Medical Management of Children with Developmental Disabilities

Carlos A. Bacino, M.D.
Texas Children’s Hospital
Baylor College of Medicine
Causes of Developmental Disabilities

- Chromosome problems: Down syndrome, VCFS, Williams syndrome, etc.
- Cerebral palsy
- Rare genetic problems: Fragile X syndrome, Angelman syndrome, Prader-Willi syndrome, etc.
- Prenatal exposures, infections
- Metabolic diseases
- Unknown causes in 50% of cases
How can a genetics doctor help you and your child?
Typical Questions for the geneticist

- What is the diagnosis?
- What is the cause?
- What is going to happen in the future?
- What is the treatment?
Diagnosis and Counseling

• If a diagnosis is made, provide information about the condition, medical problems that may be seen and form a treatment plan
• Offer counseling about chance for condition to occur in future children
• Discuss reproductive options
• Provide information about preconception care such as taking folic acid, maintaining diabetic control, alcohol and smoking cessation
• Help with family support and coping
Medical problems seen commonly in children with developmental disorders

- Sleep problems
- Psychiatric illnesses
- Eating problems
- Gastrointestinal disorders: celiac disease, constipation
- Planning for the future
Sleep Problems are very common

Between 44% and 83% of children with developmental disabilities have sleep problems
Sleep Problems

• One of parents’ most common concerns

• Sleep is needed for children to function best

• Typical problems: difficulty going to and staying asleep, early awakening, morning drowsiness (difficulties wakening)
Results of Poor Sleep

- Mood: irritability
- Poor attention, learning problems, poor academic performance
- Accidental injuries
- Behavior: poor impulse control, overactivity
- Organic: impaired cardiovascular, immune and endocrine function
How to evaluate and findings

- Actigraphy (watch recording movement)
- Sleep study:
  - Records electroencephalogram (EEG), oxygen saturations (oxygen consumption), electromyogram (muscle movement recordings)
- Findings: epileptic activity, reduced rapid eye movements (REM), periodic leg movements, breathing problems: apnea (obstructive and/or central)
Obstructive sleep apnea

- Brief pause in breathing during sleep
- Can be due to face structure, low muscle tone, large tonsils, and obesity
- Symptoms: snoring, insomnia, excessive sleepiness, and changes in behavior during the day
Medical Care

- Treat medical problems: heart disease, thyroid problems, etc.
- Treat sleep problems: apnea
- Sleep hygiene
- Behavioral approaches
- Medications
Obstructive sleep apnea: Treatment

- Typical treatment is to remove the tonsils and adenoids
- Use of night time breathing mask (C-PAP)
- Sleep position
- Weight management
- In some cases give growth hormone
Medications

- Melatonin 3 mg (up to 25 mg) 30 minutes before bedtime
- Clonidine 0.025-0.05 mg before bedtime
- Risperidone 0.5 mg before sleep or BID
- Zolpidem (Ambien) 5-10 mg
- Antidepressants: Trazadone 25-50 mg
• Psychiatry definition: The branch of medicine that deals with the diagnosis, treatment, and prevention of mental and emotional disorders

• Psychiatric Problems: the chance to have a psychiatric problem is higher in children with developmental disabilities:
  - 18% to 38% in Down Syndrome
  - More than 40% in VeloCardioFacial Syndrome
Psychiatric Problems

• Different issues for children before and after they start puberty (around age 12-14 years)
• May not be linked to learning disabilities
• Both diagnoses now accepted: in the past was not accepted that mental retardation or intellectual disability could be associated with other psychiatric illnesses
Common Concerns and Symptoms

- Behavior and temperament traits in young children include:
  - Inflexibility, resisting change
  - Stubbornness, repeating and repeating same activity or words – perseveration
  - Increased motor activity and impulsivity non-compliance (tantrums)
  - Agitation/anxiety/disruptive
  - Repetitive movements
  - Peculiar sensory responding
  - Thought processing not typical
  - Social impairments: withdrawal, aggression
Factors that may make it more likely for a child to have a psychiatric problem

- Medical problems
- Frequent seizures
- Many hospital stays
- Hearing and vision are impaired
- Sleep problems
- Obstructive sleep apnea
- Thyroid problems
Disorders before Puberty (before age 13)

- Oppositional-Defiant and Disruptive Behavior Disorders:
  - Seen sometime before 36 months, the child becomes disruptive and aggressive over time.

- Stereotypy Movement Disorder:
  - Repetitive acts that impair function
  - Symptoms: speech difficulties, self-injury, disruptive, anxious, makes unusual speech sounds, refuses foods, unusual sensory seeking/responding, sleep disturbance.
Disorders after Puberty

Causes: More awareness of being different and having different abilities, lack of acceptance by peers, losses, grieving.

