COMMONWEALTH of VIRGINIA
Department of Health
Karen Shelton, MD
State Health Commissioner
P O BOX 2448
RICHMOND, VA 23218
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January 7, 2026

MEMORANDUM

TO: The Honorable 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: 2025 Rare Disease Council Report

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/KB
Enclosure

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

RARE DISEASE COUNCIL

REPORT TO THE GOVERNOR AND THE
GENERAL ASSEMBLY

2025

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 assisted the Council with preparing this document, which serves as the Council's 2025 report.

RARE DISEASE COUNCIL MEMBERS

Current Members of the Council as of June 1, 2025 (Gubernatorial Appointees)
Ms. Gwen Traficant, Chair
Ms. Hermionne Johnson, Vice Chair
Ms. Ijeoma Azubuko, individual diagnosed with a rare disease
Mr. John Feore, representative from the biopharmaceutical industry
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. Peter Kasson, representative from an academic research institution
Dr. Tiffany Kimbrough, physician with expertise in rare diseases
Dr. Sharon Kopis, registered nurse or advanced practice registered nurse
Ms. Leslie Mehta, representative of rare disease patient organization
Dr. John Michos, representative from health plan companies
Dr. Angela Olmsted, licensed pharmacist
Dr. Christina Peroutka, geneticist licensed and currently practicing
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

Ex Officio Members of the Council as of June 1, 2025
Mr. Marcus Allen, Department of Health
Dr. Rachel Cain, Department of Medical Assistance Services
Dr. Samantha Hollins, Department of Education

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

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LETTER FROM THE CHAIR

Dear Governor Youngkin, Senator Lucas, and Speaker Scott –

On behalf of the Virginia Rare Disease Council, I am honored to present the annual report for your review. Rare diseases, sometimes referred to as “orphan diseases” are categorized as a disease or condition that impacts less than 200,000 people in the United States. Individually they are rare, but have broad implications in delayed diagnoses, limited funding for research, and treatments.

As members of this Council, we have personal connections to the rare disease community that fuel our passion to advocate on behalf of the people we serve in this great Commonwealth. We are dedicated to understanding the specific rare disease needs in Virginia in an effort to bring some relief to those who are suffering and the families that lovingly care for them.

We established a strategic plan for 2025 goals and initiatives:

- Identify and quantify the unique challenges and needs for rare disease patients and family caregivers across state health regions.
- Recommend legislative actions for patients and family caregivers.
- Increase awareness of our Rare Disease Council and resources available.
- Deliver impactful 2025 Council plan results.

This year we completed a survey for dissemination, established a funding document, and adopted a policy to initiate Council social media platforms. We will have a booth at the 2025 Virginia State Fair as another opportunity to create Council awareness, meet the people we serve, and access needs. We established partner groups within our Council to utilize the talent and network of our members to advance our goals and initiatives.

The Council has two recommendations for your consideration. We recommend Virginia’s participation in the Interstate Medical Licensure Compact (IMLC) to enable rare disease patients to have specialty care outside of limited health region resources. This is a vital aspect in providing telehealth access for rare disease Virginians. Most notable is the impact this would have to our rural communities.

The Council also recommends that the General Assembly appropriate $20,000 in General Funds to conduct a large-scale dissemination of the Rare Disease Survey and subsequent analyzing of the qualitative and quantitative data. This will enable our Council to be informed advocates for policies that fit our unique statewide needs.

It is my great honor to serve as Chair, and I am grateful for the opportunity. As a Council, we join with you in your commitment to make Virginia the best place to live, work, and raise a family.

Respectfully,
Gwen Traficant
Chair, Rare Disease Council

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 from July 2024 through June 2025, for which the Virginia Department of Health (VDH) provided staff support. Findings and recommendations from the Council are listed below. An update on the Rare Disease Survey and Council funding is provided prior to the findings and recommendations sections at the end of this report.

FINDINGS

Based on public comments, presentations, and discussions from the Council's quarterly meetings, the Council identified several findings in 2025 related to the needs of individuals with rare diseases, challenges such individuals face, and the role of the Rare Disease Council in supporting individuals with rare diseases. A summary of the Council's findings are as follows; additional details on these findings are included later in this report:

1. Individuals with a rare disease(s) who are in need of additional resources or assistance with managing their disease(s) may benefit from contacting their medical insurance company and inquiring about speaking to a case or disease manager.

2. Adjusting cutoffs for homocystinuria (HCU) on the newborn dried bloodspot test could aid in fewer missed diagnoses and better outcomes.

