DUCHENE MUSCULAR DYSTROPHY [DMD]

Duchene described this condition first and then Erb in 1884 called it dystrophy 
X linked Recessive 
3/10000 live male; 1/3rd spontaneous mutation 
Genetic defect: Dystrophin protein in the muscle [Xp21.2 for both DMD and BMD]

Pathology

Normal muscle at birth but lack muscular protein “Dystrophin”
There is progressive loss of muscle mass and it is replaced by fibro fatty tissue 
DMD manifest at the age of 3-5 years and BMD [Becker’s] at 8-12 years 
Dystrophin is important for cell membrane permeability. When absent, there is leakage of CPKinase enzyme and this leakage may cause inflammation and fibrosis

Initial signs

1. Late walker: More than 18 months in boys 
   They need screening for DMD

2. Toe walkers

3. Unable to run 
   Walk with wide base stiff knee gait

4. Clumsiness

5. Hypertrophy of the calf

6. Positive Gower’s sign: Ask the child to stand from sitting position. He can do it only by climbing on his own limb.

7. Sensation Normal; Weak muscles

8. Tendon reflex last to be lost

9. Achilles tightness

10. Lumbar Lordosis and positive Trendlenburg test

EMG

Absence of f waves
Presence of Low amplitude polyphasic waves.
Nerve conduction tests are normal
CK
- Normal: < 200 U/L
- Dermatomyositis: 200-5000 U/L
- DMD or BMD: > 5000 U/L

DNA Analysis
Differentiates DMD/BMD.
70% with 90% accuracy.
21 chromosome for both

Muscle Biopsy
Muscle selection: Vastus lateralis, gastrosoleus
Fix the muscle sample to the spatula to maintain the length of the muscle fibers
Specimen to be sent straight to the lab
Sent in a sterile bottle and not in formalin
Ask for ATPase staining, antidystrophin AB

Treatment
1. Counseling: great deal of sensitivity
2. Support group
3. Genetic counseling
4. When Maternal DNA +ve: Prenatal chorionic villous biopsy
5. Corticosteroid:
   Recently corticosteroid [Deflazacort] given at the age of 7-12 yrs
   Appear to dramatically improve in terms of pulmonary function at 15 years
   Good to excellent results been noted in 89% in treated group and 40% in untreated.
   Experimental
7. Orthotic: Not very popular
   They are cumbersome
   In late stages: need special wheelchair to support the spine

Surgical

<table>
<thead>
<tr>
<th>Diagnostic phase</th>
<th>Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>[0-5Y]</td>
<td>No intervention</td>
</tr>
</tbody>
</table>

Quiescent Phase [5-8 Y]
Mild-Moderate equinus shouldn’t be corrected as it supports the weak quadriceps.
Prevent severe equinus: Stretching Exercises and AFO at night

Active phase [9-12 Y]
Contracture: prevents ambulation: release:
   Ilio-Tibial Band release with Hamstrings release
   Power wheelchair
Stage of Spinal deformity C curve apex in the TL junction [T12- L1] [>12 Y] 95% in DMD [unusual in BMD]
Surgery at COBB angle of 20° ie., early surgery
No place for bracing [All curves progress]
Fusion from T2-L5 (Sacrum when there is Pelvic obliquity)

**Scher:** Soutter’s[when Ober’s test is positive] release and Yount’s to release iliotibial band near the knee and Tibialis posterior transfer as well as aponeurosis release of Gastrocnemius and Orthosis.
And early weight bearing. However, this has not shown to improve motor function.

