

Rare Diseases in Pennsylvania

*Preliminary Report Issued by
the Rare Disease Advisory
Council Pursuant to Act 14 of
2017*

July 2018



pennsylvania
DEPARTMENT OF HEALTH

Letter from the Chair

July 2018

Health and Human Services Committee, Senate of Pennsylvania
Health Committee, Pennsylvania House of Representatives

On behalf of the members of the Pennsylvania Rare Disease Advisory Council (Council), it is my pleasure to submit this report to these committees as required by Act 14 of 2017. This Preliminary Report summarizes 10 months of work and collaboration outlining six ambitious yet attainable priority goals from now through 2025. Additionally, it showcases innovative approaches to achieve these goals through a methodic and strategic plan.

According to the National Organization for Rare Disorders, there are more than 7,000 rare diseases, each of which affects 200,000 or fewer individuals in our country. A year ago, the Council was established and, in a relatively short timeframe, has become a unified voice for the estimated 1.2 million Pennsylvanians affected by rare diseases who are often not heard or understood. The uncertainty of the actual number of Pennsylvanians affected by rare diseases is a key gap being addressed by the Council's work.

Uncertainty is also evident in the faces of the individuals suffering from those rare diseases, their families and their caregivers. Members of the Council represent health care providers, advocacy groups, researchers, clinicians, and the insurance and pharmaceutical industries. More importantly, the Council is also comprised of individuals with, and parents of children with, rare diseases. This is a dedicated and committed group with no misconceptions of the difficulties that lay ahead. They are inspiring, and aspire not to fame or recognition, but to an improved quality of life for their family members, patients and absolute strangers. The members of the Council have all agreed on the goals in this document with the specific intent to improve the quality of life for those affected by rare diseases in Pennsylvania. Those affected face a difficult journey, fraught with uncertainty and complications. The goals herein will not remove all that uncertainty; however, they represent meaningful actions that have the potential to improve the day-to-day issues such individuals encounter, with their unique needs that today create a labyrinth from which there appears to be no definite exit.

The Council recognizes one of the greatest obstacles to its success is the lack of available resources to meet this challenge. We will strive to be innovative, form partnerships and leverage resources to the greatest extent possible. The members are available to you as subject matter experts. As Chairman of this Council, I look forward to continuing to work with its members and with each of you.

Sincerely,



Tomas J. Aguilar
Chairman, Pennsylvania Rare Disease Advisory Council
Director, Bureau of Health Promotion and Risk Reduction
Pennsylvania Department of Health

Council Agreement

We, the undersigned, as members of the Pennsylvania Rare Disease Advisory Council, by our signatures below, hereby forward this report pursuant to Act 14 of 2017. The report contains a summary of accomplishments of the Rare Disease Advisory Council since its first meeting on October 3, 2017 and the status of its work in preparation of the comprehensive report required under Section 6b within two years of the effective date of Act 14. As a body, we agree these assessments and goals provide a starting point to fulfill the intent of Act 14 for a statewide approach to improve and protect the health of individuals with rare diseases of the Commonwealth of Pennsylvania.



Tomas J. Aguilar, Chairman
Department of Health, Harrisburg



Patrick Collins, Senior Director
CSL Behring, King of Prussia



Marie Conley, Founder, Conley Cushing's
Disease Fund, Hummelstown



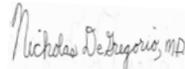
Joseph Coyne, Executive Director
Garrett The Grand Batten Fighter, Gilbertsville



Elaine Davis, RN, Clinical Coordinator
Penn State Hershey Medical Center,
Hummelstown



Jessica Deary, MBA, Independent Patient
Advocate & Health Care Advisor, Harrisburg



Nicholas DeGregorio, MD, Senior Medical
Director, UPMC for You, Pittsburgh



Connie Deline, MD, Vice President
Spinal CSF Leak Foundation, Camp Hill



Can Ficioglu, MD, Associate Professor
Pediatrics, University of Pennsylvania,
Philadelphia



Andrew Herlich, MD, Pediatric Anesthesiologist
University of Pittsburgh, Pittsburgh



Robert N. Jinks, PhD, Professor of Neuro
Science, Franklin Marshall College, Lancaster



David Kelley, MD, Chief Medical Officer
Department of Human Services, Harrisburg



Ann Kriebel-Gasparro, DrNP, Assistant
Professor of Nursing, Alvernia University,
Reading



Kathryn Lavriha, Director
Sanofi, US, Stevensville, MD



Sharon O'Shaughnessy, MA, Advocacy Chair,
Narcolepsy Network, Downingtown



Barbara Ostrov, MD, Professor of Pediatrics &
Medicine; Rheumatology, Penn State University,
Hershey



Anna Payne, Member, Cystic Fibrosis
Foundation Great Strides, Leadership
Committee, Langhorne



Elizabeth Rementer, Deputy Director
Communications, Insurance Department,
Harrisburg



Nicholas Slotterback, Health & Education
Advisor, Pennsylvania Department of Education,
Harrisburg



Evelyn O. Talbott, DrPH, Epidemiologist and
Professor, University of Pittsburgh, Pittsburgh

William Welch, MD, Professor of Neurosurgery
University of Pennsylvania, Philadelphia



Jennifer H. Wescoe, M.Ed, Founder, Executive
Director, Wescoe Foundation for Pulmonary
Fibrosis, Coopersburg



Bret Yarczower, MD, Senior Medical Director
Geisinger Health Plan, Danville

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Executive Summary

The Rare Disease Advisory Council is submitting this preliminary report in accordance with Act 14 of 2017 (Act).

The Council has adopted the definition of rare disease as a disease or condition that affects fewer than 200,000 people in the U.S. It is estimated that there are 7,000 rare diseases affecting 30 million Americans, or 7 to 8 percent of the population, according to the National Organization for Rare Disorders (NORD). While the clinical manifestations of these diseases are manifold, there are similarities that span the wide variety of diseases and conditions. These may include a prolonged diagnostic odyssey, uncoordinated interactions with the health care system, lack of access to necessary expertise, availability of effective treatment, and struggles with cost and coverage barriers.

Preliminary Baseline Data Assessment – Highlights

Pennsylvania is one of five states that has established a Rare Disease Advisory Council. The other states are Connecticut, Illinois, North Carolina and Rhode Island. As of July 2018, several other states were in the process of considering similar bodies.

There are clinical resources in Pennsylvania that provide expertise in the diagnosis and treatment of rare diseases, but the journey to access these resources can be long and convoluted.

The percentage of rare diseases that have an FDA-approved pharmaceutical to treat them is in single digits. However, in 2017, of the 77 new drugs approved by the FDA, 18 were orphan drugs, including a breakthrough treatment for Batten Disease and another for RPE65 mutation-associated retinal dystrophy. Since we also know costs associated with treatment are very high, a goal of the Council is to make treatment more affordable and accessible.

There is a growing knowledge gap between specialists with expertise in a rare disease and those who care for those affected. Information resources are available to help bridge these gaps, but they are not easily accessible or coordinated.

Those with an undiagnosed disease require attention. Since 2002, the Genetic and Rare Diseases Information Center (GARD) has received 66,000 total inquiries. The topmost reason for outreach was for an undiagnosed condition (4,854).

There is much that is unknown, for example:

- We don't know the incidence, prevalence and trends of most rare diseases in Pennsylvania.

- We lack a comprehensive list of rare disease experts in Pennsylvania and nationally. This is especially true for those who are undiagnosed and for unnamed rare diseases.
- We don't know the financial impact of rare diseases in the commonwealth.
- What do patients (and their families) need, and what can we do to help?

Priority Goals

The work of the Rare Disease Advisory Council over its statutory life through 2025 will organize its efforts around six priority goals defined through consensus this first year of its existence. Strategic objectives in support of the following goals have been established and are described herein:

1. Develop an epidemiologic structure to characterize incidence and prevalence of rare diseases;
2. Develop a comprehensive and flexible framework for rare disease needs assessment;
3. Provide a comprehensive and accessible source of information on rare diseases in Pennsylvania;
4. Develop systems to streamline the diagnostic evaluation, management and support for those with rare diseases;
5. Promote collaborations with rare disease organizations, advisory councils, the medical community and academic medical centers; and
6. Enhance access to new and existing therapies, programs and services to improve quality of life.