3. The Council gained anecdotal insight into the needs and challenges of those who are caregivers for individuals with rare diseases. Caregiver needs include: the need for compensation as a caregiver, access to respite care, support with housekeeping and food preparation, and access to and coordination between specialists. Challenges include: financial strains due to caregiving demands and limitations for securing employment, and the inability of daycares/schools to handle medically complex children and their equipment.

4. The Council determined that while there are multiple research programs on rare diseases across universities in Virginia, a major challenge is that research programs and grant funding are not categorized under "rare disease" for research or funding opportunities.

5. The Council's presence in the rare disease community has grown over the past year, but additional work needs to be done to raise awareness of the Council and its work among the public and state legislators.

6. Medical insurance benefits are dictated by federal and state law or employer groups, and most individuals do not know their benefits. For those that receive initial denials from

their insurance, 70% are due to insufficient information, but individuals are not aware of the multiple processes they can take to appeal denials. Understanding insurance coverage and how to navigate it is vital, especially for the rare disease community.

7. Activated PI3K Delta Syndrome (APDS), a rare primary immunodeficiency that is generally inherited and was first discovered in 2013, is only diagnosable through genetic testing. APDS symptoms can include frequent and severe infections of the ears, sinuses, and upper and lower respiratory tracts, as well as gastrointestinal tract issues, nodules in the airway, enlarged tonsils, developmental delay, among others (IDF, 2025).

8. The National Organization for Rare Disorders (NORD) publishes an annual state report card, which focuses on nine distinct issue areas. From 2024 to 2025, the Commonwealth's state report card remained unchanged in all nine issue areas. The Commonwealth received a C or fail in medical nutrition, protecting patients in state regulated insurance, and telehealth. The reasoning cited was "Virgina's mandate for medical nutrition having limitations, Virginia not taking enough action to mitigate the expansion of short-term, limited-duration health plans (STLDIs), and Virginia's lack of participation in the Interstate Medical Licensure Compact (IMLC), which provides access to telehealth services across states."

RECOMMENDATIONS

The Council's recommendations remain the same as last year. These recommendations are based on public comments, presentations, and discussions from its quarterly meetings this year, as follows:

1. The Council recommends that the Governor and the General Assembly reconsider Virginia's participation in the IMLC. Participation in the IMLC would require the General Assembly to pass legislation to amend the Code of Virginia to authorize the state to join (IMLC, N.D.). The language of the compact must be consistent in each state that joins. Previous legislation to join the compact has been unsuccessful. However, Virginia is now one of only 10 states that have not joined the IMLC; to date, 40 states, Guam, and the District of Columbia have joined the IMLC (NORD, N.D.). Virginia's participation would not only benefit Virginians but would also result in a "pass" on the NORD state report card for telehealth. VDH will consider working with the Department of Health Professions (DHP) to discuss legislative action on this topic, as joining the IMLC would require a change to DHP's section of the Code.

2. The Council recommends that the General Assembly appropriate $20,000 in General Funds to support the work of the Council. Specifically, the funding would be used to conduct a large-scale dissemination and analysis of the Rare Disease Survey to better understand the prevalence of rare disease in the Commonwealth and the needs of rare disease patients in Virginia A large-scale survey dissemination may consist of mailers, flyers, paper surveys, events hosted by the Council, radio ads, and/or newspaper ads and is estimated to cost $10,000-$19,000. Analysis of the survey data by a VDH epidemiologist upon closure is estimated to cost $2,100.

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 fourth year of service to the Commonwealth on June 30, 2025. During this year, the Council met four times, offered public comment periods at each meeting, and undertook the following major activities:

- Solicited presentations, held discussions, and invited public comments to learn more about the needs and challenges faced by individuals with rare diseases in Virginia.
- Established the Fund in the state treasury and on the books of the Comptroller.
- Improved its governance structure by updating the Council's Bylaws, establishing Funding Governance for the Fund, and establishing a Social Media Policy that allows the Chair and Vice Chair to administer Council social media pages in an effort to increase awareness and knowledge of the Council.
- Submitted an Institutional Review Board (IRB) application for the Rare Disease Survey and received approval.
- Launched the Rare Disease Survey that will gather more information on rare diseases in the Commonwealth. The survey link has been added to the Council webpage and the Council is working with the National Organization for Rare Disorders (NORD) to disseminate the survey link to NORD's 600+ advocacy partners in the Commonwealth in July 2025. Information to be gathered includes rare disease patient demographics; type and number of rare disease(s); knowledge and support of the rare disease(s); diagnosis and follow-up related to the rare disease(s); insurance and out-of-pocket costs; access to specialist centers; utilization of telehealth; and caregiving for rare disease patients.
- Identified goals for the Council and partner groups to help advance each goal, including: funding, telehealth, caregiver support, research advancement, community marketing, grant applications, standards of care, health equity, insurance, Virginia support, and social media.
- Created educational Council handouts, linked handouts to the Council webpage, and distributed handouts to 50 local health departments (LHDs).
- Secured a booth at the Fall 2025 Virginia State Fair that will serve as a fundraising and community marketing opportunity.

