A. CEREBRAL PALSY

1. “Static encephalopathy”, 7.5 per 1,000 live births, UMN

2. Clinical syndrome characterized by a chronic, non-progressive disorder of motor control, resulting in abnormality of posture, movement and tone

3. Syndrome is characterized by:
   a) fixed, non-progressive brain lesion or lesions
   b) original lesion may be a result of pre-, peri-, or postnatal trauma or anoxia; “the lesion affects the immature brain and interferes with the maturation of the CNS, which has specific consequences in terms of the type of CP which develops, its diagnosis and treatment”
      Smooth movement requires integration of sensory input, central processing of information in the brain and coordination with higher level cerebral function (motor planning of activity). The brain lesion of the cerebral palsied child can affect the part of the brain that interprets sensory information (difficulties with proprioception and sensory integration)
   c) lack of motor control results in abnormal coordination of movement and posture; may also see mental retardation, convulsions, sensory disturbance, speech impediment, defects of hearing, language or eyesight

4. Risk Factors
   a) Prematurity and low birth weight*
   b) Metabolic (hypoglycemia, hyperbilirubinemia)
   c) CVA
   d) Infections (toxoplasmosis, rubella)
   e) Cerebral malformation

5. CP Classification
   a) Spastic – most common group, includes diplegia, hemiplegia, and quadriplegia
b) **Extrapyramidal** (includes choreoathetosis and dystonia)

c) **Hypotonic**

d) **Ataxic**

e) **Mixed**

6. **Clinical Features**

   a) In infants may see hypotonia, persistence of primitive neonatal reflexes, poor development of more mature reflexes, toe walking, strong hand preference (hemi)

   b) Older children – hypertonicity, ↑ DTR’s, Babinski response of dorsiflexion, delayed gross motor milestones, abnormal gait, drooling, strabismus, difficulty with feeding and speech, mental retardation, seizures, adductor grab, clonus

7. **Spastic diplegic** – more involvement of legs than arms, spastic crouch, equinovarus foot deformity

8. **Spastic quadriplegic** – involvement of all 4 limbs, usually more severely involved than diplegic, may never walk, difficulty in feeding, speech and drooling, frequent pneumonia, mental retardation, seizures, strabismus

9. **Hemiplegic** – involvement of unilateral arm and leg, early hand preference, unilateral toe walking or circumduction of limb, unilateral Babinski

10. **Choreoathetoid** – hypotonic until age 2, slow, writhing, involuntary movements of flexion & extension, pronation & supination of fingers and hands and sometimes toes and feet, abnormal movements of face, tongue, and palate, facial grimaces, difficulties with speech and feeding, may be very late in walking (8 or 10 years of age)

11. **Dystonia** – sustained twisted posture involving trunk and limbs

12. **Ataxia** – poor coordination, wide based gait

**B. SPINA BIFIDA**

1. Incomplete closure of the bony elements of spine with or without extrusion of contents of the spinal canal through bony defect
2. **Spina bifida occulta** – defect of fusion of posterior vertebral arch without disruption of underlying meninges and cord, 10% of pediatric population, asymptomatic, low midback hairy tuft, dimple or teratotoma

3. **Spina bifida aperta** – 1 in 1,000 live births, most common in lumbar and lumbosacral area
   a) **Meningocele** – distention of meninges
   b) **Myelomeningocele** – distention of meninges and cord
   c) **Myelocoele** – distention of meninges and cord with lack of closure of cord in defective area

4. Clinical Features
   a) Paralysis and loss of sensation below lesion level (ulcerations, fractures)
   b) Lack of bladder and bowel control
   c) Hydrocephalus
   d) Hip dislocations, scoliosis and foot deformities (equinovarus most common)

C. **TETHERED CORD SYNDROME**
   1. Neurologic deterioration when distal spinal cord subjected to traction or compression
   2. Differential Diagnosis: spinal cord tumor (e.g. lipoma), myelomeningocele, diastematomyelia (congenital malformation in which spinal cord is divided longitudinally into 2 hemicords by a bony, fibrous, or cartilaginous spicule from the posterior aspect of a vertebral body (spicule transfixes cord) or cauda equina. These conditions stop the normal ascent of the spinal cord during growth of the vertebral column; neurologic deficit below level of lesion
   3. Clinical Findings: sensory changes, foot deformity, bowel and/or bladder dysfunction, back pain, scoliosis, decreased motor function (late in walking or limp, and cutaneous changes over lower back area such as dimple, hemangioma, or hypertrichosis, cavovarus feet, ulcerations of feet
   4. Early diagnosis by MRI, followed by release of cord tethering; prevents further deformity but does not reverse existing deformity
5. 75% of cases female

