Annual Report

January 1, 2021, to January 1, 2022

WEST VIRGINIA
ADVISORY COUNCIL ON RARE DISEASES

STATE OF WEST VIRGINIA
DEPARTMENT OF HEALTH AND HUMAN RESOURCES
West Virginia Advisory Council on Rare Diseases

The Honorable Jim Justice
Governor, State of West Virginia
1900 Kanawha Boulevard, East
Charleston, West Virginia 25305

Members of the West Virginia Legislature
1900 Kanawha Boulevard, East
Charleston, West Virginia 25305
Dear Governor Justice and Members of the Legislature:

It is my pleasure to submit this report on behalf of the West Virginia Rare Disease Advisory Council, as required by W. Va. Code §16-5CC-3. This report highlights 12 months of collaboration and work, laying forth the Council's priority targets for the years ahead, from now through 2023. It also demonstrates creative approaches to achieving the council objectives through a unique strategic plan.

According to the National Institutes of Health, one out of every ten Americans will be diagnosed with a rare disease at some point in their life [1], putting 179,200 West Virginians at risk [2]. In addition to extensive diagnostic journeys and misdiagnoses, West Virginians with rare diseases face additional challenges. Several critical medical strategies for rare diseases have been implemented. In 2020, the Council was established and, in a relatively short timeframe, has become a unified voice for the estimated 179,200 West Virginians affected by rare diseases who are often not heard or understood [3]. The uncertainty of the actual number or the incidence of rare disease in West Virginia is a key gap being addressed by the Council's work.

The members of the council represent health care providers, patients or caregivers, researchers, and advocates. We are a dedicated and committed group who are fully aware of rare disease challenges. We are inspirational, and our goals are for a better quality of life for their family members, patients, and to the community. The Council members have all agreed on the goals in this document, with the specific intent of improving the quality of life for those in West Virginia affected by rare diseases. The goals outlined here will not eliminate all uncertainty; however, they represent meaningful actions with the potential to improve the day-to-day issues that rare disease individuals face.

The Council recognizes that one of the most significant barriers to its success is a lack of available resources. To the greatest extent possible, we will strive to be innovative, form partnerships, and leverage resources. You can consult with the members as subject matter experts. We, the council members, look forward to continuing to work on this mission and with each of you.

Sincerely,

Dr. Nadia Falah, Member

West Virginia Advisory Council on Rare Diseases Members, 2021-2023

Fernando Andrzejevski, Morgan County
Council President
Representative from a patient-based organization or advocacy group for rare disease with preference given to organizations based in West Virginia

Christy Glass, APRN, Monongalia County
Council Vice President
Registered nurse or advanced practice registered nurse licensed and practicing in the state with experience treating rare diseases

Dr. Ayne Amjad
Commissioner and State Health Officer
West Virginia Department of Health and Human Resources, Bureau for Public Health
Designee for the Cabinet Secretary of the West Virginia Department of Health and Human Resources

Raymond K. Brooks, Jr., Berkeley County
Representative from a patient-based organization or advocacy group for rare disease with preference given to organizations based in West Virginia

Amy Brown, Kanawha County
Person over the age of 18 who either has a rare disease or is a family member of a person with a rare disease

Erik L. Carlton, RPh, Monongalia County
Person with an advanced degree in public health or another health-related field

Jodi Chadwell, Berkeley County
Person over the age of 18 who either has a rare disease or is a family member of a person with a rare disease

Juliana Frederick Curry, Kanawha County
Representative from a patient-based organization or advocacy group for rare disease with preference given to organizations based in West Virginia

Kimberly Earl, Kanawha County
Person over the age of 18 who either has a rare disease or is a family member of a person with a rare disease

Dr. Nadia A. Falah, Monongalia County
Physician licensed and practicing in the state with experience researching, diagnosing, or treating rare diseases

Dr. Sarah A. McGuire, Monongalia County
Physician licensed and practicing in the state with experience researching, diagnosing, or treating rare diseases

Dr. Kathryn S. Moffett, Monongalia County
Physician licensed and practicing in the state with experience researching, diagnosing, or treating rare diseases

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Background

The West Virginia Advisory Council on Rare Diseases was established by the West Virginia Legislature to advise state agencies on research, diagnosis, treatment, and education relating to rare diseases. The Council consists of 12 voting members who are appointed for three-year terms [3].

