Transitions in Care in DMD

Patient #3: No Transition Plan

23 y/o Male

(Duchenne muscular dystrophy [DMD] associated with exon 12 deletion)

Case contributor and commentary:
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Pediatric Care

Early Life

Background:

- Knowledge of early care limited
- Diagnosed in early childhood

Age 8

Ambulation status:

- Lost ambulation

Age 8 - 17

Multidisciplinary team:

- Followed at a private neurology clinic.
- Cardiology and orthopedic care at separate hospital systems.

Overall health status:

- Stable for years

Caregiver:

- Patient's father was his primary caregiver and legal guardian

Commentary: A family member is often the most trusted source of caregiving, but caregiver fatigue, aging of the caregiver, and altered family dynamics can occur. It is therefore recommended to establish a respite plan for caregivers beginning in the patient's teenage years and to consider the hiring of a personal care attendant.[1]

Age 17

Cardiac status:

- Cardiovascular function was stable on lisinopril

Musculoskeletal status:

- Planned for surgical intervention of scoliosis

Transition in Care

Age 18

Change in caregiver status:

- Father died and brother became legal guardian
- Patient did not go to school for six weeks because of family's inability to dress him

Commentary: To prepare a young person with DMD for a transition in care, it is important to encourage them to practice instructing others on how to assist with activities of daily living (ADLs). This will lead to confidence when it is time to independently manage a personal care attendant or in the case of a sudden change in caregivers.[2]

Age 18

Unmet medical needs:

- Following the death of his father, the patient had the following unmet needs: no cardiology follow-up, lack of primary preventative care including immunizations, wheelchair in need of repair, no appropriate durable medical equipment (DME) for self-care after a decrease in upper extremity function

Multidisciplinary team workup:

- Referrals for social work, home healthcare, and physical/occupational therapy were made and DME prescriptions were written. Patient was advised to follow up with cardiology and orthopedics

Patient follow-up:

- Patient did not receive follow-up care for 14 months

Commentary: Poor compliance to routine follow-up guidelines including neuromuscular, cardiac, and respiratory specialist visits, physiotherapy, and access to medical devices and aids has been observed in a study of patients with DMD in a number of countries, including the United States.[3] Engaging a social worker to screen for barriers to care early in the patient's disease course and utilizing a nurse coordinator to facilitate scheduling of multidisciplinary appointments could support patients in compliance to follow-up care.

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% of patients in the US meeting guideline recommendations

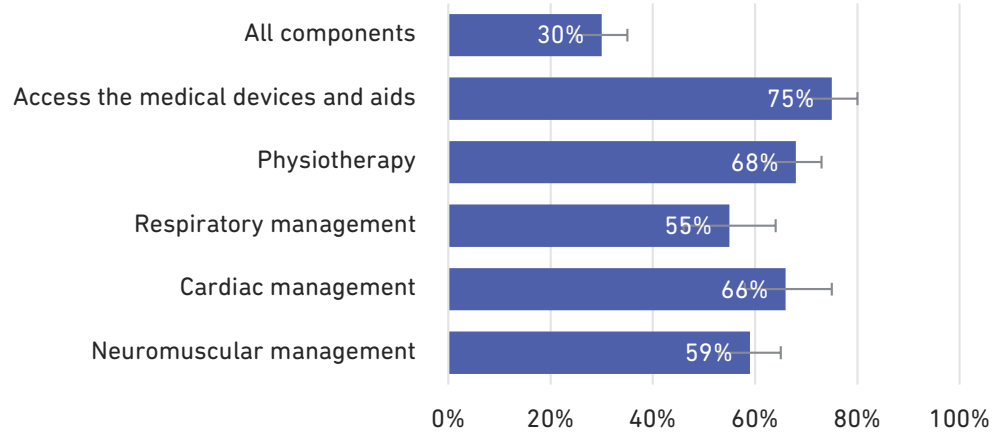

Figure adapted from reference #3.

Age
19 - 23

Change in clinical setting:

» The patient's care was transferred to an MDA Care Center

Patient status:

» Patient had not received follow-up care from cardiology and had been out of lisinopril for months

» Patient had not followed up with orthopedics and there was concern that surgical intervention would be prohibited by obesity

» Patient had no established pulmonary care, although denied symptoms of dyspnea or air hunger

» There continued to be unmet DME needs

Commentary: One study identified factors that contribute to the non-adherence of patients with DMD to physician recommendations. Identified factors included patient demographics (primarily age), financial barriers, and lack of access to specialists, among others.[4] Internalized behavioral issues, such as anxiety and depression, have also been identified as factors influencing adherence to recommended therapies.[5] Patients with DMD should be screened regularly for psychological disorders with scales such as the Strengths and Difficulties Questionnaire for pediatric patients and the Patient Health Questionnaire 9-item depression scale (PHQ-9) for adults.[6] Referrals to mental health providers should be made when appropriate.

