


“I Want That Life a Lot...How on Earth Do I Get That?” Examining Challenges for Men With Barth Syndrome in Their Transitions to Adulthood

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Abstract

For youth with life-limiting chronic illnesses, transitioning to adulthood in line with age-norms may be difficult due to symptom severity and shortened survival. This study explores whether individuals with Barth syndrome (BTHS), a condition uniquely characterized by extreme prognostic uncertainty, experience similar or different challenges compared to youth with other conditions. During focus groups with adults with BTHS ($n = 12$) and caregivers ($n = 13$), participants reported that the ability to independently manage one's health condition, the social/emotional impacts of BTHS, and the ability to set goals in the context of future uncertainty challenge their transition to adulthood. This aligns with prior research, indicating that prognostic uncertainty may hinder long-term goal setting in BTHS. Implications of these findings include providing strategies for identifying meaningful alternative goals for individuals with chronic illnesses to target, promoting increased autonomy earlier in youth, and fostering coping strategies to manage non-disease related impacts.

Keywords

chronic illness, patient perspectives/narratives, qualitative methods, quality of life, transitions of care

Introduction

Historically, chronic pediatric illnesses affected a relatively small number of young adults. However, improvements in survival with some diseases, along with increased prevalence of others, now mean that approximately half a million Americans with chronic pediatric illnesses turn 18 years old each year (1). Thus, attention to how these individuals make the transition into adulthood is important to an increasing number of youth and their families.

Members of the U.S. public can readily identify ages at which they feel people should have completed transitions to adulthood like finishing school, starting a career, and marriage (2). Scholars have also focused on being “off-time,” the experience of consequences such as social judgment, narrowed opportunities, and financial loss, for individuals whose life courses did not meet prescriptive age norms. Compared to their healthy peers, youth with chronic illnesses often fall off-time, especially in the domains of school and work, leading to socioeconomic disparities in adulthood (3).

People with chronic pediatric illnesses such as cystic fibrosis, spina bifida, and Duchenne muscular dystrophy

(DMD) have limited life expectancy (4–6). Therefore, these illnesses alter the very concept of adulthood. When people perceive their future time horizons to be limited, they target short-term, emotionally-based goals that are consonant with their current abilities (e.g., fostering relationships), as opposed to long-term, knowledge-based goals requiring greater investment of time and money (e.g., educational attainment) (7). Young men with DMD, for example, report little motivation to work towards adult goals because of their foreshortened future (4). Therefore, there is remarkable consistency in the perspectives of youth with life-limiting illnesses on three barriers to transition to adulthood (4–6,8,9).

The first barrier is capacity for independent health management. Level of psychological maturity and ability to

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perform self-care affect the transition from pediatric care to adult health services, and the ability to live independently (10). Second, the trauma of illness and treatment may leave youth with mental health comorbidities such as anxiety, depression, post-traumatic stress disorder, and/or an openness to engage in risk-taking behaviors such as substance abuse (8). Navigating these challenges can delay school completion, independent living, and financial stability. Third, while youth with chronic illnesses espouse the same broad goals as healthy youth do, they often cannot meet those ideals (8). Parents and chronically-ill youth face the challenge of determining whether goals like college completion are feasible, or whether they are a worthwhile investment in the face of shortened life expectancy (5,11).

The present research extends the investigation of these barriers to adulthood to males with Barth syndrome (BTHS), an early-onset genetic condition that involves extreme prognostic uncertainty. BTHS is rare, with an estimated incidence of 1 in every 300,000–400,000 live births (12), and is characterized by weak cardiac and skeletal muscles, fatigue, infection, and growth delay. Cardiac complications pose the greatest threat to survival, such that death is often sudden and unpredictable. Approximately one-third of individuals with the condition die before the age of 5 (13). Yet over the last 20 years, survival has improved due to advancements in disease diagnosis and management, such that a third of known individuals with BTHS are over 15 years of age (14).

However, all individuals with BTHS thus far have died prematurely, with none on record as living longer than their 50s. We aim to learn whether this lack of clarity in progression and prognosis make the transition to adulthood different for males with BTHS as compared to youth with other conditions that are equally life-limiting, yet more predictable. Individuals with BTHS may similarly struggle with adulthood transitions due to disease severity and physical limitations, or alternatively, may leverage the hopeful aspect of prognostic uncertainty to set and meet future-oriented, on-time goals.