A "rare disease" means any disease which affects fewer than 200,000 people in the United States and is known to be substantially under-diagnosed and unrecognized as a result of lack of adequate diagnostic and research information, including diseases known as "orphan diseases" for research purposes.

Duties of the Council

Per W. Va. Code §16-5CC-3, the Council is charged with the following duties to the degree that resources are available, including, but not limited to:

(1) Coordinate statewide efforts for the study of the incidence of rare disease within the state;

(2) Act as the advisory board to the secretary and the West Virginia Legislature on research, treatment, and education relating to rare diseases;

(3) Research and identify priorities relating to the quality of, and access to, treatment and services provided to persons with rare diseases in the state;

(4) Develop, in conjunction and cooperation with the state's medical schools, policy recommendations relating to the quality of, and access to, treatment and services provided to persons with rare diseases in the state;

(5) Advise, consult, and cooperate with other offices of the department, other agencies of state government, and patient-based organizations in the development of information and programs of benefit to the public and the health care community relating to the diagnosis, treatment, and awareness of rare diseases;

(6) Identify best practices for rare disease care as implemented in other states and at the national level that will improve rare disease care in the state;

(7) Develop recommendations for effective strategies to raise public awareness of rare diseases in the state;

(8) Develop recommendations for best practices for ensuring that health care providers are sufficiently informed of the most effective strategies for recognizing and treating rare disease; and

(9) Report to the Governor, secretary, and West Virginia Legislature not later than January 1, 2021, and annually thereafter on the activities of the advisory council and its findings and recommendations regarding rare disease research and care in West Virginia, including any recommendations for statutory changes and amendments to the structure, organization, and powers and duties of the advisory council. The advisory council shall terminate on January 1, 2023.

Powers of the Council

Per W. Va. Code §16-5CC-4, the Council is able to pursue and accept gifts, grants, and bequests of funds from individuals, foundations, corporations, federal government, government agencies, and other organizations or institutions to fund the activities of the Advisory Council. In conjunction with W. Va. Code §16-5CC-6, the Council has partnered with the West Virginia Department of Health and Human Resources' (DHHR) Office of Budgets to establish a mechanism for accepting funds (Rare Disease Advisory Council Fund) [3].

The Council also has the power to schedule and conduct meetings, which are held monthly in a virtual format and posted on the West Virginia Secretary of State's website in accordance with the Open Meetings Act.

Additionally, to the degree that funds are available, the Council has the power to publish findings, recommendations, and reports on diagnosis, treatment, research, and education for rare diseases for the use and benefit of DHHR, other agencies of the state, the medical community, general public, and organizations representing the patients affected [3].

Acknowledgment

Although the Council is charged with identifying the unmet needs of individuals with rare disease, it is important to recognize that there are many other agencies working together with the Council to ensure access to resources and provide support to rare disease patients in West Virginia. For example, DHHR has a contract with the West Virginia University (WVU) to provide newborn screening through WVU's genetic and metabolic specialists, who provide the majority of the state's genetic counseling [5]. Due to a lack of therapy, management, and access to specialists, patients with rare diseases in West Virginia continue to endure increased health disparities and inequities.

The Council thanks the Everylife Foundation for providing funding from the RareGIVING Grant Program for the years 2021-2022. This funding was utilized to develop the Council's website. The Everylife Foundation also helped the Council distribute and promote a survey created by the Council (detailed below) by posting an announcement on their website and sending action alerts to West Virginia residents.

The Council also thanks the Statewide Centers for Independent Living and the Fair Shake Network for their help with Council project promotion statewide.

Council Activities

As specified in the authorizing legislation creating the West Virginia Advisory Council on Rare Disease (W. Va. Code § 16-5CC-1), the Council is tasked with submitting an annual comprehensive report identifying, as best as possible, the incidence and prevalence of rare disease in West Virginia, the needs of the rare disease community, and feasible actions to address those needs [3].