The Council has already made progress by establishing this overall direction, meeting on a regular basis and identifying ways to raise awareness and educate the public. Pennsylvania Rare Disease Day occurred on June 19 in Harrisburg with a legislative lobby day for patients and advocates as well as a press conference.

In its second year, FY 2018-19, in accordance with Act, the Council will provide a follow-up report that addresses incidence and prevalence of rare diseases in Pennsylvania, the needs of the rare disease community and feasible actions necessary to address those needs.

There will be biennial reports with Council findings and recommendations. These will include follow-up on the long-term goals articulated within the activities in Year One. The Council will develop a structure to characterize incidence and prevalence. The Council will determine best practices for data collection and compare what is available in other states and at the federal level. There will be collaboration with NORD, patient organizations and other stakeholders.

The Council recognizes the lack of funding to pursue its activities. The Act allows the Council to seek public and private funding. The Council will work with its agency representatives to navigate the funding streams and will likely make recommendations to the legislature for funding.

It is nearly a certainty that many of those who encounter these words are or will be affected in some manner by a rare disease. There is reason to be hopeful. These are exciting and promising times. Advances in diagnostic and therapeutic tools are accelerating.

To our friends and neighbors, members of our commonwealth who struggle with a rare disease and to family members who support their loved ones, taking joy where they can find it and tackling obstacles when they are encountered, we dedicate our efforts.

Introduction

In 2012, researchers at the Archeological Museum in Zagreb, Croatia reported results of a diagnostic analysis performed on a mummified Egyptian man who died in his 20s approximately 2,900 years ago. Radiographic analysis revealed lesions throughout his spine and skull. They arrived at a diagnosis of Hand-Schuller-Christian disease (Langerhans cell histiocytosis), a rare disease with an incidence estimated to be 3 to 5 per million children and 1 to 2 per million adults. This would meet our definition of rare disease, which is a disease or condition that affects fewer than 200,000 individuals living within the United States.¹ Given its rarity, it's likely his physician had never seen such a case and had little to offer other than symptomatic relief from bone pain and other manifestations of this rare disease. One can imagine the consternation of the physician and the despair of the patient who succumbed to the illness at an early age.

While there have been breathtaking advances in medical care in the last three millennia, it is also true that, for many of those who suffer from a rare disease today, their experience may likely be similar to this rare disease victim in ancient Egypt. Whether due to violence caused by a misstep in the dance of genes or misfortune otherwise acquired, the burdens placed upon those affected are profound and are multiplied by delays in diagnosis, absence of curative treatments and struggles with barriers placed before them. In recognition of the desperate need of these members of our community and with a sense of compassion and urgency, the Pennsylvania Legislature passed House Bill 239 to establish the Rare Disease Advisory Council.²

The Rare Disease Advisory Council was established by the Pennsylvania General Assembly to:

- Coordinate statewide efforts to study the incidence and prevalence of rare diseases within Pennsylvania and the status of the rare disease community;
- Serve as an advisory body on rare diseases to the General Assembly and to all relevant state and private agencies that provide services to, or are charged with the care of, individuals with rare diseases; and
- Coordinate the performance of the Council's duties with those of other rare disease advisory bodies and community-based organizations and other public and private organizations within the state to ensure greater cooperation between entities within the state and federal agencies regarding the research, diagnosis and treatment of rare diseases.

Purpose of the Preliminary Report

This report fulfills the mandate detailed in Section 6a of the Act, requiring the Council to issue a preliminary report within 12 months of the effective date of the Act to the Senate Health and Human Services Committee and the House Health Committee. This includes a summary of the accomplishments of the Council since its first meeting on

October 3, 2017, and the status of its work in preparation of the comprehensive report required under Section 6b within two years of the effective date of the Act.

Statutory Authority and Direction

The Rare Disease Advisory Council Act, Act 14, was enacted on July 7, 2017, by Pennsylvania Governor Tom Wolf. The Secretary of Health, Rachel L. Levine, M.D, designated Bureau Director Tomas J. Aguilar from the Department of Health as the Chair. Members were then appointed as outlined under section “Council Membership.”



The first Council meeting was held October 3, 2017, and additional meetings have taken place both in person and by teleconference. Three working groups were established to focus initial efforts on communications, surveillance, and advocacy

The Council has been authorized to apply for and accept grants from the federal government, a private foundation or any other source that may be available for programs related to rare diseases. The Act expires June 30, 2025.

Mission and Vision

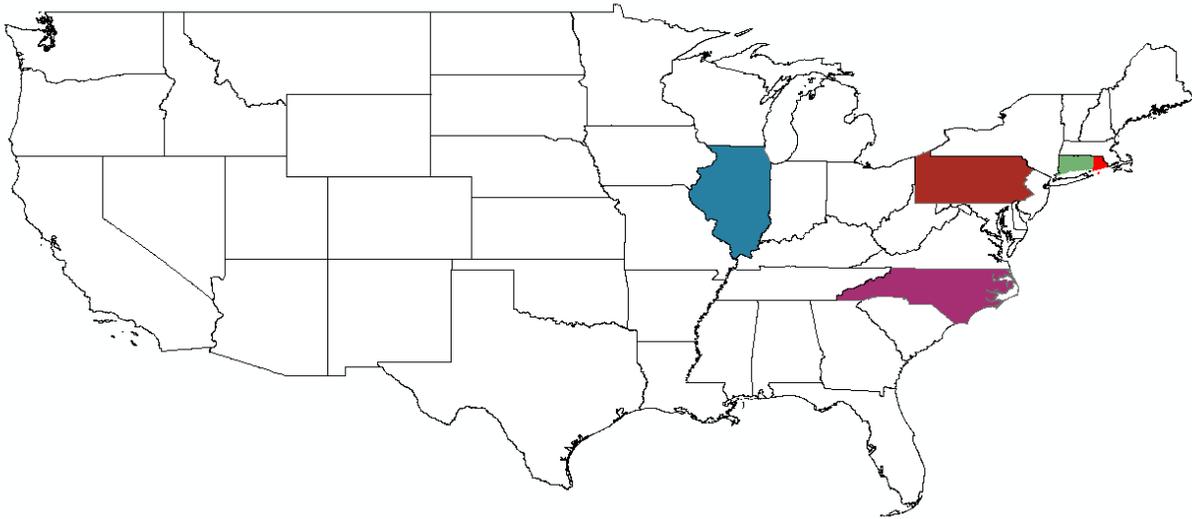
The Council strongly believes that any effort to advance an important purpose requires focus and direction. To that end, the following mission and vision statements are adopted by the Council. The mission reflects the Council's reason for existing, in line with Act 14 of 2017. The vision reflects the Council's aspiration for the coming years.

Mission:

To improve the quality of life for all those affected by rare diseases in Pennsylvania.

Vision:

To be a national leader in transforming the lives of those affected by rare diseases through collaboration, support, education and advocacy.



States with Rare Disease Advisory Councils

Preliminary Baseline Data Assessment

1) Purpose

Pennsylvania Act P.L. 285, No. 14 established this Council to help improve knowledge and management of rare diseases in Pennsylvania. Data from the Genetic and Rare Diseases Information Center (GARD) showed that, of the 66,000 total inquiries received since 2002, the topmost number of inquiries were for undiagnosed conditions (N=4854).

Patients with rare diseases and their families are often isolated and have little support; we know this because we are caring for patients with rare diseases in our communities. Those with undiagnosed conditions and rare diseases in Pennsylvania need our help.

2) Present Challenges and Barriers

Current state of fragmented data

We do not have the system(s) for communicating with patients and families to develop a clear sense of needs for these populations as described below.

There are insufficient resources to provide rare disease expertise to all those who might benefit from them.

There is a growing knowledge gap that exists between those with expertise in specific diseases and those providers who care for individuals who may have a rare disease.

