



COMMONWEALTH of VIRGINIA

Department of Health
P O BOX 2448

RICHMOND, VA 23218

Karen Shelton, MD
State Health Commissioner

TTY 7-1-1 OR
1-800-828-1120

December 27, 2024

MEMORANDUM

TO: Glenn Youngkin
Governor of Virginia

The Honorable L. Louise Lucas
President Pro Tempore, Senate of Virginia

The Honorable Don Scott
Speaker of the House, Virginia House of Delegates

FROM: Karen Shelton, MD
State Health Commissioner, Virginia Department of Health

SUBJECT: 2023 Report to the Governor and General Assembly Rare Disease Council

This report is submitted in compliance with the Code of Virginia § 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.

KS/AJ
Enclosure

Pc: The Honorable Janet V. Kelly, Secretary of Health and Human Resources

RARE DISEASE COUNCIL

REPORT TO THE GOVERNOR AND THE
GENERAL ASSEMBLY

2023



VIRGINIA DEPARTMENT OF HEALTH

PREFACE

The Rare Disease Council (Council) is tasked by the Code of Virginia (§§ 32.1-73.14 through 32.1-73.17) with advising the Governor and the General Assembly on the needs of individuals with rare diseases in the Commonwealth, identifying challenges that such individuals face, funding research related to rare diseases, and funding supports for persons with rare diseases. The Council is to provide a report to the Governor and the General Assembly each year by October 1 summarizing the activities and recommendations of the Council, and the status of funding available to the Council. The Virginia Department of Health (VDH) is tasked with providing staff support to the Council and assisting the Council with preparing this document, which serves as the Council's 2023 report.

RARE DISEASE COUNCIL

Governor Appointed Members of the Council as of June 1, 2023

Ms. Ijeoma Azubuko, individual diagnosed with a rare disease
Mr. Wes Fisher, individual diagnosed with a rare disease
Dr. Michael Friedlander, hospital administrator
Dr. Stephen Green, physician with expertise in rare diseases
Mr. Gregory Josephs, representative of a rare disease patient organization
Dr. Lisa Kaplowitz, chair
Dr. Peter Kasson, representative from an academic research institution
Dr. Tiffany Kimbrough, physician with expertise in rare diseases
Ms. Jana Monaco, vice-chair
Dr. Richard Nicholas, pharmacist
Ms. Megan O'Boyle, representative of a rare disease patient organization
Ms. Sarah Paciulli, registered nurse
Ms. Elissa Pierson, caregiver of a person with a rare disease
Dr. Stephen Rich, member of the scientific community
Ms. Elisabeth Scott, caregiver of a person with a rare disease
Dr. Samantha Vergano, geneticist
Ms. Susan Klees, representative from the biopharmaceutical industry

Ex Officio Members of the Council as of June 1, 2023

Dr. Samantha Hollins, Department of Education
Ms. Jennifer Macdonald, Virginia Department of Health
Dr. John Morgan, Department of Medical Assistance Services

Virginia Department of Health Staff Supporting the Council

Lauren Staley, Critical Congenital Heart Disease Program Coordinator and Rare Disease Council Coordinator, Office of Family Health Services
Katherine Crawford, Birth Defects Surveillance Program Coordinator, Office of Family Health Services
Christen Crews, Newborn Screening and Birth Defects Surveillance Programs Manager, Office of Family Health Services

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EXECUTIVE SUMMARY

The Rare Disease Council (Council) is tasked by the Code of Virginia (§§ 32.1-73.14 through 32.1-73.17) with advising the Governor and the General Assembly on the needs of individuals with rare diseases in the Commonwealth, identifying challenges that such individuals face, funding research related to rare diseases, and funding supports for persons with rare diseases. The Council is to provide a report to the Governor and the General Assembly each year by October 1 summarizing the activities and recommendations of the Council, and the status of the Rare Disease Council Fund (Fund). The Council held four quarterly public meetings and one public hearing during this year's reporting period (July 2022 through June 2023), in which the Virginia Department of Health (VDH) provided staff support. Findings from the Council are listed below.

