ELIMINATING GAPS:

Healthcare Transitions From Pediatric to Adult Care In the Rare Disease Community

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Introduction

Rare diseases affect an estimated 400 to 700 million people worldwide with approximately 25-30 million people affected in the United States and 30 million people in the European Union.[1]

The definition of a rare disease can vary between different regions due to a rare disease being more common in other countries. In the United States, Congress passed the Orphan Drug Act in 1983 to define a rare disease as "a condition that affects fewer than 200,000 people".[2] However, the European Union passed legislation in 2000 to define a rare disease as "a disease that affects 1 in 2,000 people".[2] Researchers and scientists of patient advocacy groups have identified approximately 10,000 known rare diseases with 80% of rare diseases involving genetics.[2] Rare diseases disproportionately affect the pediatric community due to age of onset, shortened projected lifespan, and limited drug therapy options.[1] For example, cardiomyopathy is a heart condition that is rare in infants and children and affects the musculature of the heart.[3] In the United States, the incidence of pediatric cardiomyopathy is 1.1 cases per 100,000 person-years, in which most cases involve genetics.[3] The average age of onset in pediatric patients is 1 year of age with nearly 40% of these children requiring a heart transplant or dying within 2 years of diagnosis.[3] In addition to cardiomyopathy, sarcoidosis is a rare systemic inflammatory disease that can cause multi-organ damage, especially in the lungs.[4] The average age of onset in pediatric patients is 15 years of age and is far less common in children and adolescents than adults.[4] Currently, only 10% of rare diseases have an FDA-approved drug therapy treatment.[5] The rapid advancement in evidence-based medicine and technology has allowed for increased survival and life expectancy in pediatric patients with rare diseases, however, these patients do face challenges within the healthcare system and the complexity of their condition(s) that can require more detailed attention and care. This paper will discuss the challenges of pediatric patients with rare diseases and potential solutions healthcare systems can adopt to improve overall health outcomes in the pediatric rare disease population.

Rare diseases disproportionately affect the pediatric community due to age of onset. shortened projected lifespan, and limited drug therapy options.[1]

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Challenges of Pediatric Patients with Rare Diseases

Transitioning from Pediatric Care to Adult Care

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(defined as 18 years of age or older)

13%

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Most rare diseases are commonly associated with life-threatening complications or spontaneous death after birth; however, a significant number of pediatric patients diagnosed with rare diseases survive into adulthood.[1] For example, patients with rare conditions like pediatric sarcoidosis or pediatric cardiomyopathy are now surviving as late as their 30s.[3,4] Nevertheless, the medical complexity of rare diseases increases with advancing age.[3,4] The presentation of pediatric cardiomyopathy is like cardiomyopathy in adults which includes persistent cough and shortness of breath, however, patientscan be asymptomatic for years or throughout their lifetime.[19] The subtypes of cardiomyopathy have different methods of treatment that require detailed attention to prevent poor health outcomes. Conversely, the presentation of sarcoidosis is remarkably distinct between adults and children, in which children tend to have more organ involvement and frequent symptoms.[6] The progression of sarcoidosis is poorly reported within the pediatric population into adulthood and most patients have a varying duration of follow-ups with their primary care pediatricians.[6] The need for an appropriate care transition from pediatrics to adult medicine for patients with rare diseases is an unmet demand and an observed burden on the patient and the caregiver.[1] Inadequate transition in care, such as failures to schedule follow-up appointments and incomplete patient-specific medical summaries, have been associated with an increased risk of poor health outcomes.[2] Approximately 90% of pediatric patients with rare diseases in the United States survive into adulthood (defined as 18 years of age or older) and 13% of pediatric patients are identified to have special health needs progressing into adulthood that will require care from a parent/guardian or caregiver.[7] In addition, the disposition of a pediatric patient to transition into adult care as well as the preparedness of physicians to receive these types of patients should be evaluated when there is a change in the need for specialized care. Irrespective of the severity or complexity of the condition, a smooth and ongoing transition into adult healthcare systems is essential for the improvement of a patient's quality of life and disease burden.

