DIAGNOSIS AND MANAGEMENT OF DUCHENNE MUSCULAR DYSTROPHY

Part 3: Primary care, emergency management, psychosocial care, and transitions of care across the lifespan

OVERVIEW

Duchenne muscular dystrophy (DMD) is a lethal X-linked recessive neuromuscular disorder caused by mutations in the dystrophin gene, leading to progressive muscular damage and degeneration.

The 'DMD care considerations' were first published in 2010. They have been updated to reflect several important developments, including the improved survival of patients with DMD, evolving diagnostic and therapeutic approaches, an increasing emphasis on quality of life and psychosocial management and experience with existing therapies, and emerging therapies.

Part 3 focuses on primary care, emergency management, psychosocial care, and transitions of care across the lifespan. With the exception of psychosocial care, these are new additions to the care considerations.

PRIMARY CARE

NEW TOPIC

The primary care provider is often the first medical professional to hear a family's concern about their child's muscle weakness, initiating the process that leads to a diagnosis of DMD.

The goals are for the primary care provider to provide first-line care for acute and chronic medical issues, coordinate care with appropriate specialists, provide trusted advice and continuity of care across the lifespan, and optimise the wellbeing and quality of life of patients and their family members.

Considerations for primary care include:

- Immunisation
- Nutritional counselling
- Dental care
- Safety counselling regarding fall and injury prevention
- Monitoring for adrenal insufficiency
- Psychosocial care for patients and their families
- Screening that includes hearing, vision and cardiovascular risk

EMERGENCY MANAGEMENT

NEW TOPIC

The following provides an overview of key issues related to the emergency care of patients with DMD, and is intended by the guidelines as a guide for emergency medicine providers, primary care clinicians and neuromuscular specialists.

Key issues related to emergency care of patients with Duchenne muscular dystrophy

Advance directives, history and contacts

- Determine whether there are restrictions on resuscitation
- Ask for the patient's emergency card and baseline test results, including electrocardiogram results
- Obtain a brief history with a focus on baseline respiratory and cardiac status, including use of relevant devices and medications
- Determine whether the patient is treated with chronic steroid therapy
- Contact the patient's neuromuscular specialist

Breathing problems

- Ask about respiratory symptoms and home equipment
- Monitor blood oxygen saturation (SpO_2) levels via pulse oximetry; even mild hypoxaemia (SpO_2 <95% in room air) is a concern; do a blood gas analysis if necessary
- Treat with non-invasive ventilation and frequent application of a cough assistance device (or manual assisted coughing if device is unavailable); use the patient's home equipment when available
- Obtain a portable chest radiograph
- Obtain early consultation with a respiratory therapist and respiratory physician

Cardiac problems

- Ask about cardiac symptoms
- Monitor heart rate and rhythm
- Obtain an electrocardiogram (this is typically abnormal and Q waves might be expected) and portable chest radiograph
- Measure blood levels of B-type natriuretic peptide or troponin I, or both, as indicated
- Consider worsening cardiomyopathy, congestive heart failure, and arrhythmias
- Obtain an echocardiogram when necessary
- Obtain early consultation with a cardiologist

Orthopaedic problems

- Assess for long-bone or vertebral fractures as indicated
- Review critical precautions related to sedation and anaesthesia if applicable
- Consider fat embolism syndrome if individual has dyspnoea or altered mental status
- Obtain consultation with an orthopaedic specialist early in the process

Endocrine problems

- Determine whether stress steroid dosing is necessary
- For critical adrenal insufficiency, administer intravenous or intramuscular hydrocortisone: 50 mg for children <2 years old; 100 mg for children ≥2 years and adults
- In less critical situations, consult the PJ Nicholoff Steroid Protocol*
- Obtain early consultation with an endocrinologist

Disposition after discharge from emergency care

- Be aware that most patients will need hospital admission (e.g. to initiate or intensify respiratory or cardiac therapy or to manage fractures)
- Early in the process, initiate emergency transport by skilled personnel to a centre specialising in the care of patients with Duchenne muscular dystrophy, in cooperation with the individual's neuromuscular specialist

*Kinnett K, Noritz G. The PJ Nicholoff Steroid Protocol for Duchenne and Becker muscular dystrophy and adrenal suppression. *PLoS Curr.* 2017; 9: DOI: 10.1371/currents.md.d18deef7dac96ed135e0dc8739917b6e.

PSYCHOSOCIAL CARE

Comprehensive psychosocial care should address social and cognitive development and functioning across environments, including home, school and work.

The care team should include a mental health clinician (that is, psychologist, psychiatrist, social worker or psychiatric nurse) who has specialised training and experience in assisting families and individuals with chronic medical or neurodevelopmental conditions. At each neuromuscular clinic visit, mental health and quality of life should be screened.

Components of psychosocial care for patients with DMD include:

- Care coordination between the patient and members of the multidisciplinary team
- Routine mental health screening
- Psychological care
- Educational support

High rates of intellectual disability (17–27%), learning disabilities (26%), autism spectrum disorder (15%), attention-deficit hyperactivity disorder (32%) and anxiety (27%) have been reported in people with DMD. The care considerations have been updated to emphasise appropriate screening and assessment of these conditions

TRANSITIONS OF CARE ACROSS THE LIFESPAN

NEW TOPIC

Transition planning should begin in early childhood. Individuals with DMD should be encouraged to participate fully in future planning and assisted in the transition from paediatric healthcare which is family-centred, to adult healthcare which is patient-centred and focused on independence. A care coordinator or social worker should be responsible for transition planning.

Components of young adulthood to be addressed during transition planning include:

- Healthcare
- Activities of daily living
- Education or employment
- Relationships with others
- Housing
- Transportation

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Reference: Birnkrant DJ, *et al. Lancet Neurol.* 2018;17:445–455.

TAKE ON DUCHENNE
Make every day count