REHABILITATION APPROACH TO
PATIENTS WITH MUSCULAR DYSTROPHY

Klemen GRABLJEVEC
prof. Anton ZUPAN

University Rehabilitation Institute
Ljubljana, Slovenia

„Haim Ring“ PRM School, Siracusa (I), Oct 24th 2011
- University (National) Institute for Rehabilitation
- Independent setting, not attached to acute hospital
- Government – non private & non profit hospital
- International Research and Educational hub
- 200 beds, 1,550 in-patient cases per year
- Adult and pediatric population
prof. Anton Zupan, MD, PhD
Specialist in pediatrics and PRM
Serving actively as leader of the MD rehab programe, Licensed driving assesment service
Mentor and tutor to trainees and PhD student, active professor at the PRM chair at UL
Author of 51 scientific articles and 2 books
„Normal citizen“, husband, independent driver, assistive technology freak & F1 fan…..
Drawings from the Tomb of Beni Hasan
Egypt 2,800 – 2,500 BC

Relief painting from the 18th dynasty
Egypt cca 1,500 BC, showing
Nubian queen

(or was she simply obese?)
Transfiguration – Raphael 1520
Pseudohypertrophic MD?
(as proclaimed by Duchenne himself !)
Epileptic seizures?
Gaetano Conte (1798-1858), a physician of the Santa Maria del Popolo degli Incurabili Hospital in Naples, described the first case of muscular dystrophy - that he called "scrofola muscolorum" - in two brothers in an article published in the journal "Annali degli Ospedali Incurabili".


- First clinical description of DMD – but mostly ignored!
- Description of two brothers, who both manifested calf and deltoid hypertrophy
- "...the hypertrophied muscles had indeed lost that palpability tipically of their fibers, yet they seemed to be invaded by a hard earth like substance, heterogenous to their structure..."
- The older boy "...departed his life with the signs of heart muscle hypertrophy..."
Guillaime B Duchenne

„De L`Electrisation Localisee son application a la pathologie et a la therapeutique“ (1862)

„An album of clinical photography“

„Inventor of needle biopsy“
“This disease is one of the most interesting, and at the same time most sad, of all those with which we have to deal; interesting on account of its peculiar features and mysterious nature; sad on account of our powerlessness to influence its course, except in very slight degree, and on account of the conditions in which it occurs.

It is a disease of early life and of early growth. Manifesting itself commonly at the transition from infancy to childhood, it develops with the child’s development, grows with his growth – so that every increase in stature means an increase in weakness, and each year takes him a step further on the road to a helpless infirmity, and in most cases to an early and inevitable death.”

