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.

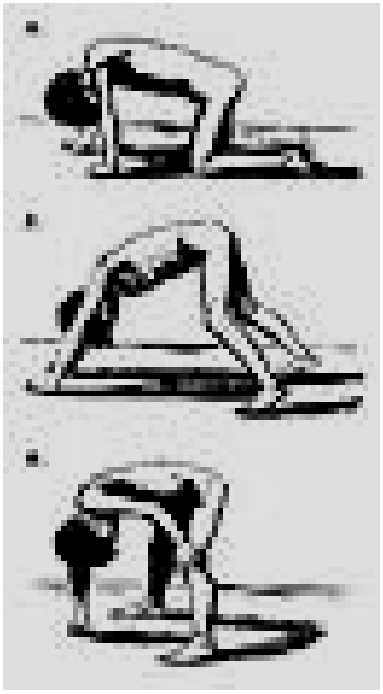
7. Sensation Normal; Weak muscles

8. Tendon reflex last to be lost

9. Achilles tightness

10. Lumbar Lordosis and positive Trendlenburg test

Gower's sign



EMG Absence of f waves
Presence of Low amplitude polyphasic waves.
Nerve conduction tests are normal

CK	Normal	< 200 U/L
	Dermatomyositis	200-5000U/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 ATP ase 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 treate group and 40% in untreated.
6. Genetic treatment: Introduction of normal dystrophin gene into muscle cells.
   Experimental
7. Orthotic: Not very popular
   They are cumbersome
   In late stages: need special wheel chair to support the spine

Surgical

Diagnostic phase	Treatment
[0-5Y]	No intervention
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 wheel chair

Stage of Spinal deformity [>12 Y]

C curve apex in the TL junction [T12- L1]
95% in DMD [unusual in BMD]
Surgery at COBB angle of 20º 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

FASCIOSCAPULOHUMERAL 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-cerebllar 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 protrein 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

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

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