New York State Department of Health Rare Disease Workgroup:

Report to the Governor and Legislature

December 2024

NEW YORK STATE | Department of Health

Prepared by the Office of Public Health

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EXECUTIVE SUMMARY

Overview

A rare disease is defined by the federal Orphan Drug Act as a disease or condition that affects fewer than 200,000 individuals in the United States.[1] The National Institutes of Health (NIH) estimates that 30 million people in the United States are living with one of the 10,000 known rare diseases.[2] An estimated 1 in 10 people, or 1.8 million New Yorkers, have a rare disease.

People with rare diseases face unique challenges and health inequities. Patients with undiagnosed rare diseases often face a long, challenging, and unpredictable journey, referred to as a "diagnostic odyssey." For one-third of those with rare diseases, obtaining an accurate diagnosis can take from one to five years.[1] Once diagnosed, people with rare diseases often struggle to find treatment; unfortunately, only about 5% of rare diseases have treatments that are approved by the Food and Drug Administration.[3] The complex and disabling nature of many rare diseases also leads to significant unmet health and social needs for people living with rare diseases and their families. These challenges and barriers are compounded for people with rare diseases who are from historically marginalized and underserved communities.[4]

In response to the significant issues facing the rare disease community, Chapter 74 of the Laws of 2020, as amended by Chapter 57 of the Laws of 2022, directed the Department of Health (DOH), in collaboration with the Department of Financial Services (DFS), to convene a workgroup on rare diseases to identify best practices that could improve the awareness of rare diseases and referral of people with potential rare diseases to specialists, and to evaluate barriers to treatment, including financial barriers on access to care. DOH was directed to prepare a written report summarizing the opinions and recommendations from the workgroup and to develop a list of existing, publicly accessible resources on research, diagnosis, treatment, coverage options, and education relating to rare diseases.

Rare Disease Workgroup Recommendations

The Rare Disease Workgroup developed the following recommendations, organized around four priority areas. Additional details about each recommendation can be found in the report.

Awareness and Diagnosis

1. Increase awareness about rare diseases.
2. Increase the awareness, availability, and accessibility of resources about rare diseases for healthcare providers, patients, and families seeking a diagnosis.
3. Develop educational programs for healthcare providers, patients, families, and the broader community about rare diseases.

Access to Care and Referral to Specialists

1. Promote the recruitment, training, and retention of needed specialists in the workforce.
2. Support the recruitment, training, and retention of behavioral health providers and homecare providers.
3. Support the recruitment, training, and retention of nurses, particularly those with experience in pediatrics, to administer pediatric infusions at home and in smaller community hospitals.
4. Promote systems that support communication between patients and healthcare providers.
5. Continue support and coverage of telehealth by commercial and state-sponsored insurance.
6. Provide resources to help with travel and childcare that will allow a person with a rare disease or a parent of a child with a rare disease to attend provider appointments.
7. Decrease barriers to utilizing health insurance and provide resources to help with the cost of medications and treatments.
8. Provide resources to help people enroll in health insurance, including Medicaid.

Patient and Caregiver Support

1. Create a centralized, “one stop” directory of information and resources for people with rare diseases and their caregivers.
2. Conduct a Support Needs Survey to develop a detailed understanding of unmet needs, as well as barriers to accessing information and support.
3. Create a standing advisory group, consisting of rare disease patients and caregivers, to collaborate with other stakeholders about community needs and ways to address them.
4. Foster greater collaboration among agencies and organizations serving people with rare diseases and their caregivers to promote greater community outreach.
5. Conduct a media campaign to increase awareness of the information, support, and assistance available to the rare disease community, including the resource directory on the DOH website.
6. Use Artificial Intelligence to help patients and caregivers access and utilize information and resource support.

Emergency Preparedness

1. Develop a checklist of important health information and make it widely available to be used by individuals with a rare disease when they are experiencing a personal health emergency.
2. Provide information on the DOH website that includes contacts for local health departments to guide families regarding specific emergency preparedness tools and processes for their local region.
3. Ensure that schools have an individualized emergency preparedness plan for children with special health care needs in case of emergencies due to external or environmental conditions such as a weather-related event, terrorist event, or communicable disease outbreak.

List of Publicly Accessible Resources

Per the legislation, the Workgroup compiled a list of existing, publicly accessible resources on research, diagnosis, treatment, coverage options, and education relating to rare diseases. The list of resources can be found on page 18 of this report and on the DOH website at health.ny.gov/diseases/rare_disease/.

Conclusion

We thank all the members of the Workgroup for their significant contributions of time and expertise. The Workgroup brought together a diverse group of stakeholders with different perspectives, but with a shared commitment to improving the lives of rare disease patients and their families. The recommendations of the Workgroup involve many complex issues and, as such, will require multisector collaboration with state and federal governments, healthcare providers, payors, scientific organizations, patient and advocacy organizations, individuals, and families.

OVERVIEW OF RARE DISEASES

A rare disease is defined by the federal Orphan Drug Act as a disease or condition that affects fewer than 200,000 individuals in the United States.[1] The National Institutes of Health estimates that 30 million people in the United States are living with one of the 10,000 known rare diseases.[2] An estimated 1 in 10 people, or 1.8 million New Yorkers, have a rare disease.

Rare diseases can affect anyone at any point across the lifespan; they can be acute or chronic; and they may have many different causes. Eighty percent of rare diseases are thought to be genetic disorders for which genetic testing is often the only way to diagnose them.[5] Rare diseases include some infections, autoimmune diseases, forms of cancer, diseases caused by exposure to toxins or other environmental factors, and some conditions with multiple causes.6 For some rare diseases, the cause is not known.

People with rare diseases face unique challenges and health inequities. Typically, they will seek medical care in healthcare settings where the providers have never seen their condition before and may not recognize that their symptoms are associated with a rare disease. Some diseases may have different presentations or be more difficult to diagnose in certain populations or ethnic groups, causing inequities in diagnosis or referral. Patients with undiagnosed diseases often face a long, challenging, and unpredictable journey, referred to as a "diagnostic odyssey." For one-third of those with rare diseases, obtaining an accurate diagnosis can take from one to five years.[1] An accurate diagnosis is essential to allow people to receive appropriate clinical care and counseling. It may also allow patients and families to connect with others with similar conditions and to the rare disease community.