Limited research in BTHS has quantitatively demonstrated impacts to psychosocial, physical, emotional, and school functioning (15,16). Studies have primarily evaluated participants younger than 18, finding that younger age related to poorer physical, emotional, and social functioning. One qualitative study reported that the physical symptoms of BTHS (e.g., fatigue, weakness) negatively affect physical, school/work, social, and emotional functioning, self-perception, and activities of daily living for individuals 2–34 years of age (17). However, the existing research lacks an in-depth exploration from the perspective of young adults regarding how BTHS impacts their life transitions.

In this study, we asked individuals with BTHS and their family caregivers to identify the challenges to on-time transitions to adulthood. This project adds to the qualitative research exploring the lived experiences of individuals with BTHS, and contributes more broadly to the literature on becoming an adult while coping with a chronic pediatric illness.

Methods

Data were collected in July 2018 at the Barth Syndrome Foundation (BSF)'s International Scientific, Medical & Family Conference, a biennial conference of BTHS community members, clinicians, and researchers. Although there are fewer than 500 diagnosed individuals in the BSF global registry, in 2018, 228 patients and family members from 12 countries attended (18,19). The conference represents an important opportunity to conduct research with a significant proportion of a small and far-flung population.

Individuals with BTHS aged 17 and older and their caregivers were invited to participate in focus groups during the conference. Three focus groups, each containing between 10 and 17 participants, were held: One combined session of adult men and caregivers, one session for adult men, and one session for caregivers. Twelve individuals with BTHS and 13 caregivers participated in at least one of the sessions. Participants could attend an individual/caregiver specific session and the combined session. Seven individuals with BTHS attended two sessions and five attended one session. Six caregivers attended two sessions and seven attended one session.

All participants provided written informed consent. Parental consent and minor assent were collected for one seventeen-year-old participant. The first author and a second volunteer, both trained in qualitative research or social work with experience in interview/focus group methodologies, moderated the sessions. This training, as well as the co-moderation technique, enabled successful facilitation of these large focus groups. The sessions lasted approximately an hour, and the facilitators used a discussion guide consisting of open-ended and targeted questions regarding adulthood transitions following high school, including making educational, employment, and/or social decisions; caregiver relationships and communication; goal setting; and preferences regarding medical care. Participants did not complete any demographic forms or surveys. Each session was audio-recorded, transcribed, and anonymized.

Directed content analysis served as the methodology for data collection and analysis (20). This deductive approach allows for replication and expansion of social scientific knowledge on a topic, acknowledging that researchers have access to prior theory and empirical literature to inform their analysis. The first author was responsible for all coding. Coding followed an iterative process, whereby the researcher read the transcript from the combined focus group line-by-line, identifying transcript text and coding instances where participants reported any of the three main themes in the literature on adulthood transitions in chronic, life-limiting pediatric illness (i.e., independent health management, mental health, and goal-setting), adding themes as relevant. The researcher applied these preliminary codes across each transcript and making modifications as needed (e.g., consolidating/renaming codes) to arrive at a list of salient themes across the sessions.

Results

The three key themes that prior studies in chronic pediatric conditions (4–6) identified also emerged in this sample: (1) transition to independent disease management; (2) social and emotional barriers; and (3) goal setting uncertainty.

Transition to Independent Disease Management

Disease management is complex in a severe, multi-system condition such as BTHS. Individuals reported wanting to take greater responsibility for their medical care, such as self-managing their medications. Some men preferred that their caregivers no longer participate in their interactions with healthcare providers. Yet, individuals acknowledged that they might not be able to effectively transition to fully independent disease management and continued to rely on caregiver support.

Adult with BTHS: “She [caregiver] tends to exaggerate things... we just had, I had a cardiac arrest...the meeting afterwards she tends to make things sound worse... it’s hard to say whether I’m trying to make things sound not as bad or... where it falls, but to me she is making things sound worse than they really are.”

Adult with BTHS: “...my mom...shook this pill box and she was like, “You missed a day!” I’m like oops.”

