Purpose: This course will review specific considerations for practitioners who care for patients with neuromuscular diseases, particularly Duchenne muscular dystrophy (DMD) and Spinal Muscular Atrophy (SMA). While many of the challenges faced by these patients are similar to those with other childhood disabilities, there are specifics of care for DMD and SMA with which the practitioner must be familiar.

Target Audience: Physicians and therapists who care for patients with DMD or SMA

Course Summary: This course will review the genetics and pathophysiology of DMD and SMA, and then turn to specific scenarios and recommendations for clinical care. Chief among these are cardiopulmonary care and support; other topics that will be covered include management of corticosteroid complications, nutrition and bowel issues; bone health; and prevention and treatment of nephrolithiasis. We will discuss orthopedic and rehabilitative aspects of care, as well as issues of transition to adult providers. We will close with an overview of current trials to treat DMD and SMA.

Learning Objective 1: Understand the genetics and pathophysiology of DMD and SMA

- DMD: Know the mutation [www.duchenneconnect.org](http://www.duchenneconnect.org)
  - In frame
  - Out of frame
  - Nonsense Mutation

- SMA: Copy Number
  - Type 1-4

Learning Objective 2: Develop expertise in the holistic medical care of children and adults with these disorders

Considerations for Primary Care of Individuals with DMD

<table>
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<tr>
<th>Immunizations</th>
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<tr>
<td>- Administer all non-live vaccinations recommended by CDC (<a href="https://www.cdc.gov/vaccines/schedules/">https://www.cdc.gov/vaccines/schedules/</a>)</td>
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<tr>
<td>- Aim to give live-virus vaccines before steroid initiation</td>
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<td>- Live-virus vaccines are contraindicated in individuals with DMD on high-dose daily corticosteroids (&gt;20 mg/day or &gt;2 mg/kg/day)</td>
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<tr>
<td>- Administer injectable influenza vaccine annually to individuals with DMD and all close contacts (do not give the live-virus nasal vaccine; it is contraindicated)</td>
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<tr>
<td>- Follow CDC pneumococcal vaccination schedule, integrating PCV-13 with PPSV-23 (<a href="http://www.immunize.org/askexperts/experts_pneumococcal_vaccines.asp#ppsv23_rec">http://www.immunize.org/askexperts/experts_pneumococcal_vaccines.asp#ppsv23_rec</a>)</td>
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Nutrition (see Nutritional, Swallowing, and Gastrointestinal Management section, Part 1)
- Ensure individuals with DMD receive nutritional counseling to prevent obesity and malnutrition
- Encourage adequate nutrient intake (especially calcium and vitamin D)
- Refer to registered dieticians for nutritional counseling

**Dental Care**

- Ensure that individuals with DMD have regular dental care
- PCP or dentist should apply fluoride varnish per protocol\(^43\)
- Fluoride should be supplemented for individuals with DMD with unfluoridated water

**Safety Counseling**

- As weakness progresses, individuals with DMD are prone to falls, especially when they are beginning to lose ambulation
- Consult a physical or occupational therapist for appropriate safety practices, such as use of a wheelchair in school, and for advice on safety devices, to minimize risk of falls
- Seatbelts should be worn in motor vehicles at all times; individuals with DMD with poor trunk control may require special positioning devices
- Individuals with DMD who sit in their wheelchairs in motor vehicles should be restrained according to manufacturer guidelines

**Monitoring for Adrenal Insufficiency (see Neurology and Endocrinology—Growth/Puberty/Adrenal Insufficiency sections, Part 1)**

- For individuals with DMD taking corticosteroids:
  - Educate the family not to miss any doses of the prescribed corticosteroid and to be vigilant for signs of adrenal insufficiency (such as lethargy) in association with febrile illnesses, vomiting, surgeries, and other physiological stresses
  - Supply the patient/family with a stress dose of steroids at home for symptoms of adrenal insufficiency and ensure that they seek immediate medical attention at those times

**Psychosocial Care for Patient and Family (see Psychosocial Care section above)**
Monitor physical and developmental milestones and be aware of DMD-specific neurodevelopmental and neuropsychological issues, such as the increased prevalence of intellectual disability, attention deficit/hyperactivity disorder and autism spectrum disorder.

Refer to a psychologist for psychological and neuropsychological assessments and interventions when appropriate (see Psychosocial Care section).

Refer to a speech-language pathologist for suspected delays.

Help the family with special educational needs (e.g., in the U.S, Individualized Education Programs [IEPs] and 504 plans).

Identify community resources that may enhance patient and family functioning and coping, such as local social service agencies and patient advocacy organizations.

Help to initiate discussions about transition of care (see Transitions section).

Ensure that adults with DMD have completed advanced directives, when appropriate, and that they have appointed a health care power of attorney.

Other Screening

Conduct standardized screenings on the usual schedule, such as hearing and vision screening and screening for mood disorders and substance abuse.

Conduct screening for cardiovascular risk factors, such as hypertension and hypercholesterolemia.

ABCs of Duchenne MD Emergency Care

Advanced Directives, History and Contacts

Determine if there are limitations on resuscitation.

Ask for patient’s emergency card, if available.

Obtain brief history with focus on baseline respiratory and cardiac status, including relevant devices and medications.

Determine if patient is treated with chronic steroid therapy.

Contact the patient’s neuromuscular specialist.