• Obsessive-Compulsive Disorder:
  - Repetitive, compulsive acts: rearranging objects, opening closing doors, switches, blinds.
  - Hording of worthless objects.
  - Perseveration - repeating words or a gesture

• Depressive Illness:
  - Depressed mood, decreased interest, slowing, fatigue, sleep disturbance, sense of guilt, poor appetite and weight loss.
When to Seek Help?

When to treat?

- Interfere with daily living
- Social withdrawal, apathy, slower movements, lose self care skills.

- Psychotic Illnesses:
  - Delusions (not to be mistaken for talking to imaginary friends)
  - Paranoia
Treatment: SSRI’s
(Selective Serotonin Reuptake Inhibitors)

- Certain brain chemicals called neurotransmitters are associated with depression: serotonin
- SSRIs help symptoms of depression by blocking the reabsorption of serotonin by nerve cells in the brain
- This leaves more serotonin available in the brain which helps to improve the child’s mood
- Depression: Prozac and dual re-uptake inhibitor (norepinephrine and dopamine): Bupropion
- Psychosis:
  - Delusions-hallucinations: Neuroleptic
  - Catatonia: Benzodiazepines
Feeding Problems

- Swallowing problems: due to neuromotor coordination in Down syndrome, Angelman or Prader-Willi syndromes.

Swallow studies needed to check for problems

- Anatomy: cleft lip/palate, large tongue, velopharyngeal incompetence. Can check with X-rays, MRI, CT, endoscopy

- Behavioral: child does not like certain food textures

- Problems are often due to a combination of swallowing problems, the child’s physical features and behavioral issues
Feeding Management

- Position the infant upright
- Breast feeding: improving seal
- Bottle feed, special nipples: Haberman nipples
- Occupational therapy to help the child with low muscle tone
- If child at risk to choke or inhale food into lungs make the formula or fluids thicker
- Solid foods: make a pureed or blended diet
- NG feeds, G-tube feeds (G-Jejunal feeds)
Gastrointestinal Problems

- Esophageal dysmotility: incomplete relaxation of the upper sphincter, reduced amount of movement to push food into stomach
- Gastroesophageal reflux disease (GERD) 15-75%: peptic acid esophagitis, burning pain
  - Child shows chronic irritability, crying, arching of the back
  - Vomiting or regurgitates food
What problems can happen with Gastroesophageal reflux disease (GERD)?

- Periods when stop breathing, laryngitis, asthma/wheezing, chronic cough, aspiration pneumonia.
- Duodenal dysmotility: relaxation of entrance to stomach and when food is pushed back into stomach from duodenum
- Retching: part of the vomiting reflex, associated with vomiting, sometimes after fundoplication
- Dumping syndrome: liquids rapidly go from stomach into small intestine. Also seen in children with Nissen:
  - Abdominal discomfort, sweating, fast heart rate, low blood sugar
Treatment

- **Medicines:** Prilosec, Prevacid
- **Surgery:**
  - Nissen fundoplication
  - Gastrostomy
- **Gastrojejunal tubes sometimes needed**
Constipation

• Chronic constipation: more than half of children

• Due to: long periods when they are not moving, children with low muscle tone, and low gastrointestinal movement

• To Diagnose: Barium enema, manometry, rectal biopsy
Constipation

• To Treat: increase fiber in diet and fluids.
  ■ Softener: lactulose or mild stimulant such as senna.
  ■ Disimpaction or remove stool by hand
  ■ Laxative: Sodium docusate or a Dietary Fiber: psyllium

• Avoid mineral oil and polyethylene glycol.
Planning for the Future

- At age 18, all U.S. residents are legally free of parental control
- Guardianship petition: establishes a legal guardian who can act in the parent’s stead if parent unable to continue care.
- Health Care Proxy or someone who will make health care decisions for the patient
- Establish a will
Other Issues (resources)

- Sexual-Reproductive
  - [http://aappolicy.aappublications.org/cgi/content/full/pediatrics;118/1/398](http://aappolicy.aappublications.org/cgi/content/full/pediatrics;118/1/398)
- Health Insurance
  - Medicaid and Medicare
- SSI (Social Security Income)
  - Provides monthly check if child medically eligible and family income low enough: [http://www.ssa.gov/pubs/10026.html](http://www.ssa.gov/pubs/10026.html)
Other Resources

- Early Intervention, preschool and school planning
- AAP Policy Statement June 2007: Provision of Educationally Related Services for Children and Adolescents With Chronic Diseases and Disabling Conditions. A multidisciplinary assessment within the school system is required in the initial evaluation of children to determine their eligibility for services within the educational system:
  http://aappolicy.aappublications.org/cgi/content/full/pediatrics;119/6/1218

- Family Support: Helping families raise children with special health care needs at home:
  http://aappolicy.aappublications.org/cgi/content/full/pediatrics;115/2/507
Thank you