The Council planned and initiated a variety of activities in order to meet its charge and achieve its mission. The Council:

- Scheduled and conducted monthly virtual meetings the first Friday of each month to seek input and gain information about rare disease patient issues in West Virginia. The first meeting was February 5, 2021.
- Established a mission statement: *To improve the quality of life of all individuals living in the state of West Virginia affected by rare disease.*

- Established a vision statement: *To lead in efforts to support care for individuals living with rare disease through collaboration, education, support, and advocacy.*

- Created a logo: WEST VIRGINIA ADVISORY COUNCIL ON RARE DISEASES

- Met with representatives from the National Organization for Rare Disorders (NORD) and the Rare Disease Council from the State of Pennsylvania. Both organizations were helpful is establishing priorities for the Council to work on.
- Developed a user-friendly website, www.wvrare.com, as a centralized source for persons with rare disease to communicate with the community, share specific disease information, patients' story, community events and resources available [7].
- Initiated a project entitled "**Improvements Needed to Support Patients with Rare Disease in West Virginia."** The Council received an West Virginia University (WVU) Institution Review Board (IRB) approval to share a voluntary survey with the rare disease community in order to determine rare disease disparities, community needs that can help with future directions. The survey is available online at https://wvu.qualtrics.com/jfe/form/SV_8p07RgVaIuxqwS2. As of June 1, 2022, over 50 responses have been received and data is under review by the Council.
- Informed local media, nonprofit organizations, medical facilities and community members about scheduled meetings and the objectives of the Council and sought their input in developing Corrective Action Plans.
- Reviewed and commented on proposed policies, regulations and procedures affecting Rare Disease individuals
- Participated in rare disease education activities.
- Interacted with nonprofit agencies, foundations, and other stakeholders
- Assigned members to committees (medical team that established WVU's Institute Review Board (IRB), outreach team sharing survey to appropriate constituents).
- Provided information to families with rare disease about resources available in the community.
- Participated in meetings with other groups associated rare disease missions.
- Served as a stakeholder group for the development of the State Performance Plan and the Annual Performance Report, both required by federal law.
- Cooperated and collaborated with DHHR's Office of Maternal Child, and Family Health and its West Virginia Newborn Screening Program to address the needs individuals with rare disease.

Incidence and Prevalence of Rare Disease in West Virginia

Understanding the incidence of rare diseases in West Virginia is critical for recognizing the rare disease health burden and directing research resources. However, measuring the incidence of rare diseases is a challenge since patients with rare diseases may not be diagnosed or may be diagnosed late, even if the problem was recognized at birth. Another potential challenge is lack of consistency in diagnostic vocabulary to describe rare diseases. Only 500 out of 7,000 rare diseases have disease specific ICD-10 codes (International Classification of Diseases). Lack of budget and staffing, specifically expert epidemiologists, is an additional barrier to attain this goal.

Improvements Needed to Support Patients with Rare Diseases in West Virginia

In order to understand the magnitude and complexity of the true needs in the rare disease community in West Virginia, the Council has developed a rare disease community needs assessment survey (101 questions) that explore topics such as:

- Diagnostic delay;
- Access to diagnostic and specialty care;
- Access to treatment and support;
- Costs, including treatment, care, travel, time away from work or school;
- Insurance coverage and disability benefits; and
- Barriers and other difficulties.

This is the first statewide survey of the needs of the rare disease community. The project was approved by WVU's IRB under protocol number 2105319302 and all methods in this research study were carried out in accordance with the Declaration of Helsinki on human research participants.

The survey was distributed via flyers [see figure 1.], the Council website www.wvrare.com, and by the Council members in their respective organizations. The survey results will develop a comprehensive and flexible framework for rare disease needs assessment as well as inform the Council's future policy and resource recommendations. The survey results will also help shape future Council initiatives and strengthen its advocacy for the rare disease community.

Survey Results

Participant Characteristics

In total from March 1, 2022, to June 15, 2022, 72 participants (parents of children or patients ≥ 18 years of age) with rare disease completed the survey. All data provided for this analysis were obtained as of June 15, 2022. Of the 72 participants with rare disease analyzed, 39 were female, 22 were male, and 1 was unspecified. Data collection began in March 2022, and analysis was performed in June 2022. Most patients were Caucasian (79%). See the Table which outlines the demographic of the participant.

