

**December 20, 2022**

**MEMORANDUM**

**TO:** Dave Burhop  
Director, Division of Legislative Automated Systems

**FROM:** Alexandra Jansson  
Senior Policy Analyst, Virginia Department of Health

**SUBJECT:** 2022 Report to the Governor and General Assembly  
Rare Disease Council

This report is submitted in compliance with § 32.1-73.15 (4), which states:

Submit annually by October 1 a report to the Governor and the General Assembly for publication as a report document as provided in the procedures of the Division of Legislative Automated Systems for the processing of legislative documents and reports. The annual report shall (i) describe the activities and recommendations of the Council and (ii) describe the status of funding available to the Council, including information regarding any grants applied for and received by the Council.

Should you have any questions or need additional information, please feel free to contact me at (804) 864-7002.

Sr /

Enclosure

Pc: The Honorable John Littel, Secretary of Health and Human Resources

**2022 Report to the Governor and General Assembly**

**Rare Disease Council**

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## **Executive Summary**

The Rare Disease Council (the Council) is established in the Code of Virginia (§32.1-73.14) to advise the Governor and General Assembly on rare diseases. The Council is required to report to the Governor and General Assembly by October 1 of each year the following:

1. Activities and recommendations of the Council.
2. Status of funding available to the Council, including grants applied for and received by the Council.

The Council completed its first year of service to the Commonwealth on June 30, 2022. Throughout this period, the Council worked to understand and address its purpose, powers, and duties as defined by the legislation. This document serves as the required 2022 annual report to the Governor and General Assembly.

## **Introduction**

According to the National Institutes of Health's (NIH) Genetic and Rare Diseases Information Center, a rare disease is defined as a condition that affects fewer than 200,000 people in the United States, and there are an estimated 7,000 known rare diseases (National Institute of Health, 2022). The NIH adopted this definition from the Rare Diseases Act of 2002. Per a fact sheet published by the National Organization for Rare Disorders (NORD), it is estimated that there are nearly 30 million, or one in ten, Americans living with a rare disease (NORD, 2019).

## **Background**

The Rare Disease Council is established in the Code of Virginia (§32.1-73.14). The purpose of the Council, as defined by the legislation, is to do the following:

- Advise the Governor and the General Assembly on the needs of individuals with rare diseases in Virginia;
- Identify the challenges that such individuals face, including delays in obtaining a diagnosis or the receipt of a misdiagnosis, shortages of medical specialists who can provide treatment, and lack of access to therapies and medication used to treat rare diseases;
- Fund research related to rare diseases and the development of new treatments for rare diseases; and
- Fund supports for persons with rare diseases in the Commonwealth.

The law required the Council to hold its first meeting within ninety days of July 1, 2021, and to meet quarterly. Additionally, the legislation assigned the Council the power and duty to hold public hearings and to make inquiries of and solicit comments from the public, within the

first year, to assist the Council in understanding the scope of rare diseases and the impact of rare diseases on individuals in the Commonwealth. This report focuses on the work of the Council during its first year.

### **Status Report on Council Activities**

The Rare Disease Council became effective on July 1, 2021. The Governor's office finalized appointments for the eighteen non-legislative members, including the chair and vice-chair, in September 2021. The original appointee for the required hospital administrator seat declined appointment, and this seat remained vacant during the Council's first year. The other appointed seventeen and three ex-officio council member seats remained occupied. The Virginia Department of Health (VDH) provides staff support to the Council. The current membership roster is included in Appendix A.

The Council held its first meeting on September 27, 2021, which was within ninety days of July 1, 2021, as required by the legislation. The chair and vice-chair established the quarterly meeting schedule for the year, and meetings occurred on September 27, 2021; December 6, 2021; March 29, 2022; and May 17, 2022. The Council held two additional workgroup meetings on April 15, 2022 and May 5, 2022. Draft meeting minutes have been prepared and are posted to the Virginia Regulatory Town Hall. The Council also convened a public hearing immediately following the May 17<sup>th</sup> meeting to solicit comments about rare diseases from the general public.

To learn more about the scope and impact of rare diseases, the Council received presentations from Rare Disease Advisory Councils representing other states and rare disease patient organizations, including NORD. The Council learned about Minnesota's rare disease needs assessment survey that was completed in 2021 and Pennsylvania's needs assessment that is currently underway, as well as NORD's Annual State Report Card (NORD, 2022).

The Council received presentations and comments from caregivers and individuals affected by rare diseases in Virginia to learn more about the needs and challenges they face. Delegate Kathleen Murphy also addressed the Council at the March 29<sup>th</sup> quarterly meeting. Council members engaged in open discussion periods during quarterly meetings to learn from each member's area of expertise and to discuss the Council's plans for the future. The Council also requested the formation of a subgroup to engage in a visioning activity to develop a draft vision statement to assist the Council with defining and prioritizing its goals. The subgroup met twice and developed a draft mission statement, draft vision statement, and proposed workgroups to present to the full Council for discussion at the May quarterly meeting. VDH staff also established a [webpage](#) for the Council to provide publicly accessible resources relating to rare diseases.

