Chapter 6
Transition from Childhood to Adult in Patients with Muscular Dystrophy

Elba Yesi Gerena Maldonado and Kathryn R. Wagner

Introduction

"We were the generation that was hoped for—and now we're here," announced L. Vance Taylor, a successful 36-year-old man with muscular dystrophy (Muscular Dystrophy Association (MDA) National Neuromuscular Transitions Summit, Washington D.C., September 23, 2011). In this one phrase, Mr. Taylor voices the current achievement and challenge that our healthcare system faces: an increasing adult population of patients with muscular dystrophy (MD) and a society that lacks the structure to support them.

In the past 30 years, we have seen an increase in the lifespan of patients with MDs especially those who have early onset of their disease [1]. The scientific and medical community has made great strides in the early care of patients with MDs and has published practice guidelines to promote better care for these patients [2, 3]. However, it has not been until recently that the community has started to address the quandary that is the transition phase from childhood to independent adulthood with complex medical conditions [4–7]. A survey done in the UK, where patients older than 15 years and their family members were interviewed from 2007 to 2009, showed that while advances in healthcare have increased life expectancy in patients with MDs, this has not been matched by an increase in support at home and in the community [4]. There is no doubt that the structured care coordination for pediatric patients with MDs is significantly different from the perceived lack of support these same individuals face once they become adults. The reason for this disparity is

---

E.Y. Gerena Maldonado, M.D. (✉)
Department of Rehabilitation Medicine, University of Washington,
1959 NE Pacific Street, Box 356490, Seattle, WA 98195-6490, USA
e-mail: egerena1@uw.edu

K.R. Wagner, M.D., Ph.D.
Kennedy Krieger Institute, Genetic and Muscle Diseases,
801 North Broadway, Baltimore, MD 21205, USA

© Springer International Publishing Switzerland 2015

R.A. Huml (ed.), *Muscular Dystrophy*, DOI 10.1007/978-3-319-17362-7_6

likely to be multifactorial; nevertheless, we need to start understanding the unique features of the transition period to develop effective strategies to maximize the likelihood of success in adult life.

This challenge is not unique to the field of MD. The National Alliance to Advance Adolescent Health estimates that chronic health conditions affect approximately 25% of the 18 million U.S. young adults ages 18–21, who should be transitioning to adult-centered healthcare. Each year, approximately 750,000 young people in the U.S. with special healthcare need transition to adult care [8]. The fields of cystic fibrosis, perinatally acquired HIV, and pediatric organ transplantation are just three examples that have similarly needed to adjust their thinking and prepare their patients for an adult life where, previously, there was none.

Our goal with this chapter is to highlight the importance of the transition period in the life of an individual with MD and to provide helpful resources available for guidance and support.

Emerging Adulthood

Emerging adulthood has been defined as "the period when often people explore a variety of possible life directions in love, work, and worldviews… where the scope of independent exploration of life's possibilities is greater for most people than it will be at any other period of the life course" [9]. It is usually during this period when people start to mold their personalities and define themselves in their community. Therefore, it is of the upmost importance that our society provides the adequate groundwork on which young adults, especially those with disabilities, can establish and design their own life stories.

There are various psychological models of development that have been put forward to explain the key stages in an adolescent's life. Some of these models describe this period as filled with change in which young adults are assuming new roles in all facets of their lives. They can experience conflicting feelings between the excitement about the prospect of autonomy and the sense of abandonment due to the previous dependent nature of their relationships with their parents and/or caregivers [10].

The literature suggests that relationships with parents remain the most influential of all adolescent relationships [11], and their support has been associated with good outcomes [12]. During this period, the parents need to talk frequently with their child about his/her interests so that a transition plan can be built upon these interests. The parents need to familiarize themselves with various local and state agencies and file for appropriate services. Most importantly, the parents need to foster and not limit their child's dreams.

