General principles of management in DMD

Katie Bushby
Newcastle upon Tyne
UK
DMD is a predictable condition

• Lack of dystrophin has effects which reflect where it is important, though with individual variability
  - Skeletal muscle
  - Cardiac muscle
  - Brain

• Important at diagnosis and beyond to have a road map of who is going to help with treatment of these different areas
  - And preparation for adulthood

• And who will play the co-ordinating role
“What can we do?”

- Exactly what you would have done yesterday
- Family life
- Lots of exciting activities
- Lots of love and encouragement

- Working with the medical team to make sure DMD has as little impact as possible
  - Prediction, prevention, active treatment
“What can we do?”

- You are the people who are always there
- Be your own expert
  - In contacts with other professionals
    - With fracture
    - Anaesthetics
    - Steroid treatment
    - Schools, social services etc
Muscle clinic doctor

Physiotherapist

Geneticist

Wheelchair services

Occupational therapist

Orthopaedics

Orthotist

Psychologist

Cardiologist

Speech therapist

Respiratory team

School

Family
Genetics

• A chance to talk and understand
  - Why this happened
  - How it happened
  - The chances of it happening again
  - The risk to other family members of being carriers
  - Point of contact for the future
Skeletal muscle- priorities in the early stages

- Maintain and prolong good independent ambulation
  - Physiotherapy (specialised, local)
  - Steroids
- Preventing and managing contractures
- Maintaining/ improving muscle strength
- To be able to enjoy home and school life with minimum disruption
Learning, behaviour support

- Brain involvement in DMD is poorly understood
  - Speech therapy
  - Psychology input
  - School provision
Approaching loss of ambulation

- Discussion about use of KAFOs (long leg callipers)
- Major role of physiotherapy and orthotics teams
- Possibly orthopaedic input
When walking is difficult and no longer possible

- Appropriate wheelchair provision
- Timely
- To promote good posture, comfort and function
- Physiotherapist
- Occupational therapist
- Wheelchair services
Maintenance of function, activity and independence
Skeletal muscle is not only for walking

- Spinal support
- Development of scoliosis may require spinal surgery
- Early involvement of experienced spinal surgeon

Analysis of the impact of spinal surgery and ventilation in patients born since 1970

- Spinal surgery + ventilation
- Spinal surgery no ventilation
- Ventilated no spinal surgery
- No ventilation no spinal surgery
Respiratory muscle strength

- In early years forced vital capacity rises with growth
- Tends to fall after loss of ambulation
- Predictable and treatable course
  - Pathway of respiratory care
  - Muscle clinic, respiratory team
Cardiac involvement in DMD is almost invariable, but rarely symptomatic until late stages.

Short PR: Q waves: Tall R in V1-2: Twaves abnormal

Reduced ejection fraction and wall motion abnormalities
Cardiac involvement in DMD / BMD

What is the real goal of intervention?

Onset of LVF symptoms

Normal range

LV Function

Age (years)
Cardiology- recommendations

• Guidelines available at enmc.org
• ECG and echo at diagnosis annually from age 10
  - Or before any surgery
• Treat as soon as there is any deterioration on echo
• ACE inhibitor/ beta blocker
Multidisciplinary treatment of DMD has changed the natural history

- Specialist care + 5 years
- Home nocturnal ventilation + 7 years (+)
- HNV plus spinal surgery + 9 years
- Management of cardiac failure
- Long term steroid treatment with benefit to mobility, heart and lung function
Future treatments are also likely to be additive

- Other pharmacological treatments
- Gene therapy
- Upregulation of utrophin
- Antisense oligonucleotide therapies
- Stem cell based treatments
Teamwork.....