- Launched two Council social media pages.

A summary of this year's meetings is as follows:

8/15/2024 MEETING

The meeting was held in-person at the Virginia Hospital and Healthcare Association (VHHA). The Council achieved a quorum, conducted introductions, and voted to approve prior meeting minutes. During this meeting, the Council received presentations from Gwen Traficant (Chair, Rare Disease Council) and Alan Gernhardt (Virginia Freedom of Information Advisory Council). The Chair presented a strategic plan for 2025, and the Council provided feedback. Mr. Gernhardt presented the new Freedom of Information Act (FOIA) guidelines that went into effect July 1, 2024, and pertained to Council operations. Per the new FOIA guidelines , the Council discussed their Electronic Meetings Policy (EMP) and voted to approve updates. The Chair also gave a short introduction presentation, which highlighted her experience with rare disease and motivation for joining the Council. Prior to adjournment, the Council received updates from VDH regarding the IRB process for the Rare Disease Survey and the status of guidance from the Department of Treasury on the Rare Disease Council Fund. No individuals participated in public comment.

11/19/2024 MEETING

The meeting was held virtually. The Council achieved a quorum, conducted introductions, and voted to approve prior meeting minutes. During this meeting, the Council received presentations from Ms. Traficant, VDH staff, and Liz Carter (HCU Network America). The Chair provided updates since the last meeting. The Chair presented a plan to implement partner groups, which focus on key Council goals including funding, telehealth, caregiver support, research advancement, community marketing, grant applications, standards of care, health equity, insurance, Virginia support, and social media. The Council engaged in a group discussion regarding partner group pairings, goals, and expectations moving forward. VDH and the Council discussed the draft Funding Governance document, with specific focus on developing a vetting process for accepting funds. VDH worked with the Council to iterate on the governance document and add in a vetting process. The Council voted to approve the Funding Governance. Ms. Carter discussed her personal experience with homocystinuria (HCU) and presented recommended changes to the HCU newborn screening test. Prior to adjournment, the Chair and Vice Chair provided the Council with key takeaways from the NORD Breakthrough Summit Conference that they attended in October. One individual participated in public comment.

2/25/2025 MEETING

The meeting was held in-person at VHHA. The Council achieved a quorum, conducted introductions, and voted to approve prior meeting minutes. During this meeting, the Council received General Assembly updates from VDH staff and presentations from Ms. Traficant (and Council members. The Chair provided updates since the last meeting and presented a social media proposal. The Council discussed the proposal and agreed to vote on a Social Media Policy at the May meeting. Partner groups of Council members presented on their focus areas, which was followed by a full group discussion to reflect on the information presented. Prior to adjournment, the Chair presented the Rare Disease Day Proclamation. No individuals participated in public comment.

5/20/2025 MEETING

The meeting was held virtually. The Council achieved a quorum, conducted introductions, and voted to approve prior meeting minutes, updated Bylaws and a new Social Media Policy, which will allow the Chair and Vice Chair to administer Council social media pages to increase awareness and knowledge of the Council. The Council discussed updates to their EMP but agreed to vote on the updates at the August 2025 meeting instead. During this meeting, the Council received presentations from Kathy Tuttle (Pharming), Carolyn Sheridan (NORD), Ms. Traficant , and two Council members related to grant applications. Ms. Tuttle presented on Activated PI3K Delta Syndrome (APDS) and discussed increasing awareness of APDS genetic testing. Ms. Sheridan presented the NORD Virginia State Report Card and brainstormed ways the Council could assist Virginia in improving some of their grades. The Chair provided updates from the last meeting, including difficulties scheduling statewide visits at this time. The Chair also presented a state fair proposal that focused on the Council securing a booth at the Fall 2025 Virginia State Fair, which will serve as a fundraising and community marketing opportunity. Two Council members presented their research and findings on grant applications. The Council engaged in a group discussion. Prior to adjournment, the Council received updates from VDH regarding the Rare Disease Survey IRB approval, addition of the survey link on the Council webpage, and survey dissemination plans with NORD. No individuals participated in public comment.

REPORT OUTLINE

The remainder of this report includes a summary of topics related to the Council's work, an update on funding, findings, and recommendations from this year. Appendices follow, including the relevant section of the Code of Virginia for the Council, acronyms used in the report, references cited in the report, Rare Disease Survey IRB approval letter, and the Rare Disease Council 2025 Strategic Plan.