Age
19 - 23

MDA Care Center workup:

» DME scripts were written, patient was referred back to cardiology, orthopedics, and adult pulmonology for pulmonary function tests (PFTs) and a sleep study. A modified barium swallow study and scoliosis X-rays were also ordered

» Nurse coordinator scheduled follow-up appointments for the same day to minimize transportation issues and called the day before to confirm appointments

Patient follow-up three months later:

» None of the recommendations had been pursued

» The patient lived with his brother in a two-story residence but was unable to access bedroom and bathroom on second floor. Thus, he slept in his wheelchair and exhibited a stage 1 pressure ulcer

» Patient was unable to bathe

» Patient ate only one to two meals per day. Pre-albumin level was 14.2 mg/dL (normal 18-45), reflecting low nutritional intake. Vitamin D level was 11 ng/mL (normal 25-80)

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Age 19 - 23

MDA Care Center workup:

- Vitamin D 50,000IU weekly advised, and nutritionist at MDA Care Center provided supplemental nutrition shakes
- Adult Protective Services (APS) case was filed by social worker because of concerns for neglect, and previous recommendations were made again

Patient follow-up:

- The patient did not follow up for eight months
- The patient returned to the office for a wheelchair evaluation
- The patient had not shown up to a sleep study appointment and had not followed up on subspecialty referrals
- His wheelchair could no longer accommodate him as he had lost 66 lbs since the prior visit
- Pre-albumin was 11.7 mg/dL and Vitamin D level had increased to 26 ng/mL. Patient could no longer feed himself independently

MDA Care Center workup:

- A home health referral was made but was unable to be enacted as family did not return calls to schedule intake
- APS case again filed, which resulted in community referrals. Disability Network was recommended to provide 24/7 in-home services. Unfortunately, the legal guardian did not call to set up evaluations

Commentary: Caring for a person with DMD can be associated with a substantial burden and affected quality of life for caregivers. Assistance and helpful resources for caregivers can be found in MDA's Guide for Caregivers.[7]

Age 20

Patient follow-up:

- Returned to clinic eight months later
- A lawyer had been appointed as legal guardian, although the patient continued to live with his brother, who was reported to frequently leave him alone and unattended in the house
- There was no in-home care
- Patient reported eating one to two meals per day. Pre-albumin level 10.6 mg/dL and Vitamin D 16 ng/mL
- Overall functioning had dramatically decreased; patient exhibited end-stage DMD

MDA Care Center workup:

- Patient's disease stage and code status were discussed with the patient and family present and patient opted to be "full code"
- The guardian was contacted and transition to skilled living facility was advised
- Patient follow-up:
- With help of his guardian, patient did have an initial cardiology appointment one month later and was determined to have dilated cardiomyopathy with with ejection fraction (EF) 35% and left ventricular hypertrophy (LVH)

Cardiology workup:

- Patient was started on lisinopril

Pulmonary workup:

- Had a pulmonology assessment one month later with oxygen saturation of 70%-90% when on room air
- Was noted to be hypoxic and lethargic, with oxygen saturation in the 70s-80s on room air
- Patient was immediately sent to the emergency department and ultimately admitted to the ICU for acute chronic respiratory failure. He was admitted for 23 days

ICU workup:

- During the hospital admission, palliative care was consulted to assist with ascertainment of goals and plan of care
- Patient declined tracheostomy because of impact on quality of life and opted for mouth-piece ventilation with BiPAP
- A beta-blocker was started for management of tachycardia and heart failure. He was discharged with home healthcare services
- Patient was readmitted five days later with respiratory distress believed to be related to hospital-acquired pneumonia, which responded well to a five-day course of Zosyn

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Age 20

Patient follow-up:

- Patient did not follow up with pulmonology or neurology after hospital discharge
- Patient did see cardiologist, who adjusted his medications, including the lisinopril and transition to carvedilol

MDA Care Center workup:

- The MDA Care Center was contacted by a concerned MDA camp counselor three months later because of reports of weak cough and secretion clearance leading to episodic respiratory distress
- While pulmonary DME scripts for cough assist, tubing, and suction were sent, patient did not follow up with pulmonology
- APS was contacted and stated the prior case had been closed
- Patient subsequently cancelled and rescheduled multiple MDA Care Center appointments and ultimately was not seen in clinic until 22 months after his last multidisciplinary visit, which had occurred prior to his hospitalization

Age 23

Patient follow-up:

- At this visit, patient weighed 90 lbs, having lost another 66lbs since his last appointment
- Patient was living in an apartment with a roommate with 24/7 home healthcare services
- Pre-albumin was again low at 14 mg/dL. He could eat only one meal daily
- Patient was agreeable to gastrointestinal evaluation for G-tube
- Patient was facing eviction because of financial challenges

MDA Care Center workup:

- Transition to a skilled living facility was again recommended
- End-stage status was again discussed, along with the progressive decline seen over the past one to two years
- End-of-life wishes were discussed, and patient firmly verbalized a desire to have maximum non-invasive respiratory ventilation before trach placement, and wanted full cardiac interventions (compressions, medications) with exception of defibrillator
- Patient was advised to follow up with pulmonology and cardiology as well

Commentary: It has been noted that palliative care services are underutilized in the DMD population, despite care recommendations that encourage early engagement of palliative care professionals. In one study, the majority of palliative care services were being utilized by less than 48% of patients surveyed.[8] It is important to note that palliative care is not equivalent to end-of-life care but can include case management, attendant care, respite care, homemaker services, mental health services, pastoral care, social work, transportation, dietary services, home meals, skilled nursing, and pain management, in addition to hospice care. The John Hopkins "Vision of Hope" curriculum reviews how to incorporate palliative care into the care for chronic diseases such as DMD.[6] Benefits demonstrated by the use of palliative care services include improved care coordination and family empowerment, with more informed decision-making based on pre-considered goals of care and quality-of-life considerations.[9] Resources such as "Voicing my Choices" can be helpful in facilitating advanced care planning.[6]

References

1. Yamaguchi M, Suzuki M. Becoming a back-up carer: Parenting sons with Duchenne muscular dystrophy transitioning into adulthood. Neuromuscular Disorders. 2015;25(1):85-93. doi:10.1016/j.nmd.2014.09.001
2. Trout CJ, Case LE, Clemens PR, et al. A Transition Toolkit for Duchenne Muscular Dystrophy. Pediatrics. 2018;142(Supplement 2):S110-S117. doi:10.1542/peds.2018-0333M
3. Landfeldt E, Lindgren P, Bell CF, et al. Compliance to Care Guidelines for Duchenne Muscular Dystrophy. Journal of Neuromuscular Diseases. 2015;2(1):63-72. doi:10.3233/JND-140053
4. Denger B, Kinnett K, Martin A, Grant S, Armstrong C, Khodyakov D. Patient and caregiver perspectives on guideline adherence: the case of endocrine and bone health recommendations for Duchenne muscular dystrophy. Orphanet Journal of Rare Diseases. 2019;14(1):205. doi:10.1186/s13023-019-1173-7
5. Pascoe JE, Sawnani H, Hater B, Sketch M, Modi AC. Understanding adherence to noninvasive ventilation in youth with Duchenne muscular dystrophy. Pediatric Pulmonology. 2019;54(12):2035-2043. doi:10.1002/ppul.24484
6. Birnkrant DJ, Bushby K, Bann CM, et al. Diagnosis and management of Duchenne muscular dystrophy, part 3: primary care, emergency management, psychosocial care, and transitions of care across the lifespan. The Lancet Neurology. 2018;17(5):445-455. doi:10.1016/S1474-4422(18)30026-7
7. (No Title). Accessed August 25, 2020. https://www.mda.org/sites/default/files/Guide-For-Caregivers-2018.pdf
8. Andrews JG, Pandya S, Trout C, et al. Palliative care services in families of males with muscular dystrophy: Data from MD STARnet. SAGE Open Medicine. 2019;7:205031211984051. doi:10.1177/2050312119840518
9. Carter GT, Joyce NC, Abresch AL, Smith AE, VandeKeift GK. Using Palliative Care in Progressive Neuromuscular Disease to Maximize Quality of Life. Physical Medicine and Rehabilitation Clinics of North America. 2012;23(4):903-909. doi:10.1016/j.pmr.2012.08.002

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