

# ISPO Newcastle 2010

## **STANDARDS OF CARE FOR DMD**

Michelle Eagle

# Duchenne muscular dystrophy



- Progressive muscle weakness
- X-linked recessive inheritance (1:4000 boys)
- Symptoms from age 3
- Later respiratory and heart involved
- Wheelchair from age 10
- Death at early age if untreated

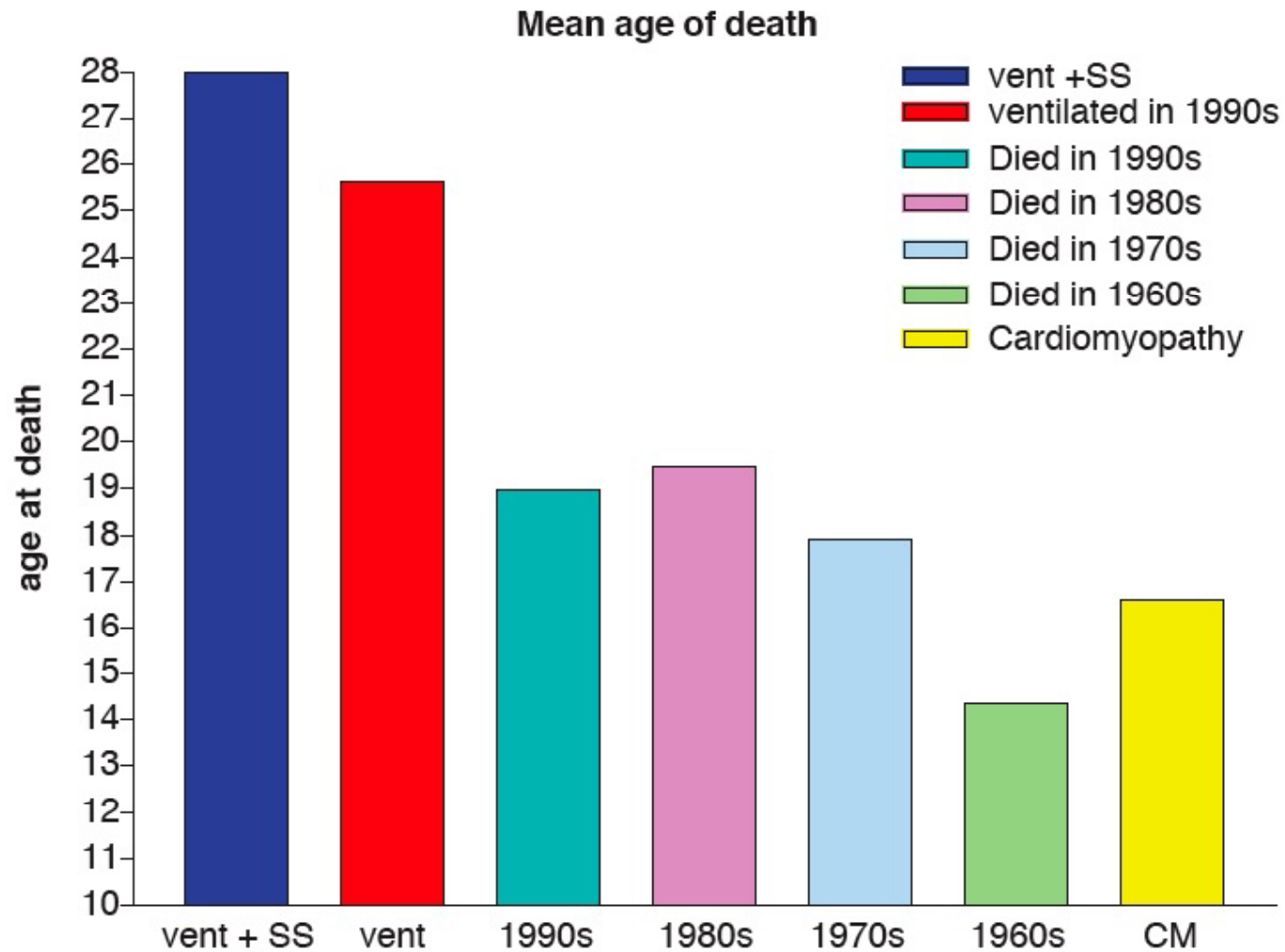
*From Erb et al, Nervenheilkd 1:13, 1891*