Since rare diseases affect a relatively small number of individuals, there is little financial incentive for clinical and pharmaceutical trials.

We do not know the fiscal impact of rare diseases in Pennsylvania. What are the costs associated with rare diseases? An economic analysis will allow us to maximize the investment made in achieving the best outcomes while eliminating waste. This should include total costs.

We do not know what new treatments are being developed for rare diseases in Pennsylvania and their future cost implications. Genomic data does not move with patients, hindering diagnosis.

We do not have a complete understanding of what rare disease research is being done within the commonwealth and nationally.

Medical benefit policies exist for specific treatments for some rare diseases but none that articulate an approach to coverage determinations for rare diseases in general. Since these diseases have few “FDA approved” treatments, some rare disease patients may be denied coverage for therapies that are on the market. The volume of such cases is unknown.

The Pennsylvania Department of Health has a data resource: Enterprise Data Dissemination Informatics Exchange (EDDIE). However, this is not focused on rare diseases, and privacy concerns may prevent EDDIE from becoming a resource for rare diseases.

We do not have the epidemiological information that we need to characterize the incidence and prevalence of rare diseases in Pennsylvania.

We do not know the incidence, prevalence and trends for most rare diseases in Pennsylvania, such as whether there is evidence for trends and/or clustering of rare diseases in certain geographic areas. This knowledge would allow for allocation of resources. Nor do we have an idea of the mortality rates within the commonwealth and if they differ from other parts of the country.

We do not have a comprehensive list of rare disease experts in Pennsylvania or nationally for the litany of rare diseases (this is especially true for less well-defined rare diseases). Specific “rare disease” medical experts are the exception. More typically, physicians with expertise in certain rare diseases are available, but this varies by the condition. There are experts in hemophilia, for example, and the state funds six hemophilia treatment centers throughout the state. However, there will not be such a reference point for those who treat alpha-1 deficiency, as an example. A comprehensive list would need to reference by condition.

What do patients (families) want and what can we do to help?

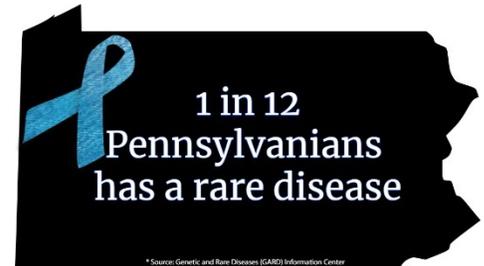
We do not yet fully understand the difficulties and challenges for those facing rare diseases. We need to know how long it takes to make a diagnosis for rare diseases, because for some diseases it is important to provide treatment as early as possible and avoid wasteful treatment that may not help or even cause harm. We do not currently know the most efficient way to reach a diagnosis and provide timely and cost-effective treatment.

We need to know what the most effective current treatment and management of rare diseases in Pennsylvania is so that gaps in best practices for those with rare diseases can be addressed.

Year Two Limitations: Funding will be needed to carry on this work if the Council is to report on best practices for advisory councils across the nation to address best practices in Pennsylvania.

3) Where We Are Today

According to NORD, there are 7,000 rare diseases affecting 30 million Americans. If Pennsylvania is representative of the U.S. population, that means, of the 12.78 million people, there are about 1 to 1.2 million people directly affected, not including family members. There is much work to be done to comply with the Act.



There are clinical resources in Pennsylvania that provide expertise in the diagnosis and treatment of rare diseases.

Pennsylvania joined Connecticut, Illinois, North Carolina and Rhode Island in establishing a Rare Disease Advisory Council.

The Children's Hospital of Pittsburgh has its own Center for Rare Disease Therapy.

There are clusters of rare diseases in certain populations and in certain regions of Pennsylvania, most notably for inherited disorders, e.g., Crigler-Najjar syndrome among Amish and Mennonite children in central Pennsylvania. The Clinic for Special Children with sites in Strasburg and Belleville is a resource for rare diseases in these populations.

NORD maintains a Pennsylvania Legislative Scorecard listing congressional representatives, as well as a chart with their voting record on bills relating to rare diseases.

In 2017, 77 novel drugs were approved by the FDA, of which 18 were for rare diseases, and an additional 59 existing drugs received FDA approval for a rare disease.^{3,4} There are an estimated 450 FDA-approved drugs that treat some of the 7,000 rare diseases, yet most remain without any treatment at all. In addition, there are few non-drug therapies with little exploration or research for new therapies.

To comprehensively address rare diseases, there remains much to be known, such as Pennsylvania's incidence and prevalence, access to diagnosis/treatment, availability and cost of medications/treatments, health insurance coverage, specialized care centers (with resources such as clinical,

infusions/therapy, gene therapy, rehabilitative and genetic counseling), as well as availability of social support for patients, families and caregivers.

Priority Goals and Strategic Objectives

At an early stage, the Council identified six priority goals and the strategic objectives for their attainment. This high-level direction setting will guide specific future activity in Year Two and beyond. The goals are:

1. Develop an epidemiological structure to characterize the incidence and prevalence of rare diseases;
2. Develop a comprehensive and flexible framework for rare disease needs assessment;
3. Provide a comprehensive and accessible source of information on rare diseases in Pennsylvania;
4. Develop systems to streamline the diagnostic evaluation, management and support for those with rare diseases;
5. Promote collaborations with rare disease organizations, advisory councils, the medical community and academic medical centers; and
6. Enhance access to new and existing therapies, programs and services to improve quality of life.

Goal 1: Develop an epidemiological structure to characterize the incidence and prevalence of rare diseases

Each of the approximately 7,000 known rare diseases is individually uncommon. However, when considered in the aggregate, rare diseases affect an estimated 7 to 8 percent of the population, making their impact relatively common (International Rare Diseases Research Consortium).⁵ In addition, more than 250 new rare disorders are identified annually. If Pennsylvania is representative of the U.S. population, there are about 1 to 1.2 million individuals directly affected in Pennsylvania. Upon completion of a comprehensive assessment, this estimate could be higher when Pennsylvania's refugee and Plain populations are included.

According to the Pennsylvania Department of Health, the incidence and prevalence of rare diseases in Pennsylvania is not known (beyond the rough estimate provided above). Act 14 of 2017 compels the Rare Disease Advisory Council (RDC) to report within two years of the Council's inception "the incidence and prevalence of rare diseases within this Commonwealth." The Council will collaborate with multiple organizations across Pennsylvania to develop an epidemiological instrument(s) to characterize the incidence and prevalence of rare diseases in the commonwealth.

Strategic Objectives

- Develop the most effective structure for accurate assessment of the incidence and prevalence of rare diseases with the assistance of an epidemiologist
- Develop a budget for implementation of the epidemiological instrument(s) developed

- Develop a plan to ensure that once established, the estimates of incidence and prevalence are updated annually
- Develop an economic estimate of the costs associated with rare diseases and recommend incentives for achieving the best outcomes while eliminating unnecessary expenses

Potential Measures of Success

- Comprehensive and sustainable assessment of the incidence and prevalence of rare diseases
- Development of a system of reporting current and new cases of rare diseases
- Development of an instrument(s) that can be adopted by other states

Potential Challenges or Barriers to Goal Attainment

- Budget and staffing
- Sustained participation from medical professionals
- Lack of consistency in diagnostic vocabulary to describe rare disease phenotypes (symptoms). Only 500 out of 7,000 rare diseases have disease-specific ICD-10 codes (International Classification of Diseases)

Future Activities

- Consult with state and local epidemiologists as well as the Pennsylvania medical community to develop an inclusive and transparent process for characterizing the incidence and prevalence of rare diseases in Pennsylvania
- Develop budget and funding plan

Goal 2: Develop a comprehensive and flexible framework for rare disease needs assessment.

Currently, there is limited information related to the health care needs of patients with rare diseases in Pennsylvania. While there are some common areas of concern, it is of critical importance that we avoid making too many assumptions regarding the needs and priorities of these individuals and their loved ones. Thoughtful needs assessments will clarify priorities.