Once diagnosed, people with rare diseases often struggle to find treatment; unfortunately, only about 5% of rare diseases have treatments that are approved by the Food and Drug Administration.[3] The complex and disabling nature of most rare diseases also leads to significant unmet health and social needs for people living with rare diseases and their families. These challenges and barriers are compounded for people with rare diseases who are from historically marginalized and underserved communities.[4]

WORKGROUP CHARGE

In response to the significant issues facing the rare disease community, Chapter 74 of the Laws of 2020, as amended by Chapter 57 of the Laws of 2022, directed the Department of Health (DOH), in collaboration with the Department of Financial Services (DFS), to convene a workgroup on rare diseases to identify best practices that could improve the awareness of rare diseases and referral of people with potential rare diseases to specialists, and to evaluate barriers to treatment, including financial barriers on access to care.

Per the legislation, workgroup members were to have expertise in rare diseases, including physicians, nurses, and other healthcare professionals with experience researching, diagnosing, or treating rare diseases; members of the scientific community engaged in rare disease research; representatives from the health insurance industry; individuals who have a rare disease or caregivers of a person with a rare disease; and representatives of rare disease patient organizations.

DOH was directed to prepare a written report summarizing the opinions and recommendations of the workgroup and to develop a list of existing, publicly accessible resources on research, diagnosis, treatment, coverage options, and education relating to rare diseases.

WORKGROUP PROCESS

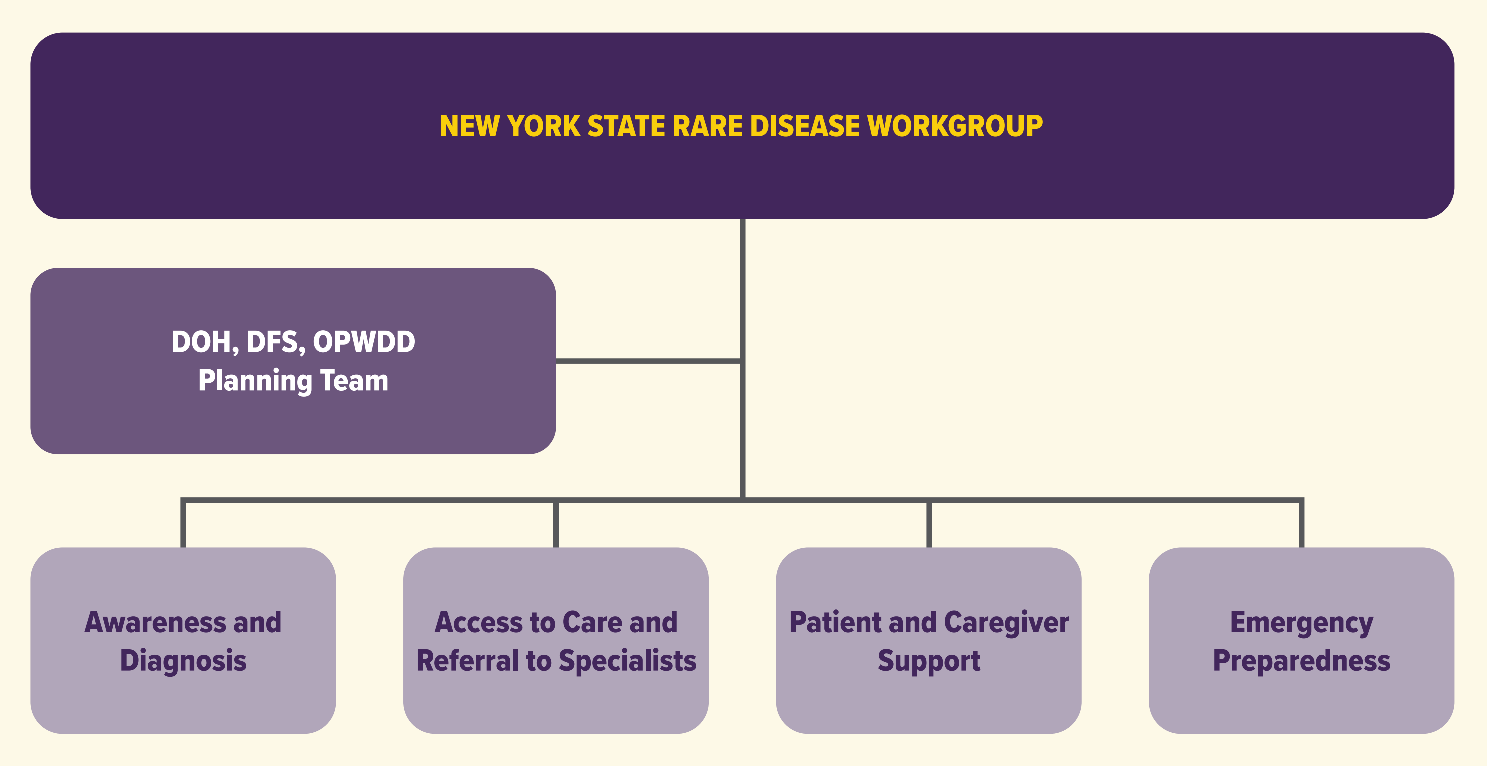
In September 2021, an internal planning team was formed consisting of staff from DOH, DFS, and the Office for People with Developmental Disabilities (OPWDD). Representatives were solicited from program areas across the state agencies that intersect with rare diseases, including newborn screening, children with special healthcare needs, chronic disease prevention, birth defects registry, New York State (NYS) Medicaid program, and commercial health insurance oversight.

The internal team met several times to discuss the process for soliciting external workgroup members for the New York State Rare Disease Workgroup (Workgroup). The team developed an introductory email and brief online application that was disseminated in December 2021 through organizations representing the rare disease community, healthcare providers, health plans, and biopharmaceutical research.

Over 50 applications were received from individuals interested in serving on the Workgroup. The internal team reviewed the applications and selected members to ensure broad and diverse representation, including individuals who have a rare disease or caregivers of a person with a rare disease. Seventeen individuals completed the vetting process and were appointed by the Health Commissioner to serve on the Workgroup. The internal planning team also established a New York State Rare Disease Workgroup page on the DOH website that provided background on the Workgroup, a list of members, meeting agendas, and resources for the public and providers.

