Neurological Comorbidity

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“Neither I nor my spouse/partner has a relevant financial relationship with a commercial interest to disclose.”
Goals

• Discuss Office Evaluation of child with Autism Spectrum Disorder (ASD)
  • Testing
  • Pain
  • Motor
  • Sleep
  • Seizures
Medical Evaluation and Exam

• Goals
  – Identify genetic disorders
  – Identify concurrent diseases
Testing: EEG

- Routine EEG abnormalities –
  - up to 66% of ASD  (Mulligan and Trauner , JADD 2014)
- Routine testing not recommended for all
- Overnight EEG for language regression- ESES
Testing: MRI

- Not recommended for ASD
- Abnormal neurological examination
- Focal seizure
- Head size

- Getting a child in scanner
- Scanner sounds: https://www.youtube.com/watch?v=9GZvd_4ot04
- MRI social story: http://nmr.mgh.harvard.edu/transcend/kid_mri.html
Metabolic Studies

• Blood
  – Amino Acids
  – Lactate
  – Pyruvate
  – Thyroid
  – Cholesterol

• Urine
  – Organic Acids
When to do further testing?

- Dysmorphism, macrocephaly- child doesn’t look like parents
- Regression after age 3
  - Loss of developmental milestones
  - Loss of skills: ADL’s
  - Increased self stimulation/ self injurious behaviors
- Failure to make progress after a year
  - Developmentally appropriate, individualized program
- Other concerning history
  - Seizures
Pain Expression - Behaviors

- Pain looks different in each child
- Nonverbal children express pain nonverbally
- Child is more autistic...
Pain Expression

- Vocal:
  - Screaming
  - Verbal tics
  - Throat clearing, swallowing
  - Echolalia or scripted speech referring to
    - body parts, doctors, or pain
  - Moaning
  - Sobbing/crying without reason
  - Aggressive vocalization
Pain Expression

- Behavior
  - Irritability
  - Sleep disorder
  - Non-compliance
  - Self injurious behaviors

- Body Movements
  - Facial Expressions
  - Dystonia
  - Mouthing
  - Gait change
  - Posture change
  - Tics
  - Dyskinesia
Sleep

• Most frequent complaint of parents of ASD children
• 53-83% of children with ASD
• More common than other psychiatric disorders
Sleep

• Disorders of:
  – Sleep onset
  – Maintaining sleep
  – Early morning awakening
Sleep

– No differences noted in ASD subtypes
– Strong behavioral component around learning to sleep
– Subjective findings lead to increase in ASD behaviors and irritability

Sleep Disorders

• Etiology:
  – Core deficits of ASD:
    • Behavior
    • Anxiety
    • Inflexible routine
    • Self regulation
    • Challenges with limit setting
  – Typical childhood sleep disorders
    • Restless leg syndrome
    • Obstructive sleep apnea
Sleep Disorders - Cause

• Biological
  – Circadian rhythms
  – Melatonin dysregulation
    • Genetic findings
    • Metabolic findings

  – Melke et al (2009)
Sleep Disorders

• Concurrent medical disorders
  – Epilepsy
  – Gastro-intestinal disease
• Psychiatric comorbidities
  – Anxiety
  – Mood disorders
• Pain
Treatments: Behavioral

- Establishing a Sleep Routine
- Provide a comfortable sleep environment
  - Temperature
  - Clothing textures
- Establish consistent bedtime routines
  - 4-6 bedtime activities
- Maintain a regular schedule
- Teaching your child to fall asleep alone in his/her bed
  - Gate or bell to alert parents that child is leaving room
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Sleep Treatments

• Behavioral
  – Autism Speaks Toolkits
  – Sleep routine: training in bedroom
  – Bedtime pass
Sleep

• Medical
  – Ensure no medications which impair sleep
    • Caffeine
    • Stimulants
  – Melatonin (1-6 mg)- short or long acting
  – Clonidine (0.05-0.2 mg)- short or long acting
  – Guanfacine (0.5 -1 mg)-short or long acting
  – Trazodone (25-100 mg)
  – Others- diphenhydramine, mirtazapine
  – Medication for comorbid illness...
Motor

- Repetitive behaviors
  - No specific treatment

- Gait
  - Toe walking, ataxia

- Dyspraxia-

- Hypotonia- low tone

- Catatonia
Motor

- Motor delays
  - Gross motor - walk, sit
  - Fine motor - imitation
  - Dyspraxia
- Gait
  - Toe walking
- Hypotonia-
- Repetitive behaviors
- Catatonia
Epilepsy

• Rate in ASD is 2.4-19-37%

• Variation depends on
  – Age: bimodal: 0-5 year old, Adolescence-Adult
  – Intellectual disability -24 vs. 8 % (Amiet’s Meta-Analysis 2008)
  – Females vs. Males – 34 vs. 18.5 % (Amiet 2008)
  – Etiology
  – Regression
  – Subtype of ASD (DSM IV)
  – Simplex vs. Multiplex
    • (Amiet 2013)
  – Other- prematurity

Francis et al. Pediatric Annals 2013
EEG Controversy

• Controversy around spikes in EEG in absence of epilepsy (Spence 2009)
  – Increased risk of epilepsy? (Kim et al 2006)
  – Relationship between spikes and ASD behaviors? ()
  – More seen during sleep (Chez et al 2006)

• Meaning of interictal epileptic discharges
  – Many studies: Spike vs Spike and Wave
  – Worse behavior / aggression(Turk et al 2009)
  – Developmental regression? +/-
  – Attention

• El Achkar et Spence Epilepsy and Behavior, 2015
Epilepsy

• There is no one seizure type in autism
  – Febrile, Benign, Focal and Generalized
• There is no one typical EEG in autism
• Effect of epilepsy on ASD?
  – Increased ASD behaviors (Mulligan and Trauner 2014)
  – Worse motor skills, Attention, (SIB) in ID and non ID population (Viscidi et al 2013)
Epilepsy

- Staring Spells look like seizures
  - Use videos to help diagnose
Epilepsy Treatments

• Medications
  – No special medications for autism
  – Increase irritability – levetiracetam
  – Pyridoxine- 100 -200 mg BID

• Diet
  – Ketogenic diet
  – Low Glycemic Index diet

• Surgery

• VNS

• Medical Marijuana
  – Cannabidiol, CBD, hemp
Take Home Message

• Pain has many faces
• Sleep disorders are common
• Behavior treatments and look for underlying cause of insomnia
• Staring vs seizures
Project ECHO Autism

- Connects primary care providers to a team of experts in autism and developmental disabilities
- Case-based learning builds confidence and knowledge in the PCP to care for children in their practices
- Improves access for families
- Email me to learn more: aneumeyer@mgh.harvard.edu
The Lurie Center

Mission

Our goal is to advance knowledge of autism spectrum and related disorders and to rapidly translate discovery into exceptional care over a lifetime.

Multidisciplinary clinic for children and adults with autism and developmental disabilities

What disciplines?

Where?