

Standards of care for DMD



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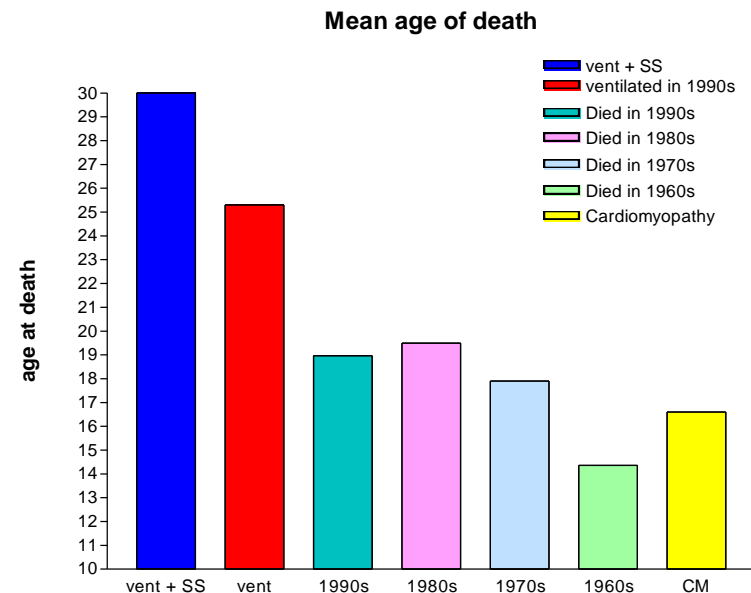
www.treat-nmd.eu

On behalf of the CDC care considerations group



Appropriate management improves survival in DMD

- DMD is a treatable disease
 - Predictable complications in different systems
- Steroids prolong ambulation and delay the onset of other complications
 - In conjunction with physiotherapy regimes
- Respiratory support is proven to improve life expectancy with maintenance of a good quality of life
- Cardiac surveillance and treatment is likely to have similar benefits
- Multidisciplinary management is key



Challenges of an adult DMD population

Best practice document for care for DMD

- Why?
 - Not all families have access to optimum management
 - The weight of international consensus can help to create a level playing field for patients and families
 - Provision of care for DMD should not be a lottery
 - To provide a tool to lobby health care providers
 - To raise standards across the board
 - To encourage excellence and further clinical research
 - To provide a basis for the introduction of trials and new treatments



Best practice document for care for DMD

- How?
 - Gather evidence from literature where this exists
 - Assemble expert panels
 - Ensure representation as broad as possible
 - Use an unbiased method for building consensus
 - RAND/ UCLA appropriateness method (RAM)
 - “Joined up thinking” for dissemination and implementation
 - Consistent message



Consensus generation via expert groups and RAM methodology

- Key areas of clinical management identified in 8 disciplines
- >80 experts went through a three stage process of generating recommendations
- Personnel and “toolkit” defined for optimal care
- Direction for care in specific scenarios generated from results of consensus
- Multidisciplinarity key issue, need for experience and co-ordination of care
- Management geared to the specific stage of condition

| Panel | ROUND 1 | | ROUND 2 | | ROUND 3 | |
|------------------------------|------------|--------------------|------------|--------------------|------------|--------------------|
| | Matrices | Clinical Scenarios | Matrices | Clinical Scenarios | Matrices | Clinical Scenarios |
| Diagnostics | 23 | 402 | 11 | 136 | 11 | 50 |
| Psychosocial | 61 | 26,075 | 19 | 325 | 18 | 148 |
| Neuromuscular | 30 | 817 | 26 | 948 | 24 | 192 |
| Rehabilitation Management | 126 | 36,015 | 29 | 1322 | 22 | 421 |
| Orthopedics / Surgical | 48 | 2,284 | 25 | 442 | 12 | 33 |
| Gastrointestinal / Nutrition | 44 | 3,479 | 25 | 479 | 20 | 129 |
| Respiratory | 36 | 606 | 33 | 269 | 27 | 115 |
| Cardiovascular | 21 | 624 | 13 | 194 | 12 | 64 |
| TOTAL | 389 | 70,302 | 174 | 4,115 | 149 | 1,152 |