**D. MUSCULAR DYSTROPHIES**

1. Duchenne MD  
   a) Most common and most severe, X-linked recessive, high mutation rate  
   b) Male to female, 9:1  
   c) Onset usually apparent by age 3  
   d) Weak fetal movements in last trimester  
   e) Delayed motor milestones, difficulty climbing stairs, use arms to rise from chair  
   f) Difficulty or unable to run or hop, frequent falling  
   g) **Gower’s sign**, waddling gait, toe walking  
   h) DTR’s disappear early except ankle  
   i) Cardiac involvement, 50% mental retardation  
   j) Symmetrical involvement first of pelvis, then shoulders, 3 to 5 years later pseudohypertrophy of calves, unable to walk by 12, death teen to third decade  
   k) Pes planus secondary to tight heel cord early stage; equinovarus later stage, lumbar lordosis  
   l) Elevated serum enzyme levels, especially CPK 30-300 X normal

2. Becker’s MD  
   a) X-linked recessive  
   b) Benign form, onset 5-20, slow progression, otherwise similar to Duchenne

3. Limb Girdle MD  
   a) Progressive weakness and atrophy of mm. of shoulder and pelvic girdle and proximal limbs during 20’s or 30’s, weakness of neck flexors and extensors
b) Autosomal recessive

4. Fascioscapulohumeral MD
   a) Autosomal dominant, onset 20’s
   b) First facial weakness (whistling, sipping, keeping eyes closed), shoulder girdle weakness and scapular winging (can’t raise arms above head), eventually anterolateral leg weakness (foot drop)
   c) Slow progression

5. Myotonic MD (myotonic dystrophy, dystrophia myotonica), varies in severity, autosomal dominant, most common form of adult onset muscular dystrophy
   a) Myotonia- inability to relax muscles at will (to test for myotonia, percuss thenar eminence or tongue, ask pt. to release a tightly clenched fist)
   b) 2 forms- infant (congenital myotonic dystrophy) and adult onset myotonic dystrophy
   c) Adult onset myotonic dystrophy- distal arm and anterior leg muscles affected (foot drop), neck, jaw and face muscles may be weak( ptosis, blank facial expression, long face), cataracts, conduction block of heart, involuntary muscles of internal organs affected leading to difficulty swallowing, cramping, and diarrhea
   d) Infant form of myotonic dystrophy- hypotonic, clubfoot deformity in infants, develop myotonia later in life, mental retardation, delay in milestones, toe walkers

**APPRAOCH TO THE CHILD WITH MUSCULAR WEAKNESS**
(Spiro, Approach to Diagnosis in the Child with Muscle Weakness Pediatric Annals March 1997)
1. Signs and symptoms
2. Genetic history
3. Biochemical tests – serum enzyme levels
4. Electrodiagnostic tests – NCV and EMG
5. Muscle biopsy
6. Checklist for history taking
   Trouble walking, running, climbing stairs, getting up from a chair? Toe walking?
Can he whistle, sip a straw, keep eyes shut?
Trips easily?
Hands weak? Trouble letting go of objects?
When was weakness first noted?
Muscle pain?
Floppy as a baby?
Easily fatigued?
Weak fetal movements in last trimester?
Family history?
Fast or slow onset?
Weakness proximal or distal?
Orthopedic abnormalities? scoliosis, congenital dislocated hip, pes planus, pes cavus, sloping shoulders, lumbar lordosis?

7. Checklist for examination

Rash? (nail beds & knuckles, face, eyelids)
Lumbar lordosis, scoliosis, pes cavus?
Weakness on manual muscle tests? Proximal, distal or global weakness?
Mental retardation?
Hypotonic? (traction response)
Paucity of limb movements as infant?
Waddling gait? Gower's sign?
Muscle wasting or enlargement?
Myotonia, fasciculations?
DTR's?
Sensory abnormalities?
Facial muscle weakness?
Toe walking? Foot Drop?
Difficulty running, hopping, going up and down stairs?