We do understand that patients with undiagnosed rare diseases often experience delays in obtaining access to specialized diagnostic consultation. Primary Care Providers may have insufficient knowledge or resources to advance the care of these patients in a timely manner, resulting in delayed diagnosis and treatment. For patients and caregivers, information on rare diseases may be fragmented or confusing. These factors often lead to prolonged physical distress, emotional trauma, and financial hardships for patients and caregivers desperate for a diagnosis.

Once a diagnosis of a rare disease is made, treatment and medication costs may be prohibitive, and health care coverage may be insufficient or nonexistent. Specialized care and treatment services are often scarce, leading to geographic challenges requiring long distance travel to obtain care. Many rare diseases have no specific treatment, leading to poor quality of life for patients and caregivers.

While specific treatment for rare diseases will vary, many rare disease patients require lifelong care. Parents of children with rare diseases are often faced with significant challenges in the provision of care. Supportive care needs of both adults and children with rare diseases include informational, physical, emotional, social, psychological and other practical needs, such as insurance coverage, financial and transportation assistance.

Strategic Objectives

- Develop a health care needs survey for children and adults with rare diseases
- Implement a health care needs survey in collaboration with other organizations
- Summarize and analyze quantitative data from a health care needs survey based on geographic areas throughout Pennsylvania

Potential Measure of Success

- Completion of a comprehensive health care survey to determine needs of patients and caregivers with rare diseases in Pennsylvania

Potential Barriers to Goal Attainment

- No funding or personnel currently exists to survey and compile information.
- Patients and/or organizations and advocacy groups may be reluctant to share information due to HIPPA concerns.
- Survey participation may be impacted by high levels of stress in affected persons and families.
- Relatively small numbers of patients with rare or ultra-rare diseases may not adequately represent needs of every patient group.

Future Activities

- Draft a health care needs survey in consultation with Pennsylvania Department of Health, NORD and other rare disease organizations
- Develop budget and funding plan

Goal 3: Provide a comprehensive and accessible source of information on rare diseases in Pennsylvania

No comprehensive source of information on rare diseases currently exists for the commonwealth. A scarcity of rare disease experts and scattered resources

contributes to diagnostic delays and challenges with access to care. Greater awareness of the scope and impacts of rare diseases by the public, health care providers and public officials is needed to improve the diagnosis, treatment, and health outcomes for persons with rare or undiagnosed rare diseases.

Strategic Objectives

- Create a user-friendly website as a centralized resource for persons with rare diseases, health care providers and the public. This should include:
 - Specific rare disease information for patients and health care providers;
 - Resources regarding insurance coverage;
 - Support for patients and caregivers;
 - Link to Rare Navigator System (see Goal 4 below);
 - Information on research and clinical trials;
 - Increasing awareness via campaigns and education of the public, health care providers, public officials and other stakeholders about many aspects and impacts of rare diseases in Pennsylvania;
 - Providing education targeted to health care providers; and
 - Reporting actionable items to the Pennsylvania legislature.

Potential Measures of Success

- Website metrics
- Survey results of stakeholders at baseline and at reassessments
- Quantify engagement of collaborating organizations and industries

Potential Challenges or Barriers to Goal Attainment

- Funding and personnel to mobilize resources, foster collaborations and centralize information about available resources
- Duplication of efforts from state to state, each with relatively small numbers of persons with each rare disease
- Costs related to awareness campaigns and educational programs

Future Activities

- Develop scope, content and budget for website
- Apply for and secure funding for website design, development, maintenance and advertising

Goal 4: Develop systems to streamline the diagnostic evaluation, management and support for those with rare diseases

Timely diagnosis of rare diseases is important to developing the most effective treatment plan and is often carried out by specialists in tertiary care centers (large research hospitals). Pre-symptomatic diagnosis through newborn screening and

other systematic approaches have proven very effective in preventing catastrophic brain damage in several rare metabolic disorders (e.g., phenylketonuria [PKU]). However, diagnosis and treatment of many rare diseases, especially those less well known to primary care physicians and not in the common screening panels, are often delayed.

This delay, often referred to as the diagnostic odyssey, can last from months to years and results in prolonged physical, emotional and financial distress for patients and families. Next-generation approaches to diagnosis of rare diseases, including exome and genome sequencing, hold great promise in reducing the diagnostic odyssey. New treatment approaches, like gene therapy, will undoubtedly positively impact the management of rare diseases. To facilitate access to these vital resources, we propose the establishment of systems to bridge the gap between primary and specialty care and streamline the diagnostic evaluation, management and support for those with rare diseases.

In our first effort in this domain, we propose to develop a system of rare disease navigators (nurses and genetic counselors, for example) who assist individuals and their support persons in navigating the diagnosis and care needed for a family member with a rare disease. The navigators will favorably impact the diagnostic odyssey and care for individuals with rare diseases by connecting expertise in specialty clinical care and translational research with individuals/support persons on a case-by-case basis.

This team-based approach, in which navigators form an essential bridge between the patient and specialized care for complex diseases, has proven very effective in the U.S. and Canada in cancer care,⁶ including neuro-oncology,⁷ in managing heart disease,⁸ in complex pediatric diseases,⁹ and in the diagnosis and management of sickle-cell disease.¹⁰ We plan to extend these successes to the diagnosis and management of rare diseases in Pennsylvania.

Strategic Objectives

- Develop and implement a statewide Rare Disease Navigator System (Navigator System) to do the following:
 - Streamline diagnostic evaluation, management and support for those with rare diseases by connecting individuals/families and their primary care provider on a case-by-case basis directly with specialist(s) with appropriate experience
 - Develop an online diagnostic database for rare diseases for primary and secondary care professionals to make direct contact with appropriately experienced specialists
 - Advocate for individuals with rare diseases, their families and their health care providers as liaisons with health insurers to ensure prompt access to cutting-edge diagnostic approaches and treatments

- Identify cost savings associated with efficient diagnosis and management of rare diseases

Potential Measures of Success

- Individual/family feedback on use of the Navigator System through interviews and surveys
- Referral physician feedback on use of the Navigator System through interviews and surveys
- Quantitative measures of time and cost savings developed by the Navigator System professionals

Potential Challenges or Barriers to Goal Attainment

- Need for collaboration with Pennsylvania hospital systems and insurers, as well as private foundations for funding
- Funding
- Participation from primary and secondary care professionals
- Full collaboration from statewide health care providers

Future Activities

- Develop a strategic plan and prospectus articulating the goals and objectives of the Rare Disease Navigator System and the budget necessary to launch the program
- Use the prospectus to solicit start-up funding

Goal 5: Promote collaborations with rare disease organizations, advisory councils, the medical community and academic medical centers

To best serve the needs of individuals with rare diseases, their families and the medical professionals who care for them, we will develop collaborations with organizations that will enable us to establish best practices for rare disease diagnosis, treatment, management and advocacy in Pennsylvania. A subcommittee of the Council has already begun this process through initiation of an ongoing conversation with the NORD. The subcommittee gained significant insight into the needs and priorities of those with rare diseases in Pennsylvania from NORD, and we will look to invite NORD to meet with the full Council to facilitate a discussion of rare disease organizations based in Pennsylvania.

Collaborations with NORD, other rare disease organizations and advisory councils, the medical community, and academic medical centers will allow us to share information, resources, best practices and advance knowledge of rare diseases. Advocacy efforts by a variety of organizations have advanced awareness of rare diseases. Acquiring knowledge, research and experience from these organizations

will be invaluable in the creation of our own model to support persons with rare diseases throughout the commonwealth, in all aspects of their lives.

Strategic Objectives

- Develop a collaborations subcommittee of the Council that will work with regional, state and national rare disease organizations to keep the Council informed about best practices and disseminate the knowledge and approaches we develop
- Develop long-term, self-sustaining relationships with individuals and organizations within the rare disease community, such as, but not limited to, researchers, advisory groups, advocates, patient groups, practices, pharmaceutical and insurance industry organizations, etc.

