On the Lookout for Zebras

Primary care's vital role in detecting illnesses that impact function

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Disclosures

- Prior individual stock options
- Ochsner PM&R physician

Objectives

Review of

- Illnesses causing functional impairment
  - Focusing on infants and adolescents
- Local/regional data, where available
- Key Findings

OK but what is PM&R

Post-WW2

- Huge number of soldiers returning with functional impairments
- *Functionally impaired persons are marginalized persons, not by definition, but by social impact*
- Began to apply these principles to other groups
  - Amputation
  - Acute and chronic MSK ailments
  - Neuropathy
  - TBI
  - SCI
  - Epilepsy

Know Where You're Hunting

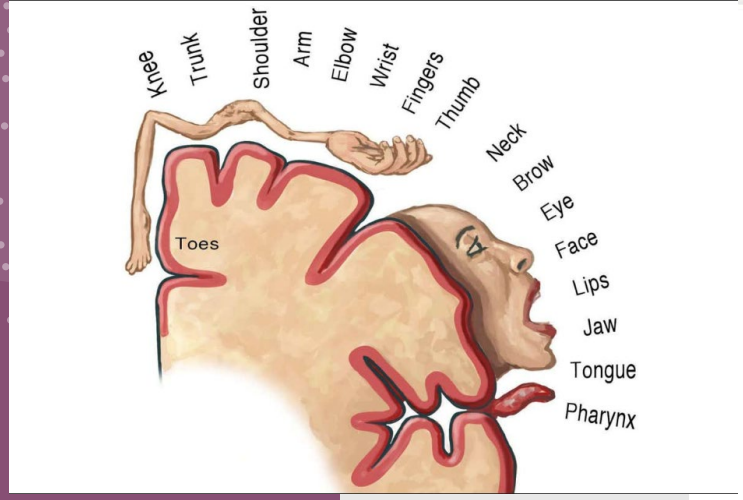
"When you hear hoofbeats..."

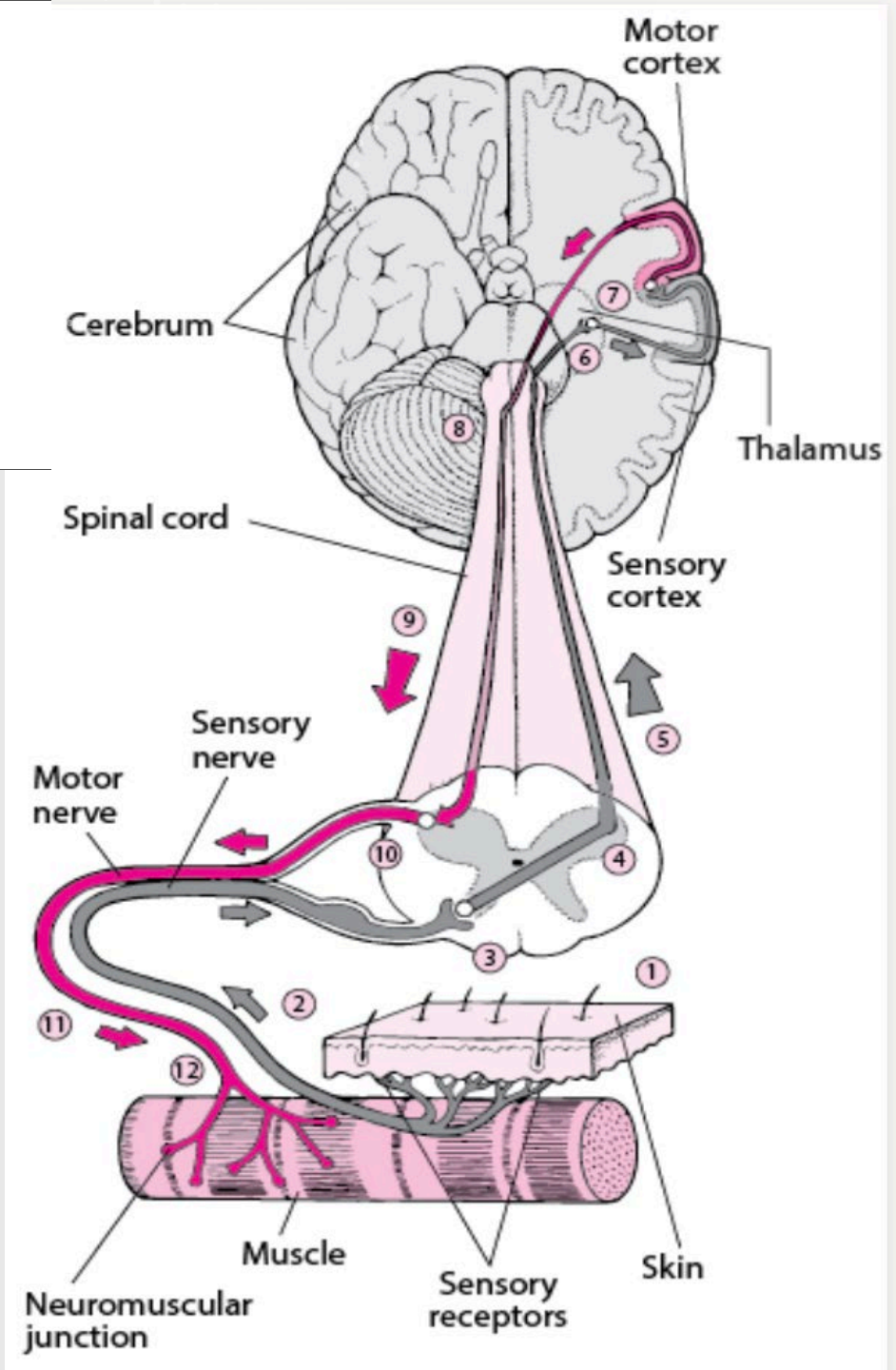
What makes a zebra rare?