Previous research has mainly focused on retrospective post-transition interviews, and less is known about younger adolescents and their families as they approach the transition period. Given the complexity of the arrangements in which these children grow up, addressing all of their needs at the same time is a challenging task. Moreover, it can be counterproductive in the end, as it can be overwhelming for these young adults to be made responsible for all of their healthcare needs in a short amount of time. Their care should be introduced as a step-by-step process where they are included in decision making, information sharing, while at the same time their parents remain informed and involved. In a study done in Denmark in 2011, 16 young adults with cystic fibrosis were interviewed about their transition experience. One of the highlighted points in this study was that these patients preferred to have a gradual transfer of responsibility of their own medical care during their young adulthood [13]. Similarly, experts in Duchenne muscular dystrophy (DMD) urge that transition not be thought of as a single event, but as a continuing process of increased choices and autonomy [5].

Timetable for Growing Up

Parents and physicians may differ in their perceptions of when to begin teaching children about self-management of their healthcare. The mean age identified by parents is 12 years, while that of pediatricians is 9.5 years [11]. So, the question remains, when should we start the transition period for children with physical disabilities, in particular those with MDs?

Due to each child's unique circumstances, it would not be prudent to stipulate a definitive age when parents should start the conversation of transition with their child. Nevertheless, preparation must begin early enough to ensure adolescents develop the knowledge and skills to take ownership of their chronic illness and disease management in an effective manner.

Some of the most common methods that are being used to start the transition stage have been the implementation of "timetables for growing up". These transition plans have some variability in their designs; nonetheless, most agree that certain "transition topics" should start to be discussed around the ages of 12–13 years. For example, the timetable provided by the Holland Bloorview Kids Rehabilitation Hospital or the MDA's "Road Map to Independence," which encourages conversations about topics of human development and social interactions when the child is at the pre-to-early adolescent stage. Once they reach the mid-to-late teens, the timetables focus on developing independent living skills (e.g. contributing to discussions and decisions regarding their medical treatments, being assigned chores around the house, driving vs. public transportation as means of mobility, etc.). Topics like future career goals should also be addressed in a timely manner, as it has been found in the literature that children with disabilities worldwide tend to be excluded from certain schooling subjects (due to lower rates of school attendance given their multiple medical appointments and lower transition rates to higher levels of education) when compared to kids without physical and/or cognitive impairments [14]. Exclusion from education has an immediate impact on a child in terms of exclusion from social participation, reduced personal well-being and welfare, and likely dependence on caregivers.

Commencement of Transition Years

The parents' approaches to promoting health and well-being for their adolescents with physical disabilities tend to be diverse. Some see themselves as the primary source of information; others rely solely on school, while a few others view friends and the media as a resource. Some parents have expressed concerns and anxiety about the fact that they believe that their adolescent with a physical disability is going to encounter greater difficulties in relationships and sexual expression [15]. All of these factors will influence the outcomes of the transition period in patients with physical and/or cognitive disabilities. Given the dual role of the parents as both progenitors and caregivers for these children, the literature has described the tendency of these parents to be overprotective [16]. However, another explanation for their "overprotection" during the transition period is that their behavior is a deliberate strategy to provide their adolescent with more time and energy for school and friends.

Nevertheless, the goal of the transition years is to promote the development of skills for independence in multiple facets of the teenager's life. One of the major concerns that parents of children with disabilities have is the development of a healthy social life and integration into their community [4]. Parents have dual worries of how their children will cope with the changes that all human beings go through while growing up and at the same time adding to the process the complexity of their physical disabilities. It is for this reason that both health providers and caregivers should address socialization of adolescents and young adults prior to them reaching adulthood.

It is important for patients with MDs to feel that they are an integral part of society. At the same time, they benefit from certain events or activities where their physical disabilities are normalized. Several organizations provide free camp experiences for those with MD. The MDA organizes a one-week summer camp for children and young adults (ages 6–17) where they can meet other youth with a variety of neuromuscular disorders and share their life experiences (http://mda.org/summer-camp). The Jett Foundation offers a week-long "Camp Promise" for those with MD (https://www.camppromise.org/). If the young adult is interested in the fine arts (music, theatre, visual arts), there are programs like the International Organization on Arts and Disability providing career building opportunities in the arts both nationally and internationally (http://www.kennedy-center.org/education/vsa/).

