

# Transitions in Care in DMD

Case contributor and commentary:

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## Patient #1: Successful "Transition-in-Place"

20 y/o Male

Case provided by M. Eileen McCormick with commentary and discussion provided by Meghan Harper-Shankie

### Pediatric Care

#### Age 6 months

##### Background:

- » Patient's older brother (now deceased) was diagnosed with Duchenne muscular dystrophy (DMD)
- » Patient's mother is a positive carrier for a DMD pathogenic variant

##### Pediatrician initial workup:

- » Patient had a positive creatine kinase (CK) test
- » Genetic testing confirmed diagnosis (deletion of exons 49 and 50 in DMD gene, consistent with the patient's brother and mother)

From diagnosis, patient received managed care at MDA Care Centers. The family moved twice, and the Care Centers facilitated the transfers.

**Commentary:** Care Centers have been utilizing electronic medical records (EMR) for at least 10 years, which has eased the burden of transferring patients between centers and enabled continuity of care.

#### Age

8

##### Treatment:

- » Steroid treatment initiated (0.75mg/kg/d prednisone)

##### Side effect:

- » Initial trial of prednisone resulted in profound weight gain

##### Management:

- » After unsuccessful dose adjustment of prednisone, patient was switched to deflazacort (daily 0.9 mg/kg) which stabilized weight and normalized growth curve
- » The patient has remained on continuous daily steroid therapy since age 8. The patient and his family have been very compliant with treatment recommendations and motivated to participate in clinical trials.

##### Early education:

- » During elementary school, the patient was assessed as learning disabled (IQ approximately 80) and was enrolled in special education

**Commentary:** Referral for neuropsychological evaluation is recommended when concerns about developmental progress arise.<sup>1</sup> A neuropsychologist can assess a child's cognitive development, academic skills, social functioning, emotional adjustment, and behavioral regulation and work with families to develop intervention plans that can be implemented at home and at school. In children with cognitive, behavioral, or learning needs, additional services should be considered, such as speech and language therapy, applied behavior analysis, or specialized academic instruction. Re-evaluations should be done every 2-3 years to monitor a child's developmental progress and response to interventions, or when acute changes in functioning or major transitions involving home or school occur.

#### Age

9 - 13

##### Loss of functional milestones:

- » Loss of ambulation occurred at age 9 resulting in full-time wheelchair use
- » Loss of upper extremity function was established prior to transition to his current Care Center

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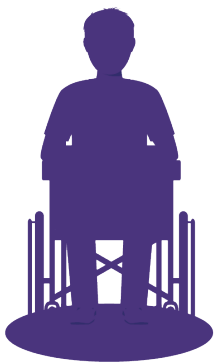
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**Commentary:** It is recommended that patients and their families are made aware of plans for health-care transition by the time the patient is 12 years old, with initiation of transition discussions and planning by age 13-14 years.<sup>1,2</sup> This plan should include services that need to be provided, providers of these services, and details of how they will be financed. The plan should be based on the needs, wants, and values of the individual with DMD and their family. The plan should be reviewed and updated annually.

### Transitions in Care



#### HOUSING

- Examine where to live (family home vs elsewhere)
- Modify home for accessibility and safety
- Use assistive technology

#### TRANSPORTATION

- Foster independent driving with vehicle modifications
- Modify family-owned vehicle
- Investigate accessible public transportation options

#### HEALTH CARE

- Transition from pediatric to adult health care
- Move from family-centered to patient-centered provider interactions
- Discuss age-related changes in health-care benefits
- Assess the need for durable power of attorney for health care

#### ACTIVITIES OF DAILY LIVING

- Explore the funding and benefits for care
- Learn to hire and train personal care attendants
- Ensure respite for family caregivers
- Consider need for guardianship or conservatorship

#### EDUCATION OR EMPLOYMENT

- Plan early for future vocation
- Consider classes online vs on campus
- Contact campus programs for students with disabilities
- Enlist employment or vocational planning resources

#### RELATIONSHIPS WITH OTHERS

- Develop skills to connect with others to manage own affairs (eg, social outing, appointments)
- Work towards desired level of autonomy and independence

Components to be addressed during transition planning. Figure adapted from reference #1.