FINDINGS

The Council does not have formal recommendations at this time, though it did note a series of findings based on presentations, discussions, breakout sessions, and comments from its meetings this year, as follows.

1. Access to care can often be a challenge for individuals and families affected by rare diseases. This can include access to primary care providers, specialists, care coordination, and insurance coverage. Access to care can be more of a challenge for those living in rural areas or areas without access to academic medical institutions. Telemedicine is one element that may provide increased opportunities for individuals with rare diseases to access specialists, especially for those with limited mobility or those living in rural areas or far away from academic medical institutions. The Council discussed potential benefits for individuals with rare diseases if Virginia were to join the Interstate Medical Licensure Compact.
2. Transitioning from pediatric care to adult care can also be a challenge for individuals and families affected by rare diseases. Individuals may lose services through Medicaid or the educational system when becoming adults and may also lose access to healthcare providers familiar with treating rare diseases. Care coordination services, especially as children age into adulthood, may assist individuals with rare diseases with a smoother transition from pediatric to adult medical care and support services. The Council discussed potential benefits of increasing access to care coordination services for individuals with rare diseases.
3. The Council is planning to conduct a survey to learn more about the needs of individuals with rare diseases in the Commonwealth and the challenges they face. The Council discussed the need to learn more about the scope and impact of rare diseases in the Commonwealth and determined that a survey of individuals affected by rare diseases would be an effective way to gather this information. The Council is currently in the planning stages of this survey, using a survey developed by the Minnesota Rare Disease Advisory Council as a reference (Bogart, K., Hemmesch, A., Barnes, E, et al., 2022). The survey will likely take some time and funding to develop, so the Council will continue to work on this effort in the coming year.

4. In order to design, conduct, and analyze the survey, the Council will likely need funding. The Council is seeking guidance on the Fund and will work with VDH staff over the coming year to develop governance for the Fund.

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INTRODUCTION

RARE DISEASE COUNCIL MANDATE

The Rare Disease Council (Council) is tasked by the Code of Virginia (§§ 32.1-73.14 through 32.1-73.17) with advising the Governor and the General Assembly on the needs of individuals with rare diseases in the Commonwealth, identifying challenges that such individuals face, funding research related to rare diseases, and funding supports for persons with rare diseases. The Council is also to conduct research and consult with experts to develop policy recommendations, publish a list of publicly accessible resources relating to rare diseases on the Council's webpage, submit a report annually by October 1 to the Governor and the General Assembly, meet quarterly, and manage the Rare Disease Council Fund (Fund). See Appendix A for the full text of the legislation.

RARE DISEASE COUNCIL ACTIVITIES

The Council completed its second year of service to the Commonwealth on June 30, 2023. During this year, the Council achieved three quorums and adopted bylaws, an electronic meetings policy, and mission and vision statements; published a list of existing publicly accessible resources on research, diagnosis, treatment, and education relating to rare diseases on the Council's webpage; received presentations, held discussions, and invited public comments to learn more about the needs and challenges faced by individuals with rare diseases in Virginia; worked to better understand funding requirements for the Council; and began discussions and development of a survey on rare diseases in the Commonwealth. With Virginia Department of Health (VDH) staff support, the Council met quarterly, as required, and provided public comment periods at each meeting. A summary of this year's meetings is as follows.

9/20/2022 MEETING

The 9/20/2022 Rare Disease Council meeting was held at the Virginia Hospital and Healthcare Association in Richmond. The Council did not obtain a quorum and therefore could not vote or make recommendations but rather discussed the Council's draft bylaws, electronic meetings policy, mission statement, vision statement, and challenges the Council faced to meeting an in-person quorum.

During this meeting, the Council received presentations from Dr. Joann Bodurtha from the New York Mid-Atlantic Caribbean (NYMAC) Regional Genetics Network, and Council member Dr. Tiffany Kimbrough. The Council learned how NYMAC focuses on improving access to quality genetics services for individuals with genetic conditions and their families, and it received information from pediatrician Dr. Kimbrough, who provides care to children with medical complexity, about gaps in care, challenges families face daily, and special considerations for pediatric versus adult complex care.