Gowers, 1879.
GENERAL FACTS ABOUT MD

- SEVERE
- CHRONIC
- DEGENERATIVE
- PROGRESSIVE
- INCURABLE
- EARLY AFFECTING QoL
- INDEPENDENCE
- LIMITATING
- DEMANDING
- SOMEHOW NIHILISTIC
- APPROACH
- YOUNG POPULATION
- MOSTLY COOPERATIVE
- WILLING TO LIVE
- ACTIVE LIFE
- TO MINIMIZE
- SECONDARY COMP`S
- TO CURE ! (?)
Fig. 1.1 Number of articles on muscular dystrophy published each year. Note the increase in the mid 1970s following evidence of a membrane defect in Duchenne dystrophy, and again some 10 years later when the gene was identified.
<table>
<thead>
<tr>
<th>Disease</th>
<th>Protein Missing/Defined</th>
<th>Age of Onset</th>
<th>Muscles Affected</th>
<th>Complications</th>
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<tbody>
<tr>
<td>Duchenne (DMD)</td>
<td>Dystrophin</td>
<td>Early childhood</td>
<td>Muscles of the hips, legs, shoulders, and spine and the heart</td>
<td>Severe muscle weakness and wasting, scoliosis, contractures, respiratory failure, pneumonia, and dilated cardiomyopathy. Death early 20s.</td>
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<tr>
<td>Becker (BMD)</td>
<td>Dystrophin</td>
<td>Adolescence or adulthood</td>
<td>Similar to DMD</td>
<td>Muscle weakness as in DMD, but slower progress and much less severe. Cardiomyopathy.</td>
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<tr>
<td>Limb-girdle (LGMD)</td>
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<tr>
<td>LGMD1A</td>
<td>Myotilin</td>
<td>Adolescence to early adulthood</td>
<td>Proximal shoulder/pelvic girdle musculature</td>
<td>Walking may not be possible within 20 years of onset</td>
</tr>
<tr>
<td>LGMD1B</td>
<td>Lamin</td>
<td></td>
<td></td>
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<tr>
<td>LGMD1C</td>
<td>Caveolin-3</td>
<td></td>
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</tr>
<tr>
<td>LGMD1D</td>
<td>Not identified</td>
<td></td>
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<tr>
<td>LGMD1E</td>
<td>Not identified</td>
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<tr>
<td>LGMD1F</td>
<td>Not identified</td>
<td></td>
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</tr>
<tr>
<td>LGMD2A</td>
<td>Calpain-3</td>
<td>Infancy to early adulthood</td>
<td>Proximal shoulder/pelvic girdle musculature</td>
<td>Type 2 LGMD is much more severe than type 1 LGMD and some result in a DMD-like phenotype. Cardiac complications, sometimes occurring in later stages.</td>
</tr>
<tr>
<td>LGMD2B</td>
<td>Dysferlin</td>
<td></td>
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<tr>
<td>LGMD2C</td>
<td>γ-sarcoglycan</td>
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<tr>
<td>LGMD2D</td>
<td>α-sarcoglycan</td>
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<tr>
<td>LGMD2E</td>
<td>β-sarcoglycan</td>
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<tr>
<td>LGMD2F</td>
<td>δ-sarcoglycan</td>
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<tr>
<td>LGMD2G</td>
<td>TCAP</td>
<td></td>
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<tr>
<td>LGMD2H</td>
<td>TRIM32</td>
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<tr>
<td>LGMD2I</td>
<td>FKRP</td>
<td></td>
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<td>LGMD2J</td>
<td>Titin</td>
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<tr>
<td>Other types</td>
<td></td>
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<tr>
<td>Emery-Dreifuss (EDMD)</td>
<td>Emerin, lamin</td>
<td>Childhood to early teens</td>
<td>Proximal upper extremity and distal lower extremities</td>
<td>Early contractures, cardiomyopathy</td>
</tr>
<tr>
<td>Epidermolysis bullosa</td>
<td>Plectin</td>
<td>Childhood</td>
<td>Generalized</td>
<td>Skin blistering with range of severity, joint contractures, dysphagia</td>
</tr>
<tr>
<td>Oculopharyngeal (OPMD)</td>
<td>Poly-A-binding protein 2</td>
<td>Age 40–60 y</td>
<td>Eyelids, throat</td>
<td>Ptosis of eyelids, dysphagia, aspiration pneumonia</td>
</tr>
<tr>
<td>Facioscapulohumeral (FSHD)</td>
<td>Not identified</td>
<td>Childhood to early adolescence</td>
<td>Face, shoulders, proximal upper extremities</td>
<td>Cardiac conduction defects, mild hearing loss, retinal abnormalities</td>
</tr>
<tr>
<td>Myotonic (DM)</td>
<td>Myotitin protein kinase, ZNF9</td>
<td>Infancy (more severe) to adulthood</td>
<td>Distal extremity muscles first, then proximal as well</td>
<td>Myotonia, cataracts, hypogonadism, cardiac arrhythmias. Adult form is mild compared to early onset.</td>
</tr>
</tbody>
</table>
MUSCULAR DYSTROPHIES

- Belong to group of *Hereditary myopathies*
together with:
- congenital myopathies,
- mitochondrial myopathies,
- metabolic myopathies,
- channelopathies
MUSCULAR DYSTROPHIES

• Group of progressive hereditary muscle disorders in which there is destruction of muscle and replacement by connective and adipose tissue.

