

Care Considerations In Duchenne Muscular Dystrophy

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Lancet Neurol 2010; 9: 177-89

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

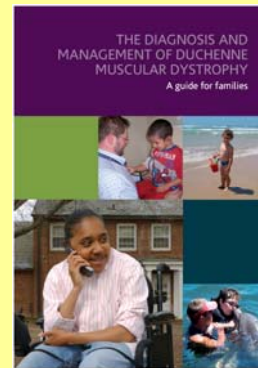
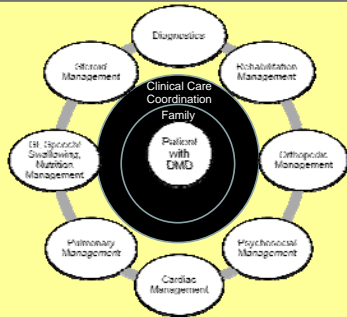
Katharine Bushby, Richard Finkel, David J Binkorst, Laura E Coze, Paula R Clemens, Linda Crisp, Ajay Kaul, Kathi Kinnett, Craig McDonald, Shree Pandya, James Poysky, Frederic Shapiro, Jean Tomezsko, Carolyn Constantin, for the DMD Care Considerations Working Group*

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

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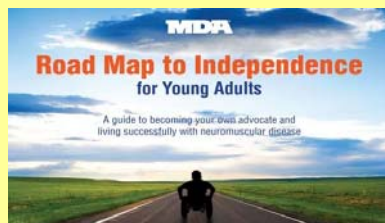
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DMD Care Considerations DMD Coordinated Care



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

Road Map



Diagnosis and Pediatric Care Considerations

Pulmonary Care Considerations

Transitioning Care From Pediatric to Adult Practitioners

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 fiancée.

<http://www.wgrz.com/video/default.aspx?bctid=49887001001>

Diagnosis and Pediatric Care Considerations

Duchenne



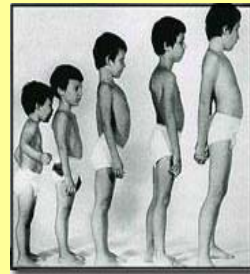
When to Suspect DMD

- 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



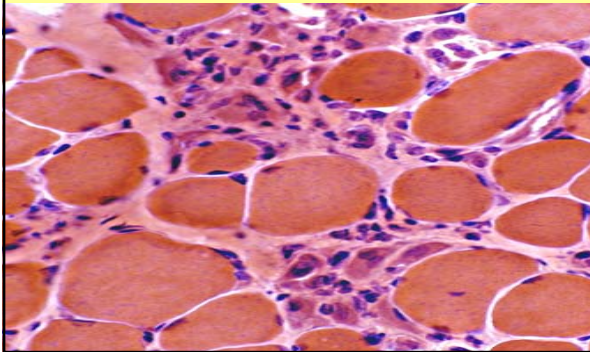
Diagnosis of DMD

- Large Deletion -2/3 Confirmed Cases
- Sequencing - Duplications, Point Mutation etc. - Increases Pick-up to 95-97 %
- Biopsy Rarely Used

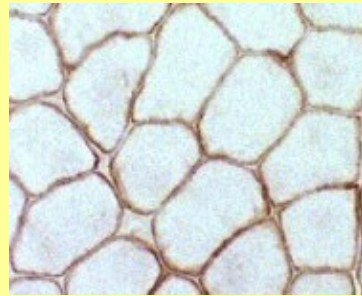
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Duchenne Muscular Dystrophy



