UCSF Benioff Children's Hospitals

Duchenne Muscular Dystrophy Standards of Care 2018

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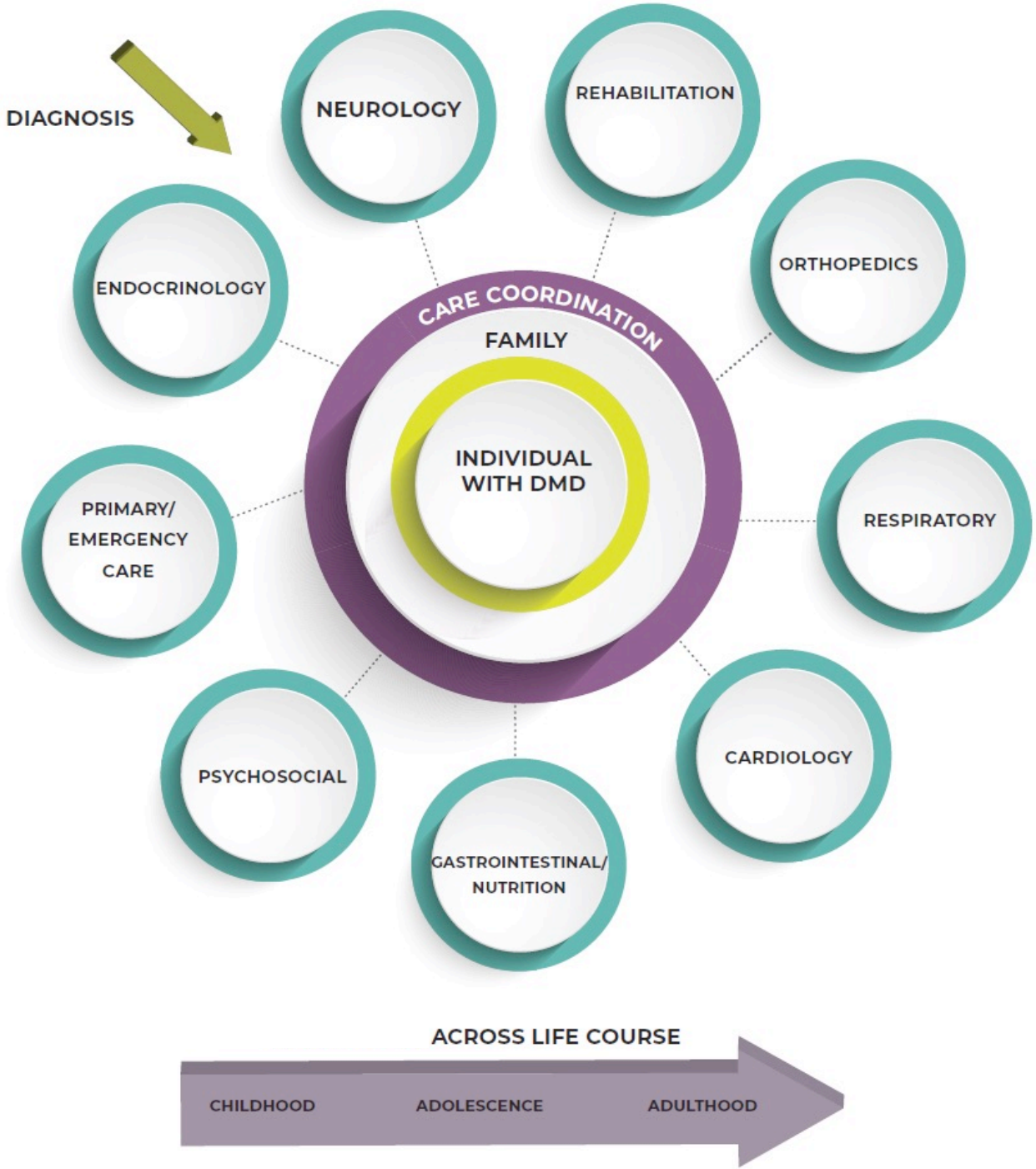

Members of the multidisciplinary care team

- Neuromuscular specialist
- Clinic coordinator
- Social worker
- Cardiologist
- Pulmonologist
- Endocrinologist
- Psychologist
- PT/OT and/or PM&R
- Dietician (RDN)
- Genetic counselor

