



## LETTER TO THE EDITOR

# The Rare Disease Advisory Council Expands its Reach into West Virginia Appalachia: A Call to Action

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## Abstract

Patients with rare diseases in rural Appalachia face an additional hurdle in getting care. West Virginia is the only state wholly within the Appalachian region, which is noted for having some of the nation's worst health disparities. The West Virginia Advisory Council on Rare Diseases was established in January 2020 as part of the Rare Disease Advisory Council (RDAC) Acts, with the goal of developing strategies and recommendations to reduce health inequalities among West Virginians with rare diseases. The major goals of the council are to coordinate research on rare disease treatment and education, enhance access to services, policy suggestions, public health, and community awareness. This project also focuses on forging close ties between rare disease organizations and state governments in order to provide advice on state activities that will raise rare disease awareness. This project aims to develop a national-level, actionable medical services strategy that will improve the delivery of high-quality healthcare to people and families living with rare diseases in West Virginia.

## Keywords

Advisory Council on Rare Diseases, Rare disease, Genetics, Appalachia, Health disparities, West Virginia, Rare Disease Advisory Council

## Background

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. For example, the state of West Virginia has a contract with the West Virginia Department of Health and Human Resources to provide newborn screening through genetic and metabolic specialists at West Virginia University (WVU), who provide the majority of the state's genetic counseling [3]. The state also has a hemophilia program, which provides adequate resources to hemophilia patients and aids in medical expense reimbursement for children with the disease [4]. 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.

Most states have orphan drug or rare disease legislation with research and service requirements, as well as some sort of plan of action and/or care for rare diseases [5]. While this does not result in consistent healthcare services across the United States, there are fewer disparities between geographic regions and hospital levels than in West Virginia [6]. Exploration of the current situation, prospective adoption of established models, and the development of new models are all critically needed in West Virginia to improve optimal prevention, timely diagnosis, and high-quality treatment of rare diseases within the current and future healthcare systems.

A Rare Disease Advisory Council (RDAC) is a statewide advisory organization that gives the rare disease community a stronger voice. Patients, caregivers, families, and physicians in North Carolina formed the first RDAC in 2015 [7]. RDACs have subsequently been formed in twenty-one states, and stakeholders in several other states have attempted to establish them in order to better represent their community and explain their challenges and demands to those in authority. Similar state-level rare disease advisory councils have been established in Alabama, Connecticut, Illinois, Kentucky, Minnesota, Missouri, New Hampshire, New York, Nevada, North Carolina, and Pennsylvania [7]. To address the issue of rare illness care in West Virginia, the Advisory Council on Rare Diseases was established in 2020 to encourage the improvement of rare disease healthcare [8].

### West Virginia Advisory Council on Rare Diseases Establishment

The council is made up of twelve voting members. The Governor appoints eleven members, including the Secretary of the Department of Health and Human Resources or his or her nominee. There are also three physicians licensed and practicing in the state with experience researching, diagnosing, or treating rare diseases; three people over the age of 18 who have a rare disease or are a family member of someone who has a rare disease; a registered nurse or advanced practice registered nurse licensed and practicing in the state with experience treating rare diseases; and a person with an advanced degree in public health or another health-related field [8].

### The Objective of the Council

The council was formed in order to coordinate statewide efforts to research and study the incidence of rare diseases, to advise the legislature and state agencies on rare disease issues, to identify priorities, and to make policy recommendations on the quality of and access to treatment and services provided to people with rare diseases in the state. The council is charged with developing strategies to raise public awareness about rare diseases, providing education, and making recommendations for best practices for healthcare providers that have been implemented in other states and at the national level. The council also reports its activities to the Governor on an annual basis [8].

### Discussion

Understanding the incidence of rare diseases in West Virginia is critical for recognizing the diseases' health burden and directing research resources. However, measuring the incidence of rare diseases is difficult since patients with rare diseases may not be diagnosed or may be diagnosed late, even if the problem was recognized at birth [9]. Since most rare diseases are

genetic, calculating the allele frequency in recessive situations (with the concept that all recessive disorders are inherited) will be necessary, which might go undiscovered in a massive publicly accessible database. Because of the rarity of the contributing alleles and the small number of affected individuals, calculating the frequency of rare diseases is complicated. Rare disease research necessitates large amounts of data that are not always readily available [9].

