CEREBRAL PALSY
&
NEUROMUSCULAR DISORDERS

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CEREBRAL PALSY

Key Points

- Classification of CP
- Etiology of CP
- Physical exam findings specific to CP
- Treatment of spasticity
- Orthopedic Treatment of hip, spine, feet

Cerebral Palsy (def.)

- An injury to the immature brain
- Non progressive
- Glialization until age 2
- To fix cerebral palsy need to fix the brain

Historical Background

- Described by Little in 1862 @London Obstetrical Society
- Noted relationship between problems with delivery and CP

Classification

- Location
  - Diplegia: legs involved greater than arms
  - Quadreparesis: all limbs “total involvement”
  - Hemiparesis: one sided involvement

Classification

- Tone
  - Spasticity: increased stretch reflex
    - Pyramidal dysfunction
  - Dyskinetic: abnormal motor movement
    - extrapyramidal
  - Ataxic: cerebellar involvement
  - Mixed

Incidence

- Not going down over time but changing from ataxia to diplegia
- Relationship between birth weight and CP

Weight (grams)	Incidence (per 1,000)
1500-2500	3.3
<1500	90.4

Incidence by GA

Term birth	60-65%
Preterm birth	35-40%
Postnatal onset	12%

Etiologies

- Prenatal
  - Maternal epilepsy
  - Severe toxemia
  - Incompetent cervix
- Perinatal
  - Low BW
  - Breech
  - Apgar<3 at 10 minutes
  - Placental complications (previa and abruption)

"MRCP"

- 50-65% of children with CP, quad has largest percentage of cognitive deficit per subgroup

Type	IQ< 50	% Seizures
Diplegia	33	31
Hemiplegia	39	67
Quadreplegia	64	56

Physical Exam

- Selective control
- Muscle tone
- Deformity and contracture at each joint
- Torsional Deformity
- Balance and Equilibrium

Special Tests for CP

- Phelps test
  - Gracilis (knee and hip) v adductors
  - Prone, knee flexed, as extend knee does hip Adduct?
- Silverskoid Test
  - Gastrocnemius v. soleus
  - Supine DF ankle knee extended than flexed
- Duncan Ely
  - Test RF from iliopsoas
  - Prone does pelvis rise with knee flexion

Fighting Spasticity

- Baclofen
- Botox
- Dorsal Rhizotomy
- Valium

Orthopedic Treatment

Needs to be specific to classification of CP

Functional level of child

Timing of intervention

Single setting multiple event

CP Hip: Incidence of Subluxation/Dislocation

- Ranges from 2-76%
- Depends on amount of neurologic involvement
  - Independent ambulators: 7%
  - Dependent sitters: 60%

Posterolateral Deficiency
Testing Concept

Pathophysiology

1. Muscular overpull

   When the abductors and extensors are overpowered the center of the hip joint shifts laterally; with flexion contractures a posterior dislocation can occur.

2. Pelvic Obliquity and scoliosis

   The high hip becomes adducted

Why operate on hips?

- Biomechanically more efficient for walking
  - Hip is concentrically reduced in the socket
- Important to maintain sitting balance
- Reduces pain (48% - 70% of patients)
  - Amount of degenerative arthritis is unknown
- Hygiene

Hip Surgery Menu

- Soft Tissue
  - Muscle: adductor, gracilis, psoas (o.n.)
  - Capsule: open reduction
- Bone
  - Femur: varus, derotation, shortening
  - Socket: acetabuloplasty
- Salvage

When to not reduce a hip ?

- Medically unstable patient
- Triangularization of the head
- Arthritis

Radiographic Findings

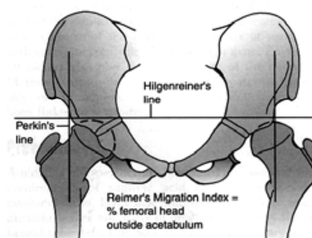


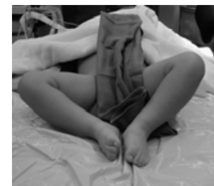
Figure 18-2 Reimer's Migration Index is a measure of the percentage of femoral head outside the acetabulum.

