Medical conditions associated with Developmental Disabilities

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Neurological Assessments of the Child with Development Disabilities

1. Obtain a medical and developmental history
2. Neurological examination and behavioral observation
3. Consider need for additional studies:
    a. Chromosomal/DNA analysis
    b. Electroencephalogram (EEG)
    c. Imaging studies (MRI, CT)
    d. Metabolic (blood/urine) studies

DD often involve more than the brain

- DD classically defined on the basis of cognitive, behavioral, language and processing modalities.
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- But DD may be more than a disorder of information processing, language and behavior.
- DD children, adolescents and adults can and often do have medical issues that have largely gone unrecognized and unaddressed.

Disruptive Behaviors

- Atypical and/or disruptive behaviors of varying descriptions and severity have often been seen as just “part of the disorder”.

- But are they???

What is the definition of “behavior”?

- The manner in which an organism behaves in reaction to social stimuli or inner need.
- Observable activity in response to an external or internal stimulus.
- Anything that the organism does that involves action or response to stimulation.

What do we know?

- Research indicates that typically developing children often show elevated rates of problem behavior in association with physical illness.
- Physical illness is common in persons with developmental disabilities (DD).
- Studies have documented significantly higher rates of acute and chronic medical conditions in DD persons as compared to the general population.

What medical conditions have been documented?

- Problem behaviors have been linked to conditions such as constipation, allergies, premenstrual syndrome, ear infections and urinary tract infections.

- Plausible explanation relates to degree of pain or discomfort experienced by the individual at the time rather than to the physical illness per se.

Monitoring pain & discomfort is a complex process

- DD persons often lack the the communicative and cognitive skills that would allow for the direct assessment of pain and discomfort using a patient scale, checklist and/or interview strategies.

- Recent data suggests that those with the most severe cognitive impairment and fewest communication skills are likely to experience the most pain over time (Breau et al., 2003)

Why have these been overlooked?

1) Longstanding assumptions about what develppmental disabilities are. Abnormal behaviors often interpreted as part of the clinical profile of the disorder.

2) DD individuals may not present with the same symptoms or “red flags” as their “neurotypical” peers. Medical history may not help us.

3) Many DD persons cannot tell us if they hurt/are uncomfortable nor accurately localize discomfort.

Weak Insights into Overall Health Issues

- Difficult to see beyond cognitive or behavioral features of the disorder
- Limited physiology research of organ systems other than brain in developmental disorders
- Limited vehicles for collaboration on health issues
- No uniform set of clinical measures or data base.

Associated Medical Concerns?

Seizures

Sleep disturbances -

Headaches

Gastrointestinal disorders

Genitourinary

Hormonal imbalance/endocrine dysfunction

Metabolic Disorders

Seizures - are they real?

- Often hard to tell - presentation may be atypical
- Routine EEG may not be helpful
- More prolonged EEG by high quality lab may help - the study is only as good as the person who interprets it.
- Use of video monitoring, MEG, other.
- Use of video taping

Sleep Disorders

- Problems with sleep onset or staying asleep
- Is this coming from the brain (centers of arousal)?
- Is this due to GI disorder? Acid reflux?
- Is this a respiratory problem? Does the child mouth breath suggesting big tonsils/adenoids?
- Sensory integration issues - needs deep pressure?
- Allergies, eczema?

Gastrointestinal Disorders

- Chronic diarrhea or constipation
- Feeding/eating disorder
- Change in sleep patterns
- Parents concerned about food allergies, need for special diet, yeast
- Possible abdominal pain/discomfort
- Behavioral changes or increased severity.

Clinical Signs of GI Disorders

- Gulping and facial grimacing
- Tapping on the chest or stomach
- Putting pressure on the abdomen
- Constant chewing on non-edible items - shirt sleeves, shirt neck lines, etc
- Frequent eating/drinking
- Any unexplained negative behavioral change, including aggression, self-injurious behavior, with or without GI symtoms.