# Standards of Diagnosis and Care

## Reasons:

1. A joint basis for diagnosis and care is a pre-requisite for well performed multinational clinical trials
2. Low prevalence of the disorders necessitates expert advice on management being collated on a multinational level
3. Families should be offered the most informed treatment and counselling regardless of where they live

## Changed treatment affects survival:



	<b>RAND Appropriateness Method</b>	<b>NIH Consensus Conferences</b>	<b>Nominal Group Process</b>	<b>Delphi Method</b>
<b>Purpose</b>	<ul style="list-style-type: none"> <li>▶ To detect when the experts agree</li> </ul>	<ul style="list-style-type: none"> <li>▶ To obtain a consensus among participants</li> </ul>	<ul style="list-style-type: none"> <li>▶ To gather and rank ideas from a group</li> </ul>	<ul style="list-style-type: none"> <li>▶ To detect when the experts agree</li> </ul>
<b>Method Description</b>	<ul style="list-style-type: none"> <li>▶ Modified Delphi focused on appropriateness with an opportunity for face to face discussion of results</li> </ul>	<ul style="list-style-type: none"> <li>▶ In-person meetings that last until the group has agreed on a statement</li> </ul>	<ul style="list-style-type: none"> <li>▶ Round robin discussion of key ideas followed by a numerical vote</li> </ul>	<ul style="list-style-type: none"> <li>▶ Silent voting process with no interaction between participants</li> </ul>
<b>Important steps/processes</b>	<ul style="list-style-type: none"> <li>▶ Literature review</li> <li>▶ Development of a highly structured list of indications</li> <li>▶ Delphi ratings</li> <li>▶ In-person discussion</li> <li>▶ Effective group facilitation</li> <li>▶ Statistical analysis</li> <li>▶ Feedback on results</li> </ul>	<ul style="list-style-type: none"> <li>▶ Literature review</li> <li>▶ Summarizing the state of knowledge</li> <li>▶ Presentations by the audience</li> </ul>	<ul style="list-style-type: none"> <li>▶ In-person meeting</li> <li>▶ Group moderation</li> <li>▶ Listing ideas</li> <li>▶ Rank ordering</li> <li>▶ Presentations</li> <li>▶ Statistical analysis</li> </ul>	<ul style="list-style-type: none"> <li>▶ Silent voting</li> <li>▶ Feedback on results</li> <li>▶ Repeat voting</li> </ul>
<b>Results</b>	<ul style="list-style-type: none"> <li>▶ Objectively- and subjectively- supported summaries of when to use specific medical tools</li> </ul>	<ul style="list-style-type: none"> <li>▶ General recommendations about how to use specific medical treatments or tools</li> </ul>	<ul style="list-style-type: none"> <li>▶ Prioritized ideas based on expert opinion</li> </ul>	<ul style="list-style-type: none"> <li>▶ Objectively-supported summaries of when to use specific medical tools</li> </ul>



## How was consensus reached?

- A VERY intensive detailed process!
- NOT just by discussion....
- RAND, -combines scientific evidence with consensus opinion among experts
- 84 experts
- 489 articles selected
- Rating of "signs and symptoms"
- Rating of "assessment tools / interventions"
- 2 anonymous ratings with in-person meeting in between



## International experts were part of one or more of these panels:

- Cardiology
- Pulmonology
- Neuromuscular
- Orthopedics/Surgical
- Gastrointestinal/Nutrition
- Physical Therapy/Rehabilitation
- Psychosocial/Family Functioning
- Diagnostics



