Muscular Dystrophy
What is “muscular dystrophy”?

- The word “dystrophy” comes from Latin and Greek roots meaning “faulty nutrition”.
- The term *muscular dystrophy* refers to a group of genetic diseases marked by progressive weakness and degeneration of skeletal or voluntary muscles.
- The heart muscles and some other involuntary muscles are also affected in some forms.
What are the causes of muscular dystrophy?

- Muscular dystrophy is caused due to:
  1. genetic mutations in the muscle protein.
  2. deficiency of the muscle protein itself (i.e. dystrophin) in the muscle
Genetic pattern of inheritance of muscular dystrophy

1. X-linked recessive inheritance pattern with carrier mother:
   - in this case a woman who is a carrier of this disorder will have 25% chances of each of having affected and unaffected males and females.
   - (i.e. 25% affected males + 25% affected females + 25% unaffected males + unaffected females).
   - This pattern is seen generally in case of Duchenne’s muscular dystrophy.
Genetic pattern of disease (contd.):

2. Autosomal pattern:

- In this case, father has 50% chance of having affected child with one mutated gene and 50% of having an unaffected child with two normal genes (i.e. two normal - 1 male, 1 female and two abnormal - 1 affected male and 1 unaffected carrier daughter).
Tests that detect the carriers of muscular dystrophy:

1. Creatinine kinase level test: this is generally raised in case of M.D. (duchenne’s n becker’s). These raised levels in blood indicates that some muscle destruction is going on.

2. DNA based detection test: this is used in case to detect families affected by DNA based carrier anomalies (in case of becker’s nd duchenne’s dystrophy).

- **Electromyography.** The doctor puts small electrodes into muscle to measure electrical activity. Changes in the pattern of activity can show disease.

- **Muscle biopsy.** The doctor removes a small piece of muscle to study in the laboratory. This can distinguish various forms of MD from other muscle diseases.
Signs in case of a typical dystrophic patient:

- Shoulders and arms are held back awkwardly when walking
- Sway back
- Weak butt muscles (hip straighteners)
- Knees may bend back to take weight
- Thick lower leg muscles (the muscle is mostly fat, and not strong)
- Tight heel cord (contracture) child may walk toes
- Belly sticks out due to weak belly muscles (child is poor at sit-ups)
- Thin, weak tights (especially front part)
- Poor balance; falls often
- Awkward clumsy if walking
- Weak muscles in front of leg cause “foot drop” and tip toe contractures
Forms of Muscular Dystrophy

1. Duchenne’s muscular dystrophy.
2. Becker’s muscular dystrophy.
4. Limb-Girdle muscular dystrophy.
5. Facioscapulohumeral muscular dystrophy.
7. Oculopharyngeal muscular dystrophy.
8. Distal muscular dystrophy.
**Duchenne Muscular Dystrophy**

- **Age of onset:** 2-8 yrs.
- **Inheritance/gender affected:** X-linked/males.
- **Muscles first affected:** pelvis, upper arms and upper legs.
- **Signs:** frequent falling, waddling gait, difficulty in getting up, enlargement of the calf (esp.) muscle and sometimes others also. Mental retardation is also seen in some cases.
- **Progression:** slow, sometimes with spurts.
- **Frequently a wheelchair will be needed by age of 12.**
BECKER MUSCULAR DYSTROPHY

- age of onset: 2-16 years.
- Inheritance/gender affected: X-linked/males.
- Muscles first affected: pelvis, upper arms, upper legs.
- Signs: the signs and symptoms are same that of duchenne.
- The differences are that becker dystrophy can first appear much later as age of 25. The boys with it have a longer life expectancy than those of duchenne.
- Progression: slow.
LIMB-GIRDLE MUSCULAR DYSTROPHY

- Age of onset: teens or early adulthood.
- Inheritance/gender affected: autosomal recessive and dominant forms/males and females.
- Muscles first affected: hips and shoulders.
- Signs: progressive weakness that starts in hips and moves towards shoulders; then include arms and legs.
- Progression: usually slow.
FACIO HUMERAL MUSCULAR DYSTROPHY

- Age of onset: teens to early adulthood.
- Inheritance/gender affected: autosomal recessive/males and females.
- Muscles first affected: face, shoulder.
- Signs: sloping of shoulders as well as raising the arms over the head and closing the eyes.
- Progression: slow.
- Disability: ranges from very mild to considerably impairments of walking, chewing, swallowing and speaking.
OCULOOPHARYNGEAL MUSCULAR DYSTROPHY

- Age of onset: 40s-50s.
- Inheritance/gender affected: autosomal dominant/M&F.
- Muscles first affected: eyelids/throat.
- Signs: drooping of eyelids, weakness of eye and facial muscles, difficulty in swallowing.
- Progression: slow.
Other Types of Muscular Dystrophy

**Facioscapulohumeral.** Scapulae are prominent and ride high, particularly on raising arms. Posterior view shows characteristic "wringing" of scapula.