Take Home Message

- Disabled children, adolescents and adults, even if they have some language/words, should be evaluated for possible GI disorders - IF they present largely or exclusively with behavioral symptoms, including sleep disorders.
- Disabled patients may not present with the usual GI symptoms.
- Do NOT assume that all behaviors are “behavioral” or pyschiatric in origin.

Endocrine/Hormonal Disorders

- Some disabled girls can have behaviors that worsens with onset or during adolescence.
- Many found to have imbalance between
- progesterone and estrogen. Can be treated.
- Small subset with Congenital Adrenal Hyperplasia
- Should we also be looking at teenage disabled boys?

Reason for GU referral

- Previously continent child becomes incontinent
- Usually a preteen
- May be a “spastic bladder”
- Treatment with Ditropan may be helpful

“Red Flags” for Metabolic Work-up

- Poor physical endurance
- Late walking (i.e. 24 months)
- Repeated regressions after age 2 1/2 years
- Dysmorphic features
- Making poor progress despite excellent services
- Qualitatively “different”
- Involvement of multiple organ systems

Mitochondrial Disorders

- Weissman, et al., December 2008
- 25 patients with ASD
- All later determined to have enzyme or mutatiion-defined mitochondrial dysfuntion.
- 21 subjects had non-neurological medical problems
- 19 subjects had constitutional symptoms, primarily excessive fatigue

Mitochondrial Disorders

- 32% - delayed motor milestones
- 40% - unusual patterns of regression
- 76% - abnormal levels of blood lactate
- 36% - abnormal levels of blood alanine
- 52% - abnormal levels liver function studies
- Most common electron transport chain disorders were Complex I (64%) and Complex III (20%)

Mitochondrial Disorders

- Although initially all subjects were identified as having Essential (Idiopathic) Autism, careful clinical and biochemical assessment identified features that differentiated them from children with Idiopathic Autism.
- This preliminary data suggests that a disturbance in mitochondrial energy production may underlie pathophysiologic mechanisms in a subset of ASD persons.

Psychopharmacology

- Approach to medication management
  - Rule out potential medical disorders first
  - Should never be first line of defense - should be used as an adjunct to other interventions.
  - Consider specific symptoms - depression, anxiety, OCD, impulsivity, ADHD, etc
  - Consider the risks and benefits of choosing and using any medication.

Psychopharmacology

- Family should find a psychopharmacologist with whom they are comfortable.
- Choice of medications may be influenced by training of provider
- Health care insurance may influence choice of medication.
- Consider medical risks, cost to the patient, potential invasive procedures (blood draws), tolerance of side effects, possible drug interactions and methods of administration.

Bullets

- Disabled individuals need/deserve appropriate medical care.
- May not present with typical symptoms.
- Changes in behavior or prolonged episodes of behavioral abnormalities merit a medical look.
- Many of these disorders are treatable.
- We need to learn the language and signs of pain/discomfort in non-verbal and sensory impaired disabled inidviduals.

Multidisciplinary Teams

- Given the complexity of the Developmental Disabilties, and the fact that theses disorders often involve many organ systems, important that these individuals are seen by doctors and therapists from differing disciplines.

Team Members

- Developmental Pediatrics, Internists, Neurologists
- Psychiatrists, Behavioral Psychologists
- Neuropsychologists, gastroenterologists
- Speech therapists, audiologists
- Occupational and Physical Therapists
- Education specialist, Family Resource Coordinator

Referral Resources

- Endocrinology
- Developmental optometry
- Allergy
- Ear, Nose and Throat (Otolaryngology)
- Medical genetics
- Urology/Nephrology
- Nutritionist/dietician

Summary
Medical Co-morbidities are important

- Improve quality of life.
- Better health leads to better outcomes.
- Subsets of disabled persons may be more specifically identified - genetically and/or metabolically.
- Understanding associated medical conditions could enhance our understanding of the neurobiology of Developmental Disabilities.

Progress

- Still many questions to be answered
- Will require researchers and clinicians with differing disciplines working together
- Heterogeneity will be a challenge. Need to define subgroups. Diagnosing females?
- Need to define biological markers to aid earlier diagnosis and treatment.