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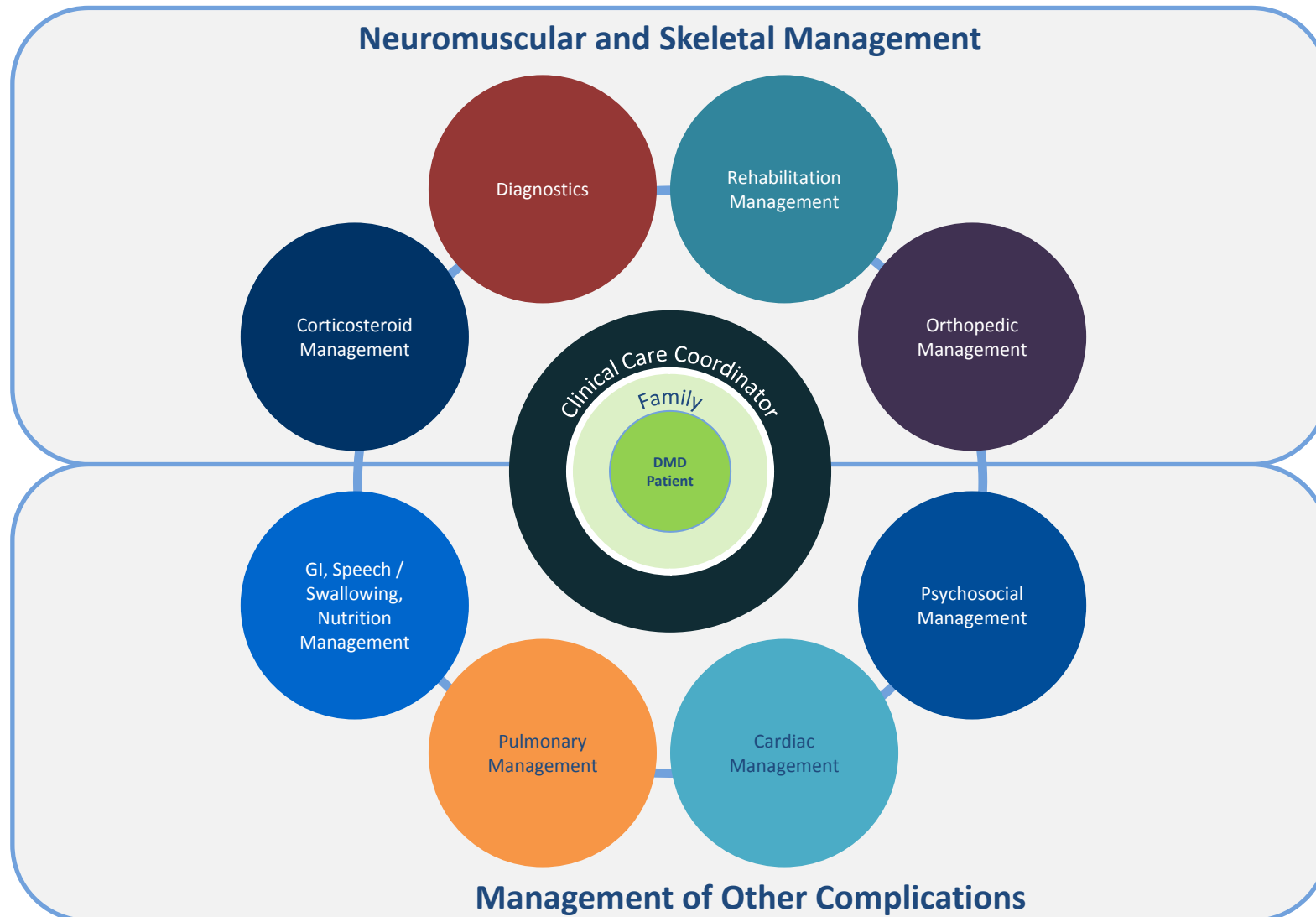
# Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management

## part 2: implementation of multidisciplinary care

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

[www.treat-nmd.eu/patients/DMD/dmd-care/](http://www.treat-nmd.eu/patients/DMD/dmd-care/)