• No associated structural abnormality in the CNS or peripheral nerves are present.
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<tr>
<th>Disease</th>
<th>Gene locus</th>
<th>Gene product</th>
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<td>X-linked recessive dystrophies</td>
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<tr>
<td>DMD/BMD</td>
<td>Xp21</td>
<td>Dystrophin</td>
</tr>
<tr>
<td>EDMD</td>
<td>Xq28</td>
<td>Emerin</td>
</tr>
<tr>
<td>Autosomal dominant</td>
<td></td>
<td></td>
</tr>
<tr>
<td><em>Limb girdle (type 1)</em></td>
<td></td>
<td></td>
</tr>
<tr>
<td>LGMD 1A</td>
<td>5q22-34</td>
<td>Myotilin</td>
</tr>
<tr>
<td>LGMD 1B</td>
<td>1q11-21</td>
<td>Lamin A/C</td>
</tr>
<tr>
<td>LGMD 1C</td>
<td>3q25</td>
<td>Caveolin-3</td>
</tr>
<tr>
<td>LGMD 1D</td>
<td>6q22</td>
<td>?</td>
</tr>
<tr>
<td>LGMD 1E</td>
<td>7q</td>
<td>?</td>
</tr>
<tr>
<td>FSHD</td>
<td>4q35</td>
<td>Myotonic PK</td>
</tr>
<tr>
<td>Myotonic dystrophy type 2/PROMM</td>
<td>19q13.2</td>
<td>ZnF9</td>
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<tr>
<td>OPMD</td>
<td>3q</td>
<td>PABP2</td>
</tr>
<tr>
<td>Autosomal recessive</td>
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<tr>
<td><em>LGMD (type 2)</em></td>
<td></td>
<td></td>
</tr>
<tr>
<td>LGMD 2A</td>
<td>15q15</td>
<td>Calpain-3</td>
</tr>
<tr>
<td>LGMD 2B/Miyoshi</td>
<td>2q13</td>
<td>Dysferlin</td>
</tr>
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<td>LGMD 2C</td>
<td>13q12</td>
<td>γ-Sarcoglycan</td>
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<tr>
<td>LGMD 2D</td>
<td>17q12-q21</td>
<td>α-Sarcoglycan</td>
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<tr>
<td>LGMD 2E</td>
<td>4q12</td>
<td>β-Sarcoglycan</td>
</tr>
<tr>
<td>LGMD 2F</td>
<td>5q33-q34</td>
<td>δ-Sarcoglycan</td>
</tr>
<tr>
<td>LGMD 2G</td>
<td>17q11-q12</td>
<td>Telethonin</td>
</tr>
<tr>
<td>LGMD 2H</td>
<td>9q3-q34</td>
<td>(TRIM/32) E3-ubiquitin ligase</td>
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<tr>
<td>LGMD 2I</td>
<td>19q13.3</td>
<td>Fukutin/related protein</td>
</tr>
<tr>
<td>LGMD 2J</td>
<td>2q31–33</td>
<td>Titin</td>
</tr>
<tr>
<td>Congenital muscular dystrophy</td>
<td></td>
<td></td>
</tr>
<tr>
<td>CMD 1</td>
<td>6q21-22</td>
<td>Merosin</td>
</tr>
<tr>
<td>CMD 2</td>
<td>12q13</td>
<td>α-7 integrin</td>
</tr>
<tr>
<td>CMD 3</td>
<td>19q13.3</td>
<td>Fukutin/related protein</td>
</tr>
<tr>
<td>CMD/Rigid Spine Syndrome</td>
<td>1p35</td>
<td>Selenoprotein N1</td>
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<tr>
<td>Fukuyama</td>
<td>9q31-33</td>
<td>Fukutin</td>
</tr>
<tr>
<td>Walker-Warburg</td>
<td>9q31-33</td>
<td>?</td>
</tr>
<tr>
<td>Muscle-eye-brain disease</td>
<td>1p32-34</td>
<td>POMGnT1</td>
</tr>
</tbody>
</table>

Inheritance patterns of muscular dystrophies

<table>
<thead>
<tr>
<th>Muscular dystrophy</th>
<th>Inheritance pattern</th>
</tr>
</thead>
<tbody>
<tr>
<td>Congenital</td>
<td>AR</td>
</tr>
<tr>
<td>Duchenne</td>
<td>XR</td>
</tr>
<tr>
<td>Becker</td>
<td>XR</td>
</tr>
<tr>
<td>Emery-Dreifuss</td>
<td>XR, AD, AR</td>
</tr>
<tr>
<td>Facioscapulohumeral</td>
<td>AD</td>
</tr>
<tr>
<td>Oculopharyngeal</td>
<td>AD</td>
</tr>
<tr>
<td>Limb-girdle</td>
<td>AD, AR</td>
</tr>
</tbody>
</table>

Abbreviations: AD, autosomal dominant; AR, autosomal recessive; XR, X-linked recessive.

_Data from_ Adams MA, Chandler LS. Effects of physical therapy program on vital capacity of patients with muscular dystrophy. Phys Ther 1974;54:494–6.
Most commonly referred types of MD

- Duchenne muscular dystrophy (DMD)
- Becker muscular dystrophy (BMD)
- Emery-Dreifuss muscular dystrophy (EDMD)
- Limb Girdle dystrophy (LGMD)
- Facioscapulohumeral dystrophy (FSH MD)
- Congenital muscular dystrophy (CMD)
Duchenne muscular dystrophy

Mutation in the dystrophin gene on chromosome Xp21, leading to absent dystrophin production

Dystrophin is a subsarcolemmal protein in skeletal and cardiac muscle

It is thought to have an important role in stabilization of the muscle membrane;

It also serves as a link between the intracellular cytoskeleton and the extracellular matrix
Duchenne muscular dystrophy

The diagnosis of DMD: blood test with DNA analysis demonstrating mutations in the dystrophin gene; however, these mutations can be detected in only two thirds of patients by routine genetic testing.

If no detectable mutation found, diagnosis requires muscle biopsy and immunohistochemistry or Western blot analyses for dystrophin in muscle.

