Paediatric Neuromuscular Disorders

May present in the first year of life; but most likely after the child walks

There are several levels where weakness may intrude;

- **BRAIN**
- **SPINAL CORD**
- **ANTERIOR HORN CELLS-** eg. spinal muscular atrophy
- **PERIPHERAL NERVE –** eg. neuropathy of diabetes
- **NEUROMUSCULAR JUNCTION-** eg. myasthenia gravis, polymyositis
- **MUSCLE-** eg myopathies, muscular dystrophy

QUESTIONS: ***family history?*** *Unkle died young in a wheelchair?*

EXAMINATION:

HYPOTONIA:

child is floppy; but floppily weak, or floppily strong?

Floppy weak = lower motor neuron problem; not moving much at all

Floppy strong = Downs syndrome, or other central problem (tone preserved)

! *dysmorphic features suggest that the hypotonia is CENTRAL*

As always in neurology. the idea is to determine where the lesion is. REFLEXES and WEAKNESS are probably the best markers.

Central features:

- **Impaired cognition, seizures, decreased intellect**
- **Poor feeding, poor sucking**
- **Micro or macrocephaly (which is a dysmorphic feature)**
- **Normal or brisk reflexes**

Peripheral features:

- **Alert baby, but not moving!**
- **Weakness**
- **Depressed or absent reflexes**

PATTERN OF WEAKNESS:

PERIPHERAL suggests **NEUROPATHY**

PROXIMAL suggests **MYOPATHY**

GOWERS' MANOEUVRE:

Get the kid to get up off the floor. TIME IT. Normal child will do it in 2-3 sec. An objective sign of proximal muscle weakness is when the kid cant get up without pushing off their knee with their hands; that's a positive Gowers.

CALF ATROPHY or HYPERTROPHY?

ATROPHY: probably neuropathy, peripheral nerve damage

HYPERTROPHY: probably a muscle issue; the calf is replaced with fatty scar tissue

GAIT:

Waddling, shifting weight on to each leg as you step- sign of gluteal weakness, eg. Duchenne's

Able to heel walk? – a test for Achilles tendon shortening

Intellect:

Duchennes means the abnormal protein is also found in the brain.

Thus: a STATIC (non-progressive) ENCEPHALOPATHY ensues;

- IQ is on average 10-15 points below peers.
- THESE ARE DULL CHILDREN.

INVESTIGATIONS:

- Serum Creatine Kinase
  - **(200 to 100 = probably myositis)**
  - **(over 1000 = probably dystrophy)**
- MUSCLE BIOPSY IS THE GOLD STANDARD;
  - take biopsy from deltoid or quadriceps.
  - NERVE tissue biopsy can also help: harvest the sural nerve, at the tibialis anterior.

ELECTROMYOGRAM: looks for abnormal response in the muscle after a normal nerve conduction.

Nerve Conduction Studies: slow nerve conduction suggests a neuropathy.

Immunohistochemistry, electrophoresis, Western Blot test, PCRs etc - *its all about identifying the dystrophin,*