The Workgroup was convened for the first time in March 2023. Over the subsequent year, the Workgroup met virtually seven times to discuss recommendations and resources that were responsive to the legislation. The Workgroup formed subcommittees around the following four priority areas: **1)** Awareness and Diagnosis; **2)** Access to Care and Referral to Specialists; **3)** Patient and Caregiver Support; and **4)** Emergency Preparedness (Figure 1). The subcommittees met periodically between the Workgroup meetings and developed recommendations and resources regarding their priority area to present at the full membership meetings. DOH, DFS, and OPWDD planning team members attended and supported the Workgroup meetings as well as their assigned subcommittee meetings.

Figure 1. New York State Rare Disease Workgroup and Subcommittees



Abbreviations: DOH, Department of Health; DFS, Department of Financial Services; OPWDD, Office for People with Developmental Disabilities.

WORKGROUP RECOMMENDATIONS

The Workgroup developed the recommendations described below, organized by the four priority areas. These recommendations will require state and/or federal resources and collaboration across partner organizations to achieve health equity and address the needs of the rare disease community.

Awareness and Diagnosis

In general, there is low awareness of rare diseases among healthcare providers, insurers, governments, organizations, and families. By definition, a rare disease is not common, so the typical pediatrician, family physician, or internist will see very few, if any, cases of a specific rare disease. The presenting symptoms may not be recognized as representing a rare disease. Many patients are initially misdiagnosed and/or the underlying etiology is undiagnosed. Some diseases may have different presentations or be more difficult to diagnose in certain populations or ethnic groups, causing inequities in diagnosis or referral. Patients are typically referred to multiple subspecialists and undergo repeated evaluation and testing before the correct diagnosis is made. Patients and families usually have limited ability or awareness to seek specialists and diagnostic centers. They often lack access to the disease-specific support groups or patient advocacy groups. Insurance companies can further complicate the diagnosis by limiting referral or coverage to specialized providers or centers.

Recommendation 1: Increase awareness about rare diseases.

Description: Healthcare providers, insurers, governments, organizations, and families are often not aware of rare diseases. Resources are usually only available in English or a limited number of languages, and there may be additional accessibility issues. Health literacy is a common problem among all stakeholders, which is exacerbated when information is not available in an individual's native language. A campaign should be developed and implemented to increase awareness of and provide information about rare diseases. It should be multicultural, in multiple languages, and accessible to individuals with any number of disabilities.

Recommendation 2: Increase the awareness, availability, and accessibility of resources about rare diseases for healthcare providers, patients, and families seeking a diagnosis.

Description: Healthcare providers (both primary care providers and specialists) have limited time with patients, which is especially problematic for those with undiagnosed conditions, unusual presentations, complicated health issues, or health problems that are not responding to usual treatments. Healthcare providers need to know when and where to refer patients for further evaluation, review of genetic or laboratory tests, or specialized treatment. Difficulties with insurance coverage of specialists, diagnostic referral centers, or Rare Disease Centers of Excellence can further delay evaluation.

Patients often have difficulties finding or accessing the appropriate healthcare provider(s) or facility. There are shortages of geneticists, other relevant specialists, and networks for undiagnosed disorders or rare diseases. There are often long waits to see certain specialists, who may be concentrated in large academic medical centers, resulting in significant geographic disparities and high transportation costs. In NYS, there are four Rare Disease Centers of Excellence, all of which are located at academic medical centers in New York City.

A web-based repository should be developed that lists diagnostic referral centers, Rare Disease Centers of Excellence, support services, and other resources in NYS. A campaign should be developed and implemented across multiple platforms to increase healthcare providers' awareness of rare diseases and resources available, including through DOH's Rare Diseases website, National Institutes of Health, National Organization for Rare Disorders (NORD), and Rare Diseases Centers of Excellence in NYS. A companion campaign should be developed and implemented in multiple languages to inform patients, families, and caregivers about rare diseases, the resources available at the above websites, other diagnostic referral centers, and patient support services.

Recommendation 3: Develop educational programs for healthcare providers, patients, families, and the broader community about rare diseases. Because education programs are needed across the United States, the NYS Workgroup and other states' Rare Disease Advisory Councils could work with organizations such as NORD to develop and implement educational programs for healthcare providers and the public to improve awareness and diagnosis of rare diseases.

Description: Healthcare providers have limited knowledge and experience with rare diseases. There is a lack of education about rare diseases in medical schools, residency training programs, and continuing medical education programs. Many patients, families, and caregivers of patients with undiagnosed or rare diseases will explore online sources for information. To counter misleading and false information, there is a need for accurate information about rare or undiagnosed diseases developed by reliable, trustworthy organizations.

Recommendation 3A: Develop an education and training program about rare diseases for healthcare providers at all training and position levels.

Description: The education and training program should be inclusive of all healthcare providers, including physicians, nurse practitioners, physician assistants, and genetic counselors. Specific modules should include information about rare diseases, such as when to consider rare diseases, when to consider that a patient may be misdiagnosed, and when to refer patients with undiagnosed conditions. Materials developed should be included in the curriculum for medical students, health professional students, and residents in pediatrics, family medicine, and internal medicine. In addition, continuing medical education programs for practicing physicians and other healthcare providers already in practice would increase the knowledge of rare diseases and facilitate earlier referral, diagnosis, and treatment. Questions about rare diseases should also be included on medical school, residency program, and medical board exams.

Recommendation 3B: Develop and expand educational materials about undiagnosed and rare diseases for patients, families, and the broader community.

Description: The educational materials should be multicultural, easy to understand, written in simple, non-medical language, available in multiple languages, and accessible to individuals with a variety of disabilities. The materials should provide explanations for patients, families, caregivers, and the broader community about what a rare disease is. The materials should include information about when a rare disease might be considered, when referral to a diagnostic center may be warranted, what patient resource services are available, and how to effectively self-advocate. Guidance could be provided for patients or families when considering or discussing with their healthcare providers further evaluation or referral to specialists, diagnostic centers, or Rare Disease Centers of Excellence. The guidance could also include approaches to facilitate health insurance coverage.

Access to Care and Referral to Specialists

Background: People with rare diseases face multiple barriers in obtaining an accurate diagnosis and accessing therapies and treatments. Payors play a large role in ensuring access to the right care at the right time. In addition, many types of resources are needed for people who have rare diseases and undiagnosed disorders to navigate the healthcare system while also maintaining financial stability in their daily lives. Subsequently, finances are a concern for many people living with rare diseases and undiagnosed disorders. People with rare diseases from lower-income households experience higher financial burden and greater barriers accessing treatment. Lastly, healthcare workforce shortages also impact access to care for people with rare disease.