Patients identified to have a gap in care were also identified to have an increase in emergency department admissions.[20]

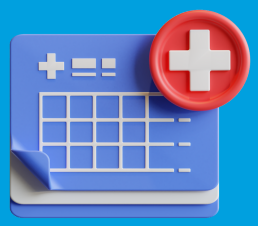
Pediatric cardiomyopathy is referred to as congenital heart disease due to genetic abnormalities involved within the heart muscles and tissue.[3] Studies have shown less than 30% of patients with at least one congenital heart disease are seen by appropriate providers, and there has been an increase in pediatric patients who are lost to follow-up when transitioned to adult care.[20] Researchers specializing in congenital cardiology conducted the Health, Education, and Access Research (HEART-ACHD) study to evaluate the gaps in care among patients with congenital heart disease. This study demonstrated 42% of patients reported a 3-year gap in cardiology care with 8% of patients having a gap in care longer than 10 years.[20] The first lapse in care was identified around the age of 19 years of age, in which transitioning to adult cardiology care would occur.[20] Patients identified to have a gap in care were also identified to have an increase in emergency department admissions.[20] When identifying key predictors to gaps in care, specialty clinic location, and disease complexity were significant predictors among patients.[20] The average age at which gaps in care were first identified in this study could reflect the lack of effectiveness of transitioning care as well as specific factors causing patients to be lost to follow up such as a change in insurance plan or a specialty clinic location.

When identifying key predictors to gaps in care, specialty clinic location, and disease complexity were significant predictors among patients.[20]

In a prospective study, researchers in France evaluated 52 patients with pediatric sarcoidosis and their transition into adulthood. The average age of onset within this patient population was 12 years of age with a duration of follow-up between 6 and 15 years.[8] At the end of pediatric follow-up, twenty-six patients were identified as stable, which was defined as no relapse for more than 1 year, whereas twenty-six patients were required to continue treatment into adult care.[8] Five pediatric patients who were defined as stable had at least one relapse in sarcoidosis transitioning into adult care which required them to undergo treatment; whereas only one patient who was undergoing treatment as a pediatric patient became stable after no relapses occurred in adulthood.[8] At the end of follow-up in adulthood, 52% of patients were stable and 48% of patients still required treatment.[8] At the first evaluation of pediatric sarcoidosis, thirty-eight patients had non-severe sarcoidosis while fourteen patients had severe sarcoidosis with the majority of cases involving the lungs.[8] The last evaluation of sarcoidosis adult care identified forty-one patients with non-severe sarcoidosis and eleven patients with severe sarcoidosis involving the lungs, nervous system, and kidneys.[8]

A multicenter study in Europe evaluated the challenges in transitions from childhood to adulthood care in patients with rare metabolic diseases.[9] Inherited metabolic diseases (IMD) are rare diseases caused by genetic defects in the biochemical pathways of the human body.[9] These conditions can vary in presentation, as newborns, to more progressive diseases where initial symptoms often occur in adulthood and overlap with other chronic metabolic conditions.[9] Approximately 90% of pediatric patients with IMD will survive beyond 20 years of age, however, challenges arise when IMD pediatric patients are moved into specialized adult care and are not provided adequate resources for transition.[9] The study observed 77 centers specialized in metabolic diseases in 23 European countries and 84% of the centers follow both adult and pediatric IMD patients.[9]

Within the centers following pediatric and adult patients with IMD, 57% of patients complete transition after the age of 18 years, 21% complete transition of care between the age of 16 and 18, and 10% of patients remain under the care of their pediatrician throughout their life.[9] In most of the centers, a transition coordinator was not identified, and only 48% of centers had an individualized transition protocol for care.[9] Further inspection of the centers' transition protocols showed the medical summary that was provided to the patient and the patient's family/caregiver was not detailed in the description of the disease, precautions, medication list, emergency plan, and the latest lab results.[9] The role of the transition coordinator is to ensure that the receiving adult care team in charge of patient management is fully prepared for the care of complex conditions, including rare diseases such as IMD. Lack of physician specialized training can hinder a patient's outcome within transitioning care, as only 11% of physicians in the 77 centers are specialized in adult IMD.[9] Physicians from all 77 centers in Europe completed a survey regarding their role in the transition of a pediatric patient with IMD into adult care. Results showed 49% of physicians found lack of time as the most difficult challenge in managing the transition process, and 90% of physicians believe there is limited financial support to allow for transition clinics.[9] Addressing the challenges pediatric patients with a rare disease(s) face within the transition into adult care will aid in the improvement of overall patient outcomes and future transitions of care.