While caregivers wanted to support the transition toward independent care management, they doubted individuals’ ability. Caregivers typically had few expectations that individuals would participate in their care in childhood, and in adulthood, were not convinced that they could be fully responsible for managing complex care needs. Some caregivers reported feeling more concerned about symptoms and medical events, such as cardiac arrests, than the men themselves. This resulted in some caregivers lacking trust in individuals’ ability to take these health concerns seriously and to self-advocate.

Caregiver: “And I think with...the complexity of some things, that he really couldn’t advocate for himself...when he had the cardiac arrest... I said call the cardiologist on-call, and he does, and he talks to them, and...you sit back, but...if he was living by himself, it makes me sick to think about dealing with what happens...”

Caregiver: “It’s a constant struggle to get [individual with BTHS] to take his medications like he’s supposed to, and he’ll sit at home and be feeling bad and blue around the mouth, and

he knows he’s going to feel bad... I don’t know if it’s laziness...”

Caregiver: “As an adult, who’s been an adult for a long time, you’ve had so much time to learn so much about Barth syndrome itself, but also the time to have the confidence to be able to ask the questions...your 18-year-old hasn’t had the time to do all of that yet, but has all the responsibility....”

Social and Emotional Barriers

Individuals reported feeling socially isolated because they felt categorically different from their peers in adolescence. They found it difficult to relate to others their age, given their physical limitations and need to focus on health-related issues. Failing to share in similar interests and experiences further cemented individuals’ sense of being different from their peers:

Adult with BTHS: “I never really compared to anybody my own age, because I couldn’t keep up with them, I couldn’t do the same stuff they did. I wasn’t even interested in the same stuff they were because I couldn’t do it, so it makes perfect sense to me that well of course...I don’t get along with them, I have no parallel with them.”

Adult with BTHS: I was always...around adults, whether it was in hospital or at home, so you learn how to relate to more things that are very adult-like and you kind of lose your almost childhood in a weird way... I wouldn’t be going and talking about what normal kids would be, right, so therefore trying to relate to them and trying to make friends, if you can’t relate to someone, you’re not friends.”

Individuals described experiencing varying levels of depression, loneliness, and self-destructive behavior (e.g., substance abuse) that hindered their motivation to pursue meaningful activities:

Adult with BTHS: “...I had some difficulties, some severe difficulties around the age of 16 and 17, around the age of finishing secondary school. I was suffering a lot physically and didn’t have any friends really almost all the way through secondary school. I didn’t have anybody I was close with...I’m not in the best state now, how many

of us are really? I'm showing all...the physical symptoms of depression as in like the sitting, shut-in, not moving or doing anything really..."

Adult with BTHS: "In my head I can do ten times what I'm physically capable of doing, and so that's just an automatic recipe for disaster. I hold myself to a far too high a physical standard and of course when I can't meet it, because there's no way I can, it's an instant recipe for depression...to cope with depression and pain and just general discontent with life, I self-medicated."

Individuals perceived their caregivers to be aware of these emotional/mental health issues, despite many reporting not openly acknowledging these experiences.

Caregivers similarly reported that these issues were mutually understood although not typically discussed. Some caregivers did not want to increase the emotional burden on the individual by raising additional issues. This was illustrated in the dialogue between parents below:

Caregiver 1: I don't talk to [individual with BTHS] about it...Because I feel like he has enough work...And he worries about me.

Caregiver 2: To talk about what the, the fear of their, their health.

Caregiver 3: Fear of them getting sick or them possibly dying.

Goal Setting Uncertainty

Individuals were not certain which "on-time" goals were feasible and worth pursuing after completing high school, given their health limitations and uncertain prognosis.

Adult with BTHS: "...do I really want to start big things like going to school for the next four years or six years or for even thinking about a PhD because am I even going to be around? A lot of the guys we've seen just go from healthy with Barth to just boof!"

Adult with BTHS: "...I want that life a lot...with the wife and the kids and the grandkids and the extended family...how on earth do I get that, because to support a family you need like a good job, you need money, you need a good career."

Some youth prioritized their health over longer-term goals. Others took incremental steps toward goals, yet unpredictable

and/or severe symptoms often interfered. One individual described tailoring his approach to identifying a more feasible, personalized goal with a greater probability of success:

Adult with BTHS: "I did go to a four-year college, and it was too much even just getting there, getting the subway, coming up the subway steps, walking to the building, taking the class, taking the two or three classes a day and then getting back home took too much out of me, so now I'm going to community college for two years."