Histopathology of the biopsy: degenerating fibers with inflammatory cells and replacement of muscle by fat and connective tissue in the later stages

Immunostaining and Western blot: markedly reduced or absent dystrophin.
Duchenne muscular dystrophy

- Presents in early childhood (within first 5 years) with difficulty walking and proximal weakness:
  - Delay of walking
  - Abnormal gait
  - Frequent falling
  - Difficulty in walking steps
- The weakness is progressive, most patients become wheelchair dependent by 12 years of age.
Duchenne muscular dystrophy

Main Clinical signs:

- Calf hypertrophy
- Positive Gower’s sign (on attempting to arise from the floor, the patient climbs up on his or her thighs using the hands).
- Waddling gait, lordotic posture
- Inability to hop
- Proximal muscle weakness (LL>UL)
Mild motor disability 23 months
Postural lordosis and moderate calf hypertrophy 9 yrs
Classical Gowers` sign. Ability to get up from the floor is mainly lost by age 10
Duchenne muscular dystrophy

Associated features:

- Cardiomyopathy
- Variable intellectual retardation
- Equinovarus
- Scoliosis after loss of ambulation
- Fixed flexion contractures after loss of ambulation.
Duchenne muscular dystrophy

Course and prognosis:

Progressive lost of function
Progressive lost of ambulation, usually at 8-12 yrs.
Cardiac muscle involvement may manifest as arrhythmia or congestive heart failure late in the course of the illness
Most patients die of respiratory complications by their early 20s.
creatinine kinase levels : 20–100 X N
EMG: myopathic
MS US: increased - hyper echogenity
Duchenne muscular dystrophy

Main specific management:

- Prevention of fixed deformities by passive stretching
- Avoid immobilisation during injury or illness
- Promotion and prolongation of ambulation with orthoses
- Prevention of scoliosis by attention to posture while W/C – bound, provision of spinal support
- Surgical treatment of progressive scoliosis
Becker muscular dystrophy

- Clinical pattern similar to DMD, but milder and slower progression
- Onset: usually after 5 yrs of age and into adolescent as well adult age
- First presenting symptoms: difficulty with running, cramps on exercise
- Investigations: CK similar to DMD
  - EMG myopathic
  - MS US: hyper echo
Becker muscular dystrophy

- **Main clinical signs:**
  - mild functional disability
  - proximal muscle weakness
  - calf hypertrophy
  - waddling gait, lordosis

- **Associated:**
  - cardiac involvement (variable ECG changes)
Becker muscular dystrophy

Course and prognosis:

- Slowly progressive, variable course
- Can be actually static course in some cases
- Ambulation beyond in adulthood
- Life expectancy dependent on degree of progression and late respiratory deficit.
Becker muscular dystrophy

Main principles of management:

• Promotion of activity
• Prevention of fixed deformities by passive stretching
• Promotion and prolongation of ambulation with orthoses
• Prevention and management of scoliosis if W/C bound
Emery-Dreifuss muscular dystrophy

- EDMD was first recognized as a distinct clinical entity by Emery and Dreifuss in 1966.
- In their seminal paper, they concluded that despite some similarities with Duchenne muscular dystrophy, such as the X-linked recessive inheritance, the disease course was milder and the distribution of weakness different from that of the more common Duchenne form.
- Emery and Dreifuss in 1966 already recognized frequent cardiac involvement and sudden death as typical features of Emery-Dreifuss muscular dystrophy.
Emery-Dreifuss muscular dystrophy

- **Onset:** late childhood, adolescence or adulthood

- **Early presenting as:**
  - difficulty running
  - rigidity of the neck and/or spine
  - Cardiac arrhythmia

- **Investigations:**
  - CK: slight to moderate elevation
  - EMG: myopathic
  - MS US: focal increase in echogenity
Emery-Dreifuss muscular dystrophy

Main clinical signs:

- Fixed deformities: equinus flexion deformity of elbows, rigidity of spine with limited neck and trunk flexion.
- Mild weakness
- Focal wasting, especially proximal in UL and distal in LL

Associated signs:
- Cardiac arrhythmia – needs 24 hrs Holter
- Nocturnal hypoventilation.
Emery-Dreifuss muscular dystrophy

- **Curse and prognosis:**
  Very slowly progressive muscle weakness and functional disability
  Cardiac involvement might be life threatening in early adult age

- **Main principles of management:**
  Promotion of ambulation
  Prevention of deformities
  Correction of fixed ankle deformities
  Close monitoring of cardiac status – might need PM
  Assessment of respiratory functions
Limb girdle muscular distrophy

- Variable severity, resembling DMD and BMD
- **Onset:** very wide period – from childhood to adolescence and adulthood
- **Presenting symptoms:**
  - Difficulty with gait, running, climbing steps
  - Cramps on exercise
- **Investigations:**
  - CK: variable levels, from mild to high
  - EMG: myopathic
  - MS US: variable increase of echogenity
Limb girdle muscular dystrophy

Main clinical signs:
- Abnormal gait, lordotic posture
- Problems with hopping and rising from floor
- Variable muscle weakness
- Joint deformities after loss of ambulation (as in DMD)
- Calf prominence – not really hypertrophy

Associated signs: none
Limb girdle muscular dystrophy

• Course / prognosis:
  Very variable
  Usually slow progressive, but in some cases as severe and more rapid than DMD!