Breathing

Ask about respiratory symptoms and home equipment.

Monitor SpO2 levels via pulse oximetry and obtain a blood gas if necessary.

Using home equipment when available, treat with non-invasive ventilation and frequent application of a cough assistance device (or manual cough assist if device is unavailable).

Obtain a portable chest radiograph.

Early consultation with respiratory therapy and pulmonology.

Cardiac
• Ask about cardiac symptoms
• Monitor heart rate and rhythm
• Obtain an ECG and portable chest radiograph
• Measure blood levels of B-type natriuretic peptide and/or Troponin I, as indicated
• Consider worsening cardiomyopathy, congestive heart failure and arrhythmias
• Obtain an echocardiogram when necessary
• Early consultation with cardiology

### Endocrine

• Determine if stress steroid dosing is necessary
• For critical adrenal insufficiency, administer hydrocortisone 50 mgs IV/IM for children < 2 years old and 100 mgs IV/IM for children ≥ 2 years old
• In less critical situations, consult the PJ Nicholoff Steroid Protocol

### Orthopedic

• Evaluate for long bone or vertebral fractures as indicated
• Involve orthopedics early in the process
• Review critical precautions related to sedation and anesthesia if applicable (see text)
• Consider fat embolism syndrome if patient has dyspnea or altered mental status

### Disposition

• 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 skilled emergency transport arrangements to a center specializing in DMD care, in cooperation with the patient’s neuromuscular specialist
## Components for Clinic-Based Psychosocial Care

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<th>Component</th>
<th>Description</th>
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| **Care coordination**             | • The care coordinator serves as a point of contact for the individual with DMD and his family. The care coordinator should be a health professional with sufficient training and/or experience regarding clinical care for DMD.  
                                          • Roles: 1) to provide information, 2) to coordinate (and possibly schedule) appointments, and 3) to facilitate communication with clinicians across disciplines. |
| **Routine mental health screening**| • Brief screening of emotional status is strongly advised at every clinic visit or at least once a year.                                                                                                       
                                          • Screening can be informal in nature and does not require comprehensive assessment.                                                                                                                       
                                          • Use of short standardized rating scales is appropriate. We strongly advise use of the Personal Adjustment and Role Skills Scale (PARS-III)\textsuperscript{19} for children and adolescents with DMD (ages 5 through 17).  
                                          • Screening can be conducted by a social worker or mental health professional or by other clinic staff with sufficient training/experience in this area (e.g., nurse, attending physician).  
                                          • If the screening is positive, then a referral should be made to a psychologist or psychiatrist for further evaluation and/or treatment.             
                                          • Every clinic should have a plan to assess/address suicidal ideation or other acute safety concerns.                                                                                                      
                                          • Caregiver emotional adjustment should be monitored and intervention/supports offered as needed.                                                                                                          
                                          • Siblings should be provided with opportunities to connect with other siblings and with access to mental health services as needed.                                                                 |
| **Psychological care**            | • For individuals with DMD who are typically developing, there should be an expectation that they will live rich, fulfilling lives and achieve a high level of independence in managing their disease but that they and their families may need occasional psychosocial supports.  
                                          • Neuromuscular care team should include a mental health professional who has training and experience in assessing and treating psychiatric conditions within the context of chronic medical/neurodevelopmental conditions (i.e., psychologist or psychiatrist).  
                                          • When mental health concerns are identified, the mental health professional should provide further evaluation of individuals with DMD and family members and provide cognitive and/or behavioral interventions to treat psychiatric conditions.  
                                          • Standard, evidence-based practices should be used for those who need more formal mental health treatment.                                                                                       |
Neuropsychological evaluations should be performed when there are identified cognitive delays, difficulties with emotional and behavioral regulation, or concerns about social skills. Re-evaluations every 2 to 3 years to monitor developmental progress and response to interventions may be needed. Neuropsychological evaluations should also be considered within the first year of diagnosis to establish a baseline or when transitioning to adulthood if state-based assistance may be necessary to promote functional independence.

Pharmacological interventions

- The neuromuscular team should include a psychiatrist or other physician with training and experience in providing medication to treat behavioral or emotional disorders in the context of chronic medical/neurodevelopmental conditions.
- Standard prescribing practices should be used.
- Selective serotonin re-uptake inhibitors should be prescribed for depression, anxiety, and obsessive/compulsive disorder.
- Alpha-agonists (first choice) or atypical antipsychotics (second choice) should be prescribed for aggression and anger/emotional dysregulation.
- Stimulants or alpha-agonists should be prescribed for ADHD.

Standards of care for SMA


Learning Objective 3: Develop expertise in the habilitative and rehabilitative care children and adults with these disorders

What we think works / know works / think doesn’t work / don’t have a clue

Stretching/Splinting for ROM maintenance or improvement

Orthotics: TLSO, AFO, KAFO

Diet and Supplements
Amino Acid Diet

Power Mobility

Learning Objective 4: Understand ongoing trials of therapies for DMD and SMA.
www.Clinicaltrials.gov

**DMD:** Mutation Specific
- Exon Skipping
- Nonsense Mutation

**Non Specific**
- Steroid type, dosing and age of initiation
- Anti-fibrotic / Anti-inflammation
- Myostatin Inhibition

**SMA:** SMN replacement
- Gene therapy
- antisense oligonucleotide (ASO)

**Neuromuscular Junction**
- Pyridostigmine