Caregivers reported being unsure when and how much they should encourage individuals with regard to goal setting and attainment after completing high school. Most adults still lived with their caregivers, and caregivers struggled with whether they should expect men to engage more in routine daily activities, such as household chores. They had further questions about life goals such as independent living, college attendance, and work for pay.

Caregiver: "We always thought [individual with BTHS]'s timeline was nonexistent. The girls were supposed to graduate high school at 18. They were supposed to get their bachelor's degree. [Individual with BTHS], it was we'll see what happens."

Caregiver: "...right now we're kind of in a holding pattern...I think we're still in a very frozen point of view as to where we go from here because we are scared to death, truthfully."

Caregivers sought to support the transitions to adulthood, yet having focused almost exclusively on individuals' survival to date, caregivers were unsure how to move forward. Some had not expected their sons to live to adulthood.

Discussion

Individuals with BTHS and caregivers described the key challenges to transitioning to adulthood as (a) difficulty taking ownership over disease management, (b) social/emotional barriers from an early age, and (c) uncertainty impacting goal setting. These themes align with prior challenges described by youth with other life-limiting illnesses (4,6).

Implications

A key implication of this work is the need to address the lack of culturally available scripts to identify alternative goals in the context of life-limiting illness. Similar research has questioned the relevance of dominant social constructions of time and adulthood, which can marginalize youth with chronic illnesses (4,6). Individuals may distance themselves from their

condition, making it difficult to tailor their adulthood transitions such that these are characterized by milestones meaningful to their circumstances (9). Some chronically-ill youth default to living in the present without longer-term goals, without endeavoring to transition to adulthood (4,6).

A second implication concerns medical self-sufficiency. To support transitions to independent adulthood, caregivers may begin shifting responsibility for managing BTHS earlier in adolescence, to build trust and competence in self-care (11,21). Medication management emerged as a key element of developing autonomy. Given the complex care needs of BTHS, caregivers require support from medical professionals and patient advocates in order to safely approach this transition.

Third, adults and caregivers discussed the unspoken but profound nature of socioemotional impacts, including the prospect of an early death. Lack of open acknowledgment of these issues may serve as a further barrier to promoting effective transitions to adulthood. Adults and caregivers require supportive services in mental health to enhance individuals' motivation for setting and meeting meaningful goals that may otherwise appear futile or overwhelming to achieve.

Limitations and Future Directions

This sample consisted of individuals attending an advocacy group conference. Although a substantial portion of the identified BTHS community attends this conference, those who do not are surely different from those who do in systematic ways. People who do not attend may, for example, experience more difficult socioeconomic circumstances, which surely compound the challenges of transitioning to adulthood amidst chronic illness.

A single focus group was conducted with each population of interest (i.e., individuals, caregivers, and combined) limiting the ability to evaluate concept saturation. Additional themes may have emerged from further focus groups.

Demographic characteristics were not collected, so we cannot characterize the sample in terms of goal attainment related to employment, education, and marriage. Finally, longitudinal research from adolescence through early adulthood could identify how goal setting and attainment evolve over time, and whether there are periods when intervention might be especially successful.

Despite these limitations, this research identified key barriers to transitioning to adulthood with BTHS elicited from a large sample of individuals relative to the rarity of BTHS. From a young age, individuals and their caregivers may be unable to gauge available time horizons. This contrasts with better-understood conditions with a more predictable course of illness. Yet people with BTHS identified themes similar to those that young adults with other serious chronic illness have identified, demonstrating that they similarly struggle to meet the "on-time" expectations which define healthy people's successful transitions to adulthood.

Authors' Note

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Declaration of Conflicting Interests

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Ethical Approval

Ethical approval to report this case series was obtained from the Boston College Institutional Review Board (IRB Protocol Number: 18.274.01).