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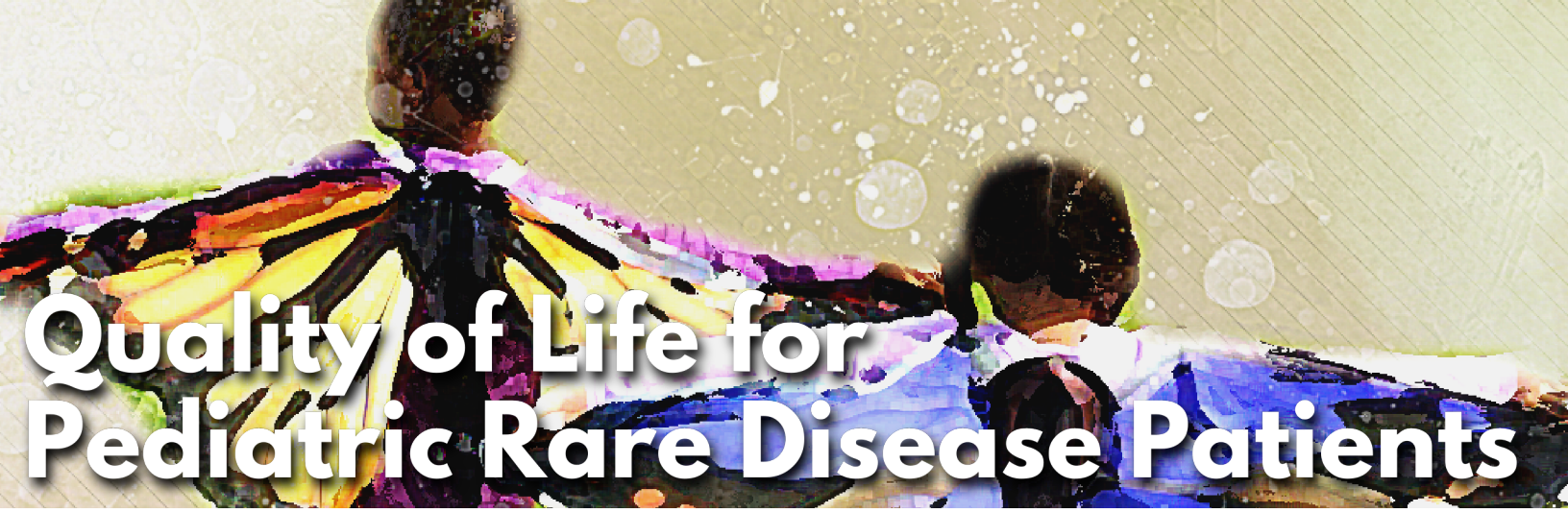
Transition readiness and providers' perspective in the transition of care was also evaluated in multiple countries across Europe. Researchers in the Netherlands evaluated 57 patients with rare endocrine diseases to assess patients' readiness to transition into adult care in endocrinology. Results showed two-thirds of the patients never talked with the doctor without the presence of their parent or caregiver.[10] All patients were aware of what kind of condition they had as well as their medications; however, only 11 patients were aware of the potential side effects of the medications.[10] Regarding practical real-world issues, 41 out of 57 patients did not understand important aspects of their health insurance such as co-pays for medications and outpatient visits.[10] Thirty-eight patients were aware of their medical rights concerning the treatment of their condition; however, the same number of patients were not aware of patient support organizations for their rare condition.[10]

Providers are essential in ensuring an effective and successful transition from pediatric care into adult care, especially in patients with complex conditions. A study examined the transition of care of adolescent and young adult patients (12-18 years of age) with rare renal diseases from a provider's perspective. Physicians completed a survey based on their center's transition protocol and current management of rare diseases in adult care. Out of 45 physicians evaluated, approximately 64% of physicians treating pediatric patients with rare renal disease provided an unwritten plan for transition and communicated the plan with their patient and/or caregiver.[11] Twenty-one physicians confirmed there was at least one transition/transfer coordinator at their center, however, it was not a fully dedicated role and was carried out by either a nurse or social worker.[11] All providers expressed their patients transitioned into adult care at 18 years of age, and the most common health institutions they transitioned into were university hospitals or community-based hospitals.[11]

Only 14 physicians provided group training courses for patients to help gain knowledge on their rare condition and improve activities involving self-management of their condition.[11] This study demonstrated there is a significant need for physicians to improve patient-provider relationships in those with rare diseases through adequate patient resources and effective transition protocols to safeguard health stability as patients transition into adult care.



Quality of Life for Pediatric Rare Disease Patients

Due to limited research and treatment availability, pediatric rare diseases can significantly impact the physical and emotional well being of a patient.

The health related quality of life (HRQoL) of a pediatric patient with a rare and complex disease can provide a better understanding of the severity of disease burden and the psychosocial impact.

Due to limited research and treatment availability, pediatric rare diseases can significantly impact the physical and emotional well-being of a patient.[7] The onset of the pediatric rare disease typically occurs at an earlier stage of development and symptoms can be debilitating in nature.[12] The health-related quality of life of a pediatric patient with a rare and complex disease can provide a better understanding of the severity of disease burden and the psychosocial impact.

