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Identifying key supports and barriers during the transition from pediatric to adult healthcare for individuals living with spinal muscular atrophy: A qualitative study

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Abstract

Individuals with genetic diagnoses require complex medical care throughout their lives. Much of this is hampered by lack of efficacious treatment or disjointed care teams. Spinal muscular atrophy type I-III (SMA) is one such example. Recently efficacious treatments have allowed more individuals to reach an age that requires transition from pediatric to adult healthcare teams (p/aHCT). A knowledge gap exists about the transition process and its impact on the biopsychosocial needs of adults living with SMA. Six telephone interviews were recorded, transcribed, and double-coded using inductive and deductive coding for thematic analysis regarding this healthcare transition process. Themes identified include: comparisons to pediatric care, transition impacts care, importance of mental healthcare, and facilitators/barriers. Interviewees highlighted positive experiences with pHCT due to the effectiveness of multidisciplinary clinics (MDC). Conversely, the lack of MDC and limited adult provider guidance and knowledge resulted in negative aHCT experiences. Interviewees felt that mental health awareness and knowledge of disability constructs were limited among both p/aHCT. While barriers to transitional healthcare exist for SMA patients, identification of support structures, visibility of disability advocates, and availability of treatments are facilitators. These interviews provide insight to patient needs and the overall impact of transitional healthcare. They highlight a need for adult MDCs, more structure from educated providers, mental health discussions, and accessibility of resources for adults with SMA. Importantly, these findings are likely to be mirrored by additional patient groups with complex healthcare needs, regardless of genetic or complex etiology.

Introduction

The development and implementation of successful gene-modifying treatments has been a significant step forward for several genetic conditions; however, a greater emphasis on continuity of care is needed. Specifically, the process of transitioning from pediatric to adult healthcare teams (p/aHCT) should be prioritized. While healthcare transition is ubiquitous, complex healthcare needs increase its complexity (White 2018). Spinal muscular atrophy (SMA) is a progressive neurodegenerative condition characterized by symmetrical muscle weakness and atrophy of skeletal muscles (First and Hurst 2017; Prior et al. 2011; Kolb and Kissel 2011). Children with a diagnosis of SMA are typically followed by relevant specialists within a multidisciplinary clinic (MDC) working collaboratively to improve healthcare management (Kolb and Kissel 2011; Ropars et al. 2020; Mercuri et al. 2018; Finkel et al. 2018).

Prior to the availability of SMN-restoring therapy, the prognosis was guarded for individuals diagnosed with a more severe type of SMA. Today, three treatment options are available in the United States and Canada, reducing progression of muscle atrophy (Kolb and Kissel 2011; Mercuri et al. 2018; Finkel et al. 2018; Reilly et al. 2022). Therapies have improved the prognosis for SMA, resulting in the need for increased awareness of SMA among adult providers (Kolb and Kissel 2011; Mercuri et al. 2018; Finkel et al. 2018; Reilly et al. 2022; Walter et al. 2021). The increased prevalence of adults living with SMA serves as a testament to the success of treatments while simultaneously highlighting the gap in knowledge about healthcare transition and its impact on the medical, psychological, and social needs of

adults living with SMA (Serra-Juhe and Tizzano 2019; Sport-Campana et al. 2024; Wan et al. 2019). A disjointed transition experience can negatively impact patient health; identifying and addressing these gaps may improve this process for individuals with complex conditions (Varty et al. 2020).

Wan et al. (2019) identified that adults with SMA often disengage from their healthcare services based on a negative transition experience. Disengagement led to a lower perceived value of care and was attributed to limited accessibility to disability services. They expressed fear and frustration with progressive loss of function, and social isolation/stigma as negatively impacting mental health (Wan et al. 2019). Our study expands upon the themes identified in Wan et al., helping to elucidate the unmet needs of individuals living with SMA within the United States and Canada during healthcare transition.

Materials and Methods

1.2.1 Study Design

This study was classified as non-human-subjects research by Augustana University Institutional Review Board under SU23.01. Individuals were eligible to participate in the study if they were 18+ years of age, had received their SMA diagnosis prior to age 18, and spoke English.

1.2.2 Sample and Recruitment Strategy

Recruitment employed a targeted and snowball approach. Research invitations through Muscular Dystrophy Canada included a brief description of the study, inclusion criteria, and a link to the demographic survey. Additionally, the study information was distributed via the social media accounts of researchers and several patient support groups. Organic distribution was also encouraged.