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References

1. Perrin JM, Bloom SR, Gortmaker SL. The increase of childhood chronic conditions in the United States. *JAMA*. 2007;297(24):2755-9.
2. Riley MW, Johnson M, Foner A. *Aging and Society: A Sociology of Age Stratification*. New York: Russell Sage Foundation; 1972.
3. Pinquart M. Achievement of developmental milestones in emerging and young adults with and without pediatric chronic illness--a meta-analysis. *J Pediatr Psychol*. 2014;39(6):577-87.
4. Gibson BE, Zitzelsberger H, McKeever P. "Futureless persons": shifting life expectancies and the vicissitudes of progressive illness. *Sociol Health Illn*. 2009;31(4):554-68.
5. Holbein CE, Zebracki K, Bechtel CF, Papadakis JL, Bruno EF, Holmbeck GN. Milestone achievement in emerging adulthood in Spina Bifida: a longitudinal investigation of parental expectations. *Dev Med Child Neurol*. 2017;59(3):311-6.
6. Moola FJ, Norman ME. "Down the rabbit hole": enhancing the transition process for youth with cystic fibrosis and congenital heart disease by Re-imagining the future and time. *Child Care Health Dev*. 2011;37(6):841-51.
7. Carstensen LL, Isaacowitz DM, Charles ST. Taking time seriously. a theory of socioemotional selectivity. *Am Psychol*. 1999;54(3):165-81.
8. Schwartz LA, Drotar D. Health-related hindrance of personal goal pursuit and well-being of young adults with cystic fibrosis, pediatric cancer survivors, and peers without a history of chronic illness. *J Pediatr Psychol*. 2009;34(9):954-65.

9. Steinberg H. Distance and acceptance: identity formation in young adults with chronic health conditions. *Adv Life Course Res.* 2020;44:100325.
10. Gray WN, Schaefer MR, Resmini-Rawlinson A, Wagoner ST. Barriers to transition from pediatric to adult care: a systematic review. *J Pediatr Psychol.* 2018;43(5):488-502.
11. Sparacino PS, Tong EM, Messias DK, Foote D, Chesla CA, Gilliss CL. The dilemmas of parents of adolescents and young adults with congenital heart disease. *Heart Lung J Crit Care.* 1997;26(3):187-95.
12. Jefferies JL. Barth syndrome. *Am J Med Genet C Semin Med Genet.* 2013;163(3):198-205.
13. Rigaud C, Lebre A-S, Touraine R, Beaupain B, Ottolenghi C, Chabli A, et al. Natural history of Barth syndrome: a national cohort study of 22 patients. *Orphanet J Rare Dis.* 2013;8(1):70.
14. Clarke SL, Bowron A, Gonzalez IL, Groves SJ, Newbury-Ecob R, Clayton N, et al. Barth syndrome. *Orphanet J Rare Dis.* 2013;8(1):23.
15. Storch EA, Keeley M, Merlo LJ, St JA, Jacob M, Storch JF, et al. Psychosocial functioning in youth with Barth syndrome. *Child Health Care J Assoc Care Child Health.* 2009;38(2):137-56.
16. Jacob ML, Johnco C, Dane BF, Collier A, Storch EA. Psychosocial functioning in Barth syndrome: assessment of individual and parental adjustment. *Child Health Care.* 2017;46(1):66-92.
17. Gwaltney C, Stokes J, Aiudi A, Mazar I, Ollis S, Love E, et al. Development and content validity of the Barth Syndrome Symptom Assessment (BTHS-SA) for adolescents and adults. *Orphanet J Rare Dis.* 2021;16(1):264.
18. Barth Syndrome Foundation [Internet]. Barth syndrome foundation FAQs; 2006 [cited 2021 Mar 20]. Available from: https://www.barthsyndrome.org/file_download/inline/8bd9d37d-6449-4687-99b3-7d78202475be
19. Larchmont, NY Patch [Internet]. 10th Barth syndrome foundation conference 1 year away. 2019 [cited 2021 Mar 20]. Available from: <https://patch.com/new-york/larchmont/20th-barth-syndrome-foundation-conference-1-year-away>
20. Hsieh H-F, Shannon SE. Three approaches to qualitative content analysis. *Qual Health Res.* 2005;15(9):1277-88.
21. Heath G, Farre A, Shaw K. Parenting a child with chronic illness as they transition into adulthood: a systematic review and thematic synthesis of parents' experiences. *Patient Educ Couns.* 2017;100(1):76-92.