Neuromuscular Care, Growth and Puberty

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Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management

David J Birnkrant, Katharine Bushby, Carla M Bann, Susan D Apkon, Angela Blackwell, David Brumbaugh, Laura E Case, Paula R Clemens, Stasia Hadjiyannakis, Shree Pandya, Natalie Street, Jean Tomezsko, Kathryn R Wagner, Leanne M Ward, David R Weber, for the DMD Care Considerations Working Group*

• Original Care Considerations published in 2010
• Updated in 2018, 3 parts
• Endocrine added
• Available on website: www.endduchenne.org
<table>
<thead>
<tr>
<th>Stage 1: At diagnosis</th>
<th>Stage 2: Early ambulatory</th>
<th>Stage 3: Late ambulatory</th>
<th>Stage 4: Early non-ambulatory</th>
<th>Stage 5: Late non-ambulatory</th>
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<td>Lead the multidisciplinary clinic; advise on new therapies; provide patient and family support, education, and genetic counselling</td>
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<td>Ensure immunisation schedule is complete</td>
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<td>Discuss use of glucocorticosteroids</td>
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<td>Refer female carriers to cardiologist</td>
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<td>Assess function, strength, and range of movement at least every 6 months to define stage of disease</td>
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<td>Initiate and manage use of glucocorticosteroids</td>
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<td>Help navigate end-of-life care</td>
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<td>Provide comprehensive multidisciplinary assessments, including standardised assessments, at least every 6 months</td>
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<td>Provide direct treatment by physical and occupational therapists, and speech-language pathologists, based on assessments and individualised to the patient</td>
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<td>Assist in prevention of contracture or deformity, overexertion, and falls; promote energy conservation and appropriate exercise or activity; provide orthoses, equipment, and learning support</td>
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<td>Continue all previous measures; provide mobility devices, seating, supported standing devices, and assistive technology; assist in pain and fracture prevention or management; advocate for funding, access, participation, and self-actualisation into adulthood</td>
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<td>Measure standing height every 6 months</td>
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<td>Assess non-standing growth every 6 months</td>
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<td>Assess pubertal status every 6 months starting by age 9 years</td>
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<td>Provide family education and stress dose steroid prescription if on glucocorticosteroids</td>
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Neuromuscular Management

• NM specialist usually serves as lead clinician
• Ensure consistent assessments (PT, OT, Pulmonology, Cardiology)
• Manage interventions
  – PT, OT, ST
  – Equipment/orthotic needs
  – Corticosteroids
  – Emerging treatments
Endocrine Management

• Monitor growth and development
• Identify and diagnose hormone deficiencies
• Provide hormone replacement therapy when indicated
  – Limited safety and efficacy data for GH and testosterone
• Prevent adrenal crisis
• Bone health (discussed by others)
Growth

- Impaired linear growth 2/2 steroids (and DMD)
- GH – not currently recommended in Care Considerations
  - Pseudotumor cerebri, glucose intolerance, worsening of scoliosis
  - Unknown how much growth gained by GH
  - (-) effect on muscle function/strength?
  - Many feel that shorter boys stay stronger longer

**Growth**

- Assessment of height every 6 months until completion of puberty and attainment of final height
- Impaired growth
  - Any of the following:
    - Downward crossing of height percentile
    - Height velocity of <4 cm per year
    - Height <3rd percentile
- Refer to endocrinologist
  - Recommended
    - Assessment of bone age with left-hand x-ray
    - Thyroid function tests
    - Coeliac panel
    - Growth factors
    - Comprehensive metabolic panel
    - Complete blood count
  - To be considered
    - Growth hormone stimulation testing
Puberty

- Delayed puberty 2/2 steroid therapy
- Testosterone: replacement tx recommended for confirmed hypogonadism
- Potential benefits generally outweigh side effects (behavior, acne, body odor, rapid growth spurt, epiphyseal closure)
- IM and topical preparations; levels need to be monitored
Adrenal Insufficiency/Adrenal Crisis

- Rare but potentially life-threatening
- Can develop if steroids are abruptly withdrawn or during severe illness/trauma/surgery
- Educate (parents and MDs) on signs, symptoms and management of AI: can be very non-specific
- Some prescribe IM hydrocortisone (100 mg) for emergency administration at home
- See “PJ Nicholoff Steroid Protocol”
- Assume nothing
- Get the PPMD app