# Interdisciplinary Management of Duchenne Muscular Dystrophy:



# The process followed stages of disease:



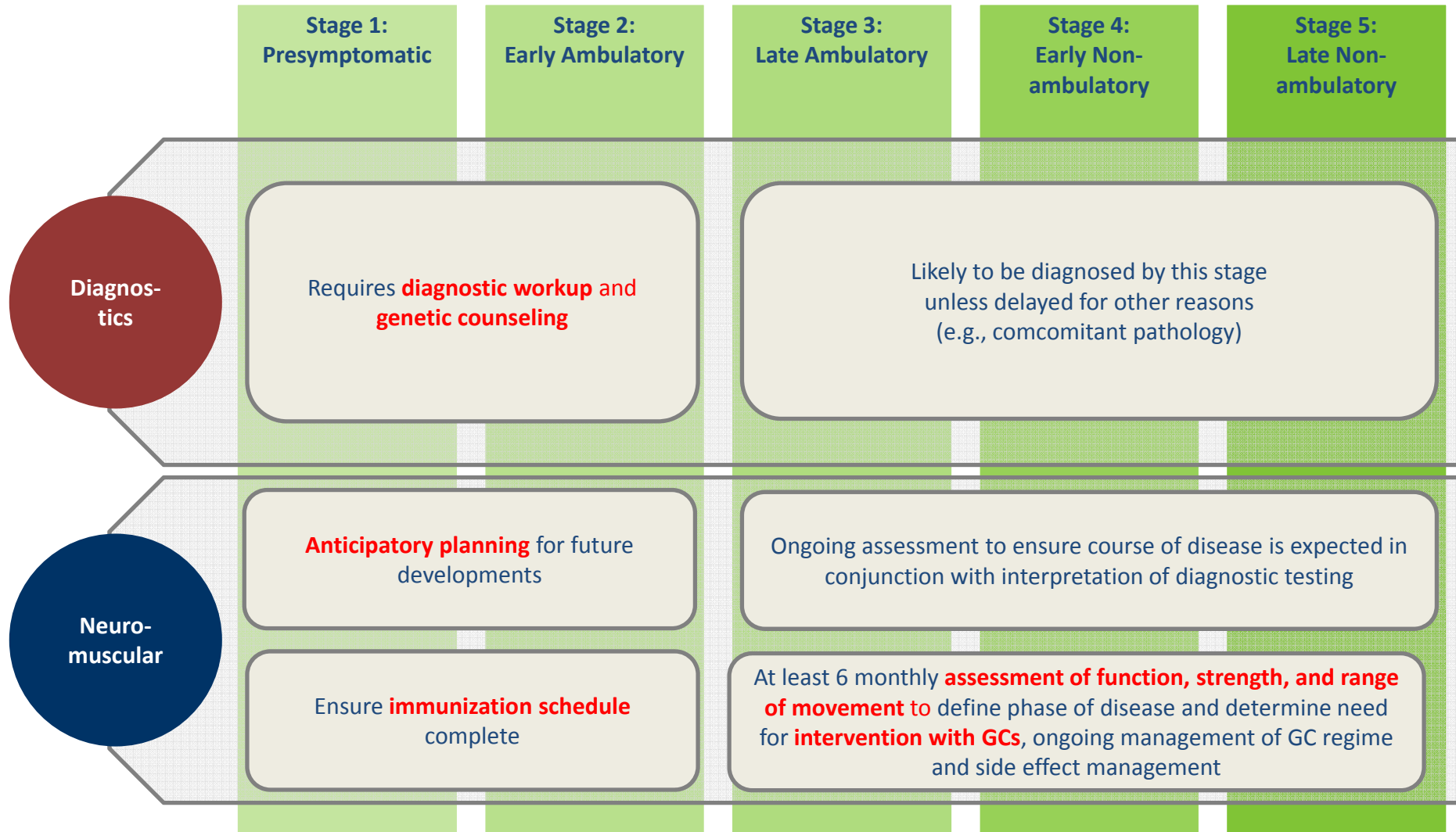


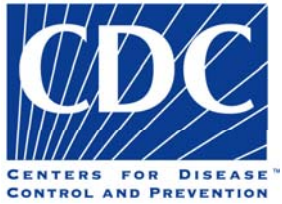
# DMD Stages of Disease:



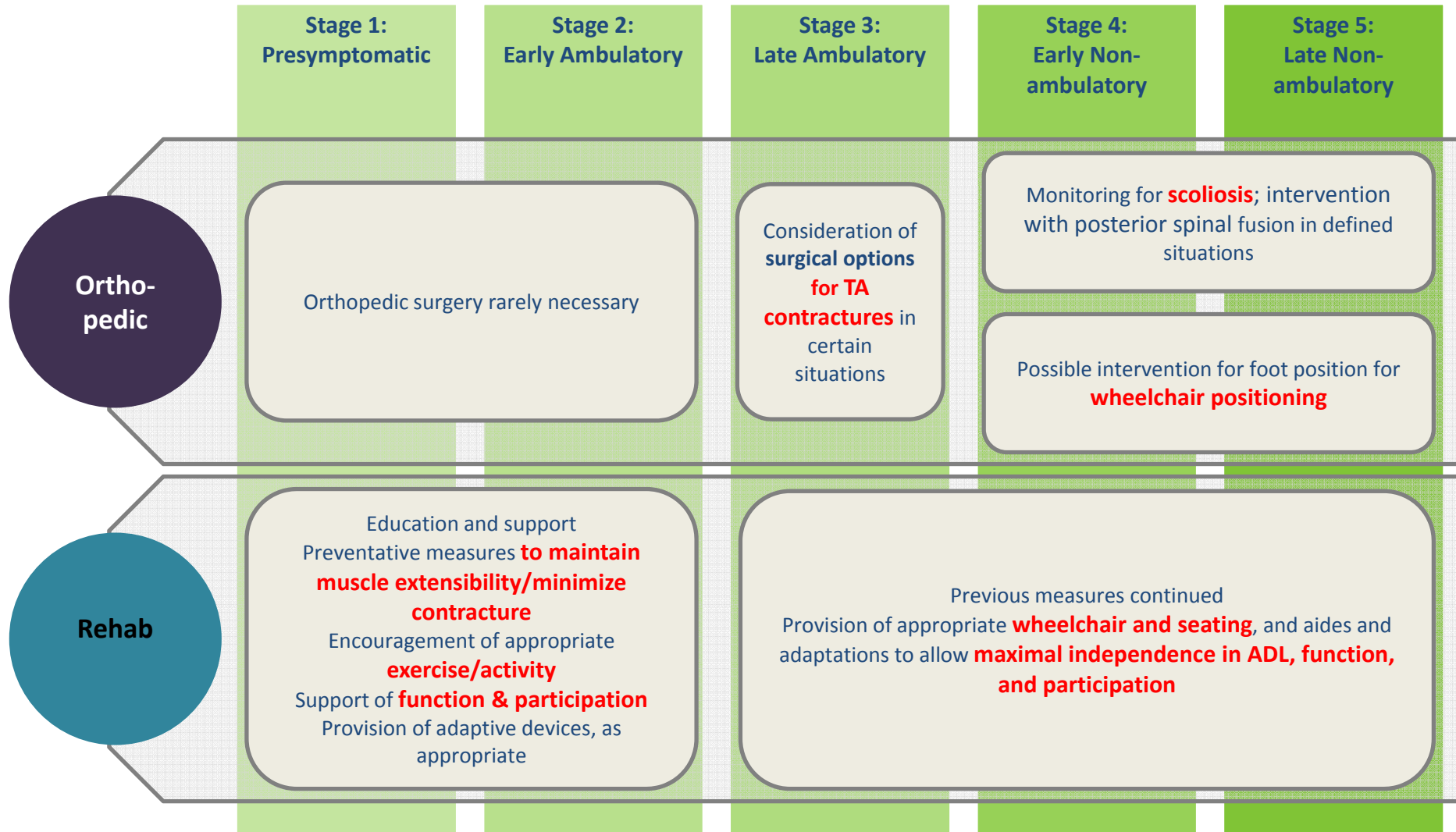


# Care Considerations by Stages of Disease:





# Orthopedic - Rehab:





# Rehabilitation Management of DMD

muscle extensibility and joint contractures

Caused by

- Loss of ability to actively move a joint through its full range of motion,
- static positioning in a position of flexion,
- muscle
- imbalance about a joint,
- fibrotic changes in muscle tissue.

# Joint contractures

## Reasons for prevention

- The maintenance of good ranges of movement and bilateral symmetry are important to allow optimum
- movement and functional positioning,
- to maintain ambulation, prevent development of fixed deformities,
- maintain skin integrity



# Methods of managing contractures

- **Stretching and positioning**
- Effective stretching of the musculotendinous unit requires a combination of interventions
- Active, active-assisted and passive stretching,
- Prolonged elongation using positioning, splinting, orthoses, and standing devices
- Active, active-assisted, and/or passive stretching to prevent or minimise contractures should be done a minimum of 4–6 days per week for any specific joint or muscle group
- Stretching should be done at home and/or school, as well as in the clinic.
- As standing and walking become more difficult, standing programmes are recommended



# Assistive devices for musculoskeletal management

## *Orthoses*

- Prevention of contractures also relies on resting orthoses, joint positioning, and standing programmes.
- Resting ankle-foot orthoses (AFOs) used at night can help to prevent or minimise progressive equinus contractures and are appropriate throughout life
- AFOs should be custom-moulded and fabricated for comfort and optimum foot and ankle alignment



# KAFOs



- Knee–ankle–foot orthoses (KAFOs; eg, long leg braces or callipers) for prevention of contracture and deformity can be of value in the late ambulatory and early non-ambulatory stages to allow standing and limited ambulation for therapeutic purposes
- Use of AFOs during the daytime can be appropriate for full-time wheelchair users.