Recommendation 1: Support the recruitment, training, and retention of specialists in rare diseases.

Description: There are limited numbers of specialists in rare diseases which, at best, increases wait times and, at worse, decreases access to care. This prolongs the diagnostic journey for many. One mechanism to increase the availability of needed specialists in NYS is to prioritize specialists' medical licensing applications and provide support to reduce turnaround time for medical license requests. A second mechanism is to support and expand the training programs for the needed workforce. Lastly, programs and incentives could be developed to recruit and retain needed specialists.

Recommendation 2: Support the recruitment, training, and retention of behavioral health providers and homecare providers.

Description: Shortages of behavioral health providers and homecare providers negatively impact marginalized rare disease populations with mental health disorders and physical disabilities. Policies and programming are needed to support the recruitment, training, and retention of individuals in these high priority jobs.

Recommendation 3: Support the recruitment, training, and retention of nurses, particularly those with experience in pediatrics to administer pediatric infusions at home and in smaller community hospitals.

Description: Some children with rare disease need intravenous medication infusions. However, in some areas and situations, there is a lack of pediatric nurses to administer these intravenous medications to children either in small hospital settings or in home settings.

Recommendation 4: Promote systems and tools that support communication between patients and healthcare providers.

Description: Poor communication between specialists, primary care providers, and people living with rare disorders negatively impacts care and can further prolong time to diagnosis. Better systems are needed, especially for at-risk patients who may already face challenges regarding communication because of their rare disorder. Examples of communication systems and tools include patient question lists, patient portals, text messaging, care coordination plans, patient education materials, and clear referral protocols.

Recommendation 5: Continue support and coverage of telehealth by commercial and state-sponsored insurance.

Description: It can be difficult and costly to attend provider appointments in person, especially for a patient with a rare disease who may face baseline mobility challenges or who needs to travel a long distance to see a specialist. Ensuring continued support for telehealth coverage can help secure access to care for people with rare diseases.

Recommendation 5A: Promote resources to ensure patients with rare diseases have stable and reliable access to the internet.

Description: Internet access can be costly; programs such as the federal Affordable Connectivity Program have allowed people access to internet connection, which enables telehealth access of provider appointments.

Recommendation 6: Provide resources to help with travel and childcare that will allow a person with a rare disease or a parent of a child with a rare disease to attend provider appointments.

Description: Lack of resources are a barrier when travel is required to see a specialist or for other provider appointments. The inability to obtain and/or afford childcare can be a barrier for some individuals to attend needed appointments.

Recommendation 7: Decrease barriers to utilizing health insurance and provide resources to help with the cost of medications and treatments.

Description: Out-of-pocket costs are a barrier that limit patient access to medically necessary medications and treatments that are denied or only partially covered by insurance. Health insurance policies vary and can change, making it difficult for patients and providers to ensure that access to treatments is financially feasible.

Recommendation 7A: Create a one-stop database for healthcare providers and patients that provides transparent information regarding formularies and enteral nutrition coverage, including out-of-pocket costs.

Description: Having multiple websites from each health insurer in NYS makes it overwhelming for patients and providers to understand benefit coverage and requirements. Having a single website that provides a centralized location with information from each health insurer reduces the burden on patients and providers to find the information they need to make decisions affecting healthcare.

Recommendation 7B: Educate providers on programs such as step therapy and exceptions for overriding step therapy; the Inflation Reduction Act for Medicare; and the Food and Drug Administration's Expanded Access Program. Create educational materials for patients to support their awareness and use of these programs. Examine potential administrative burdens and other barriers that prevent providers from utilizing these programs.

Description: Step therapy is a type of prior authorization that requires patients to try a health insurer-preferred medication prior to stepping up to a similar medication prescribed by the provider. Understanding the step therapy program and its exceptions can help providers obtain timely access to necessary patient treatments and medications.

The Inflation Reduction Act of 2022 improves access to affordable treatments for people with Medicare. Providers can help lower Medicare patients' cost sharing or out-of-pocket costs for medications by considering direct cost to patients of prescribed medications.

The Expanded Access Program (sometimes called the Compassionate Use Program) can help a patient with a serious or immediately life-threatening disease or condition obtain access to an investigational therapy which has not yet received Food and Drug Administration approval.

Recommendation 8: Provide resources to help people enroll in health insurance, including Medicaid.

Description: Health insurance plan enrollment can be confusing for people looking for coverage and for providers looking to participate in health plans.

Recommendation 8A: Create a centralized, one-stop resource for people with rare diseases to learn about different health plans to aid in choosing an appropriate plan. This can also include information on how to appeal a plan's decision.

Description: Having to navigate multiple websites from each health insurer in the state makes it overwhelming for patients to understand benefit coverage. Having a single resource that provides a centralized location containing information from each health insurer reduces the burden on patients to find the information they need for enrolling in a health insurance plan, especially if they have a rare disease and the appropriate specialists are out-of-network for some plans.

Recommendation 8B: Create a centralized, one-stop resource for providers to access information on and ease the burden of health insurance plan enrollment, including Medicaid enrollment.

Description: It can be challenging for providers to enroll in multiple health insurance plans to become a participating network provider. A centralized resource for providers can help facilitate provider enrollment, thereby allowing patients greater access to providers.

Patient and Caregiver Support

People with rare diseases and their caregivers often face many challenges to meeting their health and wellbeing needs. Managing a rare disease by itself can be a full-time job. For those with additional life needs, it can become especially overwhelming. Because health and wellbeing are so interconnected, it is essential to ensure people with rare diseases and their families and caregivers have access to information and resources that can help meet their needs.

Recommendation 1: Create a centralized, one-stop directory of information and resources for people with rare diseases and their caregivers.

Description: People with rare diseases often have difficulty finding information and resources to help meet their health and wellbeing needs. DOH should expand their website listing of organizations and websites that can help people with rare diseases and their caregivers access information and resources on a variety of topics, including specific rare disease health information; patient and caregiver support groups; obtaining healthcare coverage (e.g., Medicaid, Essential Health Plan, Marketplace); assistance with daily life essentials (e.g., food, clothing, shelter); assistance with mental health needs; obtaining financial assistance; and accessing caregiver services.