Dystrophin Present

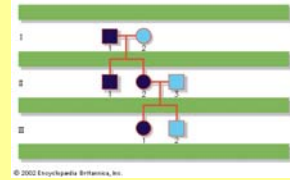


Dystrophin Not Present



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

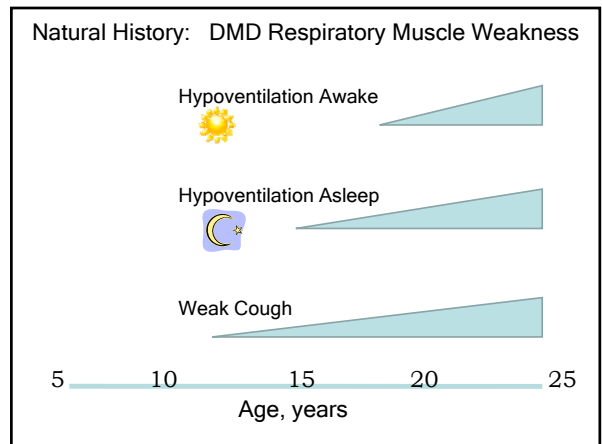
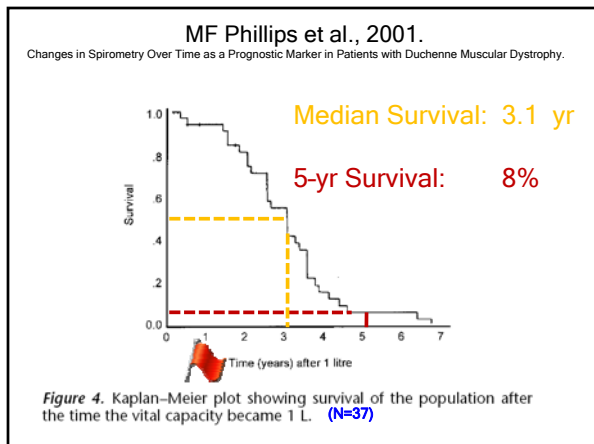
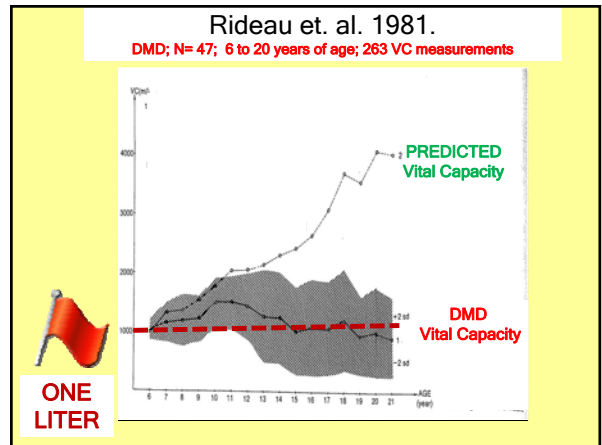
- “Old” Stats Quote Death By Early 20’s And
Places Cardio-Pulmonary Deaths At About
12%
- “New” Stats Recognize that There are
Young Adults With DMD Who Have
Problems Which Don’t Develop until an
Older Age

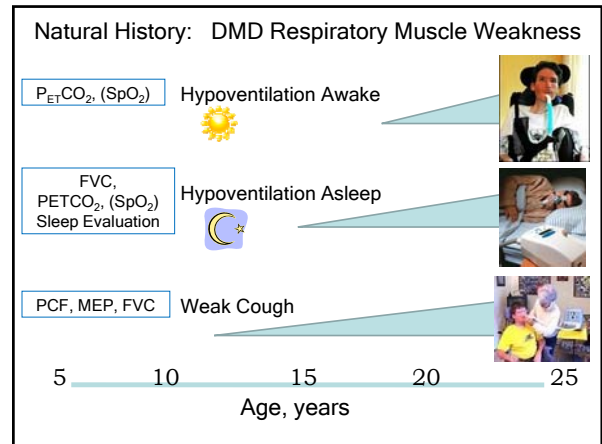
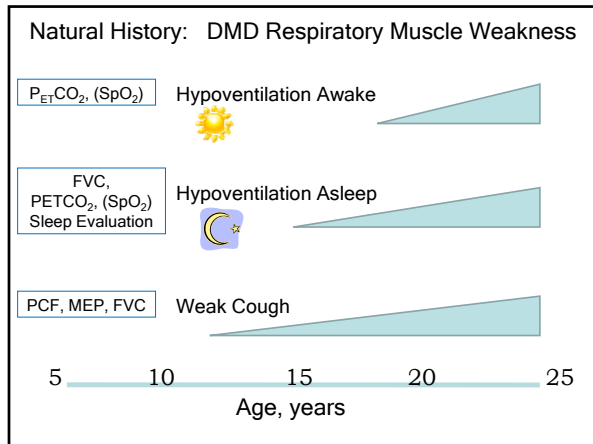


Pulmonary Care Considerations

DMD Care Considerations Pulmonary Recommendations

Stage 1: Presymptomatic	Stage 2: Early Ambulatory	Stage 3: Late Ambulatory	Stage 4: Early Non-ambulatory	Stage 5: Late Non-ambulatory
<ul style="list-style-type: none"> •Ensure usual immunization schedule includes 23-valent pneumococcal and influenza vaccines 	<ul style="list-style-type: none"> •Low risk of respiratory problems •Monitor progress 		<ul style="list-style-type: none"> •Increasing risk of respiratory impairment •Trigger respiratory assessments 	<ul style="list-style-type: none"> •High risk of respiratory impairment •Trigger respiratory investigations and interventions





INSPIRING WELLNESS

Keeping Your Lungs Healthy When You Have Respiratory Muscle Weakness

Handling Colds

During a cold, a weak cough can become weaker and it is more difficult to clear mucus from the lungs. People with a muscular dystrophy have normal lungs, so if the oxygen saturation is going down, it means more help is needed to cough and breathe (NOT just extra oxygen is needed). The following phase are very helpful in keeping the lungs clear and keeping oxygen saturations above 94%.

- At least every 4 hours:
 - Cough Assist: 8 sets of 8 breaths, [going on inspiration](#)
 - If Heat Therapy or Chest PT is available, then:
 - Allevy Clearance (Chest PT or Vaper Therapy) for 10-20 minutes
 - Followed By Cough Assist: 8 sets of 8 breaths, [going on inspiration](#)
- Use the Cough Assist whenever you hear or feel a rattle in the chest. Suction the mouth when mucus is thick in the back of throat.
- If you have them at home, use B2AP or the ventilator with @ sleep (i.e. nose and mouth), with side, w/ or w/o assist.
- Use the ventilator, once a day when well and at least 3 times a day when sick. If the screen saturation is less than 92%, use the Cough Assist to clear secretions and then suction. The oxygen saturation: If the screen saturation is 92% or less or the Cough Assist is not increasing the screen saturation to 92% or higher, please call us.