Reimers >50%
Testing Concept

- Hip dislocation approaches 100% over time
- Indication for operative treatment

Physical Exam

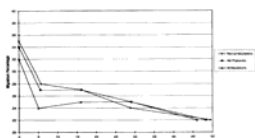
- Galeazzi Sign
- Assymetric Hip Abduction



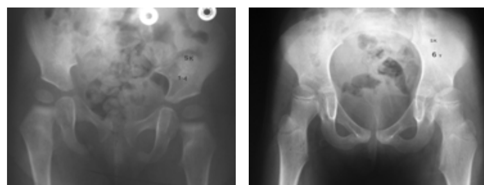
Soft tissue release

- For hip often combined with other releases for ambulatory children
- Early intervention: can the natural history be changed?

Miller et al JPO, 1997



Pre- and Post- op Patient



Soft tissue plus osteotomies Laterally based Dega Mubarek et al., JBJS

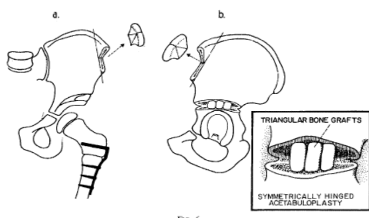
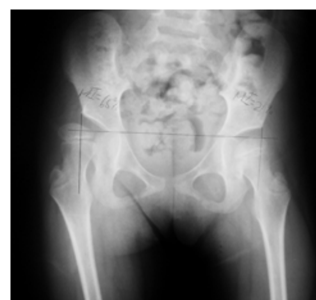
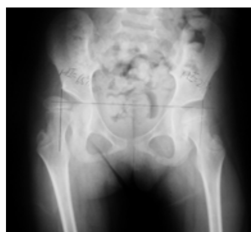


FIG. 6

6 year old ambulatory child with spastic diplegia

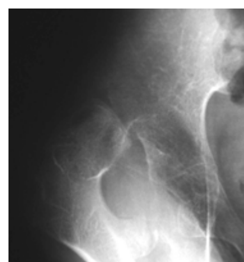


Problem List for the right hip:



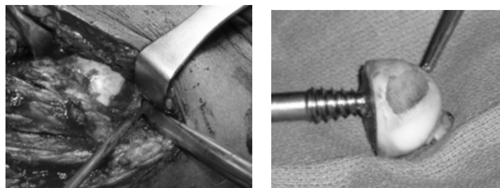
- Leg is in abduction
  - Release soft tissues
- Reimer's Migration
  - Greater than 50%
- Acetabular Index
  - Pelvic Osteotomy

Why is waiting too long a problem?

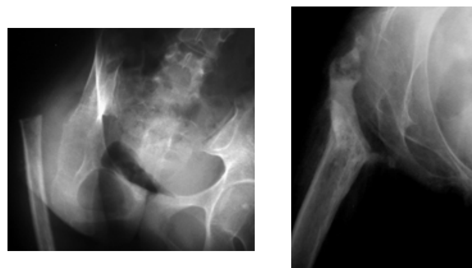


- Triangularly shaped head cannot be replaced into acetabulum
- 1-2 years after complete subluxation and reconstruction is not possible

Erosion of Head/Loss of Cartilage



Salvage Procedure: FHR



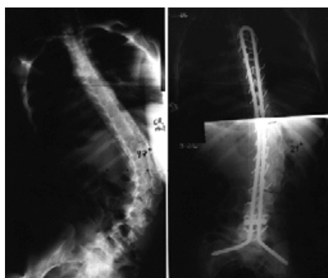
Testable Concepts

- Hips in CP can be painful
- Unlike DDH the hip goes out posterolaterally most often
- Data to show that early soft tissue releases can prevent further subluxation
- Osteotomies about femur and acetabulum will depend on x rays: look for anteversion, valgus and acetabular index

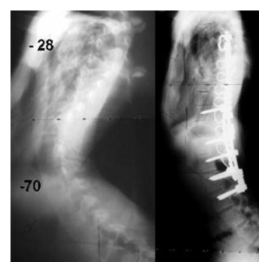
Spine in CP

- Curves are caused by muscle imbalance
- Curves do not respond to BRACING
  - Bracing however may help trunk support for children with mixed CP who low tone for head neck and spine.