Potential Measures of Success

- Council member surveys regarding value of collaborations
- Quantifying engagement of collaborating organizations and industries

Potential Challenges or Barriers to Goal Attainment

- Travel is often difficult for some, individuals/families with rare diseases
- Funding of initiatives

Future Activities

- Collaborations Subcommittee of the Council will be formed to work with regional, state and national rare disease organizations to keep the Council informed about best practices and disseminate the knowledge and approaches we develop.

Goal 6: Enhance access to new and existing therapies, programs and services to improve quality of life.

The Commonwealth of Pennsylvania does an excellent job of working with the research community, the hospital network and pharmaceutical/biotech companies to incentivize the development of medical therapies, including those for treating rare disease. However, once a product is approved for licensure by the U.S. Food and Drug Administration, residents of the commonwealth, especially those with rare diseases, are faced with difficulties in accessing the ideal therapy to meet their medical needs. These difficulties are largely driven by costs. Two major questions that typically drive such access are:

1. How can public and private insurers meet the medical needs of their beneficiaries while doing so at the lowest cost possible?

2. How can patients access rare disease therapies, which are commonly very expensive, without large out-of-pocket and co-insurance responsibilities or financially hampering insurance plans (whether private plans, state employee plans or state Medicaid)?

These questions are not limited to access for rare disease therapies. However, the rare disease patient typically faces increased barriers due to specialty tier consideration, higher out-of-pocket coinsurance, and cost containment hurdles such as formularies and expensive therapies without much competition.

Clearly, it is vital for those with rare diseases to access life-saving innovative treatments that will prolong and enhance their lives. This Council will be putting forward recommendations that we believe will improve the environment by defining a reasonable level of evidentiary support for making medical necessity decisions when populations are small. This will enable rare disease patients to have greater access to life-saving therapies without the persistent ordeal of navigating through different standards of evidence when changing insurance carriers. Additionally, a guidance document would make the process more transparent and consistent. Such recommendations, we believe, will recognize and promote the value of these therapies for positive health outcomes without adding undue cost to the health insurance system or deterring pharmaceutical innovation.

Strategic Objectives

- Advance measures that pharmaceutical/biotech companies selling product within the commonwealth can employ, which would allow for greater access to rare disease therapies
- Promote recommendations that would ease the insurance coverage burdens for rare disease therapies that can be adopted by public and private insurers within the commonwealth
- Create policy proposals for the General Assembly to consider that would improve the access environment for rare disease therapies (See Goal 4 above — Rare Disease Navigator System.)
- Advocate for initiatives that will ensure patient access to expertise that will shorten the diagnostic odyssey and reduce misdiagnosis and ineffective therapies (see Goal 4 above—Rare Disease Navigator System.)
- Recognize the value of rare disease therapies for patients instead of being focused solely on costs. Addressing the value and impact of proper treatment with innovative therapies and new technologies will bring down long-term health care costs.

Potential Measures of Success

- Commonwealth-based and national patient group endorsement of recommendations; positive feedback after enactment that the rare disease community faces a lesser burden in accessing therapies than at present; surveying patient organizations within the commonwealth in 2018, 2021 and 2024 to compare results pertaining to accessibility of expertise, products and services
- Recommendations endorsed by the major trade associations active within the commonwealth (The Insurance Federation, Life Sciences PA, PhRMA, Blue Cross Blue Shield Association, etc.); willingness of each organization to be involved in policy development
- General Assembly and administration support and enactment for the recommendations put forward by this Council

Potential Challenges or Barriers to Goal Attainment

- Collaboration between the insurance and pharmaceutical/biotech sectors will be a challenge. These two sectors typically accuse one another as the driving reason why rare disease therapy access is difficult. Navigating among these two sectors to reach a middle ground of solutions for which neither sector will drive opposition is going to be a challenge.
- A significant challenge is overcoming patients' general feeling of defeat surrounding access to therapies. Rare disease patients and citizens in general may have become jaded with the existing focus on cost containment. Convincing the impacted populations to assist in this effort and having their confidence that the Council can put forward recommendations that will improve patient access could be a challenge.
- We lack the epidemiologic infrastructure that could catalyze our goals.
- Fragmentation of care and record keeping poses a challenge to streamlining care.
- The Council currently lacks the funding to research and develop data to challenge and propose corrections to the existing norms.

While these are definite challenges, overcoming these potential barriers is well worth the time and effort. Greater access to rare disease therapies for impacted patients will result in less hospitalization, greater health outcomes, reduced overall health care costs, and productive citizens contributing to the economy and well-being of the commonwealth.

Future Activities

- Form a subcommittee of the Council to work with all stakeholders to develop and prioritize recommendations for policies that will enhance access to new and existing therapies, programs and services to improve quality of life

Collaboration Opportunities

The Council recognizes that partnerships and strategic alliances will be key to advancing the six priority goals over the next several years. Examples of such collaboration include the following:

Input and support from both the insurance and pharmaceutical/biotech sectors will be necessary for the success of Council recommendations. Engaging feedback on proposed recommendations as well as counterproposals will be sought and encouraged.

Participation from patients and patient organizations can highlight the levels of health care and treatment access difficulties at present. Feedback from patients on proposed recommendations as to whether they will have direct benefit on their abilities to access diagnosis, therapies, expertise and services will be sought.

Economic organizations can provide input as to potential costs and benefits associated with proposed recommendations. Financial analysis on budget impact will be necessary to understand likelihood of proposals being adopted.

In addition, there is no shortage of organizations with a strong stake in our work. The following list represents some of the many stakeholders that the Council expects to collaborate with going forward:

- Pennsylvania Department of Health
- Pediatric and Adult Rare Disease Patient Populations
- Patient and parent support groups
- Pennsylvania medical community/academic medical centers
- Hospital and Healthsystem Association of Pennsylvania (HAP)
- Pennsylvania Medical Society
- National Organization for Rare Disorders (NORD) and Global Genes
- Genetic and Rare Disease Information Center (GARD) of the National Institutes of Health (NIH)
- Rare disease advisory councils in other states (e.g., NC, CT, RI)
- Rare disease organizations and advocacy groups
- Pennsylvania professional societies and associations
- Health insurance industry
- Pharmaceutical industry
- Health and Human Services Committee, Pennsylvania Senate
- Health Committee, Pennsylvania House of Representatives
- Caregivers
- Primary care providers
- Rare disease medical specialists
- Homecare agencies

- Food and Drug Administration (FDA)
- Pennsylvania Health Care Cost Containment Council (PHC⁴)

Year One Accomplishments

A necessary and crucial requirement in the development and coordination of services to assist a population is its definition and an evaluation of the baseline state. This permits a measurement of outcomes, a focus on determinants of those outcomes, and the elaboration of policies and practices that can improve health, recognize and address disparities and catalyze strategies to impact safety, cost and clinical outcomes.

The Council undertook an evaluation of what statistics are available relating to rare diseases and discussed the development of a framework for addressing the expansive number of rare diseases.

The Act directs the Council to build relationships with the General Assembly and all relevant state and private agencies that provide services to, or are charged with, the care of individuals with rare diseases. The Council believes it is important for the General Assembly to be updated about our work and to engage in discussions about public policy options to address the rare disease community. However, the Council determined that the resource challenge for Council members to engage with the vast array of private agencies that provide services to individuals with rare diseases was prohibitive.

Another responsibility of the Council set forth in the Act is to coordinate the performance of the Council's duties with those of other rare disease advisory bodies, community-based organizations, and other Pennsylvania public and private health organizations to facilitate greater cooperation between and among them. Building and maintaining these relationships was identified as one of the Council's key roles.

Finally, the members of the Council are dedicated to fulfilling the Act's requirement to regularly communicate results of the Council's work, including the identification of best practices and potential policy recommendations.

Rare Disease Advisory Council Formation

A total of 25 Council members were identified and appointed by the Secretary of Health to ensure broad-based representation from patients, advocates, state government, academia, health practitioners, the insurance industry, the pharmaceutical industry and health policymakers. Each of the Council members brings perspective and personal challenges with rare disease, research insight, health care delivery expertise and health policy background. The Council has compiled a brief biographical sketch and photograph of each Council member to increase their familiarity with each other and to make available to the public the Council members and their scope of expertise.

