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Neuromuscular Disorders in Children

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Introduction

- Definition
- Clinical presentation
- Clinical assessment
- Common disorders
- Diagnosis and Investigation
- Management

Neuromuscular disorders

- A large group of diseases affecting any part of the central and peripheral nervous system and muscle
- Upper and lower motor neuron signs
- **Orthopaedic issues:**
  - Dystonia
  - Weakness
  - Delayed developmental milestones
  - Deformities

Clinical Assessment

History

- Diagnosis often already established
- From parents, patient plus physiotherapists...
- Demographics
- Presenting complaint
- Maternal history
- Birth history
- Physical and mental developmental milestones
- Performance at school, participation in sports

Clinical Assessment

Examination

- Observe child and general assessment
- Walking aids, wheelchair, orthotics.....
- Abnormal features
- Dystonic movements
- Gait assessment
- Spine
- Lower limb alignment, rotational profile, leg lengths
- joint and muscle contractures
- Joint stability
- Full neurological examination

Common disorders

- Cerebral palsy
- Myelodysplasia (Spina bifida)
- Muscular Dystrophies
- Inflammatory myositis
- Hereditary neuropathies
- Arthrogrypotic syndromes
- Myasthenia gravis
- Anterior horn cell disorders

Cerebral palsy

- Spectrum of disorders that result from non-progressive brain damage during early development (up to 2 years)
- 2 per 1000 live births
- Cause is not always identifiable
- Maternal toxaemia, prematurity, perinatal anoxia, kernicterus, postnatal brain infections or injury
- Epilepsy, perceptual problems, speech disorder , mental retardation and behavioural problems

Cerebral palsy

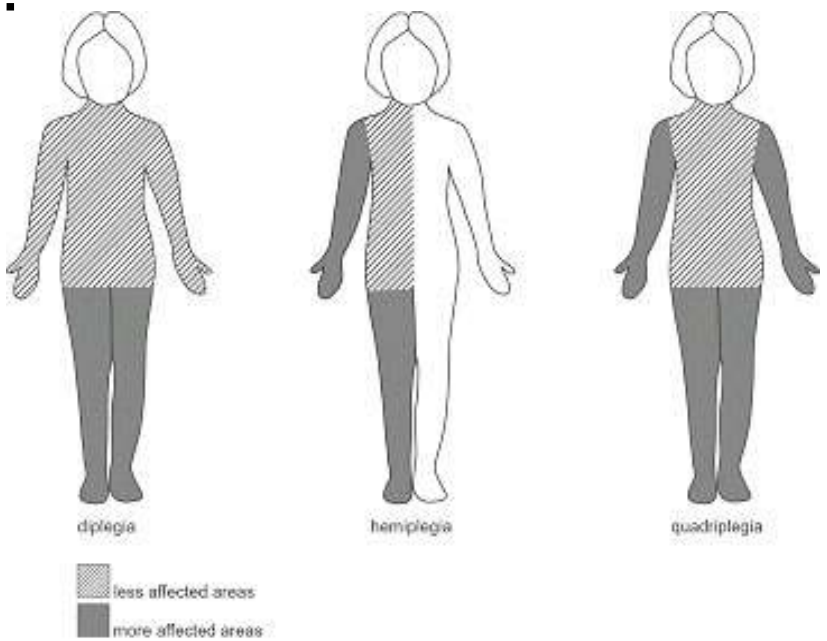
- Classification based on type of Motor Dysfunction or Topographic distribution

- Motor dysfunction:
  - Spastic palsy (commonest)
  - Athetoid (involuntary movements)
  - Ataxic (cerebellar signs)
  - Rigid palsy
  - Mixed

Cerebral palsy

- Topographic Distribution:
  - Diplegia
  - Hemiplegia
  - Quadriplegia

  (Total body involvement)
- Full clinical picture takes several months or years to develop