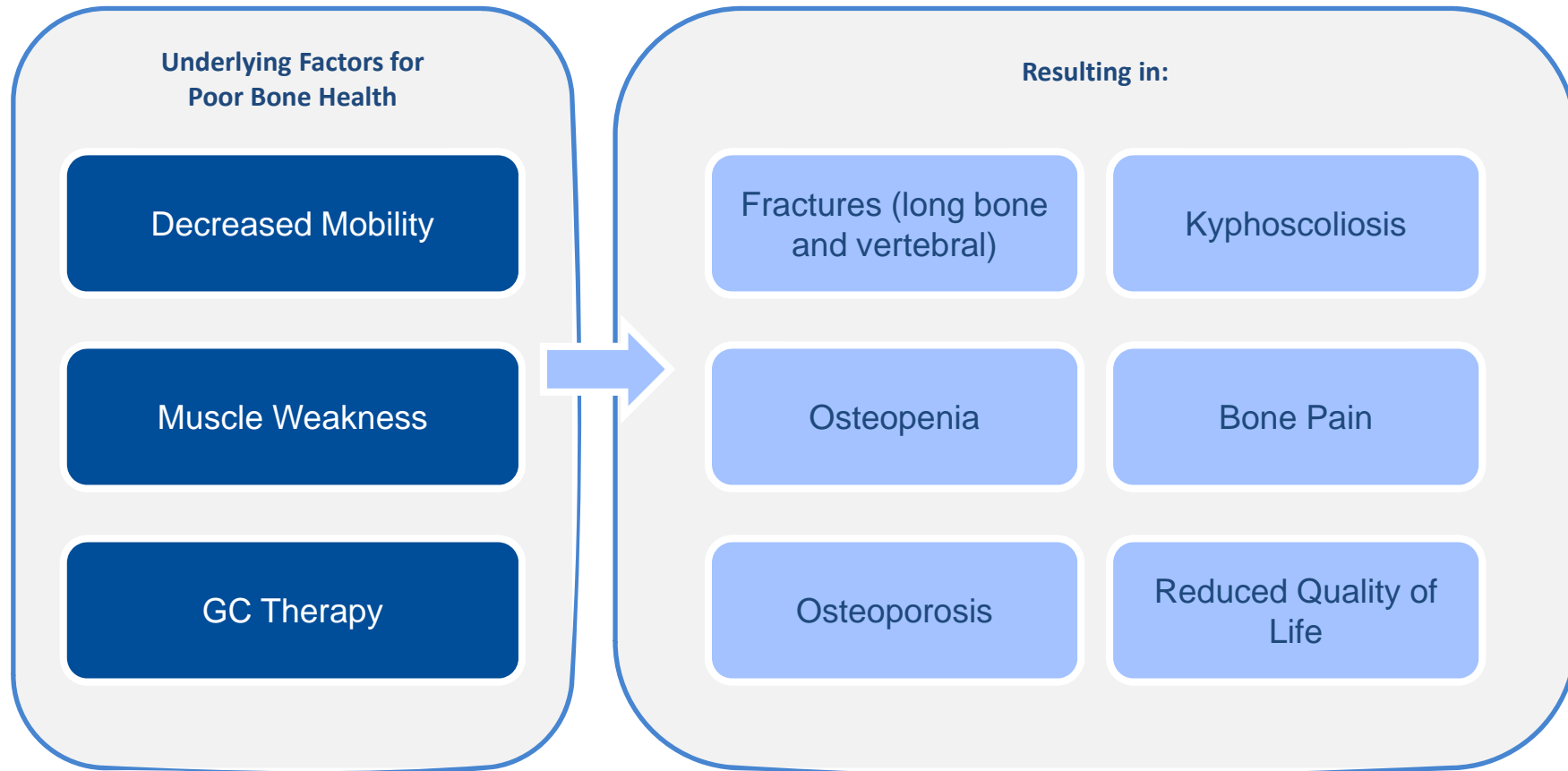


# Non-ambulant

- A passive standing device for patients with either no or mild hip, knee, or ankle contractures is necessary for late ambulatory and early non-ambulatory stages
- Many advocate continued use of passive standing devices or a power standing wheelchair into the late non-ambulatory stage if contractures are not too severe to restrict positioning and if devices are tolerable
- Resting hand splints for patients with tight long finger flexors are appropriate