Members of the Rare Disease Advisory Council

Rare Disease Advisory Council Meetings

The Council met on several occasions: October 3, 2017, February 7, 2018, April 3, 2018, May 16, 2018, and June 19, 2018. Due to inclement weather, the February 7, 2018, meeting was held via telephone. Council members deemed it important that the participants meet in person to build relationships among its diverse membership and to facilitate Council members' discussion. A telephone conference call option was provided for all meetings except for October 3, 2017 and June 19, 2018.

Strategic Direction through 2025

The members developed three main focus areas for the year's efforts. These were communication, advocacy and surveillance. These were refined from a large number of singular issues, questions and objectives. The members were grouped by these categories to define the main components of this report: a baseline assessment and long-term goals.

Members of the Council then developed a series of priority goals and specific strategies to achieve those goals that encompasses the eight-year duration of the Council's mandated tenure. This was followed by the creation of working groups based on membership expertise to develop each of the goals.

Resources

The Council recognized the significant challenge of funding to pursue these activities. The Council discussed multiple options in this regard and will work with its agency representatives to navigate public and private funding streams and, in addition to other

sources, will likely make recommendations to the legislature for funding to pursue the Council's planned activities.

Rare Disease Day—June 19, 2018

A key component of the Council's mission is to educate the public and raise awareness about rare diseases. The nationally recognized Rare Disease Day occurred on February 28, and there were events held in state legislatures across the country. Repeated snow interference led Pennsylvania to postpone its observance. Pennsylvania Rare Disease Day was celebrated on June 19 in Harrisburg with a legislative lobby day for patients and advocates at the State Capitol as well as a press conference.

Advisory Board Branding Logo

The Council secured a logo to enable a distinct and recognizable branding of the Council materials and reports. A logo will serve to distinguish Council documents to identify the Council and signal its identity. The branding logo was donated by The Conley Cushing's Disease Fund.



Future Activities

As the Council moves ahead into its second year, its mission to improve the quality of life for all those affected by rare diseases in Pennsylvania remains clearly in focus. The Council has a framework upon which to build for the future with its six goals and supporting strategic objectives. They are critical to the success of this future, and they are achievable. The need to identify and secure funding for many of those strategies and their inherent tactics cannot be overemphasized. While the Council statutorily has the authority to seek and accept most funding, mechanisms must be put in place to do so. As has been mentioned previously, the Council is acutely aware of the magnitude of this issue, and it will be prominent in every future discussion.

Further, Section 6(b) of the Rare Disease Advisory Council Act specifically states that the Council must provide a comprehensive report in July 2019 that, at least, includes the following:

1. The incidence and prevalence of rare diseases within the commonwealth

Subsequent biennial reports will follow with Council findings and recommendations related to the management, monitoring and surveillance, education, detection, diagnosis, information, and care regarding rare diseases in the commonwealth. Such findings and potential recommendations will include the quality and cost effectiveness of care, access to treatment and other relevant services for individuals affected by rare diseases.

The Year One activities of the Council resulted in the development of six long-term goals identified in this report. The pursuit of these goals will allow the Council to report back in Year Two on the incidence and prevalence of rare disease within the commonwealth.

The Council is committed to developing an epidemiological structure to characterize the incidence and prevalence of rare diseases. The Council will work to determine the best methods and practices for such data collection and compare with what is being done in other states and at the federal level. Collaboration with national patient organizations, such as NORD and known patient organizations within the commonwealth, will occur to confirm whether collected data reflects the recognized understanding of incidence or challenges the conventional wisdom. This activity will directly address the first of two items outlined by the Act for the Year Two report. Further, this structure will act to identify rare disease incidence and prevalence in the commonwealth for future years.

2. The needs of the rare disease community within the commonwealth and the actions necessary and feasible to address those needs

To identify the needs of the rare disease community, the Council is going to undertake several measures:

- Develop a comprehensive and flexible framework for a rare disease needs assessment. Such an assessment will specifically identify the needs of the rare disease community in the commonwealth. The items listed below will assist in this regard.
- Provide a comprehensive and accessible source of information on rare diseases in Pennsylvania. This resource will serve to educate citizens, health officials and those with rare diseases about resources that are available within the commonwealth. This source will also help to identify current needs and knowledge gaps in addition to potential means to address.
- Develop systems to streamline the diagnostic evaluation, management and support for those with rare diseases. This will begin to address an area of significant need within the rare disease community, which is the delay in diagnosis and management of a treatment protocol for someone with a rare disease. The Council will explore methods to streamline this process. As with most diseases and conditions, an earlier diagnosis and treatment results in greater health outcomes for the individual and reduced costs through less hospitalization.
- The continuous, systematic collection, analysis and interpretation of NBS data is essential to monitor and clarify the epidemiology of health problems detected by NBS, which will enable the establishment of priorities and inform public health policy and strategies. This will also allow for some evaluation of the success of NBS with respect to rare diseases. Coordination of the Council's efforts with the Pennsylvania Department of Health Newborn Screening Advisory Board will ensure timely updates to newborn screening for medically actionable rare diseases.
- Promote collaborations with rare disease organizations, advisory councils, the medical community and academic medical centers. With engagement of other organizations, the Council will better understand the needs of individuals with rare diseases and best practices that can be replicated in the commonwealth.
- Enhance access to new and existing therapies, programs and services to improve quality of life. The Council will explore methods that can ensure therapies treating rare diseases have as smooth a pathway as possible for citizens of the commonwealth to access. The Council will explore ways to build upon the excellent work of the commonwealth in supporting biotech and pharmaceutical development within the state and recommend methods to ease barriers for accessing rare disease therapies once those therapies are developed and ready for market. Such barriers can include, but are not limited to, insurance coverage and reimbursement of therapies, overall cost of therapies, patient out-of-pocket requirements and patient assistance programs.

Closing

To conclude, the Council is excited to begin to achieve the goals outlined in this preliminary report. The pursuit of identifying the incidence and prevalence of rare diseases within this commonwealth is necessary. Identification is only the beginning. The needs of the rare disease community and the actions necessary and feasible to address those needs are what will improve the lives of those with rare diseases living within the commonwealth.

The Council does not have misconceptions regarding the entirety of issues to be addressed, nor does the Council presume to have all the solutions to these issues. However, the Council does resolve to work toward making Pennsylvania a national leader in transforming the lives of those affected by rare diseases through collaboration, support, education and advocacy.

Knowledge is power and the Council will endeavor to provide it to both the commonwealth and its citizens. The Council will hope to act upon a common phrase within the rare disease community: “Out of the darkness ... into the light.”

Council Membership

Council members come from myriad backgrounds across the commonwealth. What follows are the members, followed by member biographies:



Dr. David Kelley, Chief Medical Officer for the Pennsylvania Department of Human Services' Office of Medical Assistance Programs

– Dr. Kelley oversees the clinical and quality aspects of the Medical Assistance Programs that provide health benefits to over 2.5 million recipients. The office includes oversight of eight managed care organizations and the access fee-for-service program. In the past 10 years, the office has lead multiple, significant processes and patient-oriented improvements to maximize efficiencies and improve the health of the health benefits recipients. Prior to joining the department, Dr. Kelley worked for Aetna Health Inc. as the medical director responsible for utilization and quality management in Pennsylvania, served as assistant professor and director of Clinical Quality Improvement at Penn State University's College of Medicine, and clinically practiced at a FQHC, private practice and a community-based team approach to diabetes care in a Medicaid hospital clinic. Dr. Kelley received his B.S. degree at Elizabethtown College, completed medical school at the University of Pittsburgh and his residency training at Baylor College of Medicine in Houston, obtained his MPA at Penn State University, and is board certified in internal medicine and geriatrics.