For users to be able to access and use the tool, they will need to know the directory exists and have access to a device with reliable internet connection. In addition, the directory must be understood by those of different reading skills and languages and accessible to people with visual/cognitive disabilities.

Recommendation 2: Conduct a Support Needs Survey to develop a detailed understanding of unmet need, as well as barriers to accessing information and support.

Description: To best help people with rare diseases and their caregivers meet their needs for information and support, it is essential to develop a detailed understanding of what those needs are, as well as the barriers to accessing that information and support. A survey of people with rare diseases and their caregivers should be conducted to determine what supports people with rare diseases and caregivers already have and know about, what supports they still need, what barriers and challenges people encounter when attempting to seek care and obtain needed support, and whether any groups have unique needs that are not being met.

Recommendation 3: Create a standing advisory group consisting of rare disease patients and caregivers to collaborate with other stakeholders about community needs and ways to address them.

Description: Helping people with rare diseases and their caregivers meet their health and wellbeing needs requires an ongoing effort based upon consistent input from people with rare diseases and their caregivers. The Rare Disease Workgroup should be built upon to establish a standing advisory group to discuss emerging community needs and ideas for addressing them, work with DOH to collect data on patients and their needs, and advise other state agencies on the needs of people with rare diseases.

Recommendation 4: Foster greater collaboration among agencies and organizations serving people with rare diseases and their caregivers to promote greater community outreach.

Description: People with rare diseases and their caregivers often require assistance from a variety of agencies and organizations, which can be difficult for many to find and access. Identify agencies that offer information and/or resources for people with rare diseases, learn more about what the agencies specifically do and offer, add the agencies to the list of resources, and invite the agencies to participate in discussions and efforts related to what they do. This would be most successful as an ongoing effort, which further supports the recommendation for a standing advisory group to coordinate and support such an effort.

Recommendation 5: Conduct a media campaign to increase awareness of the information, support, and assistance available to the rare disease community, including the resource directory on the DOH website.

Description: People with rare diseases and their caregivers need to know what information and resources are available. Awareness about available resources should be promoted using a variety of media, including creating QR code-based materials for distribution in print and online, reaching out through social media (e.g., Facebook, Instagram), and networking through providers and patient organizations. This will take money, time, and the cooperation of patient organizations.

Recommendation 6: Use Artificial Intelligence (AI) to help patients and caregivers access and utilize information and resource support.

Description: People with rare diseases and their caregivers often need assistance finding and utilizing information and support, regardless of where they are in their rare disease journey. Providing them with tailored assistance in their search would be enormously helpful. However, such assistance can be time and labor intensive. To make this more accessible and efficient at scale, an interactive tool should be developed in collaboration with AI experts that can essentially function as an assistant to help patients and caregivers do such things as assess their needs, direct them to appropriate information/support, assist them in accessing and using that information/support, and collect data on patient needs and how best to meet them.

Reasonable concerns with this effort are that it will take money and time. There will be barriers to utilization such as the availability of online access and people's willingness to use AI. There will also need to be safeguards to ensure the accuracy of information provided and to prevent the AI tool being substituted for human expertise when inappropriate (e.g., addressing medical questions, etc.). However, offsetting these concerns is the reality of rare diseases. Rare disease communities are, by definition, small and typically lacking in resources. AI can bridge the gap between information/resources and patients/caregivers, especially those who don't know what they don't know and are unable to effectively search on their own. Properly programmed, AI can assess a user's needs and direct them accordingly to the proper websites, organizations, professionals, or agencies.

Emergency Preparedness

Emergencies can be particularly challenging for individuals with rare diseases. There are two categories that someone living with a rare disease needs to be prepared for: **1)** medical emergencies related to their rare diagnosis, and **2)** emergencies due to external or environmental conditions such as a weather-related event, terrorist event, or communicable disease outbreak.

Recommendation 1: Develop a checklist of important health information and make it widely available to be used by individuals with a rare disease when they are experiencing a personal health emergency.

Description: Individuals with a rare disease who are experiencing a personal health emergency are at risk of having first responders and other medical personnel be unfamiliar with the person's medical condition, symptoms, and specialized treatment or medications. Having a checklist completed that includes critical information will help ensure a person is treated correctly during a medical emergency. The checklist template should include information such as clinical team contacts, pharmacy contacts, caregiver contacts, allergies, updated medication lists, and "special information" from a clinician regarding emergency management of rare disease.

Recommendation 2: Provide information on the New York State Department of Health website that includes contacts for local health departments to guide families regarding specific emergency preparedness tools and processes for their local region.

Description: This may include a list of recommended topics for individuals to request information about such as: local emergency shelters with capability to care for those with medical complexity; processes for notifying local emergency service teams that an individual within the district may require special or time-sensitive assistance in the case of a power outage or natural disaster; and information for contacting local energy suppliers to initiate the appropriate process for notifying those providers of important medical and safety equipment in the home that relies on power supply.

Recommendation 3: Ensure that schools have an individualized emergency preparedness plan for children with special health care needs in case of emergencies due to external or environmental conditions such as a weather-related event, terrorist event, or communicable disease outbreak.

Description: The individualized plan should address the specific needs of each child in an emergency, including the ability to evacuate and the need for individualized support for the duration of the emergency on school premises.

CONCLUSION

We thank all the members of the Workgroup for their significant contributions of time and expertise. The Workgroup brought together a diverse group of stakeholders with different perspectives, but with a shared commitment to improving the lives of rare disease patients and their families. The recommendations of the Workgroup involve many complex issues and, as such, will require multi-sector collaboration with ongoing engagement from state and federal governments, healthcare providers, payors, scientific organizations, patient and advocacy organizations, individuals, and families.