• Main principles of management:
  Promotion of ambulantion
  Prevention and treatment of deformities
Facio-scapulo-humeral dystrophy

- Dystrophy affecting primarily facial and shoulder girdle muscles
- **Onset**: variable – from early childhood to adult
- **Presenting symptoms**: Disability relating to shoulder and facial muscles
  In some cases trunk and pelvic girdle weakness and difficulty with locomotion.
- **Investigations**:
  CK: normal or mildly elevated
  EMG: normal or myopathic
  MS US: variable
Facio-scapulo-humeral dystrophy

Main clinical signs:
- Facial weakness
- Shoulder girdle weakness
- „Terracing of shoulder on abduction"
- Lordosis and pelvic weakness in some families.

Associated clinical signs:
- Deafness (variable)
- Fundal changes (variable)
Facio-scapulo-humeral dystrophy

• **Course / prognosis:**
  Very variable. Might be mild and very slowly progressive, with normal lifespan.
  Might happen marked progression of LL weakness with lose of ambulation in adult age
  Variable degree of facial muscles weakness
  Variable respiratory deficits in later stages.

• **Main principles of management:**
  Promotion of activity
  In some cases benefit from op. fixation of the scapulato facilitate abduction in shoulder
Congenital muscular dystrophy

- Heterogenic group presenting with clinical weakness or deformities in early infancy
- Variable dystrophic changes in muscles
- **Onset:** At birth or in infancy / early childhood

- **Presenting symptoms:**
  - Hypotony and weakness
  - Fixed deformities – arthrogryposis
  - Sucking, swallowing and respiratory problems
  - Delayed motor milestones in later onset cases
Congenital muscular dystrophy

- **Investigations:**
  CK: normal or mildly elevated
  EMG: myopathic
  MS US: marked increase – hyper echogenity

- **Main clinical signs:**
  Generalised hypotony and weakness
  Fixed deformities related to intrauterine posture
  Variable weakness and contractures in later presenting cases
Congenital muscular dystrophy

Associated signs:

- Intellectual retardation (Fukuyama)
- Dislocation of hips
- Secondary deformities, scoliosis
- Hydrocephalus and fundal changes (Santavuori)

Course / prognosis:

- Variable, many cases relatively static
- Might show functional improvement with time

" Might be fatal due to respiratory deficit and infection
Congenital muscle dystrophy

Main principles of management:
- Active PT to encourage mobility
- Passive stretching for soft tissue contractures
- Avoid immobilisation, which promotes deformities
- Supportive treatment for respiratory problems
- Surgical correction of residual deformities at right stage – eg. Equinovarus correction when able to stand.
REHABILITATION APPROACH TO PATIENTS WITH MUSCULAR DYSTROPHY
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<th>CRITICAL PERIODS</th>
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<td>WHEN CHILD CAN'T START WALKING</td>
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<td>PARENTS/FAMILY</td>
<td>THE LOSS OF WALKING ABILITY</td>
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<td>CHILDHOOD</td>
<td>RESPIRATORY FAILURE AND VENTILATOR ACCEPTANCE</td>
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<td>PUBERTY</td>
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<td>PHYSICAL DISABILITY</td>
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<td>INTELECTUAL CAPABILITY</td>
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<td>ENVIRONMENT</td>
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</table>
GENERAL RECOMMENDATIONS FOR REHABILITATION OF MD PATIENTS

Maloney FP (ed). Interdisciplinary Rehabilitation of Multiple Sclerosis and neuromuscular disorders. JB Lippincott, 1985; 239-58

1. Rehabilitation interventions should be run in specialised and MD dedicated institutions

2. Undefined/unclear diagnostic process should not be an obstacle for immediate rehab interventions and complications limitations
GENERAL RECOMMENDATIONS

3. Rehabilitation of MD patients must run in comprehensive and complex approach. Rehab team does not need to be completely consisted, although specially MD oriented.

4. Due to severity and chronic progress of diseases relatives are necessary to be involved in rehab process. Therefore, rehab medicine team is obliged to perform theoretical and practical education and counselling for them.
5. Early manifestation of MD demands appropriate educational and counselling approach, Schooling and vocational guidance, emphasised attention to intelectual abilities and cooperation with educational institutions—as a part of comprehensive rehabilitation.

6. Rehabilitation team members should actively participate in elimination of architectural barriers in local society to enable as free as possible ambulation for MD patients.
# EAMDA Recommendations

