Assessment for conversion into an electronic survey format.

The final Assessment[16] drew inspiration from toolkits by NORD and needs assessments from rare disease advisory councils in Pennsylvania, Nevada, and Florida, as well as from Council member research and discussion. Once finalized, the Council submitted the Assessment to MDH's Institutional Review Board (IRB) for approval. The IRB determined the Assessment exempt from further review and able to proceed, so the Council will distribute the Assessment in the Fall 2025.

In addition to developing the Assessment, the Council engaged in multiple discussions on potential statutory changes that could improve health outcomes for the Maryland rare disease community. Specifically, the Council discussed the Rare and Expensive Case Management (REM) Program. The REM Program assists individuals with specific medical conditions and their families by providing case management and other eligible, medically necessary services.[17] To be eligible, individuals served by the REM program in Maryland must meet a multitude of requirements, including but not limited to being eligible for Medicaid, and having one of the diseases listed on the Rare and Expensive Disease Diagnosis list provided in COMAR 10.09.69.17.

The Council held discussions over the course of multiple meetings on the REM Program, exploring ways to improve and expand the program's purview. As a result of these conversations, the Council wrote a letter to the Secretary to request a meeting with REM leadership. The letter detailed Council thoughts on sections of the REM Program that could be reevaluated, including conditions covered, age limits for coverage, and the necessity of Medicaid eligibility to receive REM benefits. Ultimately, the Council's goal for the letter is to expand the REM program to positively impact those in Maryland with rare diseases who are not currently covered by the program's benefits. Further, this work aligns with the Council's

[16] Final RDAC Needs Assessment

[17] Maryland Department of Health Maryland Medicaid Administration. (2022). Rare and Expensive Case Management (REM) Program. Maryland.gov. https://health.maryland.gov/mmcp/Pages/remprogram.aspx

legislatively mandated goals to make recommendations on behalf of individuals with rare diseases regarding treatment of disease and coverage of services.

Council Recommendations

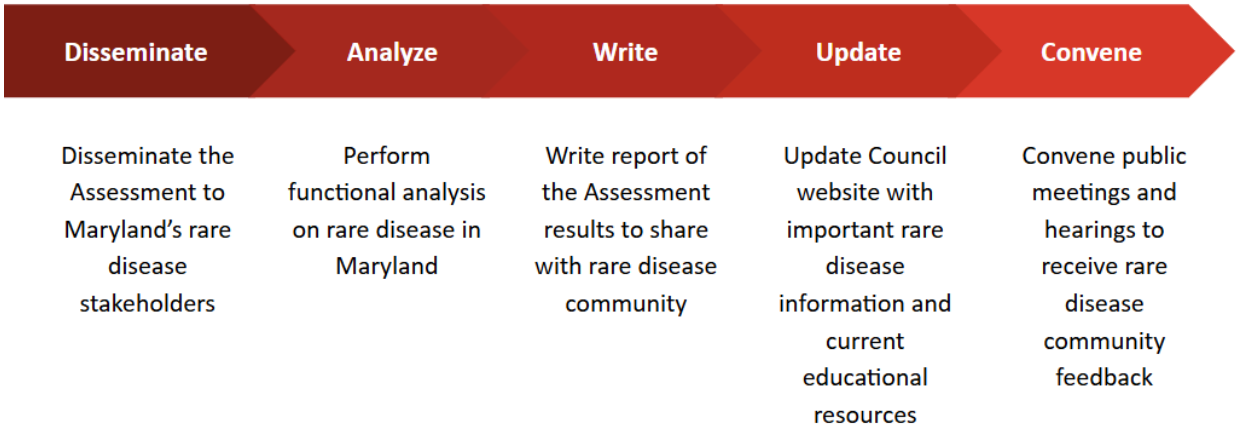
The Council aims to make recommendations on behalf of stakeholders in the rare disease community. To inform these recommendations, the Council has developed and made plans to distribute the Assessment. The research that went into creating the Assessment, as well as its deployment, are crucial steps in foundational research to understand the rare disease community in Maryland. Results from the Assessment will allow for the Council to focus future recommendations, but prior to receiving results, the Council has created the following recommendations to focus efforts:

1. Disseminate the Council Assessment to better understand the needs of those in the Maryland rare disease community;
2. Analyze critical rare disease data in Maryland on the overall prevalence of rare diseases, the most prevalent rare diseases, healthcare gaps experienced by the rare disease community, and more;
3. Raise awareness of rare diseases in Maryland by sharing the results of the needs assessment with state constituents, policymakers, healthcare systems, providers, and nonprofit organizations;
4. Reevaluate how the Maryland REM Program could better serve the Maryland rare disease community; and
5. Publish informational content on the Council's public website, to educate community stakeholders and public entities on rare disease.

Future Work

The Council has made significant progress in its second year by establishing priorities, researching needs assessment practices, collaborating with peers, developing the Assessment, evaluating statute, and identifying future goals. The chart below depicts the Council's work to be completed in the next year.

Figure 1: Future Work Through 2026



Conclusion

The Council worked diligently this year toward several of its legislatively mandated tasks. Once the Assessment is disseminated and results are gathered, baseline functional analysis can be performed, and a report of findings can be written. The data gathered from the Assessment will enable the Council to make focused legislative recommendations with the goal of advocating for and improving the health of Marylanders living with a rare disease.

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, MEd
Caregiver of an individual with a rare disease	Jeneva Stone, PhD
Representative of the biopharmaceutical industry	Matthew Meehan, 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	David Mann, MD, PhD
Representative of the Maryland Medical Assistance Program	Elisdel Garcia-Bousquet, MD, FAAP
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	Gerald Raymond, MD
Secretary of Health or Designee	Intentionally Vacant
Academic research institution that receives rare disease grant funding	Phillip Iffland, PhD
Geneticist practicing in Maryland	Ada Hamosh, MD, MPH, Chair
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	Celinda Carr, LCSW-C, CCM