NM curves are Different: Long C and S shapes



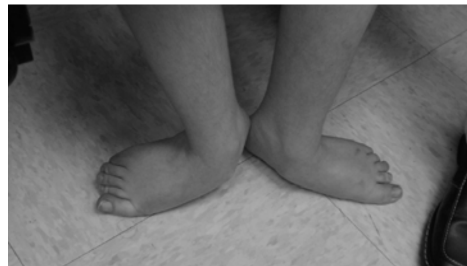
Lumbar lordosis common too



Treat with Fusion once curves are 50 degrees and rigid Testable Concepts

- Often from T3 to the pelvis
- Sublaminar wires, hooks, pedical screws all used
- Iliosacral screws, Unit rod, Galveston
- Allograft
- Indications for going anteriorly????
  - Crankshaft
  - Curve magnitude

Feet



Feet we see….

- Planovalgus
  - Os calcis lengthenings
  - Subtalar arthodesis
- Cavovarus
  - SPLATT
  - SPLOTT
  - Calcaneal slide
  - Triple

What to do depends on:

Testable Concept

- Rigidity of the foot: if not passively correctable than soft tissue releases are not the answer!

Gait cycle

- Swing Phase
  - 40%
- Stance Phase
  - Double support to double support
  - 60%

Prerequisites of normal Gait

- Stability in stance
- Foot clearance in swing
- Proper prepositioning of the foot in terminal swing
- Adequate step length
- Reduction of Energy

Gait Analysis

- Approx. 20 gait labs in US
  - Kinetics and kinematics
  - Force plates
  - Joints defined by sensors info. analysed by computer
- Revolutionary in demonstating the differences between compensations and true pathology.
- Should all kids get a gait analysis?
  - Difficult interpretation

NEUROMUSCULAR DISORDERS

Three Common Disorders

- Muscular Dystrophy
  - Duchenne's
  - Becker's
- Spinal Muscular Atrophy
- Charcot Marie Tooth Disease (HMSN)

Physical Examination

- Skin, facies
- Drooling, nasal speech
- Tongue fasciculations (LMN)
- Ophthalmologic examination
- Muscle testing
  - Proximal>distal (myopathic)
  - Weakness>atrophy (myopathic)
  - Atrophy>weakness (neuropathic)

Physical Examination

- Neurologic
  - Vibratory sense (CMT)
  - DTR's (absent – CMT or SMA)
  - Babinski (UMN)
  - Romberg (cerebellar)
  - Mental function evaluation

Diagnostic Studies

- Hematologic studies
- Electromyography (EMG)
- Nerve conduction studies
- Muscle biopsy
- Nerve biopsy
- Molecular diagnostic studies

EMG

- Differentiate between neuropathy/myopathy
- Rarely establishes specific diagnosis

- Myopathic
  - +/- Abnormal spontaneous activity (fibrillations)
  - Small amplitude, polyphasic motor unit potentials
- Neuropathic
  - Abnormal recruitment of voluntary motor units
  - Abnormal spontaneous activity (acute to chronic)
  - High amplitude, long duration voluntary motor units (chronic)

Nerve Conduction Studies

- Aid in diagnosis of peripheral neuropathy
- Normal values
  - Child>3 years = 45-65 m/sec
  - <3 years lower due to incomplete myelinization
- Normal in anterior horn cell, nerve root disease, myopathies Testable Concept
- May detect CMT before clinical deficit
- Can determine single nerve (mononeuropathy) vs disseminated process (polyneuropathy)

Duchenne Muscular Dystrophy

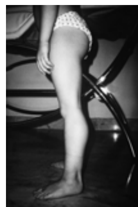
- Most common X-linked locus Xp 21
  - 1/3,500 boys
  - High new mutation rate (1/3)
  - > 90% cases now first in family
- Early stages most like polymyositis
  - Can present with toe walking

Clinical Presentation

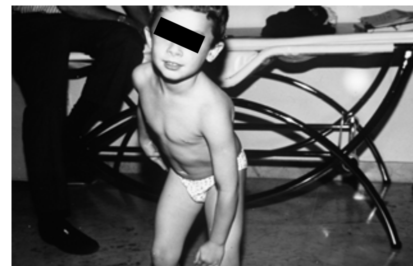
- Present 3-6 years
- Delayed ambulation/toe-walking
- Difficulty climbing stairs, hop, jump
- Proximal muscles > distal
- Lower limbs > upper
- Sensation, DTR's intact
- Global developmental delay in 50%
- Asymptomatic elevated ALT/AST/CPK

Clinical Features

- Pseudohypertrophy of gastrocsoleus muscle
- Trendelenburg's sign
- Gower's sign
- Contractures of LE
  - Hip, ankle, foot
- Scoliosis