**E. POLYMYOSITIS**

1. Non-hereditary, inflammatory myopathy

2. In presence of skin rash, dermatomyositis

3. **Dermatomyositis** – symmetrical weakness of proximal limb mm. and neck flexors, rash (butterfly distribution, heliotrope, erythematous, scaly extensor surfaces of joints especially ankles, (medial malleolus), elbows, knees, knuckles and dorsal finger near nail bed; later stage – skin tight and glossy with subcutaneous calcium deposits

4. In children, no association with malignancy
F. NEUROFIBROMATOSIS (VON RECKLINGHAUSEN’S DISEASE)

1. autosomal dominant genetic disorder that causes tumors to grow along various nerves and causes skin and bony abnormalities

2. 8 subtypes
   a) NF1- 1/ 3,500, most common subtype, mutation of gene affects body’s ability to suppress tumors/ 3 classic signs in childhood include café au lait spots, multiple soft tissue tumors (mainly neurofibromas), and skeletal changes
   b) NF2- rare 1/ 40,000, patients develop bilateral acoustic neuromas on vestibulococchlear nerve/ hallmark of disease is hearing loss due to acoustic neuromas between ages 18-22/ early symptoms are headache, vertigo, facial paralysis, deafness, and tinnitus
   c) Schwannomatosis- rare 1/ 40,000 multiple schwannomas, not neurofibromas
   d) Other 5 extremely rare

3. Signs and Symptoms of NF1
   a) 6 or more café au lait spots measuring 5 mms. in children and 15 mms. in adolescents and adults
   b) 2 or more neurofibromas (develop in tissue surrounding peripheral nerves) or 1 plexiform neurofibroma (involves many nerves)
   c) freckling in arm pit or groin
   d) 2 or more Lisch nodules (tumors on iris of eye, no clinical significance)
   e) optic glioma
   f) abnormal development of spine (scoliosis), sphenoid bone of skull, or tibia (often anterolateral bowing of tibia and pseudoarthrosis)
   g) 1st degree relative with NF1

4. Other conditions associated with NF1: large head circumference, short stature, hydrocephalus, epilepsy (due to intracranial tumors), congenital heart defects, hypertension, vasculopathy, and learning disabilities

5. Treatment-control symptoms
   a) surgery for bony deformity and some tumors (although tumors can grow back in greater numbers)
   b) surgery, radiation, and chemotherapy for malignant tumors
   c) intervention for children with learning disabilities
6. **Prognosis** - in general, most patients with NF1 have mild to moderate symptoms and a normal life expectancy

G. **PERIPHERAL NEUROPATHIES**

1. **Guillain Barré syndrome** (post-infectious polyneuritis)
   a) Rapid onset following an illness
   b) Distal weakness that ascends
   c) 50% cranial nerve involvement, DTR’s absent, sensory deficit minimal
   d) Most children recover completely

2. **Charcot Marie Tooth (Peroneal Muscular Atrophy)**
   a) A heterogeneous group of disorders characterized by progressive muscle wasting and weakness and sensory deficit (HMSN- hereditary motor and sensory neuropathy)
   b) Symmetric involvement of the distal extremities
   c) Most common inherited disorder of the peripheral nervous system
   d) 1 in 2,500 people; 3 to 5 X more common in males than females; accounts for 50% of pediatric cases of hereditary peripheral neuropathies
   e) Most common cause of neurologic cavus foot
   f) Chief complaints: falling, clumsiness, gait abnormalities, high arched foot, difficulty in fitting shoes, corns, calluses, metatarsalgia, and ankle sprains

   **Subgroups of CMT include CMT IA, CMT IB, and CMT II**

   **[A] CMT IA**
   1. Autosomal dominant and most common CMT group
   2. Presentation in first and second decade of life (average age of 12)
   3. HYPERTROPHIC, DEMYELINATING PERIPHERAL NEUROPATHY; degeneration of posterior columns of the spinal cord, spinocerebellar tracts and nerve roots; loss of anterior horn cells
   4. Decreased or absent ankle reflex, later absent patellar reflex
5. Mild distal sensory loss {EARLY SIGN – decrease in vibratory sensation}
6. Enlargement of post auricular nerve
7. **EMG & NCV HALLMARK TESTS FOR PERIPHERAL NEUROPATHY**; NCV is slowed suggesting demyelination
8. Peripheral nerve biopsy reveals onion bulb formation and segmental demyelination
9. Gait – HIGH STEPPAGE AND DROP FOOT
10. Genetic defect is a large duplication of proximal portion of the short arm of chromosome 17. The gene encoding a protein found mainly in Schwann cells, peripheral myelin protein {PMP22} has been mapped to this area
11. DNA BLOOD TESTING – detection of CMT IA duplication confirms the diagnosis and makes it possible to diagnose and rule out other family members

[B] CMT IB
1. Autosomal recessive, much rarer than CMT IA
2. Gene for CMT IB on the long arm of chromosome 1. Peripheral myelin gene [PO] is localized to this sight.
3. Early onset in the first or second year
4. Clinically similar to CMT IA

