FACT SHEET

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Duchenne Muscular Dystrophy (DMD)

- Duchenne Muscular Dystrophy (DMD) is an X-linked inherited disorder with a worldwide incidence of 1 in 3,500-6,000 males.[1] The genetic defect is a deletion, duplication, or a point mutation on the XP-21 region. This defect leads to an absence or decrease of dystrophin, a cytoskeletal protein resulting in progressive weakness.
- The natural progression of children with DMD is well documented and characterized. Boys who are untreated lose the ability to walk by age 10-12 and 80% develop a scoliosis after loss of ambulation and standing, with death historically occurring in the late teens, due to respiratory (70%) or cardiac (30%) complications.[2]
- In addition to musculoskeletal complications, up to 30% of children have intellectual and behavioral problems.[2]
- Over the past 3 decades advances in care have changed the natural history of the disorder with survival now into the twenties and thirties, improved quality of life, and greater participation increasingly possible.[3]
- Stretching, splinting, knee-ankle-foot orthoses (KAFOs), and contracture release surgery can prolong ambulation and standing by 1-3 years.[4,5]
- Surgical correction and stabilization of scoliosis improves sitting comfort, upper-extremity function, and delays respiratory decline.[6]
- Use of non-invasive ventilation and assistive devices for coughing have decreased and delayed the morbidity and mortality from respiratory complications.[7]
- Proactive preventive use of ACE (angiotensin-converting enzyme) inhibitors seems to be having an impact on developments of cardiomyopathy.[3]
- Corticosteroids (Prednisone and Deflazacort) have delayed the loss of ambulation to the teens, decreased the incidence of scoliosis, delayed the need for non-invasive ventilation, and contributed to prolonged survival, with increased level of function into the twenties and thirties.[3]
- These advances have created expanded opportunities for individuals with DMD and an accompanying need for updated skills for optimal management over time. DMD is no longer a fatal pediatric disorder, but a chronic progressive disorder. Optimal care is vital in allowing full participation into young adulthood.

Early Signs and Symptoms

Early signs and symptoms as reported by families and care providers include[8]:

- Delayed walking
- Delayed speech
- Delayed motor development
- Neck flexor weakness when pulled to sit
- Inability to hop and jump
- Difficulty getting up from floor (Gower's maneuver)
- Difficulty running
- Difficulty with stairs
- Frequent falls
- Toe walking
- Characteristic gait: anterior pelvic tilt, lumbar lordosis, posterior and lateral lean during stance, "hip waddling" gait from hip abductor and extensor weakness, compensatory movement patterns (such as excessive arm swing for momentum), and foot/ankle pronation and eversion

DMD should be considered in any male child with unknown etiology of:

- Low muscle tone/hypotonia
- Developmental delay

Tests and Measures

Health-Related Quality of Life:

- Pediatric Quality of Life Inventory (Peds QL) and Neuromuscular Module (PedsQL 3.0 NMM)[9]

Participation:

- Enderle-Severson Transition Rating Scale-3[10]
- Pediatric Evaluation Disability Inventory[11]
- School Function Assessment[12]
- Functional Independence Measure (FIM or the FIM for Children (WeeFIM)[13]

Self Determination:

- American Institute for Research (AIR) Self-Determination Scales[14]
- Arc Self-Determination Scale[15]

Function:

- Egen Klassifikation (EK) Scale[16,17]
- Jebsen Hand Function Test[18,19]
- Modified Vignos Lower-Extremity Scale[20]
- Motor Function Measure[21]
- North Star Ambulatory Assessment (NSAA)[22]
- Timed tests[23]
- Gait, Stair, Gower, and Chair Assessment (GSGS)[24]

Impairments:

- Muscle strength (manual muscle testing [MMT][25] protocol, Medical Research Council [MRC] Scale)[26,27]
- Range of motion and measures of muscle extensibility (goniometer)[28]
- Scoliosis screening and postural assessment

If your state, clinic, or school district requires standardized assessments, assessments to consider include:

- Battelle Developmental Inventory 2[29]
- Peabody Developmental Motor Scales (PDMS2)[30]
- Bruininks-Oseretsky Test of Motor Proficiency (BOT-2)[31]