11/29/2022 MEETING

The 11/29/2022 Rare Disease Council meeting was held at the Virginia Hospital and Healthcare Association in Richmond and was followed by a public hearing. The Council achieved a quorum; approved prior meeting minutes; and voted to adopt bylaws, an electronic

meetings policy, a mission statement, and a vision statement. The Council also discussed Council funding and edits to the Council webpage, including a list of publicly accessible resources relating to rare diseases.

During this meeting, the Council received presentations from Nickie Brandenburger from the Partnership for People with Disabilities and Jackie Yetka, Sandy Hermann, and Anne Wienke from Care Connection for Children (CCC). The Council learned from the Partnership for People with Disabilities about the services CCC offers individuals and their families, as well as difficulties faced by parents of children with rare diseases. No one participated in the public hearing.

2/15/2023 MEETING

The 2/15/2023 Rare Disease Council meeting was an all-virtual meeting. The Council achieved an all-virtual quorum and held breakout sessions to discuss resources/education, data collection, and strategic planning. From these breakout sessions, the Council expressed an interest in developing and disseminating a survey and identifying an academic institution to assist with the process. The Council also received updates about the Council's webpage and the need to draft governance for the Council's Fund.

During this meeting, the Council received presentations from Ann Bevan from the Virginia Department of Medical Assistance Services (DMAS), Heather Norton from the Virginia Department of Behavioral Health and Developmental Services (DBHDS), and Annessa Reed from the National Organization for Rare Disorders (NORD). The Council received information from DMAS and DBHDS on Medicaid waivers offered to individuals and the process for obtaining a waiver, and it learned about Rare Disease Day 2023 from NORD.

5/16/2023 MEETING

The 5/16/2023 Rare Disease Council meeting was held at the Virginia Hospital and Healthcare Association in Richmond. The Council achieved a quorum and held breakout sessions about the survey discussed in the prior meeting: survey development, survey dissemination, and survey assistance. The Council also performed general Council business, including adopting an updated electronic meetings policy and approving prior meeting minutes.

During this meeting, the Council received presentations from Alan Gernhardt from the Virginia Freedom of Information Advisory Council, and Dr. Andrew Campbell from Children's National Hospital. The Council learned about Freedom of Information Act (FOIA) requirements for public meetings from Mr. Gernhardt as well as the importance of coordinated and comprehensive care for Sickle Cell Disease from Dr. Campbell, especially as children transition into adulthood.

REPORT OUTLINE

The remainder of this report includes a summary of topics related to the Council's work and findings from this year. Appendices follow, including the relevant section of the Code of Virginia for the Council, acronyms used in the report, and references cited in the report.

WHAT IS A RARE DISEASE?

According to the National Institutes of Health's (NIH) Genetic and Rare Diseases Information Center (GARD), a rare disease is defined as a disease or condition that impacts less than 200,000 people in the United States (NIH GARD, 2023). Currently, there are an estimated 10,000 known rare diseases (NIH GARD, 2023) and, of those diseases in which a molecular cause is known, only about 500 have approved treatments (NIH, 2023). Per the National Organization for Rare Disorders (NORD), it is estimated that there are nearly 25-30 million Americans living with a rare disease (NORD, 2023).

RARE DISEASE COUNCIL RECOMMENDATIONS, FUNDING, AND FINDINGS

RECOMMENDATIONS

The Council does not have formal recommendations at this time. The Council aims to have recommendations and to develop governance for the Fund by its next report.

FUNDING

The Council did not apply for nor receive any gifts, grants, or donations during its second year. The Council worked to better understand the scope of funding procedures and processes for requesting gifts and donations, applying for grants, and using received funds in alignment with the legislation. The Council and VDH staff will work to draft governance for the Fund in order to move forward with the Council's established goals and action plan of developing and disseminating a survey to learn more about rare diseases in the Commonwealth.

FINDINGS

The Council does not have formal recommendations at this time, though it did note a series of findings based on presentations, discussions, breakout sessions, and comments from its meetings this year, as follows.