Monday through Friday from 9-4: 878-7524
Other times: 878-7000 and ask to speak with the Lung Center physician on-call.

A Assisted
B Breathing **C Center**

Changing Natural History of DMD

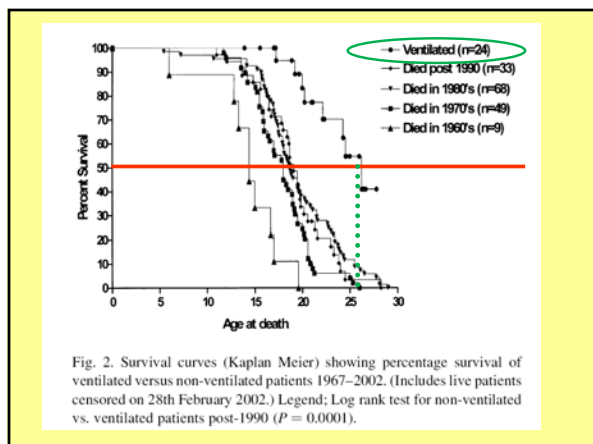
PERGAMON
Neuromuscular Disorders 12 (2002) 926-929
www.elsevier.com/locate/nmd

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

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^dDepartment of Mathematics and Statistics, University of Northumbria at Newcastle, Newcastle upon Tyne, UK
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Received 13 March 2002; received in revised form 28 May 2002; accepted 7 June 2002



Duchenne Muscular Dystrophy

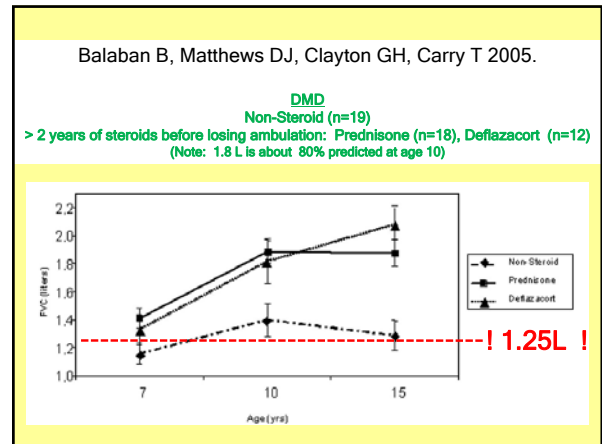
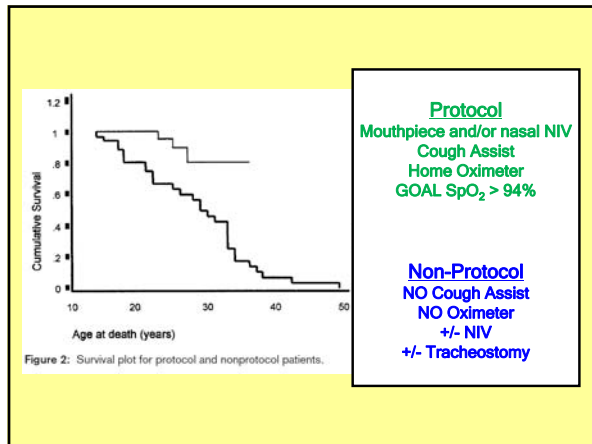
Prolongation of Life by Noninvasive Ventilation and Mechanically Assisted Coughing

ABSTRACT

Gomez-Merino E, Bach JR: Duchenne muscular dystrophy: Prolongation of life by noninvasive ventilation and mechanically assisted coughing. *Am J Phys Med Rehabil* 2002;31:411–415.

Management of Patients with Neuromuscular Disease

John R. Bach, MD



Transitioning Care From Pediatric to Adult Practitioners

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

- 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

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
Bushby K, Finkel R, Birnkrant D, et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management. *The Lancet Neurology*. 2010;9(1):77-93.

Bushby K, Finkel R, Birnkrant D, et al. Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care. *The Lancet Neurology*. 2010;9(2):177-189.

Podcast

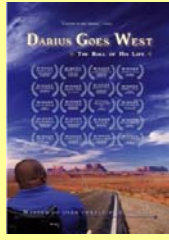
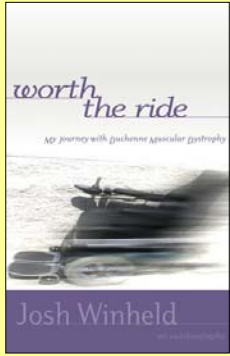
Katharine Bushby (2009, December 12). Retrieved from http://podcast.thelancet.com/audio/lancet/2010/lancet_january.mp3

Duchenne Partners

Muscular Dystrophy Association
<http://www.mda.org>

Parent Project Muscular Dystrophy
<http://www.parentprojectmd.org>

TREAT-NMD
<http://www.treat-nmd.org>



www.dariusgoeswest.org