

Recognizing Co-Occurring Conditions in People with Intellectual and Developmental Disability



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Course Goals:

1. Identify atypical presentations of medical illness in persons with Developmental Disability
2. Evaluate medical conditions from the perspective of common developmental syndromes
3. Increase awareness of medication side effects and interaction in the presentation of medical illness.

Thinking Inside and Outside the Box

- Individual
 - Age, Sex, Race
 - Common things are common
 - Symptoms
 - Diseases
- Syndrome Related-Atypical presentations
- Medication Implications

History

Get it where you can!

- The Patient
- Parents
- Residential staff
 - Nonmedical direct care professionals
 - Nursing
 - Day program staff
 - Therapists- OT,PT and Speech
 - Behaviorists

- Other Medical Providers
- Collaboration is KEY

Amy W.

Amy is a 32 year old female with the diagnosis of Autism. She lives at home with her parents and has a community worker who takes her out several times per week. She usually enjoys this but has been more reluctant to go out for the last few weeks.

She is minimally verbal and repeats a few words over and over. She has recently been noted to have **increased rocking and hand biting**. Her appetite has been good but she has a limited repertoire of foods she will eat. She has a history of seasonal allergy. She has a history of anxiety and aggression which has been treated successfully with Zoloft and Zyprexa. Several family members have colds.

- **Individual**
- Syndrome level
- Medication perspective

Individual

24 year old female with:

Anxiety

- Is it menstrual related?
- Are there changes in her life, stressors?
- Any concerns for abuse?

Seasonal allergy or cold?

- Individual
- **Syndrome level**
- Medication perspective

Autism and Autism Spectrum Disorders

Common Neurodevelopmental disorder
characterized by

- Impairment of social interactions and communication
- Restricted, repetitive and stereotyped behaviors and interests
- Multifactorial inheritance but is considered heritable
- Occurs in about 1 in 150 children
- Approximately 50% of patient with ASD have intellectual disability
- Often concurrent with other genetic disorders such as Down syndrome, Fragile X syndrome, Prader-Willi Syndrome and others.
- Associated with other medical disorders including
 - Seizure Disorder
 - AD/HD
 - Sleep Disorders
 - GI dysmotility
 - Frequent ear infections

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Autism and Autism Spectrum Disorders

Associated Medical Disorders:

- Seizure Disorder
- GI Dysmotility
- Anxiety
- AD/HD
- Intellectual Disability
- Sleep Disorders
- Frequent Ear infections

Behavioral Manifestations of Illness in Autism and ID

- Fist Jamming in the Mouth GERD, Dental Pain, Asthma, Nausea, Anxiety, Hand Pain
- Biting Side of Hand Sinus Problems, Eustachian Tube Dysfunction, Dental problems, Hand pain
- Intense Rocking Visceral Pain, headache, Depression, Anxiety, Medication Side Effects
- Head Banging Headache, Seizure, Dental Pain, Sinus problems, Tinea Capitis, Depression
- Waving of fingers in Front of Eyes Migraine, Vision problem, Seizures

- Individual
- **Syndrome level**
- Medication perspective

Syndrome perspective

- Dental
- Ear Pain
- Sinus Pain
- Hand Pain
- Abdominal Pain
- Anxiety/Depression
- Allergy

- Individual
- Syndrome level
- **Medication perspective**

Atypical Anti-Psychotics

- **Possible Side effects**
 - Weight gain
 - Diabetes
 - Hyperlipidemia
 - Prolonged QTc interval
 - Cataracts
 - Elevated Prolactin- menstrual disorders
 - Tardive Dyskinesia
 - Extrapyrarnidal Side effects
 - Parkinsonianism
 - **Akathisia- a state of agitation distress and restlessness**

Case of Alan

- 48 year old male with, learning disability, history of alcoholism in remission, and cerebral palsy with spastic diplegia. He is employed part time in a sheltered workshop. He uses public transportation and lives alone. He complains of left sided abdominal pain, on and off, sometimes quite severe. It is partially relieved by voiding or defecating. He denies fever. He has a long history of constipation which is generally managed with diet. He reports he has not been as good at drinking water lately because of his fear of incontinence on the bus. He reports he has not had a drink in over one year and attends AA as regularly as he can, depending on transportation. He used to smoke but does not any longer. He recently saw his psychiatrist for his spasticity who increased his baclofen dose.
- Family History
- Medications:
 - Baclofen
 - Miralax
 - Gabapentin

- **Individual**
- Syndrome level
- Medication perspective

Individual Perspective

45 year old male with history of alcohol abuse with abdominal pain:

–Pancreatitis

- Individual
- **Syndrome level**
- Medication perspective

Cerebral Palsy

- Is the most common childhood physical disability,
- Affects 2.5 children per 1000 born in the US
- STATIC neurological condition resulting from brain injury that occurs before complete cerebral development (age 2).
 - NON-progressive , so changing in underlying neurological deficit should prompt testing for genetic, metabolic, muscular or tumor related disorders
- 70-80% thought to be acquired pre-natally
- Approximately 6% are thought to be related to birth complications
- Remaining occur after birth from encephalitis and meningitis, hyperbilirubinemia, intracranial trauma from MVAs, falls or child abuse

Cerebral Palsy

- May or may not include intellectual disability
 - Approximately 2/3 have some degree of ID
- Characterized by abnormal muscle tone and posture
- Muscular hypertonicity
- Increased deep tendon reflexes
- Tremor
- Weakness
- Athetoid or Dyskinetic type occurs in 10-20%
- NON-progressive

Cerebral Palsy

Associated Conditions

- Seizure Disorder
- GI Dysmotility
 - Oral Incoordination- speech impairment
 - Dysphagia
 - Oral dysmotility
 - Excessive drooling
 - Aspiration
 - GERD
 - Chronic constipation
- Urological Disorders
 - Urinary Retention
 - Kidney stones
- Visual and Hearing Impairment
- Autism
- AD/HD
- Disuse Osteoporosis
- Respiratory Issues
 - Recurrent Aspiration
 - Asthma
 - Scoliosis with structural impingement
- Hypertension

Syndrome Perspective

- Constipation flare
- Kidney Stones
- Rib Fracture
- Urinary Retention

Medication Perspective

Baclofen

- **Urinary retention**
- Urinary frequency
- Constipation
- Dry mouth

Gabapentin

- Constipation
- Depression

The Case of Patrick

Patrick is a 45 year old with Down Syndrome, and seizure disorder who presents with his residential staff who report gradual decline in function over the last year. He appears less interested in activities at home and at day program. Patrick was ambulatory but he fell last time he walked and has since refused. He has gained weight and has a good appetite. He frequently falls asleep during the day which he is spending mostly in his wheelchair. He was previously ambulatory.

Medication List

- Vimpat
- Clonazepam
- Depakote
- Miralax

FH:

- Father had diabetes, recently passed away from MI,

SH:

- He has lived in residential housing for 5 years, Dad visited regularly and was very involved.

- What are we thinking?
- What might we ask?
- What might we pay special attention to on exam?
- What diagnostic tests might we perform?

- **Individual**
- Syndrome level
- Medication perspective

45 year old man with recent loss with social withdrawal and decrease in function:

- Individual
- **Syndrome level**
- Medication perspective

Down Syndrome

- Down Syndrome is the most common genetic cause of Intellectual Disability which can range from very mild to profound.
- Most commonly caused by Trisomy 21, an extra 21st chromosome

Down Syndrome

Down syndrome predisposes an individual to multiple medical conditions involving nearly every organ system

- Early Onset Dementia
- Visual Impairment
- Hearing loss
- Hypothyroidism
- Diabetes
- Hematologic abnormalities
- Atlantoaxial instability
- Congenital heart disease
- GI motility disorders
- GERD
- Sleep Apnea

Syndromic Perspective broadens the differential

- Early Onset Dementia
- Visual Impairment
- Hypothyroidism
- Diabetes
- Hematologic abnormalities
- Atlantoaxial instability
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- Individual
- Syndrome level
- **Medication perspective**

Medication Perspective

Vimpat

Depakote

Medication Perspective

Vimpat

- Blurred vision
- Sleepiness
- Vertigo
- Ataxia
- Depression
- Impaired memory
- Bone marrow suppression

Depakote

- Pancreatitis
- Hyponatremia
- Bone marrow suppression
- Hyperammonemia
- Psychosis
- Parkinsonism
- Dizziness
- Depression
- Blurred vision
- Abnormal Gait

What Next?

Lessons Learned

Providers

- Common things are still common
- Common things can happen at a younger age
- Get history anywhere you can
- Think about the person as an individual, and as a person with a syndrome
- Talk to your specialists!
- Don't forget about medications and their side effects
 - Keep list updated
 - Consider reduction of unused medication
 - Not everything needs a prescription

Patients, Parents and Support Staff

- Plan your visit, Don't bury the lead
- Bring or send history, We depend on you!
- Think about details-
 - when did it start,
 - when does it occur,
 - how does it impact behavior,
 - what is baseline,
 - what changes in medication, lifestyle or stressors has happened.
- Bring an accurate medication list
- Careful about what you ask for- all medications have potential side effects

Resources

- Psychiatry of Intellectual Disability, A Practical Manual, Julie Gentile and Paulette Marie Gillig
- Genetics Home Reference <https://ghr.nlm.nih.gov>
- Down Syndrome Dementia Questionnaire kcdsg.org
- Cerebral Palsy: An Overview, Karen Krigger MD, Am Fam Physician, 2006 Jan1;73 (1) 91-100
- www.aacpdm.org

Thank you!