Self-Advocacy

Being a strong self-advocate is necessary for the youth with MD. No one will understand his/her particular manifestations of disease and resulting needs as well. Being able to effectively communicate these needs, propose solutions for how they should be met, and engage others in these solutions are key factors in success.

As emerging adults, a balance should be struck between establishing independence and receiving support from parents/ caregivers. These young adults need to have a good understanding of their condition, be it a fast or slowly progressive MD. Health literacy is a priority.

There are multiple opportunities for the youth to become an advocate for the MD community. Local groups sponsored by the MDA provided supportive environments for discussion and activities. If the adolescent is interested in groups that are focused on their own specific diagnosis, there are various groups which provide information and support: PPMD for Duchenne and Becker muscular dystrophy, FSHD Society and the Friends of FSH for FSHD, and the Myotonic Dystrophy Foundation, for myotonic dystrophy, to name a few (websites provided below).

Social

In this era of globalization that has risen from the advances in the telecommunications infrastructure, it is easy to acquire large amounts of information, some of which might not be necessarily correct or appropriate for certain age groups. It is for this reason that certain topics should not be taboo at home or school and should be addressed by a trustworthy adult. Children are exposed to subjects like smoking, drug use, and alcohol on a daily basis. Topics related to sexuality, contraception, and preconception counseling and surveillance should also be addressed. The existing literature is scarce in research concerning sexual health and MDs [17]. Openness and knowledge are integral for a successful rapport with the adolescent with MDs seeking counsel in these matters. Common barriers that affect the success of these talks are the sensitivity of the subject matter, the parents' insecurity in how they can be supportive, or the parents' abdication of the discussion of sexual health to someone else.

The adolescents themselves might be reluctant to initiate communication about sexual health problems, especially if they believe that the adult is unavailable to offer support or if they believe that they shouldn't be sexually active due to their disability. It is for this reason that they should have a support group identified which can reliably provide additional accurate information and advice.

Young adults are faced with a world that increasingly is more available through different communication devices. Although these devices, such as computers and mobile devices, can be used as assistive devices for communication, they might also promote isolation. The social integration in the community cannot be overemphasized, given the fact that the natural history of growing up is that certain childhood friendships grow apart as kids become teenagers and eventually adults.

Education

The Individuals with Disabilities Education Act (IDEA) provides federal financial assistance to state and local educational agencies to ensure that students with disabilities receive an education "designed to meet their unique needs and prepare them for employment and independent living" (http://idea.ed.gov/explore/home). IDEA was reauthorized in 2004 with very specific language about transition planning. One of the primary methods by which IDEA is enacted is through Individual Education Programs (IEPS). It is imperative that children with MDs be gradually integrated in the parent–teacher meetings about their education. They should be aware of what their IEP entails, and how it is being tailored to not only their physical needs but also their academic interests. They should talk openly about the careers that they want to pursue and explore opportunities with volunteer work or part-time jobs.

By the time students are starting high-school, they should be contacting the Disability Support Services Office of various post-secondary schools. Students need to self-identify and self-advocate whether they choose a vocational school, community college, or university. There is a wide range of accommodations made for post-secondary students with disabilities from schools such as Edinboro University in Pennsylvania (www.edinboro.edu), which has dozens of students making use of attendant care, meal aides, homework aides, and on-campus wheelchair repair services to the majority of colleges which are barely wheelchair accessible [18]. Several universities and colleges provide opportunities for students with disabilities to have exposure to future employers. Programs like the DO-IT (http://www.washington.edu/doit), based at the University Of Washington College Of Engineering, help in bringing together students and employers in a setting that can provide career advancement for these patients. The process for university students to acquire or receive services tends to be determined by how proactive students are to advocate for themselves and provide clear documentation of the disability. A self-advocating student in a college with a strong Disability Support Services Office is a combination that yields a safe and rewarding new home transitioning from the parents' home to complete independence.

Self-Care

Mastering normal developmental tasks, such as household chores or an after-school job, can help boost the self-confidence and independence of an adolescent with MD. As the child begins to manifest more autonomy, a clinical checklist updated on each visit prompts review and discussion of the patient's progress toward self-management and eventual transition.

