Care Considerations In Duchenne Muscular Dystrophy

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Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management

Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care

Supported by:
Centers for Disease Control and Prevention
TREAT-NMD

DMD Care Considerations
DMD Coordinated Care

Depression
Care Coordination
Stuttering Management
Pulmonary Management
Cardiac Management
Urinary Management
Nutritional Management

Road Map

Diagnosis and Pediatric Care Considerations
Pulmonary Care Considerations
Transitioning Care From Pediatric to Adult Practitioners

http://www.treat-nmd.eu/patients/DMD/familyguide/

Objectives

At the end of this Grand Rounds, the attendee will be able to report:
- Epidemiology and genetics of DMD
- When to suspect and how to diagnose DMD
- Multidisciplinary care considerations and options
- Transition challenges and adult DMD issues as young men with DMD live into their 20s and 30s
wgrz MDA Video: 9/2009

Young Man from Lockport is a Muscular Dystrophy Survivor

Video: Samuel Prentice has lived with Duchenne Muscular Dystrophy for nearly his entire life. His story of hope and survival is an inspiration, as he looks toward his future with his new fiancee.


Diagnosis and Pediatric Care Considerations

Duchenne

When to Suspect DMD

- Developmental Delay
- Elevated CPK
- Elevated SGPT/SGOT
- Child Runs Awkwardly Or Never Really Ran
- Sits At Top Of Stairs And Won’t Come Down

Gower’s Sign

Lordosis
Diagnosis of DMD

• Large Deletion - 2/3 Confirmed Cases

• Sequencing - Duplications, Point Mutation etc. - Increases Pick-up to 95-97 %

• Biopsy Rarely Used

Duchenne Muscular Dystrophy

Dystrophin Present

Dystrophin Not Present

Dystrophin – Largest Gene In Body
**X Linked Recessive**

**DMD**
- Known Protein Product - Dystrophin
- Know Gene And Can Detect Abnormalities
- Have Animal Models (eg DMX Mouse)

**Multidisciplinary Approach**
- Neurology
- Rehabilitation/Orthopedics
- Pulmonary
- Cardiology
- Nutrition
- Social Work/Patient Representative/Support Groups

**Treatment**
- Maximize Function of Affected Areas
- Steroids
- Future Work - Replace Missing Protein Or Allow Gene To Protein Normally

**“Old” vs. “New” Stats**
- “Old” Stats Quote Death By Early 20’s And Places Cardio-Pulmonary Deaths At About 12%
- “New” Stats Recognize that There are Young Adults With DMD Who Have Problems Which Don’t Develop until an Older Age
Pulmonary Care Considerations

DMD Care Considerations
Pulmonary Recommendations

Stage 1:
Presymptomatic
- Education and support
- Preventive measures to maintain

Stage 2:
Early Ambulatory
- Low risk of respiratory problems
- Monitor progress

Stage 3:
Late Ambulatory
- Increasing risk of respiratory impairment
- Trigger respiratory assessments

Stage 4:
Early Non-ambulatory
- High risk of respiratory impairment
- Trigger respiratory investigations and interventions

Stage 5:
Late Non-ambulatory

DMD; N= 47; 6 to 20 years of age; 263 VC measurements

Changes in Spirometry Over Time as a Prognostic Marker in Patients with Duchenne Muscular Dystrophy.

Natural History: DMD Respiratory Muscle Weakness
Hypoventilation Awake
Hypoventilation Asleep
Weak Cough
Natural History: DMD Respiratory Muscle Weakness

<table>
<thead>
<tr>
<th>Pco2, (SpO2)</th>
<th>Hypoventilation Awake</th>
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<tr>
<td>FVC, PETCO2, (SpO2)</td>
<td>Sleep Evaluation</td>
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<td>Weak Cough</td>
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Age, years

5 10 15 20 25

Hypoventilation Asleep

PETCO2, (SpO2)

Sleep Evaluation

PCF, MEP, FVC

Weak Cough

Age, years

5 10 15 20 25

Changing Natural History of DMD

Survival in Duchenne muscular dystrophy: improvements in life expectancy since 1967 and the impact of home nocturnal ventilation

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Received 16 March 2002; accepted 29 May 2002

Duchenne Muscular Dystrophy
Prolongation of Life by Noninvasive Ventilation and Mechanically Assisted Coughing

ABSTRACT

Transitioning Care From Pediatric to Adult Practitioners

Challenges of Transition
- When to transition? Best if based on:
  - Specific age?
  - Clinical severity of disease?
  - Hospital regulations?
  - Physician (both pediatric and adult) comfort?
- Regardless of above, need to develop a plan months-years ahead of time to ensure a safe and smooth transition

Scope of the Problem
- MMWR 10/9/09 indicated that ~58-85% of patients with Duchenne and Becker Muscular Dystrophy (DBMD) are surviving into adulthood
- “...As males with DBMD increasingly survive into adulthood, practitioners who care for adults are increasingly challenged to manage patients who have rare disorders, who historically did not survive to adulthood...”

Challenges of Transition
- May occur when cardiac and/or pulmonary status is most vulnerable
- Involves multiple specialties and subspecialties (Pediatrics, Internal Medicine, Neurology, Rehabilitation Medicine, PT/OT, Orthopaedics, Cardiology, Pulmonology, etc.)
- Transition from pediatric to adult MDA Clinic
- Adult practitioners less familiar with DBMD, NPPV, Duchenne cardiomyopathy, and masking of hypoventilation by oxygen
Why is Transition Important?

- As children with DBMD become adults with DBMD, it is imperative that adult practitioners become familiar with manifestations of the disease and methods best suited to treat them.
- Adults with DBMD may have other medical comorbidities better treated by adult physicians (just as pediatricians better suited to treat common pediatric disorders).

Disease Manifestations in Adulthood

- In addition to muscle weakness, cardiac, pulmonary, and psychosocial manifestations, adults with DBMD are at risk for secondary manifestations of the disease and/or its treatment.

Concept of Prolonged Survival as Exposure to Risk

- Risks of aspiration and malnutrition due to oropharyngeal dysphagia
- Risks of necessary procedures - e.g. PEG, tracheostomy, scoliosis surgery
- Risks of immobility - e.g. DVTs, cholelithiasis, nephrolithiasis
- Risks of treatment with glucocorticoids - Diabetes, osteoporosis, hypertension, cataracts, etc.
- Risks of prolonged survival on psychosocial well-being

Carrier Females

- Daughters of men with DBMD and mothers of affected children with family history of DBMD - obligate carriers of mutated dystrophin gene
  - Sisters of affected patients also at risk
- Rarely develop muscle weakness due to skewed inactivation of normal X chromosome
- May develop cardiac dysfunction similar to DBMD and must be monitored (starting in as a teen and periodically throughout adulthood)

Our Goal

- Identify adolescent patients with neuromuscular disorders (including DBMD) suitable for transition to adult practitioners and develop an individualized plan months-years ahead of time
- Work with members of the medical community (both primary care physicians and sub-specialists) to ensure smooth transition of care

DMD Care Considerations

References

Lancet Neurology

- Articles

Podcast

Duchenne Partners

- Muscular Dystrophy Association http://www.mda.org
- Parent Project Muscular Dystrophy http://www.parentprojectmd.org
- TREAT-NMD http://www.treat-nmd.org