1. Access to care can often be a challenge for individuals and families affected by rare diseases. This can include access to primary care providers, specialists, care coordination, and insurance coverage. Access to care can be more of a challenge for those living in rural areas or areas without access to academic medical institutions. Telemedicine is one element that may provide increased opportunities for individuals with rare diseases to access specialists, especially for those with limited mobility or those living in rural areas or far away from academic medical institutions. The Council discussed potential benefits for individuals with rare diseases if Virginia were to join the Interstate Medical Licensure Compact.
2. Transitioning from pediatric care to adult care can also be a challenge for individuals and families affected by rare diseases. Individuals may lose services through Medicaid or the educational system when becoming adults and may also lose access to healthcare

providers familiar with treating rare diseases. Care coordination services, especially as children age into adulthood, may assist individuals with rare diseases with a smoother transition from pediatric to adult medical care and support services. The Council discussed potential benefits of increasing access to care coordination services for individuals with rare diseases.

3. The Council is planning to conduct a survey to learn more about the needs of individuals with rare diseases in the Commonwealth and the challenges they face. The Council discussed the need to learn more about the scope and impact of rare diseases in the Commonwealth and determined that a survey of individuals affected by rare diseases would be an effective way to gather this information. The Council is currently in the planning stages of this survey, using a survey developed by the Minnesota Rare Disease Advisory Council as a reference (Bogart, K., Hemmesch, A., Barnes, E, et al., 2022). The survey will likely take some time and funding to develop, so the Council will continue to work on this effort in the coming year.
4. In order to design, conduct, and analyze the survey, the Council will likely need funding. The Council is seeking guidance on the Fund and will work with VDH staff over the coming year to develop governance for the Fund.

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APPENDIX A – CODE OF VIRGINIA §§32.1-73.14 THROUGH 32.1-73.17

ARTICLE 19. RARE DISEASE COUNCIL.

§ 32.1-73.14. Rare Disease Council; purpose.

There is hereby created in the executive branch of state government the Rare Disease Council (the Council) for the purpose of (i) advising the Governor and the General Assembly on the needs of individuals with rare diseases in the Commonwealth; (ii) identifying 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; (iii) funding research related to rare diseases and the development of new treatments for rare diseases; and (iv) funding for supports for persons with rare diseases in the Commonwealth.

2021, Sp. Sess. I, c. [303](#).

§ 32.1-73.15. Powers and duties of the Council.

The Council shall have the power and duty to:

1. Within the first year, hold public hearings and make inquiries of and solicit comments from the public to assist the Council in understanding the scope of rare diseases in the Commonwealth and the impact of rare diseases on individuals in the Commonwealth.
2. Conduct research and consult with experts to develop policy recommendations related to:
 - a. Improving access to health care and other services for individuals with rare diseases, including access to health insurance, specialists, health care services, and other necessary services for individuals with rare diseases;
 - b. The impact of health insurance coverage, cost sharing, tiers, or other utilization management procedures on access to health care and other necessary services; and
 - c. The impact of providing coverage under the state program for medical assistance services for approved health care services and medications for rare diseases.
3. Publish a list of existing publicly accessible resources on research, diagnosis, treatment, and education relating to rare diseases on the Council's webpage.
4. 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.
5. Apply for, accept, and expend gifts, grants, and donations from public or private sources to enable the Council to better carry out its objectives.

2021, Sp. Sess. I, c. [303](#).

§ 32.1-73.16. Membership; terms; quorum; meetings; staffing.

A. The Council shall have a total membership of 21 members that shall consist of 18 nonlegislative citizen members and three ex officio members. The Governor shall appoint a chairman and vice-chairman who shall be residents of the Commonwealth and shall not be employed by any federal or state government. Nonlegislative citizen members shall be appointed by the Governor and shall include, in addition to the chairman and the vice-chairman, one representative from an academic research institution in the Commonwealth that receives any grant funding for rare disease research; one geneticist licensed and currently practicing in the Commonwealth; one registered nurse or