Know Where You're Hunting

- PCP often first to encounter neurological and muscular disease
- Incidence in Louisiana (or USA)
- CP[1-3]:
  - Significantly more common among black children
  - 764,000 children and adults living with CP in the US
  - 8000 – 10000 US babies born with CP/annum*
- DBMD[4,5]
  - 2010; prevalence of 1 in 3,500-5,000 males aged 5 to 9 yo
  - DMD 3x > BMD
- SMA
  - Type 1: ~5-6 births/yr
- Ages at diagnosis
- Once ID'd, siphoned away into complex care paradigm

When in Doubt, Map it Out





Newborn Screening

State-Based

- Louisiana
  - Disorders recommended by the HHS
    - (ACHDNC)

3-methylcrotonyl-CoA carboxylase deficiency
3-OH 3-CH3 glutaric aciduria
Argininosuccinic aciduria
Biotinidase deficiency
Carnitine uptake defect
Citrullinemia
Congenital adrenal hyperplasia
Critical congenital heart disease
Cystic fibrosis
Galactosemia
Glutaric acidemia type I
Glycogen storage diseases type II (Pompe disease)
Hb S/C disease
Hb S/β-thalassemia
Hearing loss
Holocarboxylase Synthase Deficiency (Multiple Carboxylase Deficiency-MCD)
Homocystinuria
Isovaleric acidemia
Long-chain L-3-OH acyl-CoA dehydrogenase deficiency
Maple syrup urine disease
Medium-chain acyl-CoA dehydrogenase deficiency
Methylmalonic acidemia - Cbl A B
Methylmalonic acidemia – mutase deficiency
Mucopolysaccharidosis type I
Phenylketonuria
Primary congenital hypothyroidism
Propionic acidemia
Severe combined immunodeficiency
Sickle cell anemia
Spinal muscular atrophy
Trifunctional protein deficiency
Very long-chain acyl-CoA dehydrogenase deficiency
β-ketothiolase deficiency

Neurological

Brain

- Cerebral palsy/Stroke[7]
  - Muscle tone
  - Dev delay
- Neoplasm

Spine

- Spinal dysraphism / Spina bifida / MMC
- Tethered cord[9]
  - 40-60% a/w spinal dysraphism
  - Mean age 6.5yr
  - Bowel/bladder, weakness, edema, back/leg pain
- AFM[8]
  - Peak/even years – median ~5 yo
  - Off-peak/odd years – median ~8 yo
- SMA
  - Anterior horn cell

Neonatal
- Epilepsy
- Disordered consciousness
- Gross motor impairment
- Apnea

Infantile
- Early hand preference/fisting
- Focal seizures
- Impaired consciousness

Toddler
- Sensory disturbance
- Psychomotor agitation

Musculoskeletal Disorders

Do we wait until Gower sign for diagnosis?

- 1st muscle group affected is neck flexors
- Treatments as early as possible
  - Pre-symptom onset?

Musculoskeletal Disorders

Muscular dystrophy[4]

- Onset-dx 2.5yr
- Avg age at dx 5yr
- Evolving Tx

Gait abnormality

- Toe walking
- In-toeing
- Out-toeing
- LLD

Myopathies

Skeletal dysplasia[6]

- Skeletal shape and size
- >350 distinct skeletal disorders
- Joint Laxity

Congenital deformity

- Arthrogryposis
- ABS
- TEV

After Discovery

Work-up

Referral

PM&R, Neuro, Ortho, Complex Care

Therapies*

Medical travel

Admissions

Specialized testing

Long-term

Coordination

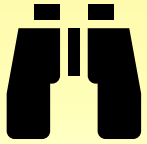
Specialized schooling

Care facilities

Outcomes

- It’s up to all of us!
  - There is no I in “team”
  - But there is a u & me in “Ochsner Medical Center in Louisiana”
- Time-limited treatment
- Screening updates

Keep a sharp eye!



Citations and Valedictions

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