# Bone Health Issues:



Note: use of standing and mobilisation not specifically mentioned in SOC but prolongation of standing and walking i.e. Increased mobility is promoted

# exercise

- Swimming is highly recommended from the early ambulatory to early non-ambulatory phases and could be continued in the non-ambulatory phase as long as it is medically safe.
- Additional benefits might be provided by low-resistance strength training and optimisation of upper body function.
- Significant muscle pain or myoglobinuria in the 24-h period after a specific activity is a sign of overexertion and contraction-induced injury, and if this occurs the activity should be modified



# Exercise

- Submaximum, aerobic exercise/activity is recommended
- especially early in the course of the disease
- avoid overexertion
- High-resistance strength training and eccentric exercise are inappropriate due to contraction-induced muscle-fibre injury
- To avoid disuse all ambulatory/early non-ambulatory boys should participate in regular submaximum functional activity Including a combination of swimming-pool exercises and recreation-based exercises in the community.



# wheelchairs

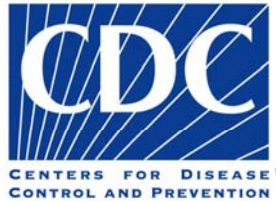
- early ambulatory stage, a lightweight manual chair
- late ambulatory stage, an ultra lightweight manual wheelchair supportive seating
- early non-ambulatory stage, a manual wheelchair with custom seating and recline as a necessary back-up to a powered wheelchair.
- As functional community ambulation declines, a powered wheelchair is advocated



# Electric chairs

- Custom seating and power positioning components recommended for the initial powered wheelchair,
- Including headrest lateral trunk supports, power tilt and recline, power-adjustable seat height, and power-elevating leg rests (with swing-away or flip-up footrests to facilitate transfers)
- Some recommend power standing chairs.
- Additional modifications could include a pressure-relieving cushion, hip guides, and flip-down knee adductors.





[www.treat-nmd.eu/patients/DMD/dmd-care/](http://www.treat-nmd.eu/patients/DMD/dmd-care/)

## DMD care standards

### Introduction

A major international consensus document setting out best practice in care for Duchenne muscular dystrophy (DMD) was published in the *Lancet Neurology* journal in January and February 2010. The product of an extensive review process by 84 international experts in DMD diagnosis and care, this document is a unique guide to expert recommendations on the care that all individuals with DMD should receive.

The document is available in the January and February editions of the printed journal, online from the journal's website, or here on this page, with special permission from the journal's publisher Elsevier.



**[Click here to download the full document \(parts 1 and 2 combined\)\\*.](#)**

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

### **The Diagnosis and management of Duchenne muscular dystrophy**

**Part 1: diagnosis, and pharmacological and psychosocial management\***

*Lancet Neurol.* 2010 Jan; 9(1):77-93. Epub 2009 Nov 27.  
PMID: 19945913, DOI: 10.1016/S1474-4422(09)70271-6

**Part 2: implementation of multidisciplinary care\***

*Lancet Neurol.* 2010 Feb; 9(2):177-189. Epub 2009 Nov 30.  
PMID: 19945914, DOI: 10.1016/S1474-4422(09)70272-8

\* If you downloaded your copy of the second article **BEFORE Jan 18th 2010**, please download it again! The new version contains an important correction.

### Downloads

[DMD - Interim Recommendations - Czech](#)

[DMD - Interim Recommendations - Dutch](#)

[DMD - Interim Recommendations - English](#)

[DMD - Interim Recommendations - French](#)

[DMD - Interim Recommendations - German](#)

[DMD - Interim Recommendations - Italian](#)

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[DMD - Interim Recommendations - Polish](#)

# THE DIAGNOSIS AND MANAGEMENT OF DUCHENNE MUSCULAR DYSTROPHY

A guide for families



“This is a guide to the ‘medical’ aspects of DMD, but always bear in mind that the medical side isn’t everything. The idea is that by minimising medical problems, your son can get on with his life and you can get on with being a family. It’s good to remember that most Duchenne boys are happy kids and most families do very well after the initial shock of the diagnosis.”

Elizabeth Vroom,  
United Parent Projects Muscular Dystrophy