**Surgical risk**

1. Malignant hyperthermia: Avoid Succinyl choline
2. Cardiac dysfunction: ECG and ECHO
3. Pulmonary dysfunction: Vital capacity <35 avoid surgery [high complications]
4. Increase chance of intra-op bleeding (due to dysfunction of vasa muscularis)
5. GIT: gastric emptying may be delayed [use nasogastric tube]
6. Immobilization weakens muscle strength

**BECKER’S DYSTROPHY [BMD]**

Sex linked
Dystrophin protein is less
Late onset  8-12 years
Red and green color blind
Usually live beyond 22 yrs

Treatment similar to Duchenne muscular dystrophy but usually requires equinus release.
Spinal involvement is rare

**LIMB GIRDLE TYPE**

Usually in late teens
Weakness of Hip and shoulder muscles
CPK normal
Muscle biopsy: Myopathic pattern

**FASCIOCAPULOHUMERAL MUSCULAR DYSTROPHY**

AD, Chromosome 4q87
Normal life expectancy
Slow progressive weakness of the face, shoulder girdle and arm., early adulthood.
Scapular winging, weakness of abduction, very slowly progressive
Treatment: Scapulopexy

**MYOTONIA CONGENITA**

Chromosome 7; AD
Delayed muscle relaxation. Eg., Delayed release of hand grip. Frontal baldness in men and glaucoma do not occur until the middle of adult life.
Biopsy: Type I atrophy
EMG: Dive bomber pattern
Motor strength normal and does not deteriorate with age.

**CONGENITAL MYOTONICA DYSTROPICA**

At birth, affected infants have severe hypotonia Floppy baby
Facial diplegia, problems with respiration and feeding.
There is a high prevalence of club foot and moderate mental retardation.
Very often clubfoot is analogous to an arthrogrypotic condition.

**TOE WALKERS**

**Early:** Idiopathic  
Cerebral Palsy  
Congenital short achillis  
Limb length discrepancy

**Late:** Tethered cord syndrome  
Spinal cord tumor  
Muscular dystrophy

**Assessment**

1. Onset
2. Stand: see heel touches [ in idiopathic toe walker, child can bring the heel down]
3. To demonstrate toe walking: Distract the child while walking or ask the child to run. The toe walking is more prominent
4. Check Sole for pressure and shoe wear pattern
5. Assessment for cerebral palsy  
   Silverskiold test
6. Gower’s sign
7. Check spine
SPINAL MUSCLE DYSTROPHY

Type I  Acute Wernig Hoffman  Die at 6 months
Type II  Chronic Wernig-Hoffman. Initial walk but looses ability to walk with time.
Type III  Kugelber-Welder  Initial walk but looses at teenager

FRIEDREICH’S ATAXIA

AR: Chromosome 9
Defect: Spino-cerebellar tract and Corticospinal tract
1: 50,000
Cardiomyopathy
Cavovarus deformity
Scoliosis: Surgery if >40º [DMD 20º]
Ataxia [spinocerebellar]

SPINA BIFIDA

With current recommendation for women at child bearing age to take 0.4 mg of folic acid and prenatal ultrasound. Assement have decreased incidence of spina bifida.

Alcohol and anticonvulsants are other teratogens should be avoided.

It should be noted that these children have a high latex sensitivity and malignant hyperthermia.

Classification

I Open Spina Bifida Manifest
   Myelomeningocele
   Myelocele

II Closed Spina Bifida Manifesta
   Lipomyelomeningocele
   Myelocystocele
   Simple posterior meningocele

III SBO

   Diastometamyelia
   Dorsal dermal sinus
   Intradural lipoma
   Tight filum terminale
   Hydrosyringomyelia
Diagnosis

1. Alpha Feto protein
2. Sudden change in neurology: suspect tethered cord syndrome. Need MRI
3. 70% will have hydrocephalus
4. L4: Quadriceps is the key muscle. When it is present, it is likely that the patient to walk.
5. Fractures may present like infection. Heals by non-op with excessive callus

Associated lesions

Hydrocephalus
Arnold Chiari Malformation

Treatment

1. Importance of Prenatal diagnosis with ultrasound and Alpha feto protein and abortion
2. If this is unacceptable: Counseling
3. Ventriculo-peritoneal shunts when hydrocephalus associated with spinal bifida. When shunt is blocked, the child becomes irritable; difficult in swallowing.
4. Neurosurgeons: closure in case of open Myelocele
5. Urology: clean intermittent catheterization
6. Orthopedic: CTEV and dislocated hip
7. Genetic counseling: one baby with spina bifida there 1 in 25 chances having second one with spina bifida and when 2 babies has spina bifid this incidence becomes 1:10.