### Age

14 - 17

#### Transition planning:

- » Transition planning was initiated at age 14
- » As per the wishes of the patient and family, the plan was to "transition in place"; the pediatric care team would continue to follow the patient during and after transition, and adult providers would be incorporated into care as appropriate

**Commentary:** Got Transition<sup>3</sup> has issued guidelines for three transition scenarios: transitioning in place, transitioning to adult care providers, and integrating into the adult healthcare system. Some patients and families do not wish to formally/physically transition to adult care providers and choose to transition in place.

#### Change in clinical setting:

- » The patient was an appropriate candidate for a clinical trial using eteplirsen, which was approved in Sept 2016. However, his previous Care Center institution did not endorse this therapy. Thus, the patient and family requested a transfer to a Care Center that utilized eteplirsen therapy; the Care Center facilitated his transfer without delay

**Commentary:** Practice patterns are generally consistent across MDA Care Centers. However, institutional and jurisdictional policies can limit treatments available in a particular center or area. MDA Care Centers will work with patients, families, and other stakeholders to locate and assist in the transfer of patients to centers that best meet the needs of the patient. To learn more, the MDA Resource Center can be reached by phone at 1-833-ASK-MDA1 (1-833-275-6321) or by e-mail at ResourceCenter@mdausa.org.

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### Age

17 - 20

#### Multidisciplinary care:

- » Patient received care from an uninterrupted series of MDA centers with good continuity of care. He transferred to his current clinic at age 17
- » Patient is followed by a Primary care physician (PCP) external to MDA clinic; PCP (e.g. vaccinations, wound care, ER visits) is well-integrated with MDA care

#### Treatment:

- » Patient was started on eteplirsen therapy, administered as a weekly home infusion (30 mg/kg) via mediport. No complications occurred during placement of mediport, which has since been functional with no signs of infection

#### Social factors:

- » Patient is married. He met his wife through his special education program in high school (his wife also has cognitive disability) and they married shortly after graduation with the full support of both families. The wife works part time as a nurse's aide in a nursing home, and the couple has arranged to live there full time
- » The couple is able to accomplish many of their activities of daily living but require assistance, for example, with shopping and transportation

#### Care coordination:

- » Patient and his wife receive support from their families (e.g., financial assistance, transportation to medical appointments), a Care Center social worker (to identify and obtain private and governmental support services), and county support services, as well as from their residential facility
- » A social worker provided by the county was able to match the patient's goals and functional ability (Brookes level 5: able to hold a pen and type on a keyboard) with an organization that provides part time, web-based employment in data entry

**Commentary:** It is recommended that a care coordinator or social worker oversee transition planning.<sup>1</sup> The care coordinator can serve as a central resource for questions; facilitate communication between providers and the family; navigate insurance benefit eligibility; assist with acquisition of prescribed equipment; identify community care agencies; advocate for students in their schools; ensure transmission of medical records; identify and protect financial resources; and enlist the help of social care systems, such as human service organizations, specialty clinics, and advocacy groups.

#### Social Factors:

- » Shortly after the patient got married, disagreement arose between his mother and the patient's wife over how to best care for him, causing the mother to temporarily disengage from the patient's life for approximately one year. This was a period of increased medical instability and social isolation
- » With the encouragement of the Care Center social worker, the patient and his wife came to appreciate the benefits of his family's involvement. They have since reconciled and the mother has reengaged

**Commentary:** Young adults strive for autonomy and independence from their family. This can have adverse consequences for people with complex medical needs, especially when there is overlapping cognitive impairment. It is essential that social support personnel identify these situations early and work to find solutions that lead to the best outcome for the patient. In most cases, a compromise can be found that satisfies all stakeholders.

#### Insurance and reimbursement:

- » Care Center provided updated durable medical equipment (DME), including a wheelchair, prior to patient turning 18 years old

#### Psychosocial and emotional support:

- » Patient qualified for adult disability support programs because of his intellectual disability

- » Patient's anxiety is being managed by counseling and a low-dose anxiolytic

#### Long-term planning:

- » Patient wishes full resuscitation and support and does not wish for palliative care

