Objectives

- Get an appreciation of the spectrum of the Pediatric Neurologist
- Discuss some of the common disorders seen in Peds Neuro
- Discuss some common Peds Neuro emergencies
- Appreciate some of the diagnostic and therapeutic strategies used by Pediatric Neurology
- Have fun doing all these!!

What do we do?

- Provide Neurologic care for children
- Have residency training in both Pediatrics and Neurology
- Get to see an amazing breadth of human disease, primarily as outpatients
- Participate in a revolution of new neurologic diagnoses and therapies

Breadth of Disorders

- Benign
  - Headache, tics
- Life-threatening
  - Epilepsy, metabolic disorders, tumors
- Devastating
  - Epilepsy, autism, mental retardation

Working with Children

- Have fun with them!!!
  - History
  - OBSERVATION
  - Examination
  - Testing
    - Imaging
    - Genetic
    - Metabolic/Biochemical
Where is the problem?

Localization within the neural axis

- Brain
- Spinal Cord
- Nerve root
- Peripheral Nerve
- Neuromuscular Junction
- Muscle

Types of Seizures

- Partial
- Complex partial
- Partial, secondarily generalized
- Primarily Generalized

Complex partial seizure, secondarily generalized

14 year old boy swerved to avoid a bus while riding his bicycle and struck his head on a tree. Twenty minutes later he developed confusion, jerking in his left thumb that over the next minute spread to involve his left arm, then shoulder and face, then all extremities. Localized seizure onset, then Jacksonian march with contiguous spread through the right and then left cerebral cortex.

Afterwards, he had a headache, asymmetrical face and was unable to use his left arm. Postictal Todd’s paralysis due to exhaustion of involved cortex.

Generalized Seizures

- Discharge affects the entire cortex simultaneously and causes unconsciousness at the outset.
- There is usually no warning before seizure
- No head or eye deviation or focal post-ictal weakness
- Generalized tonic-clonic seizures can start as partial OR generalized
- Myoclonic jerks, head drops, simple staring or sudden falls are common “minor motor” sz.
### Features of Absence Seizures

- Abrupt impairment of consciousness = “staring”
- Brief duration, rarely > 30 seconds
- Variable motor components
  - None
  - Slight blinking and eye deviation
  - Subtle clonic movements
  - RARE Automatisms
- Often occur multiple times per day
- Post-ictal drowsiness is RARE

### Febrile Seizures

- About 4% of children will have a febrile seizure
- Seizures are typically brief, generalized tonic-clonic convulsions at the start of an illness
- Age of onset: 5 months to 3 years.
- A family history of febrile seizures is common.
- EEG is normal (if ordered)
- Atypical febrile seizure-focal, duration
- 2-3% of these children will later develop epilepsy

### Evaluation of Febrile Seizures

- Always consider the possibility of meningitis; look for stiff neck, headache, photophobia.
- Consider LP in any child less than 18 months old, even in the absence of “meningeal signs”.
- Treat underlying etiology for fever as appropriate.
- Atypical febrile seizures should have a complete neurologic evaluation and imaging.
- Typical febrile seizures may not need further investigation.

### Treatment of Febrile Seizures

- Treat prolonged febrile seizure as with any status epilepticus
- Consider rectal diazepam (Diastat) for home use if prolonged seizure
- Discuss seizure precautions and first aid
- Counsel regarding recurrence risk.
- Use aggressive fever control for future illnesses. (evidence says no benefit though)
- Overall a common but benign entity

### Case

- A 6 months boy, develops “startle-like” spells in clusters, especially after awakening.
- PMH: Healthy infant
- FH: Negative
- Exam: Hypopigmented skin lesions, poor tone, brisk reflexes
- EEG: Hypsarrhythmia

- What is your differential diagnosis?
- What is hypsarrhythmia?
**Infantile Spasms**
- SEIZURE: Sudden forward (or backward) flexion of head and trunk, with extension of arms.
- ONSET: Age 3-8 months.
- ETIOLOGY: Idiopathic, Cryptogenic, Symptomatic
- EEG: Hypsarrhythmia is classic finding.
- TREATMENT: ACTH, topiramate, vigabatrin, pyridoxine
- PROGNOSIS: Poor-Guarded, many patients severely retarded. Prognosis best for idiopathic etiology.