Intervention Recommendations

- Care recommendations state that comprehensive care should be anticipatory and preventive, based on an understanding of the well-defined natural history.[32]
- Multidisciplinary care is critical, with early referral to specialists including physical therapists, occupational therapists, speech therapists, nutritionists, psychologists, social workers, orthopedists, pulmonologists, cardiologists, and gastroenterologists.
- Testing is recommended prior to starting school to allow early identification and intervention for educational needs.
- The role of physical therapist will vary based on the setting and the stage at which the child is seen. It may include evaluation, consultation, coordination, education, and/or direct treatment.
- Promote *self-advocacy*. Provide opportunities to consult with children and their families, and encourage them to advocate for themselves as well as teach others how to assist them (eg, how to transfer them, generate communication to improve transportation opportunities, request educational assistance).
- *Participation* may be optimized by the provision of adaptive equipment and assistive technology, including motorized mobility, computers and computer access including internet, voice activation (if needed), patient lifts, bathing and toileting equipment, electronically controlled beds for position changes, pressure relief mattresses, accommodations for access, and opportunities for adapted and/or wheelchair sports. Plan early—waiting until it is obvious that the young man has lost the function and opportunity to participate is too late.

- *Function*, with the primary goal of participation, should always be one of our main areas of focus during our evaluations, our treatment plans, and when working towards outcomes with the individual and their family. Consider participation in various settings: at home, in school (including higher education), in the community, and the employment market. These young men will live longer and experience better quality of life if we, as therapists, promote the larger and appropriate picture for them and their families.
- The family may require the home to be modified. This takes time and financial consideration. Become knowledgeable about simple architectural Americans with Disabilities' (ADA) requirements.
- Functional and recreational activities such as bike riding and swimming are recommended, with caution against excessively strenuous exercise. Exercise should remain submaximal and avoid resistive and eccentric exercise.[33]
- KAFOs (knee-ankle-foot orthoses) or long leg braces may be recommended to prolong ambulation. AFOs (ankle-foot orthoses) are not typically recommended for use during ambulation since they do not allow the necessary compensation of walking on toes to accommodate for pelvic girdle and knee extensor weakness.
- Power-positioning components on motorized wheelchairs allow independent change of position for more independent stretching, increased function, and maintenance of skin integrity.
- Seating systems in wheelchairs may include a solid seat and back, rigid lateral trunk supports, hip guides, and swing-away adductors to provide appropriate support; pressure relief cushion to maintain skin integrity; and swing-away leg rests to facilitate transfers. Additional options based on the needs of the child may include elevating leg rests, head rests, and a variety of different options for upper-extremity support.[32]
- When standing in good alignment becomes difficult, consider the following standing supports[32,34]:
    - ¤ Standers
    - ¤ Power "stand and drive" wheelchairs
- Prevention of contracture and deformity should *always* be one of our main areas of focus. This requires ongoing preventive care and should include the following[32]:
    - ¤ Daily active/active-assisted and/or passive:
        - ◊ stretching of plantar flexors (with knees flexed and extended), hip flexors, iliotibial bands, hamstrings, and posterior tibialis.
        - ◊ stretching of long wrist and finger flexors, and neck extensors in older individuals as well as any structures or soft tissues identified as "at risk'" in physical therapy evaluation.
    - ¤ Custom-molded ankle-foot orthoses (AFOs) for stretching and to sleep in at night if tolerated[35,36]
    - ¤ Serial casts recommended in some situations[37]
    - ¤ Wrist/hand splints/stretching gloves may be recommended in older individuals for prevention of contracture in long wrist and finger flexors and extensors[32]
    - ¤ Anticipatory, preventive care (with respect to prevention of contracture and deformity). Waiting until it is obvious that muscle or joint tightness is developing, or that positioning or alignment, puts the individual at risk for deformity.
    - ¤ In some instances, respiratory management. Respiratory management may occur in conjunction with respiratory therapy and pulmonary medicine, and should include consideration of assisted coughing (mechanically assisted coughing by caregiver as well as "Cough Assist" machines) and non-invasive ventilation, with BiPAP or a ventilator when needed.

Websites

DMD Pioneers: http://www.dmdpioneers.org
MDA: http://www.mda.org/
Parent Project Muscular Dystrophy: http://www.Parentprojectmd.org

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