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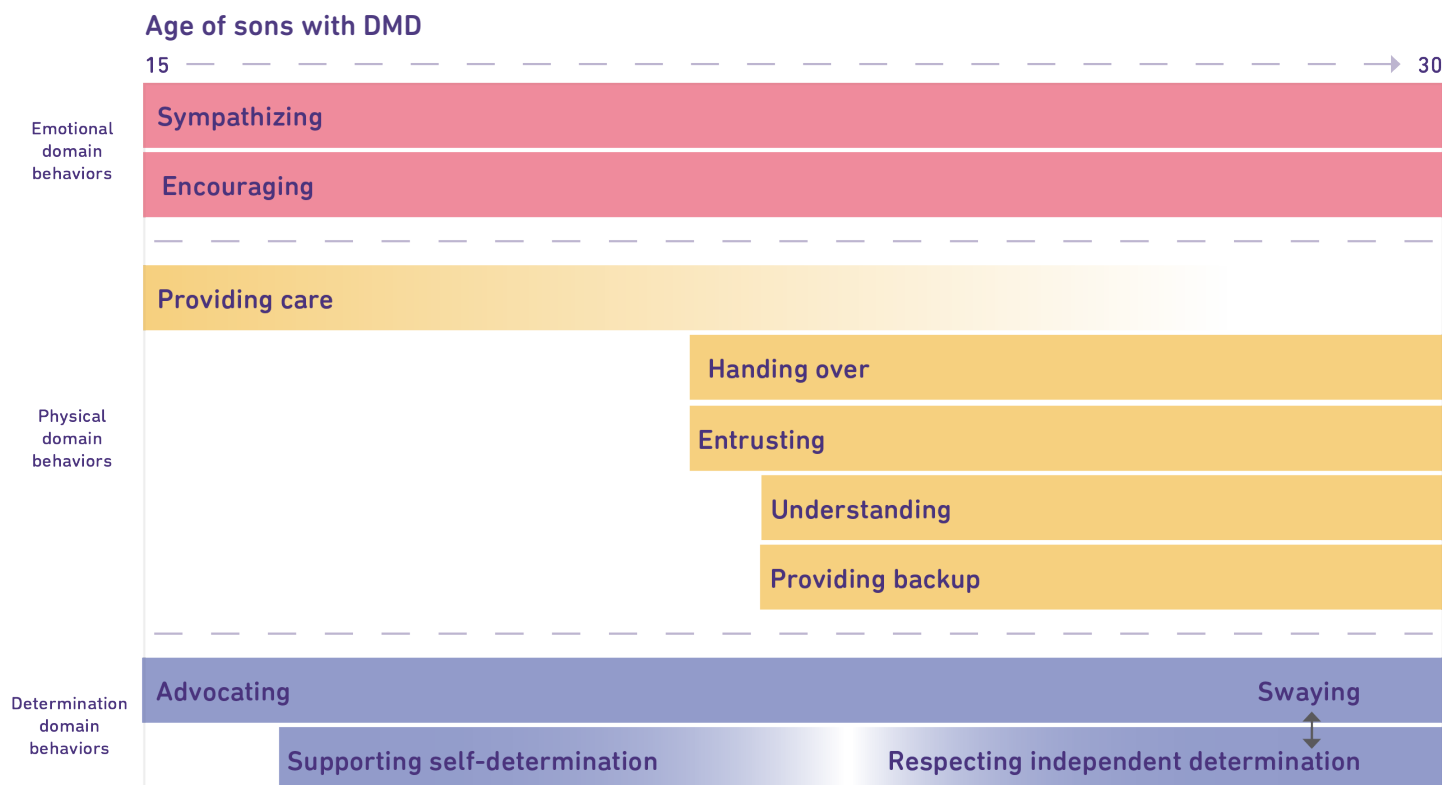


Figure adapted from reference #5.

**Commentary:** Palliative care consultation can be useful at various times, including at diagnosis, at the time of major treatment decisions, during life-threatening events, and at the terminal phase of care. The Vision of Hope curriculum<sup>6</sup> integrates the principles of pediatric palliative care into the care of individuals with DMD. Advance care planning is particularly important in DMD<sup>7</sup> and allows individuals with DMD to express how they prefer to be comforted, supported, treated, and remembered. In many places, minors can participate in formulating advance directives. It is essential that social support personnel identify these situations early and work to find solutions that lead to the best outcome for the patient. In most cases, a compromise can be found that satisfies all stakeholders.

#### References

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