Frequency of Main Issues Reported

The majority of participants (57%) were diagnosed by a specialist (28 local,13 national). It took more than three years for 27 percent of diagnoses to be made. Seventy-one percent of the participants are dissatisfied with their health care providers. Thirty-one percent have private insurance, 41 percent have had a service denied, and 32 percent have had a specialist referral denied. Thirty-four percent are unaware of any nearby specialized centers, and twenty percent say none are available. To go to a specialist facility, 45 percent said they had to travel more than 60 miles. Although 32% of people have access to telehealth, 10% have no interest in using it (see figure 2).

18% said their child's school isn't equipped to meet his or her demands, and 15% said their child needs to take specific medications while at school. Around 45 percent said the COVID-19 pandemic had a negative impact on their health and access to care.

Comments From the Council's Survey of the WV Rare Disease Community

- **"**Providers did not correctly recognize the symptoms and presentation of charge syndrome and also did not order the correct imaging (brain and inner ear CT) when my son was born that would have easily confirmed the diagnosis. Instead, he had multiple genetic tests ordered by geneticists in WV and PA that were off target, and his diagnosis was delayed many years."

- "Physicians in WV will not acknowledge a problem and continually just say to wait and see if it gets better. They delay diagnostic testing and treatment. I'm not sure if they don't recognize the issue or if they really think it is better to wait, but either way it is a massive barrier to care."

- "Not enough specialists for second opinions, difficulty with insurance approving medications."

- "Lack of awareness/ knowledge of patient before diagnosis. Lack of primary care doctors recognizing symptoms."

- "First off finding the diagnosis at a young age. Then finding the correct doctors in our area to treat the disease. Then trying to manage with medicine which ended in surgery and almost death until we started Stelara. She has now been in remission for 3 years for the most part."

- "Being told my condition was not real. I was told by a hematologist that I made up the disorder of iron overload. Which delayed my treatment. Fortunately, Dr. Clarke helped me pursue help."

- "Cost of medical services even with insurance co-pays and deductibles are causing financial hardships. Traveling out of state multiple times a year for medical care. Providers in WV do not know how to handle her care, and most cannot answer questions. It is frustrating."

- "I wish I would have had closer counseling and order of labs to monitor my son's condition."

- "It is frustrating to have to educate other physicians I see about Gaucher Disease as they, for the most part, have never heard of it."

- "Genetics testing is a wonderful tool."

- "We shouldn't have to travel 3 hours each way to visit doctors or have routine procedures. Cost is a huge barrier even though both parents have full time jobs and insurance. My credit has been ruined due to the deductibles associated with medical care. There is a never-ending cycle of medical bills. It's stressful."

- "This is a treatable disease if caught at birth. It needs added to newborn screening immediately."

- "At work I am judged without accommodation to my medical disabilities. I need the job and don't want to risk termination, or it will result in financial ruin and homelessness for me and my family. Disability determination (SSDI) is impossible to get in WV for someone with rare illnesses. My daughter was bullied in school and has failed to make transitioning from childhood to adult responsibilities of financial self-sufficiency. She also has 2 children. I support all of us on about $30,000 a year. It is unsustainable. We need socialized health care like the UK."

- "Testing at birth since this is a hereditary disease and treatments to begin earlier would slow the progression."

- “Need more specialists and better access to new medical innovations/technology/etc.... especially pediatric specialists”

- “Her late diagnosis was difficult to adjust to. She sometimes felt (everyone) expected too much from her since she did not grow up doing these treatments so often as most CF patients do. She eventually adjusted but sometimes still does not always want to do extra treatments if she has been very active and is very tired.”

- ”Try to not judge by age, try to get them mental help if needed earlier on rather than later. Give them tips on how to manage life and programs with that disease. Help groups.”

Rare Disease Council Challenges and Future Direction

It is of critical importance that the Council receives broad statewide input from those affected with rare disease to better understand and quantify the needs of the rare disease community. The Council is not aware of any published data from other states about rare disease survey to assess the difficulties and needs of those affected by rare disorders; however, the WV Rare Disease Survey was adapted from the Pennsylvania Rare Disease Survey, so data may be available in the future.

The West Virginia Rare Disease Survey is continuing to undergo further wide-range distribution and testing/validation. With statewide marketing, the Council expects to increase the number of participants and to collaborate with stakeholders for in-depth analysis of the results.

Rare disease patient organizations and their members are vital to the advancement of their healthcare. In West Virginia, there is lack a lack of patient organizations. The Council plans to look at methods to aid additional patient organizations in the state through the national collaborative rare diseases network.