Survey respondents could express interest in a semi-structured interview. Contact information was collected in a third party application, WuFoo, anonymizing survey and interview data. Invites to schedule telephone interviews were sent to the provided email address.

1.2.3 Data Collection

A 26-item Qualtrics survey was used to collect information about basic demographic information in addition to details regarding the transition process, members of their healthcare team, and unmet needs. Verbal consent was confirmed prior to beginning the interview recording. All recordings were kept on a secured laptop in a password protected folder. EL, a genetic counseling student, conducted all interviews. She received interview training from DP who has extensive interview experience. The interview guide from Wan et al., 2019 was refined through literature review and discussions between the research team and representatives of the SMA community. Updates include a greater focus on changing healthcare systems/personnel, support resources, mental health care, perceived facilitators and barriers, potential improvements, and life achievements. A debrief and concluding remarks were made after completion of the interviews; including confirmation of email for distribution of a $15 Amazon gift card.

1.2.4 Data Analysis

Interviews were recorded and transcribed verbatim using the transcription software Otter.AI. Recordings were reviewed and cleaned by EL. Themes and subthemes were identified via inductive coding using a narrative interpretationist framework. The code book was iteratively applied to each transcript, which were double-coded by EL and DP. Comparison of applied codes was discussed, identifying inconsistencies which were remedied through consistency meetings (Braun and Clarke 2023; Wainstein et al. 2023). New codes were retroactively applied to previous transcripts. Quantitative content analysis generated major themes and representative quotes are included within the results section. Demographic data of survey respondents are presented with simple descriptive statistics.

Results

1.3.1 Study Sample

A total of 14 individuals completed the demographic survey, 12 at 100% completion with one participant partially omitting a question about SMA type and another participant omitting country of residency. Due to the open nature of study recruitment, the overall response rate is unknown. Of the 14 respondents, 10 provided contact information for follow-up telephone interviews. Six interviews were completed, the mean interview duration was 35.9 ± 7.7 minutes (range: 28.7 to 45.4 minutes).

1.3.2 Participant Demographics

Demographic information for survey respondents is available in Online Resource 1. There was an insufficient number of responses to perform statistical analyses. Highlights from survey responses regarding transition experience include a majority reporting changes to their HCT during transition, feeling supported by providers, and having mixed feelings about their aHCT's ability to address their needs (Online Resource 2).

1.3.3 Interviewee Responses and Key Themes

Interviewee's lived experiences reflected four major themes, *comparisons to pediatric care, transition impacts care, importance of mental health care, and facilitators/barriers exist,* each being composed of several sub-themes (Figure 1).

1.3.4 Comparisons to pediatric care

Sub-theme 1.1

Interviewees reported positive experiences with their pHCT: *"[I] had a lot of trust in my pediatric doctors"* (Interviewee 4). When comparing pediatric and adult experiences, several expressed *"I really took that for granted"* referencing support from their pHCT (Interviewee 1).

Canadian interviewees reported a strict pHCT cutoff at age 18, while US interviewees reported the option of extending to 21. Regardless, interviewees desired to maintain their pHCT because *"They seem to care more and have more experience and expertise"* (Interviewee 5).

Pediatric providers who were aware of transition pitfalls and provided guidance during transition were described as helpful and reassuring, "*They gave me a medical passport"* (Interviewee 4).

Sub-theme 1.2

All interviewees shared similar experiences about challenges arising from disjointed aHCT *"It takes more time and energy because it's not really all coordinated"* (Interviewee 5).

Interviewees reported less public funding and reduced access to hospitals, school services, and caregivers after transitioning to aHCT (Table 1).

Table 1: Interviewee responses according to sub-themes. This table includes direct quotes from interviewees. Each response is categorized by sub-themes. Each sub-theme is derived from the four major themes identified during thematic analysis. SMA=spinal muscular atrophy.