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<th>STAGE 2</th>
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<td><strong>From ambulatory to W/C</strong></td>
<td><strong>W/C bound</strong></td>
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<tr>
<td><strong>Endurance</strong></td>
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<tr>
<td>Active strengthening exercise, ROM exc.</td>
<td>Swimming</td>
<td>As in stage 2</td>
</tr>
<tr>
<td>Resistance exercises</td>
<td>Tricycle riding</td>
<td>Intensifying Respir.ther.</td>
</tr>
<tr>
<td>Hydro/swimming</td>
<td>Use of walker</td>
<td>Assistive coughing</td>
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<tr>
<td>Bicycle...</td>
<td>Standing in tilt frame</td>
<td>Mechanical ventilation support</td>
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<td><strong>Contractures</strong></td>
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<tr>
<td>Active exercises</td>
<td>As in stage 1</td>
<td>As in stage 2</td>
</tr>
<tr>
<td>Passive stretching</td>
<td>Assessment of orthotic needs</td>
<td>Testing and prescribing</td>
</tr>
<tr>
<td>Positioning, Splinting, lying prone position</td>
<td>Spinal orthotics</td>
<td>W/C</td>
</tr>
<tr>
<td></td>
<td></td>
<td>Special seating technology</td>
</tr>
<tr>
<td><strong>Education Counselling</strong></td>
<td></td>
<td></td>
</tr>
<tr>
<td>Which position to avoid, programme compliance control,</td>
<td>As in stage 1</td>
<td>As in stage 2</td>
</tr>
<tr>
<td>Dietary consultation</td>
<td>Home adaptation</td>
<td>Nursing care advisory</td>
</tr>
<tr>
<td>Ergonomy consultation</td>
<td>School counselling</td>
<td>Position changing advisory</td>
</tr>
<tr>
<td>Healthy living promotion</td>
<td>Education about importance changing position</td>
<td>Respiratory techniques advisory</td>
</tr>
</tbody>
</table>
CATEGORIES OF REHABILITATION INTERVENTION IN MD

1. Deconditioning
2. Maintenance of muscle strength and endurance
3. Maintenance of the range of motion
4. Maintenance of the aerobic capacity
5. Scoliosis
DECONDITIONING

• Occurs when a person drops from a certain level of physical activity to a lower level, causing the body to adapt to the lesser demands.

• Near-complete inactivity (most commonly bedrest) has the most dramatic effects.

• Short periods of forced bedrest can have devastating effects, dramatically affecting function and possibly prognosis.

• DMD child who is confined to only a few days of bedrest for an acute illness can prematurely lose the ability to walk and become wheelchair-dependent
DECONDITIONING

**Immobility** by definition leads to the start of multiple musculoskeletal changes:
- multiple contractures,
- scoliosis,
- increased pulmonary restrictive disease
- cause pain and markedly change the nature of the disease as well functional state.

If **hospitalization** or surgery is necessary, MD patients should become mobilized as early as possible.
<table>
<thead>
<tr>
<th>Site</th>
<th>Prevalence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Low back</td>
<td>70%</td>
</tr>
<tr>
<td>Knees</td>
<td>53%</td>
</tr>
<tr>
<td>Ankles</td>
<td>50%</td>
</tr>
<tr>
<td>Toes</td>
<td>46%</td>
</tr>
<tr>
<td>Feet</td>
<td>44%</td>
</tr>
</tbody>
</table>

MAINTANCE OF MUSCLE STRENGTH AND ENDURANCE

• Most common complaints with neuromuscular diseases are weakness and fatigue.

• Weakness is the inability to generate sufficient contractile force or tension in a muscle (ie, a lack of strength or force),

• Fatigue is the inability to maintain tension or a decrease in force-generating capacity with repeated contraction.
WEAKNESS AND FATIGUE

• Weakness and fatigue can be caused by a deficiency in any component in the motor system (muscle, neuromuscular junction, lower motor neuron, upper motor neuron, other cortical structures).

• Weakness and fatigue are closely related.

• Maintaining or improving strength and endurance is a main goal in managing neuromuscular disorders.
WEAKNESS AND FATIGUE

- Maintaining strength and endurance requires the routine contraction of the muscle against resistance.
- Overuse injury in MD patients!
- Muscle damage and rhabdomyolysis even in healthy subjects (Ecc contraction)
- MD muscles are less adaptable to overuse.
- Muscle strength in „dominant“ UL & LL is lower than in non-dominant (Johnson 1971, Belanger 1991)!
- Submaximal exercise can improve strength and limit the progress of disease in early stages of DMD (DeLateur 1979)
WEAKNESS AND FATIGUE

• 4-months resistance exercise program improved strength in less affected antigrav muscle groups for 50%.
• There was further improvement up to 1-year (Vignos & Watkins, 1966)
• Muscle strength and endurance improved after resistance exercise program only in muscle groups with starting strength at least 20% of expected value (Milner-Brown & Miller, 1988).
WEAKNESS AND FATIGUE

• Although many other studies have shown strength gains in various neuromuscular diseases, they have been short-term and do not necessarily show the impact on final outcome


• As well numerous studies report exercise-induced injury in neuromuscular diseases!

WEAKNESS AND FATIGUE

SWIMMING / HYDROTHERAPY

• Possible in any stage of MD disease
• Allow to maximally use even the minimal strength of the muscles in limbs through, therefore to improve CVS
• Enhanced respiratory activity
• Increased load on and work of inspiratory muscles in vertical position
• Thermal effect on peripheral circulation and muscle tone

Zupan A. 1995
Contractures are common complications of neuromuscular diseases.

Efforts to improve function - using a wheelchair for all mobility, may lead to contractures in hip and knee.