For example, pediatric patients with cardiomyopathy can have different degrees of disease severity based on the type of cardiomyopathy, which can have a significant physical and psychosocial impact on quality of life.[21,22] Researchers evaluated multiple studies involving the clinical impact of pediatric cardiomyopathy on health-related quality of life and overall health outcomes. One study evaluated patients between the ages of 2 and 18 years with and without cardiomyopathy using HRQoL measurement tools. A low HRQoL score demonstrated a negative impact on a patient. Results demonstrated patients with pediatric cardiomyopathy have significantly lower scores than healthy patients, with scores related to taking multiple cardiac medications and restrictions on activity.[22] These results also coincide with a study observing pediatric patients with congenital diseases, including cardiomyopathy, compared to healthy pediatric patients and patients with other chronic diseases. The study revealed lower HRQoL scores compared to healthy patients and similar scores with children with chronic conditions.[23] Key factors identified which impacted HRQoL in a pediatric patient with congenital heart disease included increased healthcare costs, disease severity, presence of device or pacemaker, and behavioral and emotional problems.[23] Given the significant impact of cardiomyopathy on pediatric patients, effective transition protocols into adult care are vital to most patients' survivability.

In contrast to pediatric cardiomyopathy, pediatric sarcoidosis can either spontaneously resolve on its own or can become progressive and chronic throughout a lifetime.[4] Patients with pediatric sarcoidosis report higher psychological symptoms than the general population.[12] Researchers observed anxiety and depression in patients with sarcoidosis and identified chronic symptoms such as shortness of breath and cough significantly impacted psychosocial well-being. After evaluating 1,197 patients with sarcoidosis, pediatric patients who required prolonged treatment were significantly more anxious and depressed than those without sarcoidosis.[13] Among those with heightened psychological symptoms, researchers identified the severity of shortness of breath as an indicator of worsening anxiety and depression.[13] In addition, one study evaluated the symptoms of sarcoidosis and its impact on quality of life; the results discovered shortness of breath was associated with worsening health-related quality of life and increased depressive symptoms.[14] The psychological symptoms observed in pediatric sarcoidosis patients do not diminish as patients become adults. In fact, according to the previous study observing the evolution of pediatric sarcoidosis from childhood to adulthood, approximately 50% of patients still required treatment. Among the patients which were stable (no activity of the disease while without treatment) at the end of pediatric follow-up, 19% presented relapses in adulthood.[8] The study concluded pediatric sarcoidosis patients benefit from transitionary care, avoiding destabilizing gaps between the pediatric and the adult management of the chronic disease.[8]

Solutions to the Gaps in Transitioning Care

Transition Care Coordinator

Several studies have evaluated the use of transition care coordinators in specialized healthcare centers including those specialized in rare conditions. The challenge identified in most studies is that the transition care coordinator is not a specialized or dedicated role but rather a task performed by either a registered nurse or a licensed social worker.[15] Transition coordinators ensure the receiving adult healthcare team in charge of patient management is fully prepared for the care of complex conditions including medication lists, most up-to-date lab results, emergency plans, and financial health capabilities.[15] In a previous study, researchers discovered 10% of patients did not transition into adult care and continued to be seen by their pediatrician.[9] For patients with apprehension about their disease or those who may not be equipped with the knowledge of their rare condition, a transition care coordinator can provide resources and schedule individualized meetings based on the patient's health, financial, and/or psychosocial needs during the transition from pediatric to adult care.

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Solutions to the Gaps in Transitioning Care

Use of Patient Reported Outcome Measures

Clinicians suggest the use of PROMs allows for more enhanced conversations between the patient and provider regarding important matters such as apprehension of treatment regimens, psychological well-being, and comprehension of disease-specific information.[17]

If PROMs were to be implemented in rare disease management, it would require community-based disease specialization to promote patient-clinician communication and encourage patient involvement in the development of disease-specific care management.

Patient-reported outcome measures (PROMs) are quality measures that define the status of a patient's health condition coming directly from the patient.[16] PROMs help to better understand the patient, and their implementation can begin to resolve the complexity of care and facilitate shared decision-making with providers to improve overall health outcomes.

A study in the United Kingdom evaluated the use of PROMs to observe their role in inpatient and clinician views of routine management of rare diseases. Patients diagnosed with rare diseases have expressed PROMs can allow clinicians to preview their lived experiences with their condition.[17] Clinicians suggest the use of PROMs allows for more enhanced conversations between the patient and provider regarding important matters such as apprehension of treatment regimens, psychological well-being, and comprehension of disease-specific information.[17] However, PROMs can create issues in their implementation in rare disease management such as multiple variations of questionnaires which can impact the completion and accuracy of PROMs (especially in patients with learning disabilities).[16] If PROMs were to be implemented in rare disease management, it would require community-based disease specialization to promote patient-clinician communication and encourage patient involvement in the development of disease-specific care management.