**Tuberous Sclerosis**
- Autosomal dominant – variable expression
- 65% arise from new mutation
- Features:
  - Mental retardation (approx 50%)
  - Autism (25-50%)
  - Seizures (90%)
  - Infantile spasms (50%)
  - Skin lesions: Shagreen patch, Ash leaf spots, facial angiofibromas
  - Retinal tumors
  - Tuber
  - Subependymal giant cell astrocytomas
  - Renal cysts and tumors

**Tuberous Sclerosis Stigmata:**
- Facial angiofibromas (Adenoma sebaceum)
- Shagreen patch is most commonly found in the lumbosacral region.
- Hypopigmented macules

**Neurofibromatosis**
- Type I
  - Autosomal Dominant (1: 4000)
  - 50% have disease due to new mutation:
  - 10% get malignancy (neurofibrosarcoma)
  - High penetrance but variable expression
  - Café-au-lait macules
  - Axillary freckling
  - Optic nerve gliomas
  - Lisch nodules

**Neurofibromatosis 1 Stigmata:**
- Skin neurofibromas
- Nerve neurofibromas
Spinal Cord

Spinal Muscular Atrophy (SMA)
Type 1 – Werdnig-Hoffman Syndrome
- Onset at birth to 6 months, never can sit
- Exam
  - Alert, interactive infant
  - Hypotonic, lies in frog-leg position
  - Paradoxical respirations (diaphragm weak)
  - Tongue fasciculations, weak suck
  - Absent reflexes
- Death from respiratory failure by age 6-12 months
- DX – Genetic testing available (SMN gene), EMG, muscle biopsy if genetic testing negative. Autosomal recessive, variable expression

Transverse Myelitis
- Demyelinating disorder
  - Rapid onset symptoms
  - Usually thoracic level
- Symptoms
  - Sensory loss, weakness
  - Altered reflexes
  - Bladder dysfunction
- Diagnosis
  - MRI of spinal cord
  - CSF
- Treatment and Prognosis
  - IV Steroids
  - Most have an very good to excellent outcome
  - Rare to later develop multiple sclerosis

Peripheral Nerve

Brachial Plexus - Erbs Palsy
- Upper Plexus injury, C5 + C6 = proximal dysfunction
- Traction of head/shoulder during delivery
- Shoulder adducted, internally rotated, elbow extended, forearm pronated, no biceps reflex
  - Weakness of deltoids, biceps, brachioradialis
- Consider hemi-diaphragm paralysis with respiratory distress (phrenic nerve involvement)
- Treatment
  - Passive stretching/ROM
  - Surgery (nerve graft) if no improvement by 3-6 months
- Prognosis very good for most patients

Brachial Plexus Injury
Guillain-Barre Syndrome

- Common presentations:
  - May start with painful paresthesias
  - Weakness, legs>arms (ascending)
  - Absent DTRs
  - Ataxia, facial weakness, cranial nerve palsies
- May have preceding viral or diarrheal illness
  - Mycoplasma, Campylobacter
- Diagnostic testing
  - CSF: elevated protein, normal glucose, few or no cells
  - EMG/Nerve conduction testing: Absent F-waves, demyelination

GBS-Treatment

- Treatment
  - IV immunoglobulin
  - Plasmapheresis
- Complications
  - Respiratory failure-monitor pulmonary function!
  - Aspiration from bulbar weakness
- Prognosis
  - Majority recover fully
  - May take months for full recovery

Myasthenia Gravis

- Onset in adolescence girls>boys
- Fatiguing weakness on exam:
  - Ptosis, diplopia, limb weakness
- Pathophysiology: antibodies to ACh receptors at NMJ, MuSK
- Diagnosis
  - Anti–AChR radioimmunoassay
  - Edrophonium Chloride (Tension) testing (not recommended)
  - EMG/NCV: Repetitive Nerve Stimulation with decremental response, single fiber electromyography

Myasthenia Gravis

- Treatment
  - Pyridostigmine, neostigmine, steroids, immunosuppressants, thymectomy
  - Acute exacerbations may require ventilatory support, plasmapheresis
- Neonatal form
  - Passive transfer of IgG antibodies from affected mother
  - May require medications few weeks only
Muscle

Muscular Dystrophies
Duchenne and Becker

• Diagnosis
  – Massively elevated CPK is virtually diagnostic
  – Genetic testing available
  – Muscle biopsy shows absent/reduced dystrophin

• Treatment
  – Physical therapy
  – Nutrition
  – Steroids seem to improve strength and maintain ambulation, risks vs. benefits?

• Prognosis death/ventilator dependence by adulthood for Duchenne, survival into adulthood for Becker type.

Gowers’ Sign

• Gowers sign. A boy with hip girdle weakness due to Duchenne muscular dystrophy.

Questions? Comments?

LIFE IS A JOURNEY, ENJOY THE TRIP