Limited access to funding and school services (Sub-theme 1.2)	*"When I was a child, I got a lot of funding and had someone with me in school that helped. As an adult, I think it's more lacking just because I have limited funding for caregivers... When I went to university, I had to wait until I turned 19 to get my funding."* (Interviewee 2, SMA Type I).
Limited access to hospital and daily life services (Sub-theme 1.2)	*"The pediatric hospital has social workers there and they had nurses and they had people that were up to date with what programs you can get caregivers and nursing in your home and all the stuff that went beyond just receiving the care at the hospital that translated into your life outside of the hospital. Whereas in the adult system, it's not like that... just very singular focused. I would say there's no help for living your life with SMA outside of treatment."* (Interviewee 3, SMA Type II)
Willing to learn (Sub-theme 1.3)	*"She wasn't familiar with SMA but she was willing to learn... she knew that she didn't know anything so she was going to listen to me and talk and ask" (Interviewee 1, SMA Type II)*
Unwilling to learn (Sub-theme 1.3)	*"I was discharged out to a neurologist but the neurologist was probably the most unkind man I've ever met in my life... At this time, Spinraza had just become available in Canada. There was a period of time where you needed to apply quickly because there's something called a Special Access Program where you could get funded through the federal government. So my parents and I had gone to him to fill out the paperwork... He absolutely refused and he said he didn't believe in the drug. He said that there was no way that it would help an adult like me. Keep in mind, I'm 18 at this point." (Interviewee 2, SMA Type I)*
Patient-led education (Sub-theme 1.3)	*"A lot of adult providers don't know how to care for SMA patients. Historically people with SMA didn't really live into adulthood… I definitely have to educate my providers most of the time simply because most of them do not have many, if any, other patients with SMA" (Interviewee 6, SMA Type II)*
	"I would say I definitely felt like there wasn't enough resources. I had to teach. I was the one educating and trying to bring other providers up to speed." (Interviewee 4, SMA Type II)
Suggestions for improving transitional care (Sub-theme 2.1)	*"There wasn't really a person that [guided me to] switch from this person to this person... I want them to lay out the process of what the transition is like and who is taking care of what, like a caseworker" (Interviewee 5, SMA Type II).*
Additional school supports recommended by pHCT (Sub-theme 2.2)	*"I started requiring a nurse to come to school with me that the pediatric team helped set up for us. They helped set up nursing inside our home as well, which allowed my parents to get a restful night's sleep, so they didn't have to be up all night turning me and taking care of my medical needs." (Interviewee 3, SMA Type II)*
Limited resources during the transition to aHCT (Sub-theme 2.2)	*"I had classroom aides in school growing up and also in college… But as I got older, it was less and less. They did that to try and make me independent because I knew in college I wouldn't have the same support available." (Interviewees 5, SMA Type II)*
Early intervention for mental health resources (Sub-theme 3.1)	*"I wish there was other mental health supports available... I think a lot of that is having a provider, a pediatrician, that knows you well and can identify some things on their own without having to bring it up yourself. I think a lot of people have mental health issues that are very valid and normal that they just don't know about it and no one has ever told them that... Having that*

	early assessment and that early plan... helps them build that support system." (Interviewee 1, SMA Type II)
Stigma and disease-progression impacts self-esteem (Sub-theme 3.2)	*"I tried to hide my disability or accommodate it but then ever since I got into social media and seeing a lot of outspoken disability influencers, they're just so proud of who they were made to be. Society looks at it as a mistake, or a mess up or something that has to be hidden when that's not true. I feel I have a whole new sense of self and identity. I'm a disabled woman and I'm not afraid to identify that way and I'm not ashamed of it."* (Interviewee 5, SMA Type II)
Relationship with family, friends and the SMA community aided the transition period (Sub-theme 4.2)	*"Be ready to talk to other families... I didn't really meet other people with SMA until my adulthood... talking to other people in my situation was really helpful... Just having a role model who I can connect with. I never really been given that opportunity as a kid"* (Interviewee 2, SMA Type I)
The impact of the SMA community on individual's transition (Sub-theme 4.2)	*"I would have loved to know sooner that there is a lot of support to be found in the community outside of medical providers. The SMA community is so strong. I didn't know about CureSMA conferences and things like that until I was in my 20s. That was my first introduction to a community and I learned that there was this wealth of knowledge and support. People who could tell you exactly what transitioning out of pediatric care might be like." (Interviewee 4, SMA Type II)*

Interviewees reported high levels of frustration, confusion, and fear throughout the transition process, primarily when comparing their pHCT and aHCT. One interviewee enlisted financially costly measures *"I'll go coast to coast"* in their quest for an aHCT which is *"still not as good as [my] pediatric care"* (Interviewee 6).