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BACKGROUND: 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, 2025). Currently, there are an estimated 10,000 known rare diseases (NIH GARD, 2025) but, despite growing scientific knowledge about the causes and progression of rare diseases, fewer than 5% have approved treatments (NIH GARD, 2025). 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, 2025). Currently, there is not sufficient data to understand the prevalence of rare diseases in Virginia. This data gap is what the Council hopes to address through the dissemination of the Rare Disease Survey discussed in the next section of this report.

GATHERING INFORMATION ON RARE DISEASES IN THE COMMONWEALTH: UPDATE ON THE RARE DISEASE SURVEY

IMPETUS

Pursuant to § 32.1-73.14 of the Code of Virginia, the Rare Disease Council is tasked with advising the Governor and the General Assembly on the needs of individuals with rare diseases in the Commonwealth and identifying challenges that such individuals face. In order to better understand the prevalence of rare diseases in the Commonwealth and needs of individuals with rare diseases and their caregivers, the Council developed the Rare Disease Survey to gather this information. The Council undertook significant work in 2025 to develop and launch the Rare Disease Survey. The survey was launched in July 2025, outside of this reporting period. The survey launch and results will be discussed in the 2026 report.

RARE DISEASE SURVEY

During its fourth year, the Council submitted a VDH IRB application for the Rare Disease Survey, which will be completed in two stages due to current financial limitations. While IRB approval was only sought and approved for stage one at this time, information on stage two is also outlined below.

Stage one will consist of a small-scale, pilot, survey link dissemination using REDCap, a secure web application for building and managing online surveys and databases (REDCap, 2025) and will be distributed with the assistance of internal and external stakeholders identified by the Council. Stakeholders may consist of NORD, rare disease foundations and nonprofits, universities, pediatricians, physicians and specialists, healthcare systems, and Care Connection for Children (CCC) centers. Those willing to assist in the dissemination of the Rare Disease Survey will be provided with the survey link and will be asked to distribute the link to those within the rare disease community. The survey link used in stage one and will be open for responses for a duration of two years, during which time the Council will work to obtain funding to implement stage two. Upon closure of the survey in stage one, the Council and VDH staff will analyze the data and report their findings to the Governor and General Assembly, per the Code of Virginia (§§ 32.1-73.14 through 32.1-73.17).

Stage two will consist of a large-scale dissemination of the survey, using funds obtained by the Council. In addition to the REDCap survey link and the utilization of internal and external stakeholders to assist in survey link dissemination, stage two will also consist of a full-scale marketing of the survey. Full-scale marketing methods may consist of mailers, flyers, paper surveys, events hosted by the Council, social media posts, radio ads, and/or newspaper ads. A full-scale marketing of the survey would aim to reach those rare disease community individuals and caregivers who may not have access to the internet or that were missed during stage one, in order to provide more comprehensive data related to the rare disease community. Upon closure of stage two, the Council and VDH staff will evaluate the data and report their updated findings to the Governor and General Assembly, per the Code of Virginia (§§ 32.1-73.14 through 32.1-73.17).

UPDATE ON FUNDING

In 2025, with assistance from the Department of Accounts and the Department of Treasury, the Council successfully established the Rare Disease Council Fund in the state treasury and on the books of the Comptroller this year, per the Code of Virginia (§ 32.1-73.17). In addition to establishing the Fund, the Council also adopted Funding Governance for the Fund. This represents significant progress from last fiscal year. However, the Council did not apply for or receive any gifts, grants, or donations in the Fund in this reporting period. The Fund was established at the end of this reporting period, as it took time to follow the correct process for administratively establishing the Fund. Following guidance from the Department of Accounts, VDH is currently working to seek approval from the Governor to apply for and/or accept grants and donations into the Fund in the next year, as is required by Chapter 725 of the 2025 Acts of Assembly (Budget Bill), § 4-2.01.

Funds obtained after receiving approval from the Governor will go towards funding stage two of the Rare Disease Survey, in order to carry out the Council's purpose and duties, per the Code of Virginia (§§ 32.1-73.14 through 32.1-73.17).

RARE DISEASE COUNCIL FINDINGS AND RECOMMENDATIONS

FINDINGS

Based on public comments, presentations, and discussions from the Council's quarterly meetings, the Council identified several findings in 2025 related to the needs of individuals with rare diseases, challenges such individuals face, and the role of the Rare Disease Council in supporting individuals with rare diseases. The Council's findings are as follows:

1. Individuals with a rare disease(s) who are in need of additional resources or assistance with managing their disease(s) may benefit from contacting their medical insurance company and inquiring about speaking to a case or disease manager.