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APPENDIX A: LEGISLATION

Chapter 74 of the Laws of 2020, as amended by Chapter 57 of the Laws of 2022

The department of health, in collaboration with the department of financial services, shall convene a workgroup of individuals with expertise in rare diseases, including physicians, nurses and other health care professionals with experience researching, diagnosing or treating rare diseases; members of the scientific community engaged in rare disease research; representatives from the health insurance industry; individuals who have a rare disease or caregivers of a person with a rare disease; and representatives of rare disease patient organizations. The workgroup's focus shall include, but not be limited to: identifying best practices that could improve the awareness of rare diseases and referral of people with potential rare diseases to specialists and evaluating barriers to treatment, including financial barriers on access to care. The department of health shall prepare a written report summarizing opinions and recommendations from the workgroup which includes a list of existing, publicly accessible resources on research, diagnosis, treatment, coverage options and education relating to rare diseases. The workgroup shall convene no later than December twentieth, two thousand twenty-one and this report shall be submitted to the governor, speaker of the assembly and temporary president of the senate no later than three years following the effective date of this act and shall be posted on the department of health's website.

APPENDIX B: RARE DISEASE WORKGROUP MEMBERS

Co-chairs

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APPENDIX C: RESOURCES

The information below is also publicly available on the
Department of Health website at health.ny.gov/diseases/rare_disease/.

Rare Disease Resources for Individuals, Families, and Caregivers

What is a Rare Disease?

In the United States, the National Institutes of Health (NIH) defines a rare disease as a condition that affects fewer than 200,000 people. Although individual diseases may be rare, it is estimated that there are more than 10,000 rare diseases that collectively affect about 1 in 10 people, or 1.8 million New Yorkers.

What Causes Rare Diseases?

Rare diseases have many different causes, although about 80% are thought to have a genetic cause. Rare diseases also include some infections, autoimmune diseases, some types of cancer, diseases caused by exposure to toxins or other environmental factors, and some conditions with multiple causes. For some rare diseases, the cause is not known.

How Can I Get Help?

People with rare diseases often face challenges with getting a diagnosis, accessing information, affording treatment, and finding support and resources. The New York State Rare Disease Workgroup developed this list of resources to assist and support people with rare diseases, including those who are searching for a diagnosis for their condition, and their families.

The list is organized around the general topic areas of getting a diagnosis, finding information about your disease, getting medical care and health insurance, finding clinical trials and research, getting financial and transportation assistance, accessing support for patients and caregivers, and preparing for emergencies.

Getting a Diagnosis

Institute for Basic Research in Developmental Disabilities (IBR)

Institute for Basic Research in Developmental Disabilities | Office for People With Developmental Disabilities (ny.gov)

The IBR focuses on the causes of developmental disabilities and furthers the understanding of brain development and pathology. The Institute also provides extensive, specialized biomedical, psychological and laboratory services to people with developmental disabilities and their families, and educates the public, researchers and health and education professionals regarding the causes, diagnosis, prevention, and treatment of developmental disabilities.

National Genetics Education and Family Support Center

Home | National Genetics Education and Family Support Center (nationalfamilycenter.org)

The National Genetics Education and Family Support Center provides tools and resources to support family engagement and genetic services. The site contains information about genetics and resources for families on a variety of rare diseases.

New York State Department of Health's Newborn Screening Program

Newborn Screening Program | New York State Department of Health, Wadsworth Center

Newborn screening refers to medical tests, the majority of which are genetic, performed to identify babies with certain disorders, which without intervention, may permanently impact newborns and their families. Early recognition and treatment of most of these disorders leads to a better outcome for the newborn. The Newborn Screening Program's goal is to help affected babies live as long and normal of a life as possible.

New York State Physician Profile

New York State Physician Profile (nydoctorprofile.com)

The purpose of the New York State Physician Profile website is to enable the public to review information about all licensed doctors of medicine and doctors of osteopathy who are registered to practice medicine in New York State. This information can help you choose a doctor or find out more about a doctor you are currently seeing.

New York Mid-Atlantic Caribbean (NYMAC) Regional Genetics Network

Home | NYMAC, Regional Genetics Network (nymacgenetics.org)

A network of family groups, health care providers, public health groups, and other partners that work together toward a goal of making it easier for families to connect to the genetic services and information they need.

Pathways

Pathways.org | Tools to maximize your child's development

The Pathways site provides free resources to help understand, track, and encourage your baby's milestones for the best possible outcomes.

The Undiagnosed Diseases Network (UDN)

UDN | Undiagnosed Diseases Network (harvard.edu)

The UDN is a research study funded by the National Institutes of Health Common Fund. The UDN is made up of clinical and research centers across the United States working to improve diagnosis and care of patients with undiagnosed diseases. The UDN has two main goals: 1) to provide answers for patients and families affected by mysterious conditions, and 2) to learn more about rare and common diseases.

Finding Information about your Disease

Genetic and Rare Disease (GARD) Information Center

Genetic and Rare Diseases (GARD) Information Center | National Center for Advancing Translational Sciences (nih.gov) or call GARD at 1-888-205-2331 to speak to an information specialist about rare or genetic diseases in English or Spanish.

GARD is an information center designed to provide comprehensive information about rare and genetic diseases to patients, their families, health care providers, researchers and the public.

National Center for Advancing Translation Sciences (NCATS)

Resources for Rare Disease Patients and Advocates | National Center for Advancing Translational Sciences (nih.gov)

NCATS offers free materials and resources to help patients, caregivers, patient support organizations, health care providers and scientists learn about rare diseases and help advance research on them.

National Organization for Rare Disorders (NORD)

National Organization for Rare Disorders | NORD (rarediseases.org)

NORD, a 501(c)(3) organization, is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them. NORD, along with its more than 300 patient organization members, is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and patient services.

Unique

Disorder Guides | Unique (rarechromo.org)

Unique is a registered charity that provides information for families and professionals regarding rare chromosomal disorders or single gene disorders.

GeneReviews

GeneReviews® | NCBI Bookshelf (nih.gov)

International point-of-care resource for health care providers regarding clinically relevant and medically actionable information for inherited conditions, covering diagnosis, management, and genetic counseling.

Getting Medical Care and Health Insurance

Medicaid

Medicaid (ny.gov)

New York's Medicaid program provides comprehensive health coverage to lower-income New Yorkers. Medicaid pays for a wide range of services, depending on your age, financial circumstances, family situation, or living arrangements. These services are provided through a large network of health care providers that you can access directly using your Medicaid card or through your managed care plan if you are enrolled in managed care.

NY State of Health (NYSOH)

New York State of Health | Health Plan Marketplace for Individual and Small Business Health Insurance (ny.gov)

Help to find low cost, quality health insurance options available through the Individual Marketplace. Compare health plan options and apply for assistance that could lower the cost of health coverage. This includes finding out if you qualify for health care coverage from Medicaid or Child Health Plus. For help in applying, you can also call NYSOH at 1-855-355-5777 and for TTY: 1-800-662-1220, from Monday to Friday 8:00AM to 8:00PM and Saturday from 9:00AM to 1:00PM.

