Neuromuscular Care, Growth and Puberty

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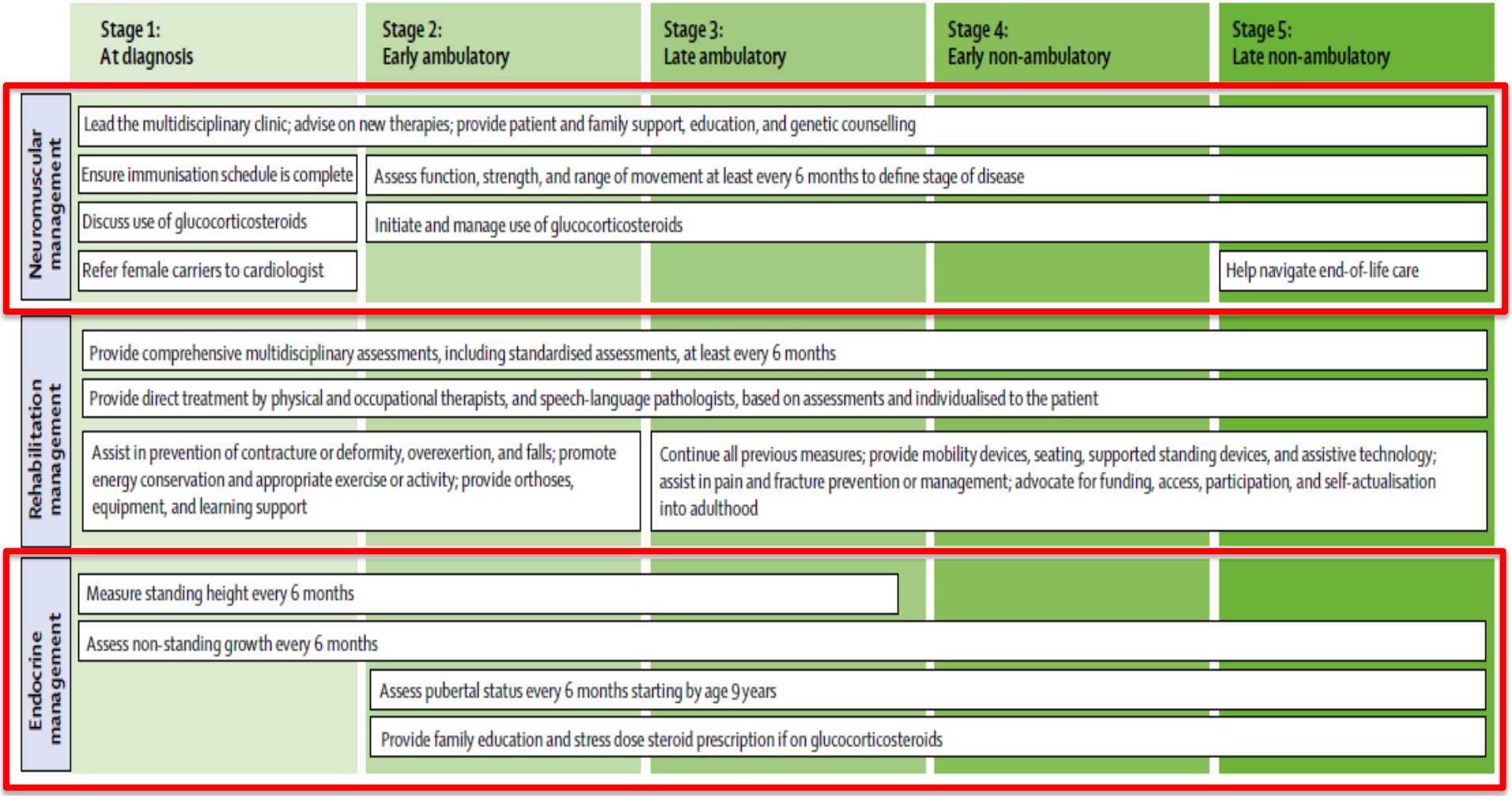
Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management

*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


Stage 1: At diagnosis
Stage 2: Early ambulatory
Stage 3: Late ambulatory
Stage 4: Early non-ambulatory
Stage 5: Late non-ambulatory
Neuromuscular management
Lead the multidisciplinary clinic; advise on new therapies; provide patient and family support, education, and genetic counselling
Ensure immunisation schedule is complete
Assess function, strength, and range of movement at least every 6 months to define stage of disease
Discuss use of glucocorticosteroids
Initiate and manage use of glucocorticosteroids
Refer female carriers to cardiologist
Help navigate end-of-life care
Rehabilitation management
Provide comprehensive multidisciplinary assessments, including standardised assessments, at least every 6 months
Provide direct treatment by physical and occupational therapists, and speech-language pathologists, based on assessments and individualised to the patient
Assist in prevention of contracture or deformity, overexertion, and falls; promote energy conservation and appropriate exercise or activity; provide orthoses, equipment, and learning support
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
Endocrine management
Measure standing height every 6 months
Assess non-standing growth every 6 months
Assess pubertal status every 6 months starting by age 9 years
Provide family education and stress dose steroid prescription if on glucocorticosteroids