2. The Council heard from HCU Network America that adjusting cutoffs for homocystinuria (HCU) on the newborn dried bloodspot test would aid in fewer missed diagnoses and better outcomes. HCU is a serious but treatable disorder, and a delay in

diagnosis causes different outcomes for patients, including missed milestones and significant cognitive issues.

3. The Council is working to acquire data on the value of caregiving and the roadblocks/ challenges that caregivers of individuals with rare diseases face. The purpose of this research is to help identify actionable solutions for caregivers of individuals with rare diseases. The Council gained anecdotal insight into caregiver needs by posting polls on four different support groups on social media asking the question "if you had one caregiver wish, what would it be?" Responses to the poll included: to be paid or compensated as a caregiver, respite care, housekeeping and food preparation, and access to and coordination between specialists. Comments and discussion from participants also spoke to the common need for assistance with routine housekeeping, financial strains due to caregiving demands and the limitations it created for securing employment, and the inability of daycares/schools to handle medically complex children and their equipment.

4. The Council is assessing the current level of research being conducted on rare diseases in Virginia including: sources and amounts of funding across the state, particular programs of excellence, and gaps and needs for additional research. During their assessment, the Council determined that, while there are multiple research programs on rare diseases across universities in Virginia, a major challenge is that research programs and grant funding are not categorized under "rare disease" for research or funding opportunities.

5. The Council's presence in the rare disease community has grown over the past year, but additional work needs to be done to raise awareness of the Council and its work. While statewide visits discussed last year have been temporarily paused due to scheduling constraints, the Council has completed the following to help bring awareness: Chair and Vice Chair attended the National Organization for Rare Disorders (NORD) Breakthrough Summit and networked with other Rare Disease Councils; developed and distributed educational handouts to local health districts that include Council and rare disease facts; uploaded educational handouts to the Council webpage for community use; implemented a Social Media Policy that allows the Chair and Vice Chair to administer social media pages for the Council; launched the Rare Disease Survey; and secured a booth at the Fall 2025 Virginia State Fair that will serve as a community marking and fundraising event. The Council also discussed the need to connect with state legislators this upcoming summer/fall to bring awareness to the work of the Council, though a formal action plan has not been established.

6. Medical insurance benefits are dictated by federal and state law or employer groups, and most individuals do not know their benefits. For those that receive initial denials from their insurance, 70% are due to insufficient information, but individuals are not aware of the multiple processes they can take to appeal denials. Understanding insurance coverage and how to navigate it is vital, especially for the rare disease community.

7. The Council heard from Pharming that Activated PI3K Delta Syndrome (APDS), a rare primary immunodeficiency that is generally inherited and was first discovered in 2013, is only diagnosable through genetic testing. APDS symptoms can include frequent and severe infections of the ears, sinuses, and upper and lower respiratory tracts, as well as

gastrointestinal tract issues, nodules in the airway, enlarged tonsils, developmental delay, among others (IDF, 2025).

8. NORD publishes an annual state report card, which focuses on nine distinct issue areas. From 2024 to 2025, the Commonwealth's state report card remained unchanged in all nine issue areas. The Commonwealth received a C or fail in medical nutrition, protecting patients in state regulated insurance, and telehealth. The reasoning cited was" Virgina's mandate for medical nutrition having limitations, Virginia not taking enough action to mitigate the expansion of short-term, limited-duration health plans (STLDIs), and Virginia's lack of participation in the Interstate Medical Licensure Compact (IMLC), which provides access to telehealth services across states."

RECOMMENDATIONS

The Council's recommendations remain the same as last year. These recommendations are based on public comments, presentations, and discussions from its quarterly meetings this year, as follows:

1. The Council recommends that the Governor and the General Assembly reconsider Virginia's participation in the IMLC. Participation in the IMLC would require the General Assembly to pass legislation to amend the Code of Virginia to authorize the state to join (IMLC, N.D.). The language of the compact must be consistent in each state that joins. Previous legislation to join the compact has been unsuccessful. However, Virginia is now one of only 10 states that have not joined the IMLC: to date, 40 states, Guam, and the District of Columbia have joined the IMLC (NORD, N.D.). Virginia's participation would not only benefit Virginians but would also result in a "pass" on the NORD state report card for telehealth. VDH will consider working with the Department of Health Professions (DHP) to discuss legislative action on this topic, as joining the IMLC would require a change to DHP's section of the Code.

2. The Council recommends that the General Assembly appropriate $20,000 in General Funds to support the work of the Council. Specifically, the funding would be used to conduct a large-scale dissemination of the Rare Disease Survey to better understand the prevalence of rare disease in the Commonwealth and the needs of rare disease patients in Virginia.