Sub-theme 1.3

Interviewees perceived members of their aHCT as lacking direct exposure to SMA, impacting their knowledge about the needs of this patient population (Table 1). In order to combat the impacts of limited provider exposure, interviewees took on the role of education: *"I have to educate everyone"* (Interviewee 5). Every interviewee expressed a need for patient-led education; however, there were significant differences in provider's perceived initiative and their willingness to learn (Table 1). Additionally, patient-led education resulted in reduced *"time and energy to focus on other parts of my life"* (Interviewee 2).

1.3.5 Transition impacts care

Sub-theme 2.1

Interviewees felt additional guidance on finding knowledgeable adult providers would be instrumental during transition. Most felt isolated and unaware of how to navigate this process, prolonging inconsistent care *"I jumped between doctors for like five years"* (Interviewee 1).

While interviewees reported high levels of trust and confidence in their pHCT, this lack of guidance led some to become *"skeptical or hesitant to rely on future [adult] doctors"* (Interviewee 4).

Interviewees provided suggestions for improvements to transitional care including a dedicated transition team/provider and a cheat sheet outlining the important steps to find knowledgeable providers, and seek out home care and additional resources (Table 1).

Sub-theme 2.2

All interviewees spoke about the importance of additional resources outside of the hospital including home caregivers, school supports, nurses, and accessible housing; highlighting that these resource requirements vary by age. As children, most received resources recommended by their pHCT through the school districts, these became limited in higher education (Table 1).

Funding for treatment was also limited for adults; with one Canadian interviewee stating *"it varies province by province whether adults get coverage or not"* (Interviewee 3).

Sub-theme 2.3

Every interviewee spoke highly about their experiences with pediatric MDC, highlighting how the MDC *"was really efficient"* (Interviewee 5).

Three interviewees reported that this type of system is not replicated well in adult healthcare and its implementation *"would be a big area of improvement"* (Interviewee 4). The lack of MDC increased patient responsibility and effort required to manage collaborative care between adult providers: *"Nobody collaborates with each other"* (Interviewee 3).

1.3.6 Importance of mental health care

Sub-theme 3.1

All interviewees reported having limited to no discussions around mental health resources with their providers *"There was never any discussion of this may be a need"* (Interviewee 4).

Similar to patient-led education, interviewees reported the need for self-advocacy regarding mental health services. Most interviewees described themselves "*feeling anxious, I was depressed… issues that have plagued me because of SMA"* (Interviewee 1). Resources to combat these *"struggles with anxiety and depression"* (Interviewee 5) were only introduced by healthcare professionals after the individual directly asked for them.

This sentiment was expanded upon by an interviewee who spoke about the importance of improving mental health resources for disabled individuals more broadly and emphasized how early intervention can impact a patient's course of treatment and transition into different phases of life (Table 1). One interviewee highlighted that mental health should be a consideration for all individuals, regardless of disability status, while simultaneously acknowledging that the process of transitioning care teams for a person with a disability involves a more significant amount of energy, time, and adaptation.

Interviewees desired access to mental health support from counselors who understand the complexities of living with SMA/disability. Lack of representation of disabled counselors, or disability informed counselors, results in the need "*to explain ableism and all my barriers to an able bodied person*" (Interviewee 5).

Sub-theme 3.2

When asked if their diagnosis impacts their sense of self or self-esteem, external barriers were cited "*The hardest part of my disability is not even the disability, it's just the external barriers I face*" (Interviewee 5). Others expressed feeling underestimated and frustrated by societal biases *"I see the shock on their face because I have a full-fledge career"* (Interviewee 6) or that increased medical needs are a *"hit on my self-esteem in my social life"* (Interviewee 3).

In order to combat society's negative perception of their disability, interviewees spoke about the positive aspects of social media representation/disability influencers (Table 1).

1.3.7 Facilitators/barriers exist

Sub-theme 4.1

All six interviewees had been receiving nusinersen and risdiplam for a number of years and attributed significant health improvements to these treatments. Improvements to breathing, swallowing, muscle strength and increased energy levels were commonly identified. These health improvements allowed for less dedication to daily health needs and greater opportunities to pursue other interests. Interviewees described treatment as creating a *"Change [in] my life plan… I began to think more about entertaining more romantic relationships"* (Interviewee 4).