Gower's sign



Molecular Analysis

- Single gene defect – dystrophin (Xp21)
- 400kD protein
  - Cell membrane cytoskeleton
  - 0.01% skeletal muscle protein
- Dystrophin testing or mutation analysis
  - Differentiate between different MDs
  - Explain variable clinical presentations

Diagnosis of DMD

- Serum CPK
  - Elevated in early stage (200X normal)
  - Decreases with time
  - Mildly Elevated in female carriers (80%)
- Dystrophin gene PCR abnormal in 60%
- Muscle biopsy
  - Muscle degeneration, loss of fiber
  - Fibrous proliferation, adipose tissue
  - Increased cellularity
  - Dystrophin staining

Gait in DMD

- Compensatory mechanisms:
- Hip extensors
  - Increase lumbar lordosis, ant pelvic tilt
  - Maintain weight posterior to hip joint
- Hip abductors
  - Increase shoulder sway
  - Increase wide-base gait

Late Features
<u>Testable Concept</u>

- Cardiac involvement (ECHO pre-op)
  - Tachycardia
  - Cardiomyopathy
  - Life threatening 10%
  - Mitral valve prolapse
- Static encephalopathy, mild MR
- Pulmonary failure (PFTs pre-op)

Treatment of DMD

- Problems
  - Decreasing ambulatory ability
  - Soft tissue contracture
  - Spinal deformity
- Goals
  - Improve, maintain function of child

Treatment of DMD

- Medical
- PT/orthotics
- Cardiopulmonary management
- Wheelchair fitting
- Psychological counseling
- Surgery

Medical Treatment

- Steroids therapy
  - Oral steroids
  - Delay loss of muscle strength up to 3 years
  - Stabilization of curve formation
  - Side effects of weight gain, osteopenia
- Gene therapy
  - Myoblast transfer – currently unsuccessful
  - cDNA in mouse model successful

Surgery: Soft tissue release LE

- Early
  - Soft tissue releases (hip, hams, ankles, PT)
  - Timing is controversial
  - ? extension of ambulatory time
- Palliative
  - Soft tissue release of ankle once WC bound

Surgery in DMD: Spine

- 95% progressive scoliosis
- Surgery improves sitting balance, quality of life
- Perform if >20-30 degrees <u>Testable Concept</u>
- Fuse to L5 or pelvis, segmental instrumentation
- PFTs
  - FVC>40% ok
  - 30-40% guarded
  - <30% may not be recommended

Post - op spinal fusion

- No pulmonary changes
- Sitting balance improves
- Overall quality of life improves
- Ability to feed may deteriorate <u>Testable Concept</u>

Becker's MD

- Similar to DMD but less severe
- Incidence ~ ½ that of DMD (1:7000 boys)
- Onset after 7 years
- Variable course
  - severe (like DMD) to minor or even normal
- Xp21 locus – dystrophin
  - In frame deletion
  - Subnormal or abnormal size dystrophin

SMA

- Hereditary group of diseases
- Degeneration of anterior horn cells
- Autosomal recessive
- 1/6,000
- Carrier state ~ 1/40

Genetics of SMA

- Chromosome 5q
  - survival motor neuron protein (SMN)
- Two copies of SMN gene, SMN1 and 2
  - All cases homozygous loss of SMN1
  - Severity ~75% related to copy number of SMN2

Diagnosis of SMA

- SMN gene test (loss of SMN 1 exon 7)
  - 95% sensitive, 100% specific
- CPK, aldolase – close to normal
- EMG – denervation
- NCV – normal
- Muscle biopsy
  - Atrophy of fibers
  - Fibers are uniform in diameter

Classification of SMA

- Type I (mostly 2 copies SMN2)
  - Acute infantile – Werdnig-Hoffmann disease
- Type II (mostly 3 copies SMN2)
  - Chronic infantile
- Type III (mostly 4 copies SMN2)
  - Milder form
  - Kugelberg-Welander disease

SMA: Type 1

- Identified first 6 months
- 30% Low activity during pregnancy
- Physical Examination
  - No DTRs
  - Tongue fasciculations
  - Face (eyes) bright, body limp
- Course – mean survival 8 months, 25% survival at 1 year

SMA: Type 2

- Onset 6-18 months
- Muscular weakness legs>arms
- DTRs absent
- Sits independently, no ambulation
- Survival duration related to vigor of care