New York State Department of Health: Children and Youth with Special Health Care Needs (CYSHCN) Program

Children and Youth with Special Health Care Needs (CYSHCN) Program (ny.gov)

The Children and Youth with Special Health Care Needs Program seeks to improve the system of care for children and youth with special health care needs from birth and up to 21 years of age and their families. New York State also provides financial supports and technical assistance to programs in most counties in the state that help families of CYSHCN by giving them information on health insurance and connecting them with health care providers. These programs will also work with families to help them meet the medical and non-medical needs of their children. For a full list of counties, visit LHD contact information.

NYS Health Profiles

NYS Health Profiles | Hospitals

Searchable database of hospitals, nursing homes, home care, hospice, adult care, and other providers in New York State. The information is available by county, region, and services offered.

NYS Office of Mental Health licensed providers

Find a Mental Health Program (ny.gov)

Allows users to search on basic, advanced, and full levels to find programs, locations, and provider information to fit their specific needs.

NYS Provider & Health Plan Look-Up Tool

NYS Provider & Health Plan Look-Up Tool

Use this tool when you are deciding which health plan to enroll in or when you are looking for a provider, for example a hospital or doctor, that works with your plan. The tool is updated with information sent to New York State directly by health plans. But to be on the safe side, you should still ask the provider if they accept your health plan and participate in your network before receiving health care services or before enrolling in a health plan.

National Organization for Rare Disorders (NORD)-Centers of Excellence

Centers of Excellence Archive | National Organization for Rare Disorders (rarediseases.org)

This resource allows the user to search by diagnosis or state to find specific Centers of Excellence. Centers of Excellence brings together medical teams experienced in diagnosing and treating a wide array of rare diseases.

Psychology Today

Psychology Today | Health, Help, Happiness + Find a Therapist

This resource allows the user to search for state specific mental health professionals, treatment centers, and support groups. Also allows for a search of online therapy. The list of mental health professionals provides a bio and picture, as well as credentials, insurance accepted, fees and contact information.

Finding Clinical Trials and Research

ClinicalTrials.gov

Home | ClinicalTrials.gov

ClinicalTrials.gov is a website and online database of clinical research studies and information about their results. It is maintained by the National Institutes of Health's National Library of Medicine. Patients and healthcare professionals can find studies that patients may be able to join.

NIH National Center for Advancing Translational Sciences
The Rare Disease Clinical Research Network (RDCRN)

Rare Diseases Clinical Research Network | National Center for Advancing Translational Sciences (nih.gov)

The RDCRN program is designed to advance medical research on rare diseases by providing support for clinical studies and facilitating collaboration, study enrollment and data sharing. Through the RDCRN consortia, physician scientists and their multidisciplinary teams work together with patient advocacy groups to study more than 200 rare diseases at sites across the nation.

ResearchMatch

ResearchMatch

ResearchMatch is a program funded by the National Institutes of Health (NIH). It helps connect people interested in research studies with researchers from top medical centers across the U.S.

Getting Financial and Transportation Assistance

RareCare Program

Financial Assistance For Patients With Rare Diseases | NORD

NORD’s RareCare program helps patients obtain life-saving or life-sustaining medication they could not otherwise afford. The program also provides financial assistance with insurance premiums and co-pays, diagnostic testing assistance, and travel assistance for clinical trials or consultation with disease specialists.

Patient Advocate Foundation: Co-Pay Relief Program

Co-Pay Relief – Patient Advocate Foundation | Co-Pay Relief (copays.org)

The Patient Advocate Foundation’s Co-Pay Relief (CPR) program provides direct financial assistance to qualified patients with co-payments, co-insurance or cost-sharing associated with prescription drugs through funds dedicated to specific disease states. In some instances, assistance with insurance premiums and/or ancillary services associated with the disease also may be available.

myBenefits

myBenefits (ny.gov)

myBenefits is an online tool for New York State residents. Users can apply online for the Supplemental Nutrition Assistance Program (SNAP), the Home Energy Assistance Program (HEAP), and/or Public Assistance; recertify for SNAP and/or Public Assistance; Report Changes; and View Case Details.

Patient Travel Assistance

About Mercy Medical Angels | Medical Transport Restore Patients’ Health

This resource’s mission is to remove the barrier to medical care with transportation on the ground and in the air. Ground transportation is provided with gas cards, bus or train tickets. Air transportation is often flown by volunteer pilots or a commercial airline pilot.

Air Travel Assistance

Free Medical Flights for Patients with Patient Airlift Services (palservices.org)

Patient AirLift Services (PALS) is a nonprofit that arranges free flights for medical patients requiring medical diagnosis, treatment or follow-up who cannot afford or are unable to fly commercially. PALS also serves veterans in need of flights.

Train Travel Assistance

Rails to Recovery | Medical Care Discount | New York by Rail

Rails to Recovery, through Amtrak partnering with Voices of Hope, offers one free companion Rail Fare with the purchase of an adult ticket, enabling a companion to travel with someone who is traveling by train within NYS to receive medical treatment. Reservations must be made and requires 3 days notice. To register for this service, visit Voices of Hope - Albany, NY - Rails to Recovery (voicesofthecapitalregion.org)

Jake’s Help from Heaven (general, travel, emergency, and technology grants)

Home | Jake’s Help From Heaven (jakeshelpfromheaven.org)

Jake’s Help from Heaven provides assistance in two different ways – through their grant application program and through their equipment exchange program. Grant applications can be submitted online for emergency support, general requests, travel reimbursement and technology requests. If in need of borrowing adaptive equipment, and within 100 miles of Saratoga, requests may be made by email.

Friends of Man (financial assistance for childcare, equipment, prescriptions)

Welcome to Friends of Man

Help with daycare/childcare, medical equipment, prescriptions: work through Referring Professionals (caseworkers, case managers, healthcare workers, social workers, school counselors, teachers, clergy) who apply on behalf of their patients and clients needing mobility equipment, prosthetics, glasses, dentures, hearing aids, etc.