**Limb-girdle.** Difficulty in arising from seated position; lordosis, wide gait.

**Oculofacial.** Protrus, ophthalmoplegia, facial weakness.
DISTAL MUSCULAR DYSTROPHY

- It is a type of rare muscle disease which have in common weakness and wasting of distal muscles (ie muscles of forearms, hands, lower legs and feet.
- Age of onset: adulthood.
- Inheritance: autosomal dominant/recessive.
- Progression: variable.
- Types of distal muscular dystrophy:
  - WALENDAR: it is affects the hands first.
  - MARKSBERY-GRIGGS: it affects the front of lower legs first.
  - NONAKA: it also affects the front of lower legs first.
  - MIYOSHI: affects the back of lower legs first.
CONGENITAL MUSCULAR DYSTROPHY

- Age of onset: at birth.
- Inheritance/gender affected: autosomal recessive/males and females.
- Muscles affected first: generalised.
- Progression: slow.
- Characteristics: involves severe weakness of facial and limb muscles. There is generalised lack of muscle tone which usually appears before 9 months. Seizures are often seen along with brain anomalies and speech disorders.
- Cause: it can be linked to gene defect on chromosome 9 or may be due to deficiency or malfunction of merosin.
EMERY-DREIFUSS MUSCULAR DYSTROPHY

- Age of onset: childhood to early teens.
- Inheritance/gender affected: X-linked recessive/males.
- Muscles affected first: arms and legs.
- Progression: slow.
- Characteristics: MS weakness starts in the shoulders, upperarms and lower legs spreading later to chest and pelvic area. Contractures appear often before the person experiences MS weakness. Here the muscle weakness is less severe than other dystrophies.
Children with DMD use the **Gower's maneuver** to stand up.

They start out on their hands and feet, planting their feet widely apart and pushing up their bottom first. Then they use their hands to push up on their knees and thighs.
TREATMENT OF MUSCULAR DYSTROPHIES

- Medications.
- Surgery.
- Physical therapy.
- Rehabilitative devices.
- Cardiac pacemaker.<in emery-dreifuss MD>.
- Respiratory care
- Speech therapy.
MEDICATIONS

- Corticosteroids to slow muscle degeneration.
- Anticonvulsants to control seizures and some muscle activity.
- Immunosuppressants to delay some damage to dying muscle cells.
- Antibiotics to fight respiratory infections.
Surgery

- **Surgical release of contractures:** A surgeon may cut through tendons to relieve contractures (tendon release surgery).

- **Spinal fusion for scoliosis:** Scoliosis in a wheelchair-dependent child with MD can become so severe that it aggravates breathing problems. Having spine surgery before this happens can help with breathing function, lessen back pain, and improve sitting balance.

- The surgery may be recommend when the spinal curve reaches a certain size (greater than 20°). A surgeon will perform a spinal fusion using metal rods to hold the back in a straighter position.
PHYSICAL THERAPY

- The goal of physical therapy to treat muscular dystrophy is to provide the patient with independence for as long as possible by focusing on movement and develop large muscle groups to make the body stronger and give it more endurance. So main emphasis is given on "MOBILITY".

- WEIGHT/STRENGTHENING EXERCISES: that strengthen and tone the muscles. Stronger muscles can help to delay the impending weakness associated with muscular dystrophy.

- RANGE OF MOTION AND STRETCHING EXERCISES: Flexibility can help ease the severity of joint contractures, a stiffening of the muscles around a joint that affects most people suffering from muscular dystrophy.

- AQUATIC THERAPY: Many experts agree that water exercises and swimming help to tone and strengthen muscles and joints without putting stress on those parts of the body that are already weakened or weakening.
BRACING

- Braces help to keep tendons and muscles stretched, avoiding painful contractures.
- Walking braces for the ankle-foot or the knee-ankle-foot can help support weak muscles and keep the body flexible, slowing progression of contractures.
REHABILITATIVE DEVICES/OCCUPATIONAL THERAPY

- Rehabilitative devices such as canes, walkers, wheelchairs, strollers, and electric wheelchairs can help maintain the child's mobility and independence.

- Sometimes it helps to make modifications to your home, such as widening doorways and installing wheelchair ramps.
Dystrophic patients with rehabilitative devices
Rehabilitation of dystrophic children
RESPIRATORY CARE

- **Non-invasive Breathing Support:** Portable ventilator with a mouthpiece attached (like a microphone) to wheelchair.

- **ASSISTED COUGHING:**
  - Manually assisted cough:
    - Abdominal thrust following deep breath (or breath assisted with ambu bag or other device)
  - Mechanically assisted cough:
    - The Emerson Cough Assist™ (In-Exsufflator, Cofflator)

- **Invasive Support of Breathing:** Tracheostomy
Some dystrophic patients
Some dystrophic patients
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