By the time the child is in the pre-teen to early teenage years, conversations about future living situations should be brought to the table. They should be slowly progressed to learn independent living skills. They should also practice budgeting and banking skills. Some clinics provide counseling with social workers or case managers that are able to guide the family in regard to community resources for their children.

Many states offer transition programs and services to help prepare individuals with a developmental disability to gain employment and independent living. These services may include employment counseling, employment training, assistive technology, and independent living skills training. There is wide variability in the number, types, and funding of these services across the nation.

Transition to Adult Healthcare

A well-organized and well-timed transition from child-oriented to adult-oriented healthcare allows young people to optimize their ability to assume adult roles and functioning in society. For many young people with special healthcare needs, this will mean a transfer from a pediatric to an adult healthcare professional; for many others, it will involve an ongoing relationship with the same provider but with a reorientation of clinical interactions. With a successful transition, healthcare is uninterrupted, function is maximized, and morbidity and mortality are reduced. There are several challenges, however, to obtaining uninterrupted healthcare services during the transition period. These include the lack of a formalized transfer linkage between pediatric and adult medical services, which frequently leaves patients and families to personally assemble their own adult medicine team. A consensus statement of the American Academy of Pediatrics, the American Academy of Family Practice, and the American College of Physicians recommended a written heathcare transition plan by age 14 but in the experience of the authors, such a plan is rare from MD providers [7]. Unfortunately, pediatric patients lose their medical safety net at the same time that they are losing a social safety net, that of their secondary school and the services that it often provides.

Pediatric and adult healthcare systems are structured differently. Children are frequently cared for at children's hospitals equipped with multiple disciplines, which not only include their physicians, but also include social services, education, vocational training, and recreation. The processes of teaching them to take responsibility for their own healthcare needs are vital, as they will need to become their own advocates once they transition to adult healthcare. Some of the adult neuromuscular medicine clinics also provide services that include assessment, consultation, and intervention from various disciplines including physiatrists/neurologists, cardiologists, pulmonologists, occupational therapists, physiotherapists, social workers, and genetic counselors. However, in a survey of patients with adult MD, they were more likely to receive services from fewer health professionals and were less satisfied with their current healthcare compared to when they were children, which was often associated with a decline in health outcomes [4].

Successful transition from pediatric to adult medicine can be achieved by multiple different strategies. Some healthcare institutions have both adult and pediatric clinics where the transition is seamless with the youth being able to meet his new team prior to full transition, records staying in the same facility, and some specialists remaining the same person. Programs such as those in children's hospitals, which include a complete transfer of care at age 18 or 21, are implementing transition care programs that involve some continuity with the pediatric physician following transition to the adult team. There are a few examples of formal transition planning that adhered to national policy guidelines [7].

Improving clinical care for young adults with MD is one of the current objectives for the adult healthcare system. Advisory committees composed of those MD patients who have already navigated the transition process and suffered the consequences of a healthcare system not equipped for them are being developed to help identify barriers to accessing appropriate healthcare services needed for future generations. Among the areas that have been identified that need to be addressed, one of the most urgent ones is the facilitation of follow-up and referrals and enhancement of community linkages for services.

Conclusion

We have all witnessed that the lack of a carefully orchestrated transition plan results in a deterioration in the quality of life of young adults with MDs. However, those few who have been able to have a successful transition have attained skills necessary to manage their own personal needs and healthcare. They are effective self-advocates and are able to pursue their own interests. They have assembled knowledgeable and responsive medical teams who work diligently to free them from recurrent illnesses, pain, and hospitalizations. They find the balance between independence and the need for social/physical support in their lives acceptable. Our goal is to make these examples the norm of all young adults with MD who are transitioning into adulthood.

It is imperative that the MD community continues to work on strengthening the transition process to ensure success of the upcoming generations. Some clear current barriers are inadequate communication between pediatric and adult providers, an adult healthcare system unprepared to meet the various needs of these often complicated patients, the lack of integration of social services at the local and state level, and the scarcity of job opportunities for those with disabilities. There are fortunately several groups, such as those listed below, as well as governmental agencies that are highly invested in improving transition for youths. In addition, with proper guidance, these young men and women are likely to positively shape the environment for themselves and for the next generation.