NYS OCFS Child Care Assistance Program (CCAP)

Child Care Assistance Program | Division of Child Care Services | Office of Children and Family Services (ny.gov)

Resource to help find and pay for quality childcare. The NYS CCAP helps eligible families with some or all of the cost of child care. Families may qualify based on income, reasons for needing day care, and a child's age or individual needs.

Good Days

Good Days | Effective Compassion | Copay Assistance (mygooddays.org)

A national non-profit charitable organization that lifts the burdens of chronic illness through assistance, advocacy, and awareness. Travel, copay, premium and diagnostic testing assistance is available for specific chronic diseases.

Elderly Pharmaceutical Insurance Coverage (EPIC)-Co-pay assistance

Elderly Pharmaceutical Insurance Coverage (EPIC) Program (ny.gov)

Program that provides seniors with co-payment assistance for Medicare Part D covered prescription drugs after any Part D deductible is met. EPIC also covers many Medicare Part D excluded drugs.

Accessing Support for Patients and Caregivers

Front Door

Front Door | Office for People With Developmental Disabilities (ny.gov)

A service of the New York State Office of People With Developmental Disabilities (OPWDD). Front Door connects eligible individuals to services such as employment, day services, and housing.

Resources for Assistive Technology, Environmental Modifications, and Vehicle Modifications

https://opwdd.ny.gov/types-services/AT-E-Mods-V-Mods

A service of the New York State Office of People With Developmental Disabilities (OPWDD). Assistive Technology, Environmental Modifications, and Vehicle Modifications are all supports eligible individuals can request through the OPWDD Comprehensive Waiver.

New York State Early Intervention Program

health.ny.gov/community/infants_children/early_intervention/

The New York State Early Intervention Program (EIP) is part of the national Early Intervention Program for infants and toddlers with disabilities and their families. To be eligible for services, children must be under 3 years of age and have a confirmed disability or established developmental delay, as defined by the State, in one or more of the following areas of development: physical, cognitive, communication, social-emotional, and/or adaptive. The Early Intervention Program offers a variety of therapeutic and support services to eligible infants and toddlers with disabilities and their families.

NYS HEARS Family Line

HEARS Family Line | Child Welfare and Community Services | Office of Children and Family Services (ny.gov)

The Office of Children and Family Services- Help, Empower, Advocate, Reassure and Support (HEARS) family line supports parents and families by providing resources and referrals to a variety of services. Caring representatives guide families to services including food, clothing, housing, medical and behavioral health care services, parenting education and childcare. Representatives are available at 1-888-55HEARS (1-888-554-3277) Monday through Friday from 8:30AM – 4:30PM.

ARCH National Respite Network and Resource Center

Home | ARCH National Respite Network & Resource Center (archrespite.org)

ARCH provides resources to help caregivers and families locate respite and crisis care programs.

Caregiver Action Network (CAN)

Caregiver Action Network

Provides education, peer support, and resources to family caregivers.

- Caregiver Action Network's Caregiver Help Desk offers free support to family caregivers across the country: Caregiver Help Desk | Caregiver Action Network.
- Caregiver Action Network also offers information tailored for family caregivers of loved ones with rare diseases: Caring for Rare Disease Caregivers (rarecaregivers.org).

NYS Council on Children and Families Multiple Systems Navigator Website

Multiple Systems Navigator Website: NYS Council on Children and Families

This resource allows users to access health, education, human service, and disability information on one website. Built for youth, parents, family members and caregivers that rely on supports from multiple child and family serving systems.

For general help: Call 211 or in NYC call 311

- Call 211 for Essential Community Services | United Way 211
  Run by the United Way, 211 is an information and referral service that is open 24 hours a day, 7 days a week, 365 days a year. It is the simplest way for New Yorkers to get the assistance they need to connect to important services such as housing, food, transportation, and health care.
- Home | NYC 311
  NYC 311 Customer Service Center provides the public with quick, easy access to non-emergency government services and information through the call center.

NYS Office of Mental Health Program Directory

Find a Mental Health Program (ny.gov)

Allows users to search on basic, advanced, and full levels to find programs, locations, and provider information to fit their specific needs.

NYC: Free, Confidential Mental Health Support, Call/Text 988

NYC 988 | Talk. Text. Chat. 24/7 (cityofnewyork.us)

NYC 988 is New York City's free, confidential support, crisis intervention, and information and referral service for anyone seeking help for mental health and/or substance misuse concerns, available 24 hours a day, 7 days a week, 365 days a year. NYC 988 also offers telephone interpretation in over 200 languages.

Courageous Parents Network

CPN | Courageous Parents Network

This resource orients and empowers parents and others caring for children with serious medical conditions, by providing tools that reflect the experience and perspective of other families and clinicians.

- NeuroJourney, an initiative of the Courageous Parents Network, is an educational resource for parents and caregivers of children with severe neurologic impairment. https://neurojourney.courageousparentsnetwork.org/

Preparing for Emergencies

American Academy of Pediatrics: Disaster Preparedness Resources

Disaster Preparedness Resources for Families (aap.org)

Information about how to prepare for a disaster, including assembling a Family Readiness Kit and completing an Emergency Information Form.

New York State Department of Health: Health Information Document

#0972_HealthInfoDoc_102121_horiz_2pp (ny.gov)

Pre-folded pocket card that helps communicate vital life-saving information during an emergency and also relays information about emergency contact. Available in multiple languages.

- PDF version can be downloaded at #0972_HealthInfoDoc_102121_horiz_2pp (ny.gov)
- Printed copies can be ordered using the publication order form (publication number is 0972) at doh_publication_order_form.pdf (ny.gov).

Ready Campaign

Plan Ahead for Disasters | Ready.gov

Ready is a National public service campaign designed to educate and empower the American people to prepare for, respond to and mitigate emergencies and disasters. The goal of the campaign is to promote preparedness through public involvement.

- Information for People with Disabilities: People with Disabilities | Ready.gov
- Home Fire Escape Plan: Practice Your Home Fire Escape Plan | Ready.gov

Red Cross: How to Prepare for Emergencies

How to Prepare For Emergencies | Be Red Cross Ready | Red Cross

Information outlining the steps people can take in advance to prepare for an emergency, including assembling a survival kit, making an emergency plan, and being informed.

- Disaster Safety for People with Disabilities: Inclusive Preparedness Resources | American Red Cross

NEW YORK STATE | Department of Health