SMA: Type 3

- Diagnosis 2-10 years
- Symptom: weakness of gait
  - Hip extensor weakness
- Pseudohypertrophy of calf may be present
- Walking at least some time in childhood
- Life expectancy normal

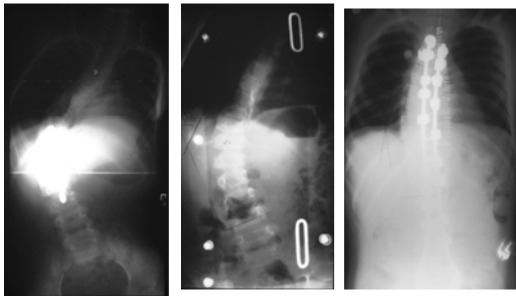
Treatment of SMA

- Development of contractures of LE
- Hip subluxation/dislocation
- Scoliosis

Surgical Treatment of SMA

- Hips – usually not indicated unless unilateral
- Spine
  - Indicated in curves >40 degrees, 10 years of age Testable Concept
  - Pulmonary is an issue, but not as with MD
  - Ant/posterior – controversial
  - Fuse to pelvis

Scoliosis Progression



Hereditary Motor Sensory Neuropathies

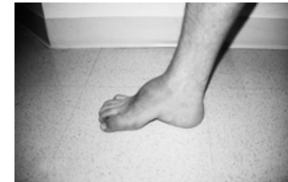
- Group of disorders
- Charcot Marie Tooth is prototype
- Major types

  CMT 1:

  demyelinating

  CMT 2:

  axonal



Classification

- Type I (demyelinating CMT)
  - 80-90% are type 1a
    - Duplication of PMP22, chromosome 17
    - Gene test is sensitive and specific
  - Type 1b: Myelin P Zero (MPZ)(gene test)
  - Type 1x: Connexin 32 (gene test)
  - Other forms (many) very rare
  - Peroneal muscle weakness, absent DTRs
  - Distal sensory loss (proprioception)
  - Slow NCVs

Classification

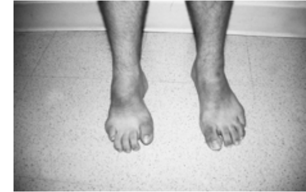
- Type 2 CMT
  - Axonal form of CMT
  - Normal reflexes where strong
  - Mild change in NCS
  - Much rarer than CMT 1
  - Wide range of genes, inheritance patterns

Diagnosis of CMT

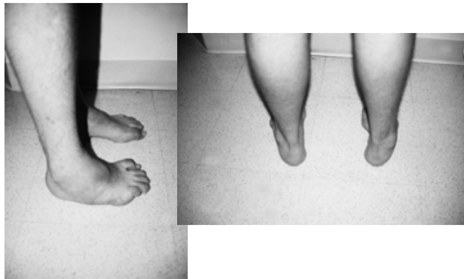
- Physical examination
- EMG, NCS
- Muscle biopsy- atrophy of fiber group
- Nerve biopsy - demyelination

Pes Cavovarus

- Etiology
  - Progressive distal to proximal muscle involvement
  - Cavus, flexion of 1st metatarsal
  - Hindfoot varus, forefoot supination



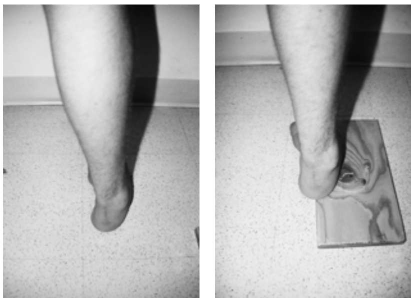
Features of Pes cavovarus



Treatment

- Depend on foot flexibilty Testable Concept
  - Coleman block test
- Plantar release
- Tendon transfer
- Calcaneal/midfoot osteotomy
- Triple arthrodesis

Coleman Block Test



Hip Dysplasia

- Rare/ Looks like DDH
- Eliminate contractures
- Approach on both sides of joint

Spine in CMT

- 30-50% of HMSN
- Similar to AIS
- Orthotic/surgical management

Hand in CMT

- 2/3 of individuals
- Occurs later on (>19 years)
- Intrinsic muscle weakness with instability
- Sensory changes occur concomitantly

Thank you for your attention!