APPENDIX A – CODE OF VIRGINIA § § 32.1-73.14 TO 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 purposes 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

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

APDS – Activated PI3K Delta Syndrome

CCC – Care Connection for Children

DHP – Department of Health Professions

EMP – Electronic Meetings Policy

FOIA – Freedom of Information Act

GARD – Genetic and Rare Diseases Information Center

HCU – Homocystinuria

IMLC – Interstate Medical Licensure Compact

IRB – Institutional Review Board

NIH – National Institutes of Health

NORD – National Organization for Rare Disorders

STLDIs – Short-Term, Limited-Duration Health Plans

VDH – Virginia Department of Health

VHHA – Virginia Hospital and Healthcare Association

APPENDIX C – REFERENCES

Immune Deficiency Foundation. Activated PI3K delta syndrome (APDS). Retrieved June 17, 2025, from https://primaryimmune.org/understanding-primary-immunodeficiency/types-of-pi/activated-pi3k-delta-syndrome-apds.

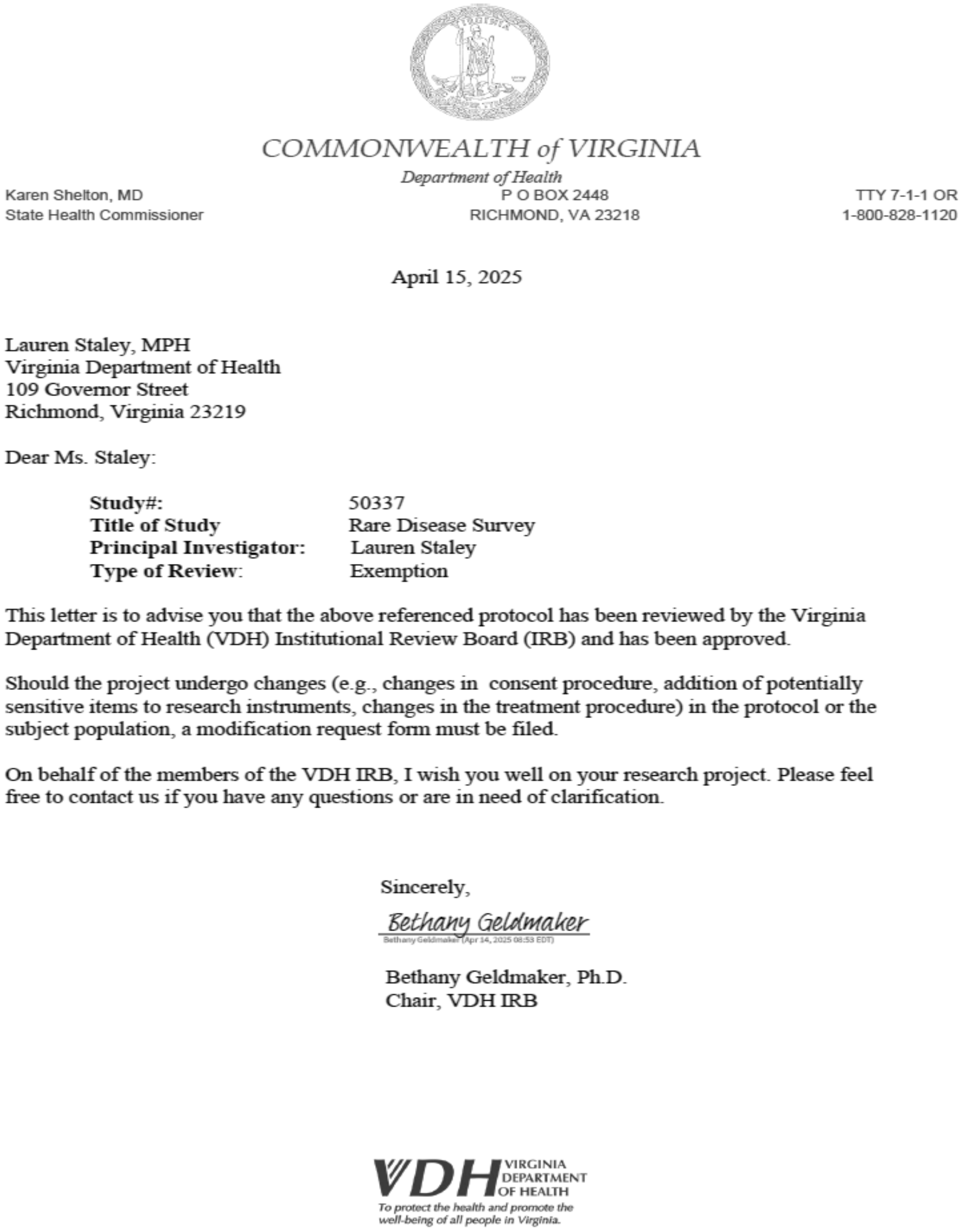
National Institutes of Health - Genetic and Rare Diseases Information Center. FAQ What is a rare disease? Retrieved June 25, 2025, from https://rarediseases.info.nih.gov/about