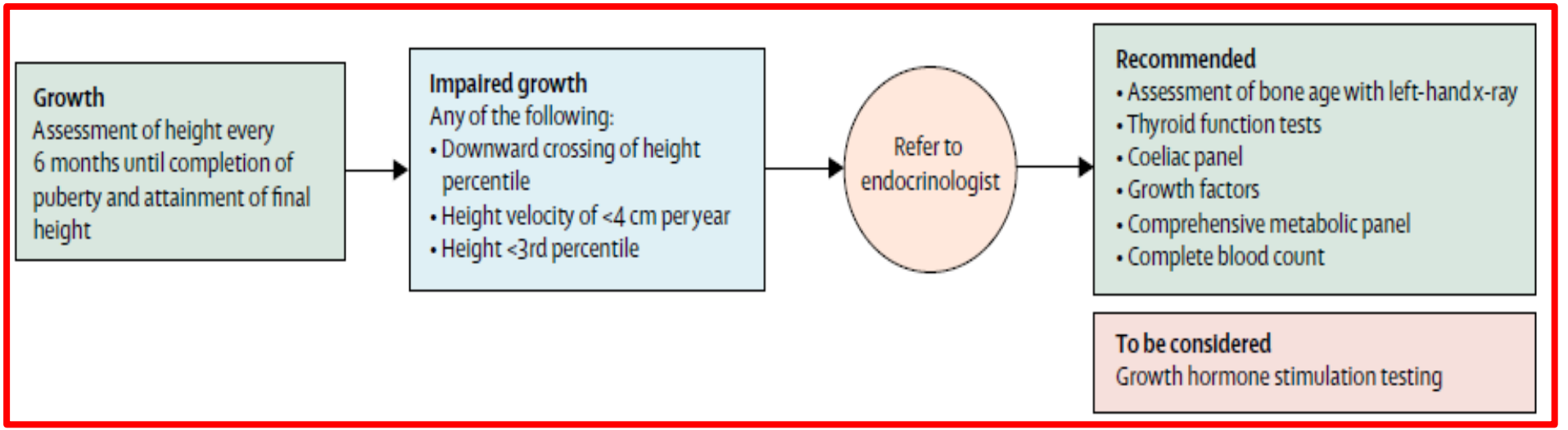
Neuromuscular Management

- 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

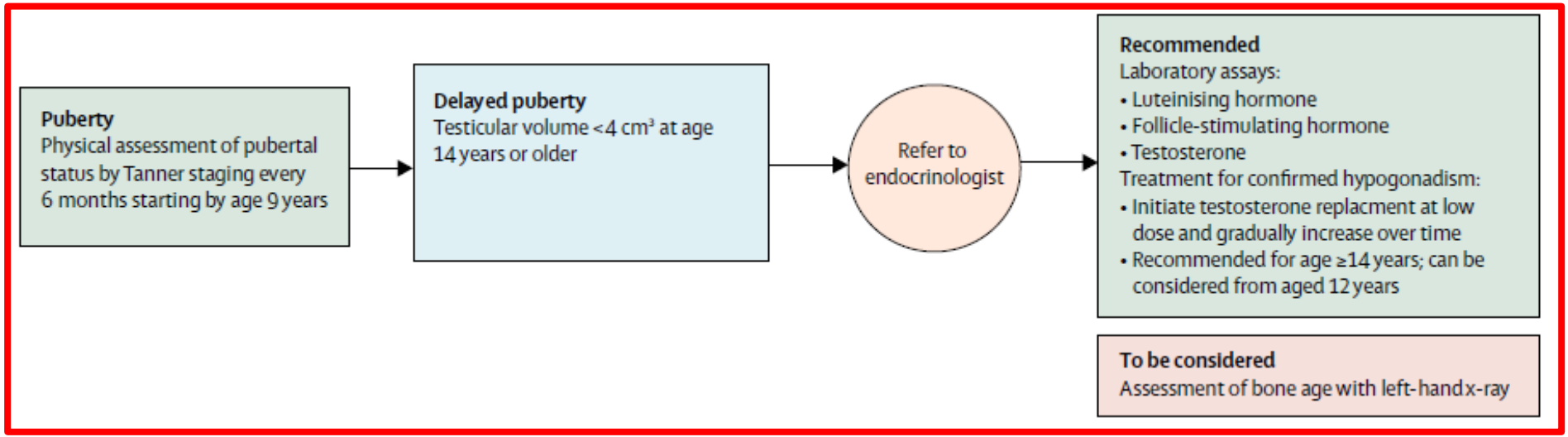
- Monitor growth and development
- Identify and diagnose hormone deficiencies
- Provide hormone replacement therapy when indicated
  - Limited safety and efficacy data for GH and testosterone
- Prevent adrenal crisis
- Bone health (discussed by others)

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

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