Core implications: the multidisciplinary toolkit

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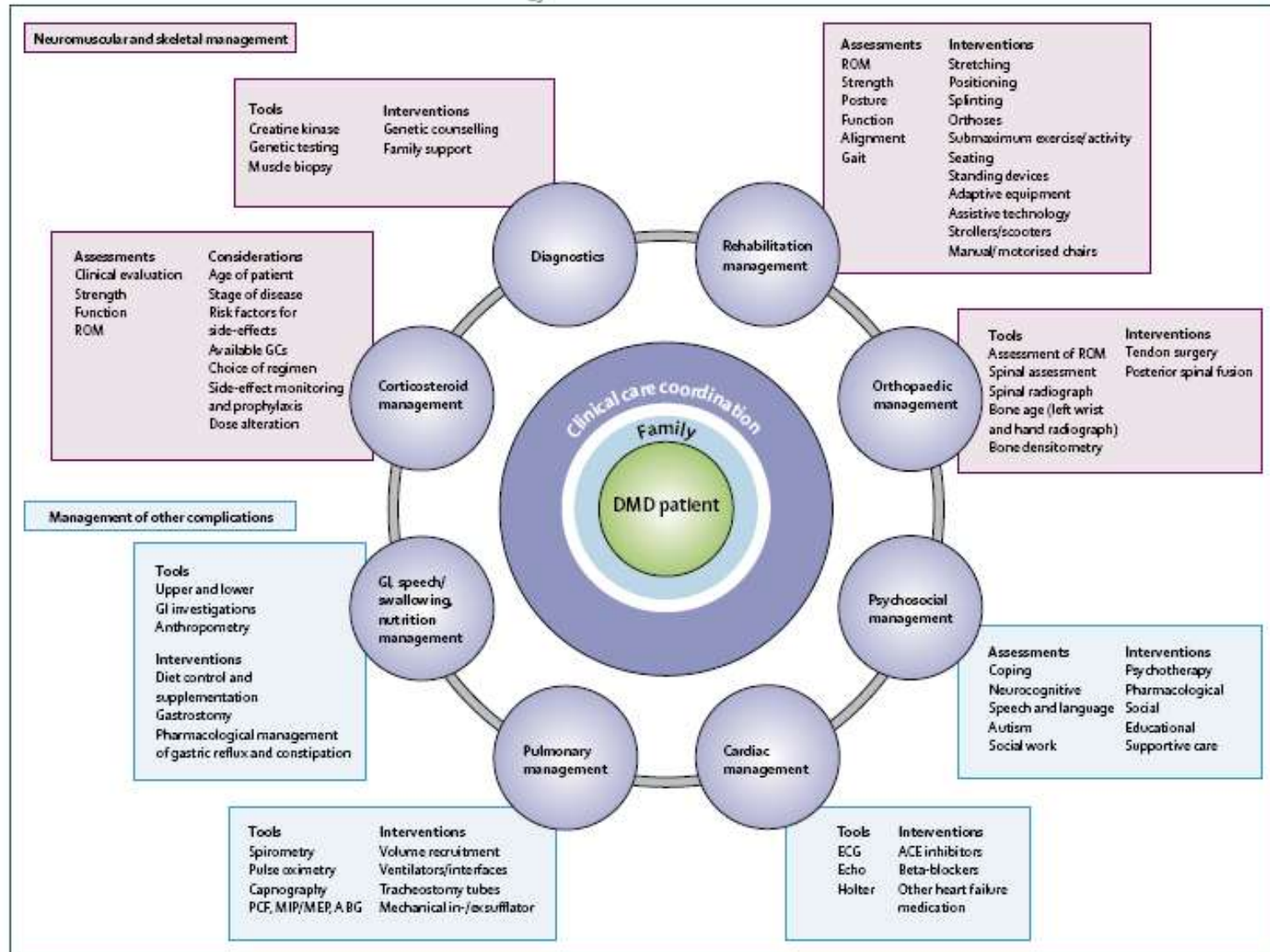