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Stages of DMD

- Stage 1: Diagnosis
- Stage 2: Early Ambulatory
- Stage 3: Late Ambulatory
- Stage 4: Early Nonambulatory
- Stage 5: Late Nonambulatory

Neuromuscular management

- Overall management
  - Team leader
  - Discuss benefits and risks of approved therapies and current clinical trials
  - Provide information for emergency care providers
  - Communicate with primary care providers

- At diagnosis
  - Assure genetic testing and/or muscle biopsy is negative
  - Offer genetic counseling and testing for family members
  - Cardiology evaluation for female carriers by early adulthood
  - Document immunizations are up to date
    - Pneumococcal and yearly influenza
  - Discuss use of glucocorticosteroids

- Throughout all stages
  - Assess function/strength/ ROM
  - Manage glucocorticosteroids
    - Discuss signs, symptoms and management of adrenal insufficiency
      - Rx for IM hydrocortisone for emergency use at initiation

  - Help navigate end-of-life
  - Palliative care can be offered any time

Rehabilitation management

- PT, OT and/or PM&R specialists
- Provide comprehensive assessments, including standardized testing
  - Strength, function, ROM, timed function tests
- Refer to speech therapy if concerned about speech and/or swallowing

- Through early ambulatory
  - Prevent contractures, falls
    - Home stretching review
    - AFOs encouraged early
    - Ask about falls, fall prevention
  - Appropriate exercise
  - Discuss possible future needs

- From late ambulatory on
  - Continue previous discussions
  - Assess need for assistive devices/equipment
  - Ask about pain

Orthopedic and bone management

- Scoliosis
  - Manual inspection annually through ambulatory stages
    - Radiograph if curve observed
      - Lateral spine radiographs for bone health every 1-2 years on steroids; 2-3 years not on steroids
  - Radiographs when first nonambulatory
    - Every 6 months after curve detected
  - Refer to Orthopedist if curve >20 degrees or progressive for possible spinal fusion
- Achilles tendon contractures
  - Consider surgery to improve gait when ambulatory or for wheelchair positioning when nonambulatory
- Refer to bone health expert at earliest sign of fracture, symptomatic or asymptomatic

Endocrine management

- Often bone health speciaist
  - Discuss IV bisphosphonate therapy if fracture
- Standing and nonstanding heights at every visit
  - Refer to endocrinology when height percentile decreases or velocity <4 cm/yr suggesting impaired linear growth
- Pubertal assessments starting at age 9 years
  - Refer to endocrinologist in absence of pubertal development by 14 years
  - Consider testosterone replacement for boys >14 or boys >12 on steroids in absence of pubertal development

Pulmonary management

- Low risk during ambulatory years
  - PFTs annually from age 5
  - Sleep study for signs of sleep disordered breathing (snoring, morning headaches)
- Increased assessments during nonambulatory years
  - Twice yearly PFTs
  - Cough assist
  - Nocturnal-assisted ventilation (noninvasive preferred)
  - Daytime ventilation during last half of late nonambulatory stage

Cardiac management

- Cardiology evaluation with ECG at diagnosis and then annually
- Echo or cardiac MRI at baseline and atleast yearly with ECG or sooner if symptoms suggest
- ACE inhibitors or ARBs by age 10 if no cardiac dysfunction or sooner if signs develop
- Holter monitor for concerns of rhythm abnormalities
- Heart failure management as needed during late ambulatory phase or later

GI management

- Dietician (RDN) every clinic visit
  - Assess nutrition and growth, develop nutrition plan
  - Monitor for overweight especially when steroids starting, or becoming nonambulatory
  - Monitor for underweight especially if swallowing dysfunction or late nonambulatory stage
- Annual 25-OH vitamin D and calcium intake
  - Vitamin D supplementation if level <30 ng/mL
  - CA supplementation if dietary intake low
- Assess constipation, GERD, gastroparesis, swallow function
- Discuss gastrostomy tube for weight loss, dehydration, malnutrition, aspiration, dysphagia starting late ambulatory stage