Useful websites for young adults with MDs:

- http://hollandbloorview.ca/programsandservices/ProgramsServicesAZ/Growingupready/TimetableforGrowingUp
- http://transitions.mda.org/
- http://mda.org/summer-camp

- http://www.kennedy-center.org/education/vsa/
- http://www.bristol.ac.uk/norahfry/research/completed-projects/ecominganadult.pdf
- http://www.doe.virginia.gov/special_ed/transition_svcs/outcomes_project/college_guide.pdf
- https://www.dshs.wa.gov/jjra/division-vocational-rehabilitation
- https://www.dshs.wa.gov/dda
- http://wid.org/publications/downloads/Latinos%20with%20Dis.%20-%20Spanish.pdf

References

1. Eagle M, et al. Survival in Duchenne muscular dystrophy: improvements in life expectancy since 1967 and the impact of home nocturnal ventilation. Neuromuscul Disord. 2002;12(10):926–9.
2. Bushby K, et al. Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care. Lancet Neurol. 2010;9(2):177–89.
3. Turner C, Hilton-Jones D. Myotonic dystrophy: diagnosis, management and new therapies. Curr Opin Neurol. 2014;27(5):599–606.
4. Abbott D, Carpenter J, Bushby K. Transition to adulthood for young men with Duchenne muscular dystrophy: research from the UK. Neuromuscul Disord. 2012;22(5):445–6.
5. Schrans DG, et al. Transition in Duchenne muscular dystrophy: an expert meeting report and description of transition needs in an emergent patient population (Parent Project Muscular Dystrophy Transition Expert Meeting 17–18 June 2011, Amsterdam, The Netherlands). Neuromuscul Disord. 2013;23(3):283–6.
6. Rahbek J, et al. Adult life with Duchenne muscular dystrophy: observations among an emerging and unforeseen patient population. Pediatr Rehabil. 2005;8(1):17–28.
7. American Academy of Pediatrics, American Academy of Family Physicians, American College of Physicians-American Society of Internal Medicine. A consensus statement on health care transitions for young adults with special health care needs. Pediatrics. 2002;110(6 Pt 2):1304–6.
8. Goodman DM, et al. Adults with chronic health conditions originating in childhood: inpatient experience in children's hospitals. Pediatrics. 2011;128(1):5–13.
9. Arnett JJ. Emerging adulthood. A theory of development from the late teens through the twenties. Am Psychol. 2000;55(5):469–80.
10. Nakhla M, et al. Transition to adult care for youths with diabetes mellitus: findings from a Universal Health Care System. Pediatrics. 2009;124(6):e1134–41.
11. Slap GB. Adolescent medicine. The requisites in pediatrics, xix. Philadelphia: Mosby/Elsevier; 2008. p. 330.
12. Kyngas H, Rissanen M. Support as a crucial predictor of good compliance of adolescents with a chronic disease. J Clin Nurs. 2001;10(6):767–74.
13. Bregnballe V, Schiotz PO, Lomborg K. Parenting adolescents with cystic fibrosis: the adolescents' and young adults' perspectives. Patient Prefer Adherence. 2011;5:563–70.
14. Kuper H, et al. The impact of disability on the lives of children; cross-sectional data including 8,900 children with disabilities and 898,834 children without disabilities across 30 countries. PLoS One. 2014;9(9):e107300.
15. Antle BJ, et al. An exploratory study of parents' approaches to health promotion in families of adolescents with physical disabilities. Child Care Health Dev. 2008;34(2):185–93.

16. Holmbeck GN, et al. Observed and perceived parental overprotection in relation to psychosocial adjustment in preadolescents with a physical disability: the mediational role of behavioral autonomy. J Consult Clin Psychol. 2002;70(1):96–110.
17. Areskoug-Josefsson K. Muscular dystrophy and sexual health. OA Musculoskelet Med. 2013;1(2):17.
18. Tiedemann CW. College success for students with physical disabilities. Austin: Prufrock Press; 2012.