Patrick Collins, Senior Director for North American Healthcare Policy and External Affairs at CSL Behring

– CSL Behring is a King of Prussia-based manufacturer of therapies for the treatment of multiple rare diseases. Mr. Collins has direct responsibility for CSL Behring's North American government affairs program and has been employed with the company for 17 years in positions of growing public policy responsibility. Prior to joining CSL Behring, Mr. Collins served as director of government relations for the National Hemophilia Foundation. He has also worked in New York City and state government. Mr. Collins lives in Exton, Pennsylvania with his wife and daughter.



Marie Conley, Conley Consulting, LLC

– Marie Conley is the founder of The Conley Cushing's Disease Fund which was established on July 17, 2014, and is a project of The Foundation for Enhancing Communities, fiscal sponsor. The funds raised are used in part to create awareness, advocacy and support for patients and their loved ones who are suffering from this disease, as well as support institutions and organizations focused on research and treatment surrounding Cushing's Disease. In 2012, Marie was diagnosed with Cushing's Disease – a disease so rare it affects less than 10 people per million each year. She is also adrenal insufficient. Marie Conley is a consultant focusing on engagement and stakeholder strategies for a variety of clients through her company Conley Consulting, LLC. Marie hails from Bucks County, Pennsylvania; she lives in Elizabethtown, Pennsylvania with her husband Chris Lammando and their son Carter.



Nick Slotterback, Health and Physical Education Advisor, Pennsylvania Department of Education

– As the representative from the Department of Education, he has a personal, common interest with the council in that his godson has a rare disease and his wife's cousin is the state ambassador for Rare Action Network in South Carolina. Their son was born with a rare disease and Mr. Slotterback is working to include rare disease education in the upcoming standard revision for Academic Standards for Health, Safety and Physical Education.



Evelyn O. Talbott, DrPH, MPH, Epidemiologist and Professor, Department of Epidemiology, Graduate School of Public Health, University of Pittsburgh – Dr.

Talbott has conducted numerous epidemiologic investigations and provided teaching in environmental epidemiology and has served on numerous committees at the Pennsylvania Department of Health. She is a founding member of the international Society for Environmental Epidemiology, serving as their secretary treasurer for four years, and is a fellow of the American Heart Association Council on Epidemiology and Prevention. She recently served as director of the CDC-funded Academic Center of Excellence in Environmental Public Health Tracking. She is dedicated to furthering ALS research in both treatment and identification of potential risk factors, as well as for all rare diseases. Most recently, she has focused on air pollution and neuro-cognitive and neurodegenerative diseases. These diseases include childhood autism and neurotoxic exposures, risk factors for ALS, and the risk of childhood lead poisoning.



Jennifer H. Wescoe, M.Ed., NCC, Executive Director, Founder, Wescoe Foundation for Pulmonary Fibrosis – Ms. Wescoe's father, Ron Wescoe, was diagnosed with

Idiopathic Pulmonary Fibrosis (IPF) in November 2003. He passed away 11 months later. There was no support, resources or education regarding this incurable lung disease. As a family, they felt lost, alone, confused and, frankly, scared of what her Dad had to endure, struggling to breathe every day. Not only did he struggle to breathe and deal with this serious lung disease, it affected the entire family dynamic, as Ms.

Wescoe's family absorbed roles of caregiver, educator, leader and observer. She and her family know how vital education and support is with any disease, let alone a rare disease. She has made it her mission to provide support, education and resources for patients and their families living with pulmonary fibrosis and created their non-profit 501(c)3 non-profit organization, Wescoe Foundation for Pulmonary Fibrosis. Today, Ms. Wescoe has the privilege of working with Pennsylvania Department of Health and an incredible group of people who have the same drive and determination to help others in Pennsylvania living with a rare disease. She believes the Council will have a profound and effective impact on patients and their families coping with a rare disease.



Joseph Coyne, M.Ed., LBSC, Executive Director, Garrett the Grand - Batten Fighter – First and foremost, Mr. Coyne is a father and advocate for his son Garrett

who is affected by Batten Disease. They started a non-profit to fund research, raise awareness and support others in the community with rare disease. Current projects consist of beginning a wellness program to run with or for those who are unable to run themselves due to disabilities. He is passionate about progressing rare disease and making Pennsylvania the leader for the rare disease community.



Elaine A. Davis, MSN, RN, CCM, Clinical Coordinator, Division of Infectious Diseases and Epidemiology, Penn State Hershey Medical Center – Ms. Davis holds a Bachelor

and Master of Science in Nursing degrees from the Pennsylvania State University. She has over 30 years nursing experience as a staff nurse, medical quality, hospital utilization and care coordinator. Her personal stake in rare diseases is the loss of her sister to Amyotrophic Lateral Sclerosis (ALS) two years ago. During the time her sister lived with ALS, her family struggled to provide the care and services she needed.



Jessica L. Deary – Ms. Deary is a patient speaker, individual patient advocate, legislative ambassador, patient advisor to organizations and a writer. Ms. Deary's education and professional experience began in the corporate sector, during which time she obtained an M.B.A. and worked in strategic management. Drawn to patient advocacy through her own experiences as a long-standing patient with a rare, complicated and incurable medical condition, her diverse background, experience and unconventional journey has blessed her with numerous opportunities to act in many differing roles -- from provider to patient and

many points in between. These differing perspectives and her own experiences as a patient in need have

resulted in an unyielding passion to improve patient care and promote a more positive workplace culture. This passion helped her to redirect her professional endeavors toward aiding other patients to obtain adequate medical care and to helping organizations create fiscally advantageous operational strategies to improve care for patients. Ms. Deary continues to expand her knowledge in patient experience and patient-centered care and recently received a certificate in patient advocacy. She is currently working towards her RN degree.



Nicholas DeGregorio, M.D., FACP, MMM, Senior Medical Director, UPMC for you –

Dr. DeGregorio is currently the Senior Medical Director for UPMC for You and provides support for UPMC Health Plan provider credentialing, opioid management and value-based payment/shared savings programs. He practiced general internal medicine for approximately 20 years before joining UPMC Health Plan in 2004, where he previously worked as medical director in the areas of medical management, network development and quality improvement. Dr. DeGregorio's areas of special interest include developing patient-centered process improvement strategies to reduce disparities while improving efficiencies and improving the health of members. In his work as a Medicaid medical director he developed experience and interest in the many issues and problems faced by members with rare conditions. In his role on the Pennsylvania Rare Disease Advisory Council, his goal is to help advance knowledge and reduce both medical and social barriers to timely diagnoses and treatment for patients with rare diseases.



Connie Deline, M.D., Vice-President, Chair, Medical Advisory Board, Spinal CSF

Leak Foundation – Dr. Deline's interest in participation with the Council is three-fold: first, as a patient with a rare disorder (spontaneous intracranial hypotension); second, as a family medicine and integrative medicine physician, now retired from clinical medicine due to disability; and third, as a representative of a rare disease health advocacy non-profit organization.



Robert (Rob) Jinks, Ph.D., Professor of Neuroscience in the Department of Biology and the Biological Foundations of Behavior Program at Franklin & Marshall College (F&M) –

Dr. Jinks earned his B.S. in bioengineering and his Ph.D. in neuroscience from Syracuse University. He has held faculty positions at the University of Pennsylvania School of Medicine and Swarthmore College in addition to F&M. For the first two decades of his career, Dr. Jinks' research focused on the cellular and molecular signaling between the brain and the retina necessary to promote normal, healthy vision. In 2009-10, as a visiting associate professor at the F.M. Kirby Center for Molecular Ophthalmology at the University of Pennsylvania School of Medicine, Dr. Jinks and the Clinic for Special Children in Strasburg, Pa. developed a collaboration focused on the use of next-generation DNA sequencing approaches to identify the genetic basis of rare neurodevelopmental disorders. The collaboration resulted in several publications that have characterized multiple neurodevelopmental disorders, many of which are now studied in U.S. and foreign labs.