National Organization for Rare Disorders. Rare Disease Facts & Statistics. Retrieved June 25, 2025, from https://rarediseases.org/understanding-rare-disease/rare-disease-facts-and-statistics/

National Organization for Rare Disorders. Telehealth. Retrieved June 11, 2025, from https://rarediseases.org/policy-issues/telehealth/.

REDCap. Research Electronic Data Capture. Retrieved June 25, 2025, from https://www.project-redcap.org/

APPENDIX D – RARE DISEASE SURVEY IRB APPROVAL LETTER



COMMONWEALTH *of* VIRGINIA
Department of Health
Karen Shelton, MD
State Health Commissioner
P O BOX 2448
RICHMOND, VA 23218
TTY 7-1-1 OR
1-800-828-1120

April 15, 2025

Lauren Staley, MPH
Virginia Department of Health
109 Governor Street
Richmond, Virginia 23219

Dear Ms. Staley:

Study#:	50337
Title of Study	Rare Disease Survey
Principal Investigator:	Lauren Staley
Type of Review:	Exemption

This letter is to advise you that the above referenced protocol has been reviewed by the Virginia Department of Health (VDH) Institutional Review Board (IRB) and has been approved.

Should the project undergo changes (e.g., changes in consent procedure, addition of potentially sensitive items to research instruments, changes in the treatment procedure) in the protocol or the subject population, a modification request form must be filed.

On behalf of the members of the VDH IRB, I wish you well on your research project. Please feel free to contact us if you have any questions or are in need of clarification.

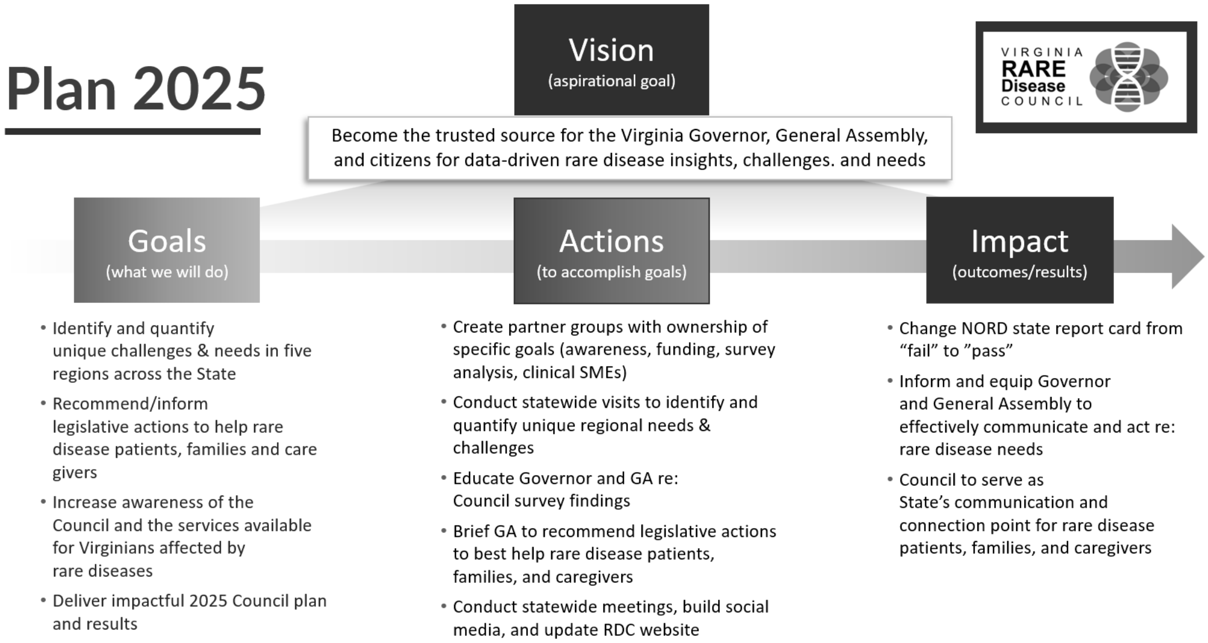
Sincerely,

Bethany Geldmaker
Bethany Geldmaker (Apr 14, 2025 08:53 EDT)

Bethany Geldmaker, Ph.D.
Chair, VDH IRB

VDH VIRGINIA DEPARTMENT OF HEALTH
To protect the health and promote the well-being of all people in Virginia.