8. Fracture

Can occur with minor trauma and not painful; may mimic infection
Can be missed in wheel chair bound patient
Heals well
Careful padding and brace [soft sheep skin wrap]. Do not immobilise joints as they can cause osteoporosis and further fracture
Fracture usually heal by 3-4 wks
### ORTHOPEDIC PRESENTATION

<table>
<thead>
<tr>
<th>Level</th>
<th>Hip</th>
<th>Knee</th>
<th>Feet</th>
<th>Orthoses</th>
<th>Ambulation</th>
</tr>
</thead>
<tbody>
<tr>
<td>L1</td>
<td>Flexion/Abduction/External rotation</td>
<td>Flexion</td>
<td>Equinovarus</td>
<td>HKFAO</td>
<td>-</td>
</tr>
<tr>
<td>L2</td>
<td>Adduction/Flexion</td>
<td>Flexed</td>
<td>Equinovarus</td>
<td>HKFAO</td>
<td>-</td>
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<tr>
<td>L3</td>
<td>Adduction/Flexion</td>
<td>Recurvatum</td>
<td>Equinovarus</td>
<td>KAFO</td>
<td>Indoor</td>
</tr>
<tr>
<td>L4</td>
<td>Adduction flexion</td>
<td>Recurvatum</td>
<td>Cavovarus</td>
<td>AFO</td>
<td>Limited outdoor</td>
</tr>
<tr>
<td>L5</td>
<td>Flexion</td>
<td>Limited flexion</td>
<td>Calcaneovalgus</td>
<td>AFO</td>
<td>Community</td>
</tr>
<tr>
<td>S1</td>
<td>None</td>
<td>None</td>
<td>Foot deformities</td>
<td>Shoes</td>
<td>Near normal</td>
</tr>
</tbody>
</table>

### DISLOCATED HIPS

**Clinical**
- L1: Flail, No dislocation risk
- L34: Flexion and adduction strong. High risk
- L5: No deformity, stable

**Bilateral:** Hips which are dislocated bilaterally at birth, in association with poor quadriceps power, should be left untreated.

**Unilateral:** Good quadriceps and unilateral dislocation, always reduce

**Surgeries**
- Adductor release or transfer, anterior Obturator neurectomy,
- Psoas transfer (Mustard) or Posteriorly (Sharrad’s procedure)
- Transfer of External oblique to Gluteus maximus
- Open reduction of hip joint, Femoral osteotomy
- Pemberton’s procedure for the hip

### KNEE

**Fixed extension**
- Lengthen quads: Quadriceps plasty and relocate Sartorius and Gracilis

**Fixed flexion**
If quadriceps is good, lengthen the Hamstring if flexion deformity >20°
If Quads poor: Lengthen Hamstrings, Posterior capsulotomy, +/- Distal femoral osteotomy, KFO
FOOT AND ANKLE

Non-walker: Correct deformity for shoe fit and appearance
**Equinus**: Stretching and orthoses [AFO]
    Tendo Achilles lengthening

**Equinovarus**: Medial Release only at 6 months
    Recurrence: repeat release+/- Tib Post transfer
    Resistant foot may need talectomy
    Triple: >14 yrs

**Planovalgus**: Grice green, Lateral column lengthening
    Supramalleolar osteotomy

SPINE DEFORMITY

**Kyphotic deformity**  Lumbar. Born with 80° deformity
    Deformity increases by 8° /year
    Early kyphectomy and bone graft

**Scoliosis**  Usually lordo-scoliosis
    Long fixation with fixation to pelvis using Galverston fixation technique