The membership of the Council includes individuals diagnosed with a rare disease, caregivers of individuals diagnosed with a rare disease, and practitioners who provide care to patients diagnosed with a rare disease. Membership furthermore includes geographic representation from across the Commonwealth. Many members opted to meet virtually versus traveling to meet in person. The Rare Disease Council did not achieve the in-person attendance requirements for a quorum at its quarterly meetings, so the Council was unable to vote or make recommendations this state fiscal year.

The Council is exploring options for learning more about the scope and impact of rare diseases in Virginia, including development of a survey and other methods of qualitative or quantitative data collection. The Council is also considering establishing workgroups to focus on specific tasks between quarterly meetings and to help the Council prioritize its efforts. For instance, the topic of telehealth was discussed extensively during council meetings, and a

workgroup could explore ways that access to telehealth services could potentially improve care for individuals diagnosed with rare diseases across the Commonwealth.

### **Status of Funding Available to the Council**

The Rare Disease Council Fund was established; however, the Council did not apply for nor receive any gifts, grants, or donations as of this report. The Council intends to establish procedures and processes for requesting gifts and donations, applying for grants, and using received funds in alignment with the legislation.

### **Conclusion**

The Rare Disease Council completed its first full year of service to the Commonwealth on June 30, 2022. During its quarterly meetings, the Council received valuable input from Rare Disease Advisory Councils representing other states, rare disease patient organizations, individuals diagnosed with a rare disease, caregivers of individuals diagnosed with a rare disease, healthcare providers, and a variety of other rare disease stakeholders. The Council's first year activities are informing its goals and plans for the next year. The unique membership composition of the Council, as outlined in the legislation (§ 32.1-73.16) and with respect to individuals who are affected by rare diseases as well as caregivers of those with rare diseases, presents a challenge to some members regarding the Council's in-person meeting requirements. The in-person quorum required to conduct business has limited the development and productivity of the Council.



## References

National Institute of Health. *FAQs About Rare Disease*. Retrieved September 20, 2021, from <https://rarediseases.info.nih.gov/diseases/pages/31/faqs-about-rare-diseases>.

National Organization for Rare Disorders. *NORD State Report Card*. Retrieved May 24, 2022, from <https://rarediseases.org/nord-state-report-card/>.

National Organization for Rare Disorders. *Rare Disease Facts*. Retrieved September 20, 2021, from <https://rarediseases.org/wp-content/uploads/2019/02/nord-rareinsights-rd-facts-2019.pdf>.

Rare Diseases Act of 2002. 42 U.S.C. §281 (2002)280. Retrieved September 20, 2021, from [https://www.congress.gov/107/plaws/publ280/PLAW-107publ280.pdf?\\_cf\\_chl\\_jschl\\_tk\\_=pmd\\_GRnKbmqPXwmJBJvpS3AgdjPcmQOQS RnWPaBuiPwwk4-1632166412-0-gqNtZGzNAjujcnBszQil](https://www.congress.gov/107/plaws/publ280/PLAW-107publ280.pdf?_cf_chl_jschl_tk_=pmd_GRnKbmqPXwmJBJvpS3AgdjPcmQOQS RnWPaBuiPwwk4-1632166412-0-gqNtZGzNAjujcnBszQil).

**Appendix: Rare Disease Council Member Roster**

Ms. Ijeoma Azubuko, Individual Diagnosed with a Rare Disease

Dr. Maureen Dempsey, Representative from Health Plan Companies

Ms. Rebecca Goldbach, Caregiver of a Person with a Rare Disease

Dr. Stephen Green, Physician with Expertise in Rare Diseases

Dr. Samantha Hollins, Ex Officio, Designee of the Superintendent of Public Instruction

Mr. Gregory Josephs, Individual Diagnosed with a Rare Disease

Dr. Lisa Kaplowitz, Chair

Dr. Peter Kasson, Representative from an Academic Research Institution

Ms. Holly Kearl, Caregiver of a Person with a Rare Disease

Dr. Tiffany Kimbrough, Physician with Expertise in Rare Diseases

Ms. Susan Klees, Representative from the Biopharmaceutical Industry

Ms. Jennifer Macdonald, Ex Officio, Designee of the Commissioner of Health

Ms. Shannon McNeil, Representative of a Rare Disease Patient Organization

Ms. Jana Monaco, Vice-Chair

Dr. John Morgan, Ex Officio, Designee of the Director of DMAS

Dr. Richard Nicholas, Pharmacist

Ms. Megan O'Boyle, Representative of a Rare Disease Patient Organization

Ms. Sarah Paciulli, Registered Nurse or Advanced Practice Nurse

Dr. Stephen Rich, Member of the Scientific Community

Dr. Samantha Vergano, Geneticist