advanced practice registered nurse licensed and currently practicing in the Commonwealth, with experience in treating rare diseases; two physicians with expertise in rare diseases who are licensed and currently practicing medicine in the Commonwealth; one hospital administrator, or his designee, from a hospital in the Commonwealth that provides care to persons diagnosed with rare diseases; two persons who are 18 years of age or older who have been diagnosed with a rare disease; two caregivers of persons with a rare disease; two representatives of rare disease patient organizations operating in the Commonwealth; one licensed pharmacist with experience with drugs used to treat rare diseases; one representative from the biopharmaceutical industry; one representative from health plan companies; and one member from the scientific community who is engaged in rare disease research, which may include a medical researcher with experience conducting research on rare diseases. The Commissioner of Health, the Director of the Department of Medical Assistance Services, and the Superintendent of Public Instruction, or their designees, shall serve ex officio with nonvoting privileges. Ex officio members of the Council shall serve terms coincident with their terms of office.

Nonlegislative citizen members of the Council shall be citizens of the Commonwealth. Appointments to fill vacancies, other than by expiration of a term, shall be for the unexpired terms. Vacancies shall be filled in the same manner as the original appointments. After the initial staggering of terms, nonlegislative citizen members shall be appointed for a term of three years.

Ex officio members of the Council shall serve terms coincident with their terms of office. Appointments to fill vacancies, other than by expiration of a term, shall be for the unexpired terms. Vacancies shall be filled in the same manner as the original appointments. After the initial staggering of terms, nonlegislative citizen members shall be appointed for a term of four years.

B. The Council shall meet quarterly and the chairman and vice-chairman shall establish a meeting schedule on an annual basis. A majority of the members shall constitute a quorum.

C. Members of the Council shall serve without compensation or reimbursement.

D. The Department of Health shall provide staff support to the Council. All agencies of the Commonwealth shall provide assistance to the Council, upon request.

2021, Sp. Sess. I, c. [303](#).

§ 32.1-73.17. Rare Disease Council Fund.

There is hereby created in the state treasury a special nonreverting fund to be known as the Rare Disease Council Fund, referred to in this section as "the Fund." The Fund shall be established on the books of the Comptroller. All funds appropriated for such purpose and any gifts, grants, donations, and other funds received on its behalf shall be paid into the state treasury and credited to the Fund. Interest earned on moneys in the Fund shall remain in the Fund and be credited to it. Any moneys remaining in the Fund, including interest thereon, at the end of each fiscal year shall not revert to the general fund but shall remain in the Fund. Moneys in the Fund shall be used solely for the purpose of (i) funding research related to rare diseases and the development of new treatments for rare diseases and supports for persons with rare diseases in the Commonwealth and (ii) supporting the work of the Council. Expenditures and disbursements from the Fund shall be made by the State Treasurer on warrants issued by the Comptroller upon written request signed by the Commissioner of Health.

2021, Sp. Sess. I, c. [303](#).

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APPENDIX B – ACRONYMS AND ABBREVIATIONS

Following is a list of the acronyms and abbreviations appearing throughout the report and its appendices.

CCC – Care Connection for Children

Council – Rare Disease Council

DBHDS – Department of Behavioral Health and Developmental Services

DMAS – Department of Medical Assistance Services

FOIA – Freedom of Information Act

Fund – Rare Disease Council Fund

GARD – Genetic and Rare Diseases Information Center

NIH – National Institutes of Health

NORD – National Organization for Rare Disorders

NYMAC – New York Mid-Atlantic Caribbean Regional Genetics Network

VDH – Virginia Department of Health

APPENDIX C – REFERENCES

Bogart, K., Hemmesch, A., Barnes, E., Blissenbach, T., Beisang, A., Engel, P., & Chloe Barnes Advisory Council on Rare Diseases (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. <https://doi.org/10.1186/s13023-022-02343-4>.

National Institutes of Health. Rare Diseases. Retrieved February 24, 2023, from <https://www.nih.gov/about-nih/what-we-do/nih-turning-discovery-into-health/rare-diseases>

National Institutes of Health - Genetic and Rare Diseases Information Center. FAQ What is a rare disease? Retrieved February 24, 2023, from <https://rarediseases.info.nih.gov/about>

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