Medical Issues in Autism Spectrum Disorders

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Goals

• Discuss medical work up of autism
• Regression in autism
• Explain comorbidities in autism
  – Neurological
  – Sleep
  – Gastroenterological
  – Other

• Lurie Center
Autism Spectrum Disorders

Social Interaction  Communication  Atypical Behaviors

Leo Kanner-1943
Hans Asperger -1944

Clinical Features

• Seizures develop in 25%–33% of cases
• 39-61% may function in the mentally retarded range (MMWR-2007)
• High incidence of non-right handers
Rett Syndrome - MECP@ Xp28 video

Management of Autistic Children

• **Diagnosis**
  • Determine need for any additional studies/evaluations
  • Identify appropriate therapies and service providers
• **Advocate** for child and family within the health care and educational systems
• Provide ongoing **monitoring** of developmental progress and support for parents, teachers and therapists.
Neurological Examination

- Head Circumference
- Muscle tone - low or high?
- Woods lamp examination for ash leaf spots
- Dysmorphology
- Genetic Screening- Microarray and Fragile X

Genetic syndromes associated with Autism

- Boston Autism Consortium Study- 15%
- Fragile X syndrome: 2%–3% of ASD cases
  - Up to 20% with ASD
- Tuberous sclerosis 17%–60% with ASD
- Angelman, Sotos, NF1, Smith-Lemli-Opitz Syndromes (5% of ASD),
- Rett Syndrome
- Mitochondrial Disorder
Autism Evaluation:

When to order additional studies/referrals?
- Examination findings-dysmorphic
- Regression after age 3
- Other concerning history
- Seizures- 25% of ASD
- Failure to make progress after a year in a ‘good’ program

Neurological Assessment

Additional studies:
- Chromosomal testing (15q, Rett, Microarray)
- Electroencephalogram (EEG):
  - Abnormal in 6%–35%
  - Sleep deprived or overnight study to rule out ESES
- CNS Imaging studies (MRI)
- Metabolic studies
  - Amino acids, organic acids, lactate, pyruvate, lead, thyroid
Regression in Autism

• A loss of skills
  – language, ADL’s, motor, social
• Change in behaviors
  – Increased self stimulatory behaviors
  – Increased self injurious behaviors

Why Regression?

• Medical Illness
• Change in environment
  – School program, staff, family changes
• Medical illness
  – Exacerbation of underlying condition causing autism (TS, PKU, mitochondrial etc)
  – Typical autism related condition
  – Pain
Expression of Pain

- Nonverbal children express discomfort nonverbally
- Exacerbation of “autistic behaviors”

Pain Expression- Non Verbal

- Motor behaviors:
  - Grimacing, wincing
  - Head, Chest, Belly banging or pressing behavior
  - Gritting teeth
  - Constant eating behaviors
  - Mouthing
  - Gait change
  - Posture change
  - Change in tics
  - Dyskinesias
Pain Expression- Vocal behaviors

• Verbal tics
• Screaming
• Throat clearing, swallowing
• Echolalia or scripted speech referring to
  – body parts, doctors, or pain
• Moaning
• Sobbing/crying without reason
• Aggressive behavior

Pain Expression- Other

• Irritability
• Sleep disorder
• Non-compliance
Autism Treatment Network

- Network of now 15 Hospitals Medical Centers dedicated to developing a model of comprehensive medical care for children and adolescents with ASD
- Patient care- comprehensive coordinated care model
- Comorbid Conditions associated with ASD
- Network Activity: Best Practices
- Database Patient Registry
- Clinical research

Medical Co-morbid conditions

- Sleep
- Gastro-intestinal
- Epilepsy
- Allergy
- Other
  - Bone?
Sleep

• Sleep disorders (Krakowiak -2008, Couterier- 2004)
  – 53-78% of children with Autism
  – 26-32% of typical children

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

Sleep Disorders - Open the Can of Worms…

• Why ask about sleep?
  – Poor sleep exacerbates autistic behaviors
    • Learning, repetitive behaviors, anxiety, aggression
  – Poor sleep impairs function during day
    • Effect on child, class, teachers, family
  – Parents don’t always tell their MD’s about sleep
  – Parents need sleep too!
Sleep Disorders:

- Concurrent medical disorders
  - Epilepsy
  - GI disease
- Psychiatric comorbidities
  - Anxiety
  - Mood disorders

Sleep Disorders: Etiology

- Sleep disorders
  - Sleep apnea
  - Restless leg syndrome
- Behavior
  - Poor sleep habits
  - ASD core deficits: emotional regulation and communication
  - Anxiety
  - Melatonin in ASD
Establishing a Sleep Routine

• Provide a comfortable sleep environment
• Establish consistent bedtime routines
• Maintain a regular schedule
• Teaching your child to fall asleep alone
• Avoid naps in older children
• Daytime activities to improve sleep

• Medication trials

Food Intolerance

• Food allergy is common in children
  – 5-8% prevalence (Sampson, 1999)
• Food allergy is reported in 36% of autistic children (Lucarelli, 1995)*

  *small study but not biased by presenting symptoms

• Lactose and sugar intolerance
Gastrointestinal Disease

- 50% of parents report GI symptoms in their autistic children (Lightdale 2001)
- Widespread GI pathology (Horvath 1999)
  - Reflux (69%), Gastritis (42%), Duodenitis (67%)
- J Pediatrics January 2010 supplements on GI and autism evaluation and recommendations
  - Chronic constipation
  - Abdominal pain +/- diarrhea
  - Encopresis
  - Gastro-esophageal reflux
  - Abdominal bloating
  - Disaccharidase deficiencies

Epilepsy

- Prevalence-
- Second decade of life
- In females: Rett Syndrome

- Meaning of abnormal eeg (Barnes 2011)
- Meaning of interictal epileptic discharges-
  - worse behavior on CBCL
  - Attention
  - Sleep Arousal Index

- When to treat?
Sandifer Syndrome

- M. Kinsbourne - 1962
- Gastro-esophageal reflux with spasmodic torticollis and dystonic body movements
- Presents as dystonia, atypical seizures, torticollis
- Positioning of head and upper extremities provides relief from abdominal discomfort

Aggression/ Self Injurious Behaviors
Head Banging: Differential Diagnosis

- Any disorder of head, face, ears
- Dental
- Cervical
- Drug reaction
- Neurological
- Gastrointestinal

Mouthing: Differential Diagnosis

- Dental: Caries, Abscess, teething
- Reflux
- Otitis/Sinusitis
- Pica
  - Colitis, Anemia
Pseudo Seizure?

Clinical Vignettes

- Facial Palsy or new expression
- Mitochondrial Dysfunction
- Drug Reaction with aggression
- Chiari Malformation- or toileting problem
- PMS- or aggression
Movement Disorder

What to do?

- Don’t rush to psychotropic medications
- Look for underlying medical illness
- Look again for underlying medical illness
- Thorough history with family or other caretakers
- Collaborative Studies: Autism Treatment Network
Resources:

- www.autismconsortium.org: local resources and transition guide
- www.aap.org: Consensus statements
- www.firstsigns.org: for M-CHAT and screening
- www.cdc.gov/ncbddd/autism/index.html: autism and vaccines
- autismspeaks.org: general information
- www.luriecenter.org

Lurie Center for Autism

- Multidisciplinary Clinic-
  - Neurology
    - Epilepsy
    - Adult and Child Developmental Pediatrics
  - Adult Autism Care
  - Psychiatry
  - Gastroenterology
  - Neuropsychology
- Educational Consultant
- Nurse practitioner
- Social worker
- Physical, Occupational and Speech therapy
- Augmentative Communication
- Resource specialists
Fun Reading List

• Thinking in Pictures by Temple Grandin
• Beyond the Wall by Stephen Shore
• Eye Contact by Tammy McGovern
• Curious Incident of the Dog in the Night-Time by Mark Haddon

Who We Are
Where work…

Boston and the Charles River
To the many children and their families who have taught me about autism.

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