APPENDIX E – RARE DISEASE COUNCIL 2025 STRATEGIC PLAN



Plan 2025

VIRGINIA
RARE
Disease
COUNCIL

Vision
(aspirational goal)

Become the trusted source for the Virginia Governor, General Assembly, and citizens for data-driven rare disease insights, challenges. and needs

Goals
(what we will do)

- Identify and quantify unique challenges & needs in five regions across the State
- Recommend/inform legislative actions to help rare disease patients, families and care givers
- Increase awareness of the Council and the services available for Virginians affected by rare diseases
- Deliver impactful 2025 Council plan and results

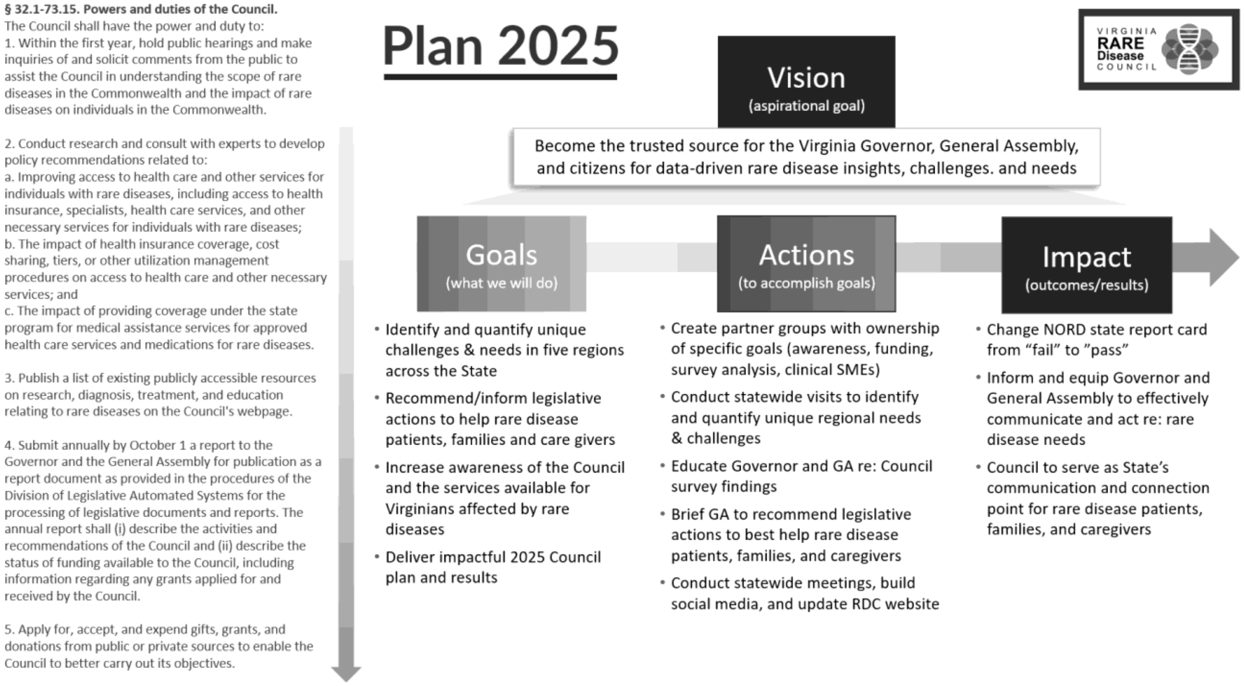
Actions
(to accomplish goals)

- Create partner groups with ownership of specific goals (awareness, funding, survey analysis, clinical SMEs)
- Conduct statewide visits to identify and quantify unique regional needs & challenges
- Educate Governor and GA re: Council survey findings
- Brief GA to recommend legislative actions to best help rare disease patients, families, and caregivers
- Conduct statewide meetings, build social media, and update RDC website

Impact
(outcomes/results)

- Change NORD state report card from “fail” to ”pass”
- Inform and equip Governor and General Assembly to effectively communicate and act re: rare disease needs
- Council to serve as State’s communication and connection point for rare disease patients, families, and caregivers

2025 Strategic Plan developed by Council Chair, Gwen Traficant



§ 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.

Plan 2025

VIRGINIA
RARE
Disease
COUNCIL

Vision
(aspirational goal)

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2025 Strategic Plan developed by Council Chair, Gwen Traficant, with § 32.1-73.15 Powers and Duties of the Council on left