In myopathies and dystrophies, fibrosis of the muscle will promote contracture development.

The simplest way to prevent contractures is to avoid joint immobility through maintaining normal levels of activity that regularly put joints through their full range of motion.
 Contractures are important factor for ambulance limitation in MD patients.

 Specific gait type and posture are further provocative factor for contracture development

 Most often in knees, hips and ankles.

 Most benefit treatment is combination of active movement, passive stretching and orthotic application during night.

 Scott OM. 1981; Brooke MH. 1989; Vignos PJ 1983
Activities that promote stretching (yoga, tai chi), solely or together with regular physical or occupational therapy are useful ways to maintain range of motion.

The amount of stretching necessary to maintain range of motion is unclear;

Two studies suggested from 10 minutes of stretching per day to 6 hours of positioning per day as necessary to maintain range of motion.

Vignos PJ, 1983; Siegel IM, 1978;
MAINTANCE OF RANGE OF MOTION

• Application of heat packs is therapeutic approach for lengthening of contracted muscle, as well for analgesic effect.

• Adding peripheral vasodilatators to heat therapy is of no sensitive use. There is no reduce of peripheral circulation in MD.

• Tissue warming will increase oxygen consumption and vessel permeability – resulting in muscle hypooxigenation and oedema!

*Fowler & Goodgold, 1988*
MAINTANCE OF AEROBIC CAPACITY RESPIRATORY CARE

• Aerobic capacity is a measure of the body’s ability to perform oxidative metabolism.
• Pneumonia and respiratory failure represent the leading cause of death and a frequent cause of morbidity for patients with DMD. *Brooke MH, Fenichel GM, Griggs RC, et al., 1989*
• Respiratory muscle weakness is found in all individuals with DMD, although it may develop at varying rates. *McDonald CM, Abresch RT, Carter GT, et al, 1995*
MAINTANCE OF AEROBIC CAPACITY
RESPIRATORY CARE

- **Caution**: Aerobic training in neuromuscular diseases poses risks.
- No studies show the long-term benefits, and the possibility exists of worsening of the disease process.
- Cardiomyopathies and conduction abnormalities could predispose to morbidity with exercise training.
- Each patient must also be individually evaluated.
- Impairments in metabolism seen with glycogenoses can make exercise painful and dangerous.
MAINTANCE OF AEROBIC CAPACITY
RESPIRATORY CARE

Zupan test of functional status in patients with MD©

Section C1: Respiration assessment

1. % FVC
2. % FEV1
3. Cough: functional (2 expulsion per expiry)
   weak functional (at least 1 expulsion per expiry)
4. Fr of respiration: less then 16 per min / less then 25 per min
5. Use of accessory respiratory muscles
6. Presence of paradoxal breathing when lying supine
Zupan test of functional status in patients with MD

Section C1: Respiration assessment (cont`d)

7. FVC ratio in supine / seated position (should be over 0.75)
8. Presence of glosopharingeal breathing
9. Morning headaches, day-sleepyness, memory and concentration problems
10. Palpitations, over tiredness, nightmares, night sweating
11. Dyspnoe at moderate exercise and at rest
12. Cianosis at moderate exercise and at rest

MAINTANCE OF AEROBIC CAPACITY

RESPIRATORY CARE
Noninvasive ventilation

• Without tracheostomy with small, portable ventilators and inexpensive patient-ventilator interfaces (masks and mouthpieces).
• Parallel to the development of devices for sleep apnea in the general population.
• Noninvasive support is now available for nocturnal and daytime use.
• Hypoventilation occurs initially at night, and almost all individuals with DMD begin using noninvasive ventilation while sleeping.
• This ventilatory support consists in almost all cases of positive-pressure ventilation applied with a nasal or oronasal mask and a pressure support or a volume cycled ventilator.
A) Diagram of setup for mouthpiece ventilation showing ventilator, connecting tubing and mouthpiece.
(B) HT-50 volume cycled ventilator with mouthpiece attachment for mouthpiece ventilation.
Invasive (tracheostomy) ventilation

- Tracheostomy with full-time ventilator support.
- Placement of the tracheostomy involves a surgical procedure and a period of hospitalization.
- Risks: infection, bleeding, and impaired speech and swallowing and tracheostomy tube occlusion with mucus.

- The ATS Consensus Statement (2004): tracheostomy should be offered to individuals when close expertise in mouthpiece ventilation is unavailable; when contraindications to NPPV exist, such as severe bulbar muscle weakness and inability to control secretions; or when the patient himself prefers tracheostomy.
Cough support and secretion management

• A crucial and often overlooked aspect of respiratory support
• Effective airway clearance is crucial to prevention of pneumonia, which can lead to respiratory failure and death.
• Early intervention seems to reduce the incidence of pneumonia and hospitalization.
• Cough flow correlates well with the ability to clear secretions.
• Many techniques are available to increase cough flow rates in individuals who have impaired cough function, including manual and mechanically assisted techniques.
Manual techniques involve increasing the breath volume by having the patient use of glossopharyngeal breathing or a self-inflating resuscitator bag to take in one or more breaths that are larger than they would be able to take in spontaneously maximally insufflation capacity (MIC).