The most applicable indicator of rare diseases is point prevalence, which gives an assessment of the disease burden in the community and can thus influence focused service delivery targeted at the special needs of patients, as well as pharmaco-economic evaluation of orphan medications [10]. Adequate epidemiological data, scholarly articles, and structured databases will be required. The diversity of the data is derived from a variety of disparate information sources that are not standardized or difficult to combine, such as published case reports or systematic reviews, patient registries, expert opinions, and other anecdotal evidence. This makes estimating the global point prevalence of rare diseases challenging [10].

The best estimate of the prevalence of numerical examples of rare diseases may be found in the inpatient medical records of significant medical centers within a range of time. West Virginia can build a de-identified patient data repository that is aggregated in a uniform manner and uses standardized nomenclature. West Virginia can also create a web-based open-source patient registry system that will be made available to the public as a service to patient organizations and others, allowing and encouraging them to create more rare disease patient registries [11,12].

Another main objective of the council is to improve the quality of and access to treatment and services for patients with rare diseases. They are known to face delays in diagnosis, access to specialists, and treatment around the world. West Virginia is one of the poorest states in the country, with one out of every five children struggling to access food. In West Virginia, 16 percent of the population lived in poverty in 2019, while 6.6 percent of citizens lacked health insurance (Kaiser, 2019). In rural West Virginia, the poverty rate is significantly higher (17.6 percent, compared with 16.3 percent in urban areas of the state) [13].

As a result, rare disease patients in West Virginia have a particular healthcare access dilemma. There is a scarcity of high-speed internet connectivity for at least some of the population, in addition to the stated dearth of nurses and physicians. The absence of broadband connectivity in West Virginia is preventing the growth of telemedicine, which would provide healthcare access to individuals in the state's most remote areas [6,14].

WVU Medicine Children's is a new project in West

Virginia aimed at addressing capacity challenges and better serving the healthcare needs of all women and children in the state [15]. In addition, we suggest that a non-profit foundation-sponsored clinic network be involved to help patients with rare diseases gain access to specialized care. The state must also create a strategy that will revolutionize medical education and exponentially enhance the workforce capacity in order to deliver best-practice specialist care while also reducing health inequities. Primary care practitioners can give de-identified case presentations to interdisciplinary specialists in the field, and the expert hub will assess them and make basic recommendations.

The most successful ways to diagnose and treat rare diseases are education and increasing knowledge and awareness of rare diseases for frontline healthcare professionals. Unprepared physicians in rare complex disorders are partly to blame for late diagnoses and misdiagnoses of rare diseases [16]. Physicians lacked basic information about how the etiology, epidemiology, and prevalence of rare diseases are documented. While genetics is an important element of medical education, there is a paucity of information about rare diseases among medical students and healthcare workers in West Virginia, which has a relatively small population. It is suggested that the schools conduct a rare illness symposium, hold rare disease discussions at conferences, hire rare disease specialists and encourage student involvement in rare disease-related activities such as ambassadors, competitions, and clubs.

Raising public awareness of rare diseases is a process that aims to enlighten and educate people about rare diseases and their causes in order to affect their attitudes, habits, and beliefs. It necessitates the creation of a targeted communications campaign that utilizes a variety of venues, including the internet and social media. The challenges of public education or awareness campaigns must be to deliver the message to the appropriate intended audience and purpose.

Rare disease patient organizations and their members play an important and active role in advancing their healthcare. There are a few patient organizations in West Virginia, although they are relatively small. Through the national collaborative rare diseases network, the West Virginia Rare Diseases Advisory Council can look into ways to help more patient organizations in the state.

An in-depth understanding of rare diseases for federal or state government officials and policymakers can address impediments to treatment resources and care within the context of the RDAC. The development of genetic testing services, as well as the analysis of data from the rare disease patient registry and repository system, will aid in determining the prevalence, regional distribution, and cost of rare diseases, and will serve as a valuable resource for health authorities when drafting

medical policies on rare diseases and orphan drugs. RDAC has the potential to raise awareness of rare diseases among the general public and professionals in low-level hospitals. This exploratory effort will hopefully result in the development of a preventative and treatment pattern that is appropriate for West Virginians. RDAC's knowledge and ideas will also be useful to other states as they address their own rare disease healthcare concerns. By objectively quantifying the burden of rare diseases, providing platforms to reach out to the rare disease community, and fostering collaboration across industries and countries, policies can be created or strengthened. These suggested measures should spark conversation and improvement in the region.

## Competing Interests

The author has no conflict of interest to disclose. The views expressed in this article are those of the authors and do not necessarily reflect the position of the authors or policy of WVU Medicine.

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