Sub-theme 4.2

Family, friends and the SMA community provided a high level of support and greatly influenced the transition process for all interviewees. For some, family and friends *"made the transition with me"* (Interviewee 4).

Most interviewees wished they had been informed about the SMA community years before starting the transition process (Table 1). Connections with the SMA community made the transition process easier, contributing to some viewing their challenging experiences as *"inspiring"* (Interviewee 1). Several interviewees noted that finding a mentor, someone who has already transitioned to an aHCT, should be strongly encouraged by an individual's pHCT.

Sub-theme 4.3

Some interviewees expressed that society and healthcare providers alike fail to recognize the added responsibilities that accompany an SMA diagnosis. Interviewees report their *"disability is a full*

time job" (Interviewee 5); the need to find, manage, and schedule daily home caregivers and support creates tension when trying to manage a successful work-life or school balance.

In contrast, one interviewee found that the successful management of their caregivers and nurses was a major life achievement because it helped them gain independence.

Discussion

Although gaps in transitional care have previously been identified, our understanding about the extent of these needs is still limited (Walter et al. 2021; Wan et al. 2019). Adult SMA patients detail common experiences and concerns that arise during the transition process. This study further explores the impact transition has on individuals' physical and mental health and emotional wellbeing.

Interviewees drew stark comparisons between their pHCT and aHCT. The reduction in support, trust, SMA-specific knowledge and accessible SMA-specific resources during transition led to ineffective healthcare. Historically, adult providers have had little to no direct clinical experience with SMA. As treatments for pediatric conditions continue to improve, medical education for adult providers about "childhood" conditions needs to improve.

Access to knowledgeable providers, resources, and seamless care teams means pHCT is more effective than the adult paradigm. Children's hospitals commonly utilize MDC. These clinics enable children to conveniently follow-up with all required healthcare providers in a single, coordinated day, increasing collaboration (Ropars et al. 2020; Salort-Campana et al. 2024; Leach et al. 2021; Wakimizu et al. 2022). Comments about absence of adult MDC were often tied to experiences of limited guidance from providers, difficulty navigating the adult healthcare system, and discrepant/disjointed messaging between providers. Institutions willing to implement adult MDCs for complex medical conditions like SMA are likely to improve patient satisfaction and positive health outcomes (Salort-Campana et al. 2024; Gwathmey and Heiman-Patterson 2023). While the potential for lost revenue has been a common rebuke for MDC implementation (Gwathmey and Heiman-Patterson 2023; Paganoni et al. 2017), interviewees in this study expressed their willingness to travel for these services - potentially leading to downstream revenue for institutions willing to offer MDC. Continuation of MDC within the adult healthcare system may alleviate emotional, physical, and mental stressors (Salort-Campana et al. 2024; Badour et al. 2022; Rad et al. 2022).

Pediatric providers will see 100's of patients navigate the transition process; however, patients will only complete this once - making them less aware of pitfalls. Sarmiento et al. highlights the perceived barriers and facilitators young adults with cerebral palsy and their caregivers face when transitioning to the aHCT. They noted the most common barriers participants experienced are loss of security and the difficulty of leaving an established provider, novel care models, and limited number of knowledgeable providers (Sarmiento et al. 2024). Experiences highlighted in this study align with Sarmiento et al.'s conclusions, reinforcing how a disjointed transition experience can negatively impact care. While not always pragmatic, institutions should be mindful of these obstacles and work to create a tangible

handoff process between p/aHCT within and between health systems. Strategies detailed in the Got Transition® Six Core Elements may aid this handoff and be particularly impactful for individuals with a disability (Varty et al. 2020; Got Transition 2026). Increasing knowledge and access to disability support programs and specialty providers should be a priority. Currently, a patient's medical home may have limited familiarity with disease pathophysiology and treatments, requiring patient-led education. This may impact new working relationships with these adult providers. Interviewees desired their aHCT lead the education about SMA-specific support groups and community organizations.

A review by Badour et al. outlined evidence from several studies highlighting the positive impact supportive parents have on transition (Badour et al. 2023). Similarly, friends and the SMA community served as important facilitators in successfully navigating transition care; including identification of and procurement of financial aid and resources. Organizations such as Muscular Dystrophy Canada, CureSMA, MDA and local community groups were cited as providing interviewees the opportunity to connect with peers at similar life stages (Chen 2020). Unfortunately, some of our interviewees were unaware of these communities during transition. Providers are encouraged to learn about patient community groups, highlighting their ability to serve as facilitators (Varty et al. 2020; Got Transition 2026).