Figure 1: Interdisciplinary management of DMD

| | Stage 1: Presymptomatic | Stage 2: Early ambulatory | Stage 3: Late ambulatory | Stage 4: Early non-ambulatory | Stage 5: Late non-ambulatory |
|----------------------------------------------------|------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|--------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|----------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------------|------------------------------------------------------------------------------------------------------------------|-----------------------------------------------------------------------------|
| Diagnosics | <p>Can be diagnosed at this stage if creatine kinase found to be raised or if positive family history</p> <p>Might show developmental delay but no gait disturbance</p> | <p>Gower's sign</p> <p>Waddling gait</p> <p>Might be toe walking</p> <p>Can climb stairs</p> | <p>Increasingly laboured gait</p> <p>Losing ability to climb stairs and rise from floor</p> | <p>Might be able to self propel for some time</p> <p>Able to maintain posture</p> <p>Might develop scoliosis</p> | <p>Upper limb function and postural maintenance is increasingly limited</p> |
| Neuromuscular management | <p>Diagnostic examination and genetic counselling</p> | <p>Likely to be diagnosed by this stage unless delayed for other reasons (eg. concomitant pathology)</p> | | | |
| Orthopaedic management | <p>Anticipatory planning for future developments</p> <p>Ensure immunisation schedule is complete</p> | <p>Continue assessment to ensure course of disease is as expected in conjunction with interpretation of diagnostic testing</p> <p>At least 6-monthly assessment of function, strength, and range of movement to define phase of disease and determine need for intervention with GCs, ongoing management of GC regimen, and side-effect management</p> | | | |
| Rehabilitation management | <p>Orthopaedic surgery rarely necessary</p> | <p>Consider surgical options for TA contractures in certain situations</p> <p>Monitor for scoliosis: intervention with posterior spinal fusion in defined situations</p> <p>Possible intervention for foot position for wheelchair positioning</p> | | | |
| Pulmonary management | <p>Education and support</p> <p>Preventive measures to maintain muscle extensibility/minimise contracture</p> <p>Encouragement of appropriate exercise/activity</p> <p>Support for function and participation</p> <p>Provision of adaptive devices, as appropriate</p> | <p>Continue previous measures</p> <p>Provision of appropriate wheelchair and seating, and aids and adaptations to allow maximum independence in ADL, function, and participation</p> | | | |
| Cardiac management | <p>Normal respiratory function</p> <p>Ensure usual immunisation schedule includes 23-valent pneumococcal and influenza vaccines</p> | <p>Low risk of respiratory problems</p> <p>Monitor progress</p> | <p>Increasing risk of respiratory impairment</p> <p>Trigger respiratory assessments</p> | <p>High risk of respiratory impairment</p> <p>Trigger respiratory investigations and interventions</p> | |
| GI, speech/swallowing, nutrition management | <p>Echocardiogram at diagnosis or by age 6 years</p> | <p>Maximum 24 months between investigations until age 10 years, annually thereafter</p> | <p>Assessment same as in the younger group</p> <p>Increasing risk of cardiac problems with age; requires intervention even if asymptomatic</p> <p>Use of standard heart failure interventions with deterioration of function</p> | | |
| Psychosocial management | <p>Family support, early assessment/intervention for development, learning, and behaviour</p> | <p>Monitor for normal weight gain for age</p> <p>Nutritional assessment for over/underweight</p> <p>Attention to possible dysphagia</p> | | | |
| | | <p>Assessment/intervention for learning, behaviour, and coping</p> <p>Promote independence and social development</p> <p>Transition planning to adult services</p> | | | |

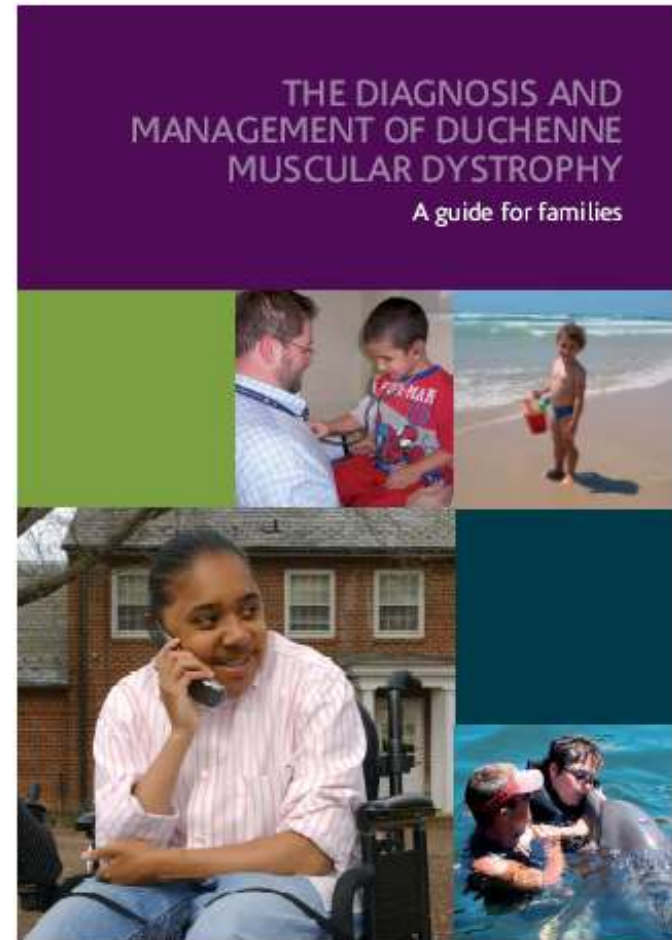
Core implications: Stage specific care

Care into the clinic



Dr J Kirschner

- Determine the current levels of care and barriers to improvement
- Introduce a programme of training and education
 - Current translation of family guide into >20 languages
- Monitor levels of improvement clinically and with patient satisfaction
- Redefine natural history to inform the development of therapies
- See CARE-NMD workshop!



What next?

- The publications reflect the content of the service definition for NMD
 - Which needs to be properly supported across the UK
 - NICE guidelines?
- We have a starting point for training, education and further research into best practice
- Family guide available and being disseminated by patient organisations
- Build on current infrastructure
- Consolidate the practice of multidisciplinary care



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