[C] CMT II
1. NEURONAL TYPE [AXONAL DEGENERATION]
2. Usually autosomal dominant
3. EMG AND NCV is consistent with axonal degeneration; **NORMAL NERVE CONDUCTION VELOCITY**. The NCV of the median nerve is used to distinguish between CMT I and CMT II. If the median nerve is less than 40 m/sec., it suggests CMT I. If the median nerve NCV is more than 40 m/sec., it suggests CMT II

[D] THE FOOT IN CMT
1. Cavus, cavovarus, tev, hammered digits, clawed digits
2. The CAVOVARUS FOOT is seen with a forefoot equinus, contracture of the fascia, and a varus calcaneus.
3. Peroneals are the first muscles to weaken; followed by tib. ant., intrinsics, and gastrocs. Hand muscles weaken years after lower leg muscle weakening

[E] TREATMENT
1. Stretching and strengthening exercises
2. Foot orthoses and ankle foot orthoses
3. Molded shoes
4. Surgery to balance the foot and reduce pain. In skeletally immature feet, soft tissue and bony procedures are performed [plantar release and calcaneal osteotomy]. In skeletally mature feet with SEVERE cavovarus, a triple arthrodesis is performed as a salvage procedure to restore hindfoot stability. A posterior tibial tendon transfer is also sometimes used to reduce a drop foot.

3. Dejerine-Sottas Disease Type III HMSN
   a) Similar to CMT
   b) Scoliosis, pupillary abnormalities

4. Refsum’s Type IV HMSN
   a) Anorexia, gait abnormalities, ichthyosis, eye problems, anosmia, hearing deficit, pes cavus
   b) Deficiency in fatty acid, metabolism, reduce dietary phytanic acid

5. Riley Day Syndrome HSN (hereditary sensory neuropathy-familial dysautonomia)
   a) Onset in infancy, autonomic dysfunction, Jewish children of eastern European descent
   b) Failure to respond to pain

6. Toxic Neuropathies
   a) Sciatic Neuropathy
      1. Result of injection into buttocks, near sciatic nerve
      2. Paralysis within days
      3. Long term effects: limb growth retardation, pes cavus, trophic ulceration
   b) Antibiotic-induced, antimetabolite, heavy metal (eg. lead exposure, seizures, headache and ataxia in children under 5, dropfoot in children over 5)

7. Friedrich’s Ataxia
   a) spinocerebellar hereditary degenerative disorder, progressive ataxia, involvement of the peripheral nerves and posterior columns of spinal cord
   b) onset in childhood, gait instability, frequent falling, pes cavus, hammertoes, kyphoscoliosis, gradual loss of vibration and proprioception, DTR’s decreased or absent
8. **Hereditary Neuropathy with Liability to Pressure Palsies**
   A] Autosomal dominant recurrent **MONONEUROPATHY** related to trauma
   B] genetic defect is a deletion of DNA at chromosome 17
   C] sural nerve biopsy reveals tomaculous or sausagelike swellings of myelin sheaths

**H. FLOPPY INFANT SYNDROME**

1. **Hypotonia** (low muscle tone), “rag doll”
   a) Infant shows little spontaneous movement, unusual postures
   b) Does not adjust to postural demands
   c) Decreased resistance to passive movement and increased range of motion of joints

2. **Clinical Features**
   a) In infant, marked head lag and floppy limbs and trunk in prone horizontal suspension
   b) Traction response – head lag
   c) Hold him under axilla “slips through” – vertical suspension test
   d) Infant supine, pick up each limb and feel resistance
   e) “Frog-legged” posture
   f) In older child, delayed motor milestones

3. **Multiple etiologies** – look at each level of nervous system
   a) **Cerebrum and diffusely acting lesions**
      1. Cerebral Palsy – most common cause of infantile hypotonia common in spastic and athetoid 6-12 months
      2. Chromosome abnormalities (Trisomy 21), hypothyroidism, inborn errors of metabolism, mental retardation syndromes (Prader Willi, Lawrence Moon Biedl)
   b) **Spinal Cord**
      1. Transecting lesion – myelomeningocele
2. Diffuse lesion – anterior horn cell loss throughout spinal cord – Werdnig Hoffman (spinal muscular atrophy), Kugelberg Welander (juvenile form, gait abnormalities, loss of bulk of thigh muscles, fasciculations)

c) Peripheral Nerves

1. All types of peripheral neuropathies (distal hypotonia, hyporeflexia, palpably enlarged nerves, distal sensory impairment)

d) Neuromuscular Junction – myasthenia gravis

e) Muscle Disease – muscle biopsy essential