The Council intends to consult with state and local epidemiologists to develop an inclusive and transparent process for characterizing the incidence and prevalence of rare disease in West Virginia, as well as to develop the most effective structure for accurate assessment of the incidence and prevalence of rare disease with the assistance of an epidemiologist. The Council also plans to develop a budget and funding resources to develop a system of reporting current and new cases of rare disease.

In the coming year, the Council plans to take a collaborative approach to achieve its goal of becoming an advocate for West Virginia's rare disease community and offering insight into breakthroughs in newborn screening, policy changes, and administration recommendations. The Council believes it has taken significant first steps by working in the best interests of the rare disease community from a fresh perspective.

Conclusion

The Council is prepared to develop deeper analysis of the needs for West Virginia and the direction the Council will work to boost confidence in the identification of rare disease patients' requirements in West Virginia. This preliminary report represents data efforts for 2021-2022, with data from a small sample size of our targeted community. The approach and protocols used in survey study should aid in expanding our knowledge to engage a wider number of participants and public health experts.

As a result, we hope to have a better awareness of the variety of demands from which policy recommendations can be developed. The difficulties that affect the rare disease population are not just about health care or insurance but have an impact on government and non-government policies, systems, and programs, and all elements of society.

To such extent, the Council would like to recommend the following actions to improve the care for patients with a rare condition in West Virginia:

1) Increase funding of the newborn screening program, specifically the genetics grant to allow for the purchase of a secondary (backup) screening machine and improved staffing to coordinate necessary tasks based on results;

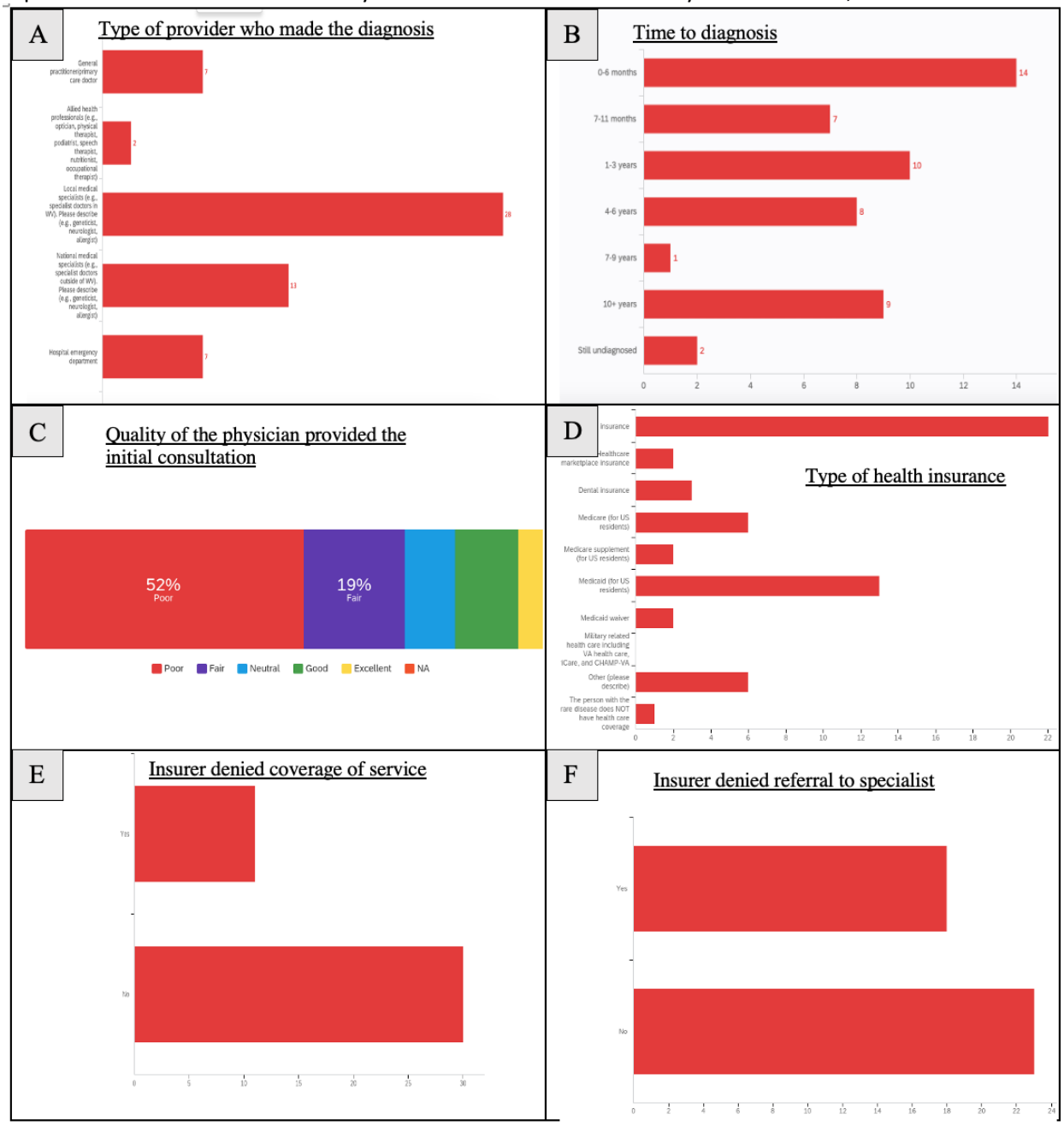
2) Ensure entrance into the Medicaid system as a secondary insurance for patients who carry primary commercial coverage within the State of West Virginia. Based on survey results, the financial burden to families impacted by a rare disease is extremely large, especially for those impacted by high out-of-pocket plans. The Council suggests developing a plan to provide secondary insurance to those with the most common rare diseases accompanied by extremely high costs such as hemophilia, cystic fibrosis, rare cancers, fabry disease, and multiple sclerosis as a trial program to study the impacts within the Medicaid system and analyze outcomes;