Solutions to the Gaps in Transitioning Care

Telehealth

The tragedy of the SARS-CoV-2- (COVID-19) pandemic caused a massive acceleration in the use of telemedicine. In April 2020, overall global telehealth utilization for office visits and outpatient care was 78 times higher than in February 2020.[24] During the rising waves of COVID-19 infections, telehealth offered a bridge to care that has been otherwise denied to patients who have difficulty accessing quality healthcare.

Researchers from the National Children's Hospital in Columbus, Ohio addressed the barriers of accessing clinical services, benefits of telehealth services in rare diseases, and considerations of using telemedicine in rare disease management. Barriers identified in patients with rare diseases include lack of local resources, difficulty in rare disease diagnosis, caregiver stress, and lack of specialized provider training.[25] Conversely, telemedicine can provide convenience, reduce costs of urgent care or emergency visits, and improve access to multidisciplinary care to diagnose and treat patients with rare diseases.[25] When providing care for rare diseases, provider-to-provider consultation through video teleconference allows for real-time advice between providers to discuss options to best fit the needs of the patient.[25] Considerations in adopting telemedicine in the world of rare diseases should include willingness from the providers and medical facilities, additional training and licensure due to disease complexity, and logistical factors such as equipment, internet availability, and device monitors.[25]

General care and accessibility to care for rare disease patients continue to be a huge challenge in developing countries like the Caribbean and Latin America.[7] Healthcare is not as readily accessible in countries outside of Europe and the United States, and accessibility to medical care has been further decreased during the COVID-19 pandemic.[18] Researchers from Argentina evaluated the implementation of information and communication technologies (ICT) in the care of telemedicine for pediatric patients with rare and complex conditions in Latin America and the Caribbean.[18] The study showed evidence that implementing telemedicine in conjunction with remote monitoring devices (in patients' homes where parents or extended family are the primary caregivers) reduced the number of transfers to medical institutions and promoted rapport with the patients' family/caregiver and healthcare team.[18] The study showed the potential for telemedicine and remote telemonitoring to improve continued quality of care and access to health care services; however, further studies need to be conducted in the use of telemedicine to gain a better understanding of its impact on medically complex and rare disease patient health outcomes.[18]

As we continue to work on filling gaps in transitioning care services, telehealth systems offer an opportunity to establish decentralized and hybrid virtual/in-person care models with the goal of improved healthcare outcomes and more access and affordability on an international level.[18]

Conclusion

Rare diseases affecting the pediatric community can have devastating consequences including a shortened lifespan.[7]

By addressing the gaps and challenges found in pediatric rare disease management, the impact of disease burden in patients may significantly reduce, creating favorable changes in patient disabilities, and improving general health, quality of life, and life expectancy.

The advancement of medicine and technology has increased pediatric patient survival into adulthood through prevention of complications, disease-specific monitoring, and improvement in overall quality of care. However, rare diseases, such as pediatric cardiomyopathy and sarcoidosis, can be complex in nature and devastating to an individual's quality of life throughout their journey from child to adult.

The absence of or inadequate transition of care services for these patients into adulthood poses life-altering challenges for patients and their caregivers. Many of these challenges include psychosocial burdens and lack of accessibility to healthcare, especially for those who live in rural areas and for those with physical mobility limitations. The burden of the rare disease and its impact generally extends beyond the patient to the caregiver.

When considering potential solutions for these challenges, identifying the specific needs of both the patients and caregivers is necessary. The use of transition care coordinators as a solution is imperative. Transition care coordinators ideally ensure that the hand-off between the pediatric and adult provider(s) leaves no gaps in communication, education, evaluation, and follow-up. Another solution includes the use of telehealth services such as virtual doctors' appointments/consultations, remote monitoring to determine changes in health status, and/or nurse care managers for ongoing support and care coordination. Utilizing telehealth services to address immediate issues as they arise ensures timely evaluations as well as helps to prevent critical exacerbations that may lead to negative outcomes. Many of these telehealth services help the patient and the caregiver feel supported between office visits. Finally, the use of PROs can aid in assessing areas the patient may be having issues that require additional support or resources. Knowing where the patient is struggling provides a basis for identifying places to focus on to ultimately improve overall health outcomes in these transitioning rare disease populations.

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