This breath stacking or MIC maneuver increases the inspiratory capacity, the volume of the chest, and the elastic recoil pressure of the chest wall and lung, which allows a more forceful cough to be performed.
Mechanical assistance for cough

• Mechanic insufflation exsufflation, a technique developed in the 1950s for polio patients.
• It mimics cough function by providing positive pressure through a mask or mouthpiece to inflate the respiratory system followed by a rapid switch to a negative pressure that clears the airway of secretions.
• Repeated applications are often needed to clear the airway adequately.
Mechanical insufflation – exsufflation technique
Can progression of scoliosis be limited, if we disrupt the continuous state of asymmetric loading, before significant bony prominence on spine occurs?


Some studies suggest that exercise-based approaches in addition to bracing may be effective in some girls who have adolescent idiopathic scoliosis


SCOLIOSIS - NONOPERATIVE TREATMENT

• The effectiveness of nonoperative treatment in children who have neuromuscular scoliosis is controversial.

• 28% success rate (defined as curve progression of less than 10° per year and good brace compliance) in 90 consecutive children who had various types of neuromuscular scoliosis.

• Better chance of improvement in ambulators with hypotonia and short lumbar curves of less than 40° and in nonambulators with spasticity and short lumbar curves. Children with longer and hypotonic curves gain worse results.
SCOLIOSIS - NONOPERATIVE TREATMENT

No benefit after 67 months of bracing in 20 children who had spastic quadriplegia observing curve magnitude, shape, or rate of progression.

Whether spinal orthoses and other conservative management techniques may be helpful in slowing the progression of scoliosis in certain subpopulations of children who have neuromuscular disease remains to be seen, but the prevailing attitude suggests that they are not. Driscoll CW, Skinner J. Musculoskeletal complications of neuromuscular disease in children. Phys Med Rehabil Clin N Am; 19 (2008) 163–194.

In boys with DMD, significant progression of scoliosis is unusual while the child remains ambulant. Rapid progression of scoliosis seems to be related to the loss of walking ability and commonly corresponds with a growth spurt in adolescence.

The primary indication for bracing is to improve postural control and seating rather than prevent progression of curvature.

ELECTROSTIMULATION OF THE MUSCLES


- In MD, mainly fast (glycolitic) muscle fibers are affected. Buchthal et al, 1974

- Can we expect, that LFES in MD increase the resistance of muscle fibers through type transformation against degenerational changes?
ELECTROSTIMULATION OF THE MUSCLES

- LFES of right m.TA 2 x 1 hour daily for 6 months
- Left m. TA served as a control group
- MVC expressed as torque in both ankles after every month of therapy.
- Improved MVC in ankle compared to control group.
- No signs of overuse disease or increased fatiguability.
- **Conclusion:** LFES can slow down the degeneration process in m. TA in MD subjects with transforming fast fibers to slow fibers.

Zupan A. Muscle Nerve 1992

Zupan A, Gregorič M, Valenčič V, Vandot S. Neuropediatrics 1993
AVERAGE TORQUES IN THE ANKLE AT MVC

Nm

1st 2nd 3rd 4th 5th 6th 7th

No.of measures

non stimulated left extremity
stimulated right extremity
COENZYME Q10

Believed to be protector from free radicals to the mitochondria
Believed to provide energy stores to the cell in the form of ATP

Double blind study of CoQ10 treatment
50 patients with different forms of MD and SMA
CoQ10 (400 mg a day) for 4 months and placebo for 4 months

No statistically significant changes in biomechanical parameters
Statistically significant fall of cell CO2 and serum glucose
Subjective improvement of physical well-being
Correctly guessed the period during which they received CoQ10 or placebo

TAKE HOME MESSAGE

Advances in genetics and molecular biology have increased awareness of the diversity of mechanisms in neuromuscular diseases.

This dramatic increase in knowledge will eventually lead to treatments that may ameliorate the pathologic cause of these disorders.

Unfortunately, these treatments are still only in the experimental stages.

Standard rehabilitation treatments remain the mainstay for treating these disorders and improving quality of life for patients.

P&R Medicine only see:

TO PREVENT MD COMPLICATIONS = TO CURE
**RECOMMENDED REVIEW LITERATURE:**

*Hornyak JE, PH Pangilinan.* Rehabilitation of Children and Adults Who Have Neuromuscular Diseases.

*Benditt JO, Boitano L.* Respiratory Support of Individuals with Duchenne Muscular Dystrophy: Toward a Standard of Care

*Srinivasan J, Amato AA.* Myopathies.


*Driscoll SW, Skinner J.* Musculoskeletal Complications of Neuromuscular Disease in Children.

Thank YouZ for all your attention!

Q&A: klemen.grabljevec@ir-rs.si