Cerebral Palsy Management

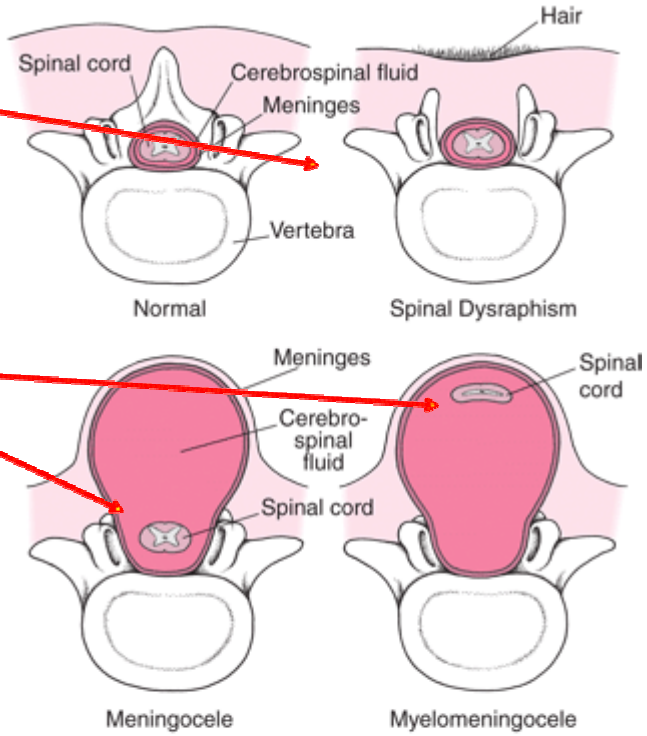
- Multidisciplinary approach
- Paediatric neurologists, Physiotherapy, orthotics, Botulinum toxin injections
- Gait analysis
- Tenotomies and tendon lengthening
- Tendon transfers for muscle balancing
- Osteotomies eg. varus proximal femoral, extension distal femoral
- Timing and decision making crucial

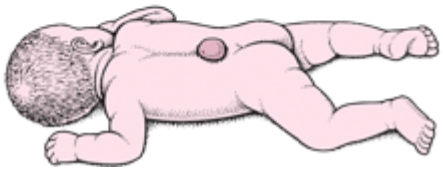
Myelodysplasia (Spina Bifida)

- Congenital disorder
- 2 halves of the posterior vertebral arch fail to fuse
- maldevelopment of neural tube and overlying skin
- ‘dysraphism’
- Cystic spina bifida -2-3 per 1000 live births
- x10 greater risk for subsequent child
- Neural tube defects associated with elevated AFP levels

Myelodysplasia (Spina bifida)

- Spina bifida occulta
- Spina bifida cystica
  - Meningocele
  - Myelomeningocele
  - Rachischisis (open myelomeningocele)
- Hydrocephalus (70% cases):
  - Distal cord tethering, raised intracranial pressure, herniation, cerebral atrophy and retardation (CP)





Myelodysplasia (Spina Bifida)

- Investigations include radiographs, myelography, CT, MRI
- With neural involvement there is paralysis, sensory loss, impaired sphincter control
- Urinary problems
- Degree of disability dependent on level of defect
- Fractures are common and can go undiagnosed
- Scoliosis can be severe and progressive
- Pelvic obliquity

Myelodysplasia (Spina bifida)

- Preservation of neural structures
- Majority cases defect should be closed within 48 hrs to prevent infection, ulceration or drying
- Hydrocephalus addressed
- Urological surgery
- Orthopaedic intervention addressing spinal, hip, knee, foot and ankle deformities
- Physiotherapy and orthotics essential
- Surgery for muscle balancing and deformity correction
- Focus on upper limb function and intellectual skills

Muscular Dystrophies – Duchenne's

- Non inflammatory inherited disorders with progressive muscle weakness
- Sex linked recessive abnormality in young boys
- Clumsy (ataxic) gait, decreased motor skills
- lumbar lordosis, calf pseudohypertrophy
- Gower's sign (hip extensors typically first muscle group affected)
- Markedly elevated CPK and Absent Dystrophin protein
- Muscle Biopsy shows foci of necrosis and connective and fatty tissue infiltration