Literature has shown that adults with SMA experience high levels of distress, anxiety and depression in response to the progressive nature of their diagnosis, specifically relating to fears of functional and physical decline (Wan et al. 2019; Budych et al. 2012; Kruitwagen et al. 2016; López-Bastida et al. 2017; Qian et al. 2015). Interviewees described conversations about mental health as limited across their pediatric and aHCT. This may adversely impact wellbeing and perceived ability to accomplish adaptive tasks (Badour et al. 2023). While many of these experiences with mental healthcare are likely applicable to the broader population, special consideration should be granted for individuals with risk factors for psychiatric conditions, such as chronic health concerns (Ropars et al. 2020; Hunter et al. 2016). Early conversations about and detection of mental health conditions can help improve a patient's quality of life and reduce complications throughout adulthood (Hunter et al. 2016; Mulvaney-Day et al. 2018). Additionally, increasing these conversations may foster greater patient rapport.

Interviewees reported that discussions about mental health were self-initiated and societal barriers felt burdensome and negatively impacted self-esteem. These psychological challenges are likely intensified by the lack of disability representation within healthcare and society, evidenced by interviewees commonly speaking about both phenomena together. Interviewees expressed a need for improved disability representation and visibility in the medical community. Interviewees stated that this would allow them to feel additional comfort when explaining their challenges and decrease feeling solely responsible for the education of the able-bodied medical community.

1.4.1 Study Limitations

Limitations of this study include a limited number of interviews and the self-selective recruitment process. It is possible that our interviewees represent a skewed cohort among the wider population of

individuals living with SMA. Individuals who were willing to participate in phone interviews may be more likely to have a strong negative or positive healthcare experience. Finally, concerns addressed by participants may have geographic boundaries which can lead to challenges in drawing broader conclusions.

1.4.2 Future Directions

Future research is needed to determine the feasibility and impact of implementing adult MDCs. Clarification on the impact of early mental health discussions to overall well-being can help providers understand their utility. Lastly, care guidelines specific to adults living with SMA may provide training and educational opportunities for adult providers.

Conclusion

Our findings suggest that pediatric healthcare for individuals living with SMA generally results in more positive experiences and a higher quality of care compared to the adult paradigm. Limited connection to adult providers, lack of mental health awareness, reduced number of adult MDC, and limited transition guidance were highlighted as impacting this transition. These themes suggest a need for the integration of MDC into the adult healthcare system, alongside improved patient guidance for successful transition. Expanding discussions about the impact of mental health during this process and the availability of resources is imperative to the overall well-being of adult SMA patients. Pediatric and adult providers need to recognize the additional stressors and responsibilities disabled individuals experience as they begin to manage their care as an adult. Provision of more direct guidance during their transition process may be achieved by: outlining how their care is likely to change, where to access resources, identifying what they desire in an adult provider, and discussions about mental health, community engagement and disability representation.

Statements and Declarations

Contribution to work:

Elisa Lapadula- Conceptualization, data curation, formal analysis, investigation, methodology, project administration, resources, visualization, writing- original draft

Sophia Ceulemans- Conceptualization, methodology, resources, visualization, writing-review & editing

Homira Osman- Conceptualization, methodology, resources, visualization, writing-review & editing

Emily Meier- Conceptualization, methodology, resources, visualization, writing-review & editing

Dylan Platt- Conceptualization, data curation, formal analysis, investigation, methodology, project administration, resources, supervision, visualization, writing- original draft, writing-review & editing

Compliance with Ethics Guidelines:

This study was classified as non-human-subjects research by Augustana University Institutional Review Board under SU23.01. All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2000 (5). Informed consent was obtained from all patients for being included in the study.

Conflicts of Interest:

Elisa Lapadula declares that she has no financial or other conflicts of interest.

Sophia Ceulemans declares that she has no financial or other conflicts of interest.

Homira Osman declares that she has no financial or other conflicts of interest.

Emily Meier declares that she has no financial or other conflicts of interest.

Dylan Platt declares that he has no financial or other conflicts of interest.