Sharon O'Shaughnessy, Narcolepsy patient with cataplexy, Advocacy Chair, Narcolepsy Network (national non-profit); member, Avadel Pharmaceuticals Patient Advisory Group –

Sharon is a parent to Rachel, 23, who has an undiagnosed genetic syndrome, despite exome sequencing. Ms. O'Shaughnessy earned her B.A. in psychology and education from Bucknell, and a M.A. in communication disorders and speech science from University of Colorado-Boulder. Using spectrographic speech analysis, her research employed reflexive vocal closure to improve vocal quality despite previous vocal failure through the course of neuro-degenerative diseases. As an advocate, Ms. O'Shaughnessy wrote legislation regarding students with special educational needs that was passed in Virginia, testified at FDA in 2013 and 2018 for treatments that are now making their way through approval, started a school for children with special needs learning alongside typical peers, and, for 15 years, served as a parent mentor for a CHOP support group for parents of children who are medically fragile. Ms. O'Shaughnessy has seen rare diseases from just about all angles, and she is thrilled to be serving on the Pa. Rare Disease Advisory Council.



Ann Marie Kriebel-Gasparro, DrNP, MSN, CRNP, FNP-BC, GNP-BC, Assistant Professor in Nursing, director, DNP Program Alvernia University, – Dr. Kriebel-Gasparro is dually credentialed as a family and gerontological nurse practitioner and has her Doctor of Nursing Practice (DrNP) degree from Drexel University. Her practice began seeing medically underserved patients in a HPSA in North Philadelphia as a primary care provider in the National Health Service Corps for four years, then as coordinator of the Penn Hemophilia and Thrombosis Center for four years; she also practiced in neurology, oncology and nursing homes in Pa. and N.J. for many years. Her clinical practice is as a family and gerontological nurse practitioner in home health and occupational medicine. Her interest in rare diseases is based on her practice and management of patients for many years with hemophilia, rare genetic thrombotic diseases and rare genetic neurologic diseases. Dr. Kriebel-Gasparro has taught in ADN, pre-licensure BSN, RN-BSN, accelerated BSN, BSN to DNP and post MSN to DNP programs. Previously, she was the director of the DNP Program at Temple University. Her teaching and practice centers on the development of innovative models of health care delivery; including the implementation of a didactic and immersion clinical experience in palliative care for nursing students.



Barbara E. Ostrov, M.D., Professor of Pediatrics and Medicine, Pediatric Rheumatology and Internal Medicine Rheumatology, Penn State College of Medicine, Secretary-Treasurer, Pennsylvania American Academy of Pediatrics – As a rheumatologist, Dr. Ostrov has cared for people across Pennsylvania affected by the breadth of rare autoimmune and genetic diseases that cause arthritis and related conditions in children and adults. Her work has focused on education and increasing the awareness, advocacy and research collaboration needed to address these disorders and their impact on the lives of Pennsylvanians. Her stated purpose on the council is to further these goals.



Bret Yarczower M.D., MBA, Senior Medical Director, Health Services Operations - Dr. Yarczower responsibilities with Geisinger Health Plan include health care services and technology. In his role, he chairs committees evaluating the safety and efficacy of new and evolving medical and surgical technologies, and pharmaceuticals. Dr. Yarczower has been with Geisinger Health System for more than 24 years, beginning as a pediatrician. He joined Geisinger Health Plan as an assistant medical director in 2001. Dr. Yarczower earned his B.A. from Temple University and received his M.D. from the Medical College of Pennsylvania, interned and completed his residency at the University of Michigan and received his MBA from the University of Massachusetts. He is a fellow and board certified by the American Academy of Pediatrics. Dr. Yarczower completed the Executive Leadership Program from AHIP, America’s Health Insurance Plans. He has received awards for outstanding patient satisfaction and community service.



Anna Payne – Cystic Fibrosis Patient - As a patient with a rare disease this is personal to Ms. Payne. She knows firsthand the struggles our community faces, as she experienced them herself. She advocates at the federal, state and local levels to help legislators and other leaders to understand how legislation will impact our community. Ms. Payne wants to improve the lives of as many patients as possible, and, for those living with a rare disease, this is a time-sensitive matter. She most recently ran for local office and was elected in November 2017 as the auditor of Middletown Township. Ms. Payne believes it is well past the time that those who are capable step up and make their voices heard.



Elizabeth Rementer, Deputy Director for Pa. Insurance Department - Elizabeth Rementer is the Deputy Director of Communications for the Pennsylvania Insurance Department. She previously worked for more than a decade as a press secretary in the Pennsylvania Senate Democratic Caucus and as a reporter for a community newspaper in Philadelphia. She is a graduate of Penn State University with a bachelor’s degree in journalism. A mother of twin sons with a rare neurological speech disorder called apraxia, Elizabeth is also an advocate and volunteer for numerous Central Pennsylvania organizations and activities that support children and adults with special needs, including their families.



Kathryn Lavriha, Director/lead of state government affairs for Sanofi, U.S. responsible for DC, Md., Pa., Va., W.Va.

Ms. Lavriha lobbies on health care industry initiatives and builds relationships with state legislators, governors and other health policy makers. She is responsible for strategically positioning the company as an industry leader both in the government and political arena. Ms. Lavriha has over 25 years' experience in state and federal government affairs. Prior to joining the Sanofi, she was senior vice president of federal and state government affairs for the International Mass Retail

Association and directed the State Government Affairs Department at the National Association of Chain Drug Stores (NACDS). Ms. Lavriha received her B.A. in political science from SUNY Geneseo and her MPA from George Washington University. Currently, she resides in Stevensville, Md., with her husband and has two grown children.



Andrew Herlich DMD, M.D., FAAP, FASA, Professor and Special Assistant to the Chair for Academic and Faculty Affairs in the Department of Anesthesiology at the University of Pittsburgh School of Medicine

Dr. Herlich is a pediatric anesthesiologist with a significant interest in anesthesia for patients with congenital syndromes, as well as for patients of all ages with neuromuscular diseases. His publications have been largely devoted to this area, as such he is well positioned for significant contributions to the

beneficiaries of the Council's work.



Can (John) Ficicioglu, M.D., Ph.D., Associate Professor of Pediatrics at the Children's Hospital of Philadelphia, Perelman School of Medicine at the University of Pennsylvania

Dr. Ficicioglu is director of the Newborn Metabolic Screening Program and Lysosomal Storage Disease Center at The Children's Hospital of Philadelphia with expertise in newborn metabolic screening, lysosomal storage disorders, intermediary metabolism defects, galactosemia and PKU. He is developing best practices in diagnostic algorithms and outcomes of inborn errors of metabolism detected through newborn screening. Dr.

Ficicioglu received his M.D. from the University of Istanbul, Cerrahpasa Medical School, completing his internship and residency in pediatrics and his Ph.D. in histology and embryology from the University of Marmara, Istanbul. Previously an associate professor in pediatrics at the Cerrahpasa Medical School, he completed his genetics fellowship at the Children's Hospital Boston, Harvard Medical School.



William C. Welch, M.D., FACS, FICS, Chair, Department of Neurosurgery at Pennsylvania Hospital

Dr. Welch received his medical degree from SUNY/Downstate, School of Medicine and completed his residency at the University of Rochester Medical Center and a fellowship at Montefiore Medical Center. Dr. Welch's memberships include multiple international and national boards, as well as colleges and associations, including the National American Association of Orthopaedic and Neurological Surgeons and the

International Neurosurgical Society of America. Dr. Welch was named to the Philadelphia magazine's 2013 Super Doctors list and has been recognized as one of America's Top Doctors and one of the Best Doctors in America on multiple occasions.



Tomas J. Aguilar, Director, Bureau of Health Promotion and Risk Reduction, Pennsylvania Department of Health

Mr. Aguilar joined the Department of Health in 2012 and leads the Department's Chronic Disease and Injury Prevention efforts, driven by dedicated public health education and interventions experts. The Bureau's programs use evidence-based interventions to implement policy, health system and environmental changes to impact health outcomes. He previously served as the chair of the Task Force

on Lyme Disease and Related Tick-Borne Diseases and is a past member of the board of directors as treasurer for the National Association of Chronic Disease Directors.

Endnotes

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