Muscular Dystrophies – Duchenne's

- Patients can lose ambulation by 10 years
- Wheelchair bound by 15 years
- Rapid neuromuscular scoliosis between 13-14 years
- Warrants early surgical intervention
- Fatal cardiorespiratory complications in 20's
- Physiotherapy and Orthotics
- Genetic counselling
- Surgery for tendon transfers and release of contractures

Muscular Dystrophies – Becker's

- Sex linked recessive
- Decrease in Dystrophin
- Red/green colour blind boys
- Similar but less severe than Duchenne's
- Prognosis and long term survival better

Muscular Dystrophies – some others

- Fascioscapulohumeral
  - AD disorder, 6-20 years old with facial muscle abnormalities, normal CPK, winging of scapula
  - Scapulothoracic fusion
- Limb Girdle
  - AR, 10-30 years old, pelvic and shoulder girdle involvement
  - Increased CPK

Scapular winging in a patient with FSH

Hereditary Sensory Motor Neuropathies

- Charcot -Marie -Tooth Disease
  - Peroneal muscle atrophy - weakness
  - Type 1 (hypertrophic) and 2 (neuronal)
  - Type 1 – onset 2nd decade, AD inheritance, slow nerve conduction, absent reflexes
  - Type 2 – onset 3rd decade, more extensive foot involvement, variable inheritance, normal nerve conduction, normal reflexes
  - Low nerve conduction velocities – peroneal, ulnar and median
  - DNA testing – duplication of portion of chromosome 17

Hereditary Sensory Motor Neuropathies

- Clinical signs
  - Intrinsic muscle wasting
  - Pes cavus
  - Hammer toes
  - Callosities
  - Peroneal weakness
  - ‘Stork legs’

Hereditary Sensory Motor Neuropathies

Orthopaedic Management of HSMN:

- Orthotics and physiotherapy
- Tibialis anterior, peroneus longus, peroneus brevis
- Tibialis posterior transfer if hind foot mobile
- Calcaneal sliding or wedge osteotomy when fixed hind foot
- Metatarsal extension osteotomies
- Plantar fascial release
- Hammer toe correction
- Triple arthrodesis
- Intrinsic procedures for Hand deformities

Hereditary Neuropathies – Freidrich's Ataxia

- Autosomal Recessive disorder
- Defect in the Frataxin gene
- Spinocerebellar disease
- Mean onset between 7 and 15 years
- Ataxic gait, nystagmus, cardiomyopathy
- Cavus foot and scoliosis
- Motor and sensory defects
- Wheelchair by 30 years
- Life expectancy 40-50 years

Arthrogryposis Multiplex Congenita

- Non progressive disorder with multiple congenitally rigid joints
- Myopathic, neuropathic or mixed
- Normal viscera, facies and intelligence

Arthrogryposis Multiplex Congenita - management

- Optimizing function – permit feeding and perineal care, possible ambulation
- Passive manipulation
- Serial casting
- Soft tissue releases
- Osteotomies and joint contracture surgery – careful planning mandatory

Anterior Horn cell Disorders

- Poliomyelitis
  - Viral infection of anterior horn cells brainstem and spinal cord
  - LMN signs
  - Rare disease with vaccination
  - Virus entry via GI tract
  - Mild influenza like illness precedes paralysis
  - Stage of recovery and convalescence (6-24 months)
  - Residual paralysis leads to deformities and growth defects

Anterior Horn cell Disorders

- Spinal Muscular Atrophy
  - Rare group of inherited disorders
  - Degeneration of anterior horn cells
  - Progressive LMN weakness
  - Werdnig-Hoffman commonest form (AR)
  - Kugelberg-Welander less severe form (AD/AR)
  - Usually adolescents and young adults
  - Limb weakness, proximal muscle wasting, scoliosis
  - Life expectancy 3rd-4th decade

Other conditions

- Polymyositis, dermatomyositis
- Myasthenia Gravis
- Acute idiopathic post infectious polyneuropathy
- Spinal cord tumours
- Post radiotherapy
- Trauma