Data Availability Statement:

In the interest of ensuring interviewee anonymity, especially considering each individual is part of the rare disease community, we are unable to provide the interview transcripts for distribution. However, a copy of the final code book is available as an online resource.

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Figures

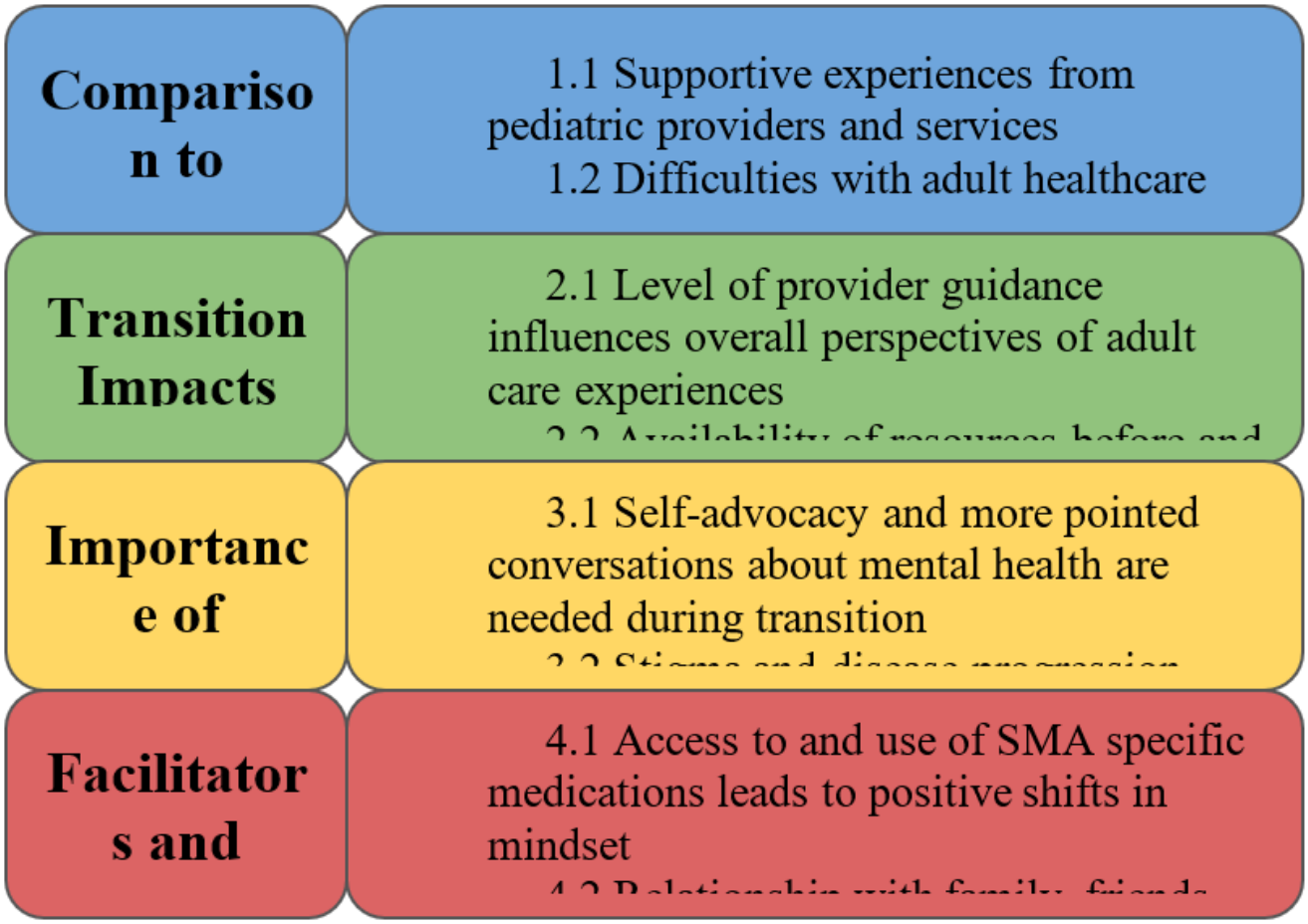


Figure 1

Major themes and sub-themes identified from interviewee responses. The interviews were analyzed and a code book was developed to identify commonalities and differences among interviewee's responses. Codes were grouped into major themes and sub-themes with direct quotes representing each theme and sub-theme.

Supplementary Files

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