Psychosocial management

- Assess mental health of patient and family from diagnosis on
  - Psychologist, social worker, psychiatric nurse
  - Informal mental health screen for depression and anxiety
    - Referral to psychiatric care if positive
- Neuropsychological evaluation for learning, emotional and behavioral problems
  - Speech and language delays, cognitive delays, autistic spectrum disorder, social communication difficulties, ADD/ADHD, OCD

Transitions

- Promote age-appropriate independence and social development
- Foster goal setting and future expectations
- Transition health care and anticipatory guidance about health changes
  - Decision-making supports or delegation of health care power or attorney
  - Advanced directives
- Transition for education and/or employment
- Mobility, transportation, housing, ADLs

Steroid management

- Steroid should be discussed at diagnosis and started some time before physical decline
- Vaccines should be up to date and varicella immunity established before starting
- Choose your regimen
  - Prednisone vs. deflazacort
  - Daily, 10-day on/off, 20-day on/10-day off, high dose weekend
  - Dosing decreased if side effects or increased if decline
  - Steroids through non-ambulatory stages
    - Consider starting later if steroid-naive

Steroid management - Cautions

- Adrenal insufficiency
  - Extreme fatigue
  - Weight loss and decreased appetite
  - Darkening of your skin (hyperpigmentation)
  - Low blood pressure, even fainting
  - Salt craving
  - Low blood sugar (hypoglycemia)
  - Nausea, diarrhea or vomiting
  - Abdominal pain
  - Muscle or joint pains
  - Irritability
  - Depression

- DO NOT STOP ABRUPTLY!
  - PJ Nicholoff steroid-tapering protocol
- IM hydrocortisone if unable to take by mouth
- Stress dose if taking >12mg/m^2/day
  - Severe illness
  - Major trauma
  - Surgery

Steroid management – Side effects 1

Side effect	Precautions	Management
Weight gain/obesity	Increased appetite	Eat sensibly
Cushingoid features	Fullness in face and cheeks	Diet, minimize sugar and salt
Excessive hair growth	Anywhere on body	None needed
Acne/fungal skin infections	Usually during teen years	Topic prescriptions
Short stature	Monitor height	Endocrinologist
Delayed puberty	Monitor from age 9	Consider testosterone

Steroid management – Side effects 2

Side effect	Precautions	Management
Behavioral changes	Often worsen in initial 6 weeks	• Treat baseline issues prior to starting steroids • Consider changing timing of steroids
Immune suppression	Address minor infections promptly	• Immunizations up to date • Varicella immunity
High blood pressure	BP should be checked regularly	Decrease salt and weight or consider additional medication management

Steroid management - Side effects 3

Side effect	Precautions	Management
Adrenal suppression	• **Inform medical personal taking steroids and carry an alert** • **Don't miss more than 24 hours**	• **Consider stress doses** • **IM hydrocortisone** • **Tapering protocol**
Glucose intolerance	• Monitor sugar in urine • Monitor blood for signs of DM • Increased thirst and increased urination	• Limit sugar • Consider diabetes treatment
Gastritis/GERD	Stomach acid going into esophagous	• Avoid NSAIDs/ASA • Antacids

Steroid management – Side effects 4

Side effect	Precautions	Management
Peptic Ulcer Disease	• Stomach pain • Check stool for blood	• Avoid NSAIDs • Antacids • GI referral
Cataracts	• Usually benign • Annual eye exam • Deflazacort higher risk	• Consider switch to prednisone • Ophthalmology consult • Remove surgically if interfere with vision
Osteoperosis	• Discuss fractures and back pain • Discuss exercise/ weight bearing program before starting	• Spine x-rays every 1-2 years • Dexa scans every 2-3 years • Yearly Vit D levels and supplement as needed • Dietary calcium/supplements

Emergency Care Considerations

- Always carry an emergency card (or PPMD App)
  - Dx
  - Meds
  - PFT/cardiac function studies
  - Past medical problems (ie. pneumonia, heart failure, gastroparesis, etc.)
- Contact NMS after initial assessment
- Advise emergency staff if taking steroids
- If oxygen levels drop be careful about getting oxygen without breathing support
- If pain, be very careful with narcotics, especially without breathing support
- If you have a broken bone insist they speak with NS and watch for signs of fat embolism syndrome
  - Confusion/disorientation, “not acting yourself,” rapid breathing and heart rate, shortness of breath
- Bring equipment with you to hospital (ie. cough assist)

Thank you

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