3) Instill coverage on a state level to exome trio genetic testing which is currently not covered by PEIA or Medicaid; and

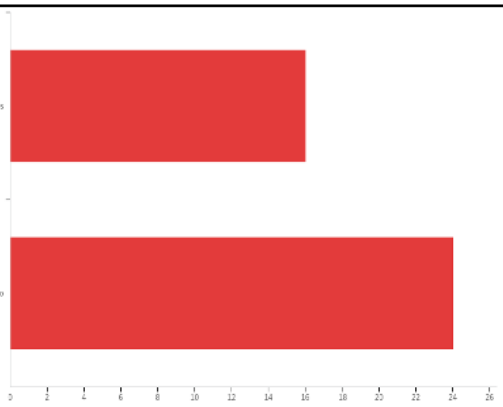
4) Provide coverage for medical foods, when necessary, based on patient diagnosis.

Figure 1. West Virginia rare disease assessment survey flyer

Figure 2. (A-N) Prevalence of issues of rare disease participants: number of the participants based on the reported issues in the shared survey who were included in the analysis as of June 6, 2022.

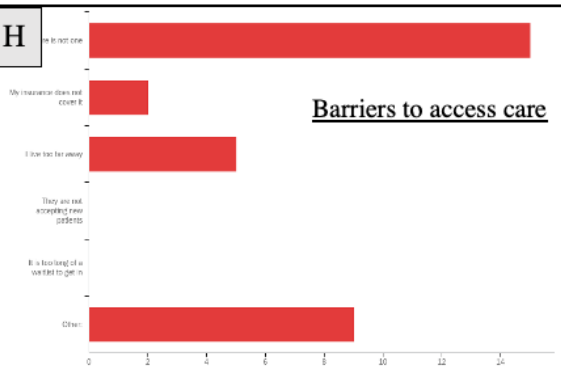


G



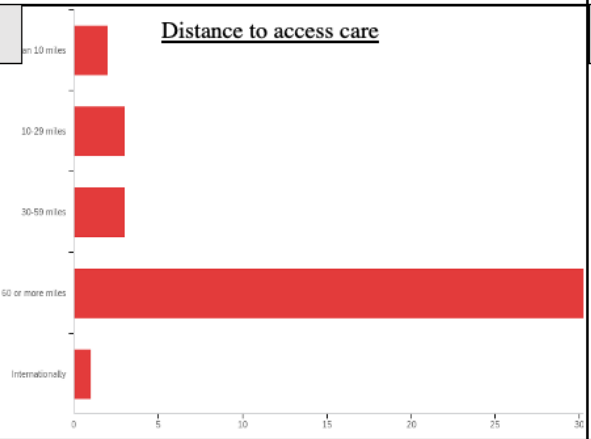
H

Barriers to access care



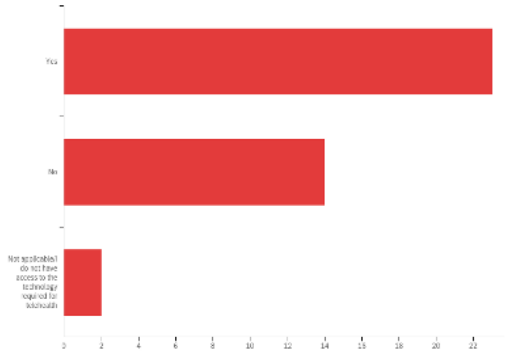
I

Distance to access care



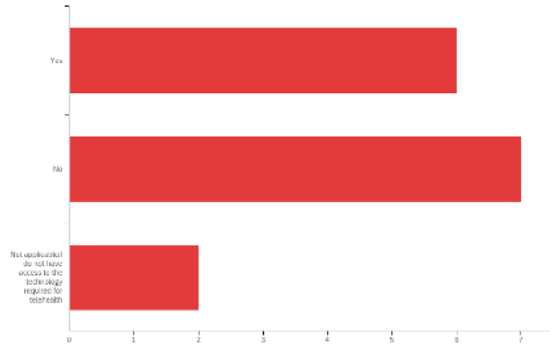
J

Participant interest to use telehealth



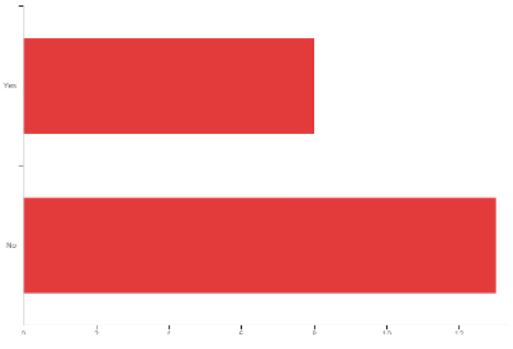
K

Your child's need was anticipated by the school



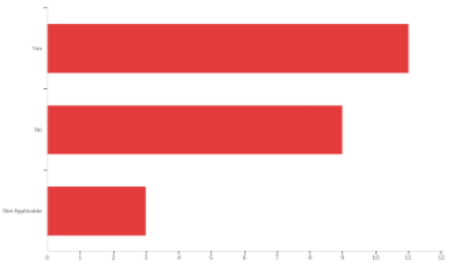
L

The school adequately quipped to care of the child



M

Your child's need of medication during school



N

COVID-19 pandemic affected access to care

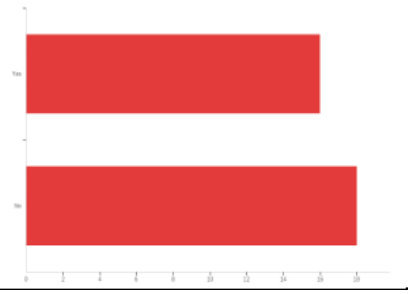


Table: Demographics of Rare Disease individuals in West Virginia residents (March 1, 2022-June 15, 2022)

		Patients (n 72)	**%**
Gender			
	Female	39	54%
	Male	22	31%
	Not identified	1	1%
Age			
	< 18	26	36%
	=> 18	36	50%
	Blank	10	14%
Ethnicity			0%
	Black	2	3%
	Hispanic	1	1%
	White	57	79%
	Other	2	3%
Marital status		1	1%
	Single	33	46%
	Married	23	32%
	Other	16	22%

References:

1. National Institutes of Health – Rare Disease. https://www.nih.gov/about-nih/what-we-do/nih-turning-discovery-into-health/rare-diseases. Accessed 20 Jun 2022.

2. U.S. Census Bureau QuickFacts: West Virginia. https://www.census.gov/quickfacts/WV. Accessed 25 Jan 2022.

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5. West Virginia Department of Human Health Resources – Newborn Screening. https://www.wvdhhr.org/nbms/. Accessed 25 Jan 2022.

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