Neurological and Neurodevelopmental Manifestations in Down syndrome

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1

Objectives

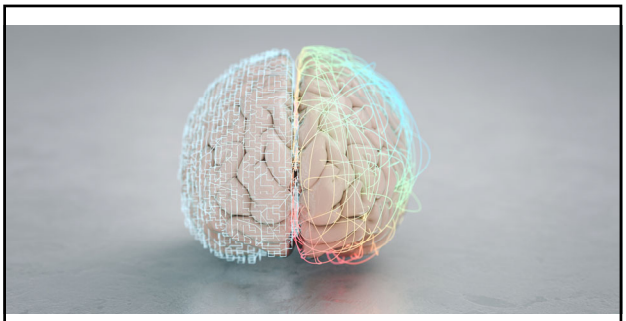
Review
- Review what is known about neurobiology in Down syndrome and the characteristic developmental, cognitive, and learning profile

Discuss
- Discuss co-occurring neurodevelopmental and mental health conditions that may co-occur in children and adolescents with Down syndrome and may sometimes be misattributed to manifestations of ID alone

Present
- Present neurologic findings such epilepsy, abnormal movements, and hypotonia and motor impairment

Examine
- Examine medical conditions that may impact development and functioning

2

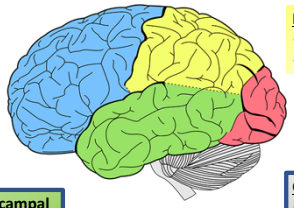


Neurobiology & Neurodevelopmental Profiles in Down Syndrome

3

Brain Differences in Down Syndrome:
May lead to common cognitive and neurodevelopmental characteristics

Frontal
- Motivation
- Expressive language
- Executive Functioning



Parietal
- Visual attention
- Integration of senses

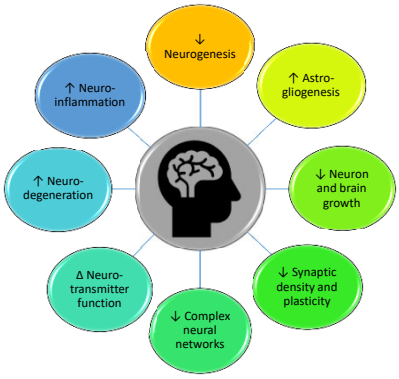
Occipital
- Vision / Visual Processing

Temporal –hippocampal
- Memory
- Emotion

Cerebellum
- Balance, motor memory / planning
- Learning, social interaction?

4

Neurobiology



5

Neurodevelopmental Profile

Strengths	Challenges
Receptive Language	Expressive language
Visual-spatial processing and visual short-term memory	Verbal/Auditory processing and short-term auditory memory
Social interest & engagement	Motivation and engagement with non-preferred topics/tasks
Recognizing emotions	Noncompliant behavior and difficulty with task persistence
Procedural learning of routines and daily living skills	Fact-based learning, executive function and organization

6

Challenging Behaviors
in Down Syndrome

7

Common Behavioral Challenges

Oppositionality	Defiance	Aggression	Non-compliance
Irritability, Agitation	Restlessness	Anger, Mood swings	Hyperactivity
Disruptive	Impulsive	Inattentive	Destructive
Tantrums	Flopping	Eloping/Bolting	

8

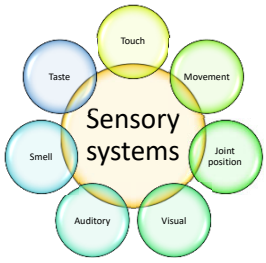
Behavior

Common	30% may have diagnosable behavioral condition, but up to 94% of parents report behavioral concerns May be unsafe
Interferes with function and outcomes	Children: Learning, daily living, and social relationships Adults: recreation, vocation, employment, community living
Many underlying factors	Communication challenges, unusual sensory responses, poor emotional regulation/coping, and rigidity
May serve many functions	To gain social attention, to obtain access to a tangible item or activity, to escape or avoid a situation, or to provide sensory or self-stimulation

9

Sensory Processing Challenges

- Sensory processing = collection, integration, and interpretation of information from multiple sensory systems
- Not socially / externally motivated, rather internally motivated
- Impacts how an individual learns, moves, explores the environment, behaves, completes tasks, and interacts with others
- May be associated with challenging behaviors in Down syndrome



10

Hypersensitive → Over-reaction	Hyposensitive → Under-reaction
Fearful/avoidant	Sensory-seeking
May be dysregulated or shut down with too much input/stimulation	May require more intense sensory input to be regulated – craves stimulation
Agitated in presence of stimulation, may meltdown	May not register low level of stimulation and by under-responsive
• Choosy about fabric texture, tags on clothing • Picky eating food aversions • Avoidant of grooming tasks and messy play • Upset by loud noises, covers ears • Difficulty with new or unexpected situations	• May seek constant motion, swinging, movement, etc. • Takes risks with play, jumping/crashing into things • May get hurt and not realize it/high pain tolerance • Mouthing of nonfood objects and overstuffing • Makes nonfunctional noises • Challenges managing multiple auditory stimuli

11

Sensory Diet:

Physical activities and tools tailored to meet sensory needs

12

Learning and Development: Key Strategies

- Capitalize on strengths / interests, good mood and momentum → Use positive reinforcement: praise and rewards
- Distraction, redirection, planned ignoring of undesired behaviors
- Use visual supports → schedules, timers, video models
- Support functional communication
- Break up tasks into small steps
- Consistent use of routine and structure
- Focus on skill development: imitation, play, patience, flexibility
- Practice, use repetition

13

New/Change in Behavior → Comprehensive Evaluation

Medical / Clinical Evaluation
- Detailed medical + behavioral history:
  - Timeline, medications, family history
- Physical Exam, Labs, Diagnostic testing as needed
- Consideration of potential psychiatric, neurodevelopmental, or behavioral disorders

Psychosocial Evaluation Assessment of stressors
- Assess / Assist in navigating supports
  - Behavioral, family, educational services
- Assess student/teacher fit, major environmental change, loss, transition

Assessment of skills → Psychological Assessment

14

Medical Assessment:

Look for Potential Organic / Medical Causes of Challenging Behaviors

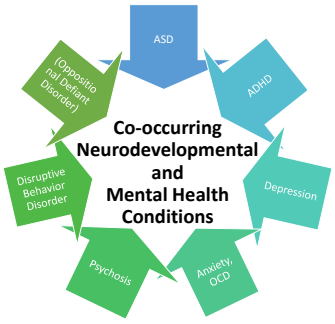
- Pain (ears, dental, GI, musculoskeletal, headache, menstrual)
- Infection
- Sleep disorders: behavioral, OSA (50-80%)
- Nutrition
- Hearing (75%) or vision (60-80%) impairment
- Celiac disease (1-5%)
- Thyroid disease (50% by adulthood)
- Gastrointestinal problems: constipation, reflux, abdominal pain

15

Neuropsychological Assessment

- Cognitive / Development
- Speech and Communication – receptive, expressive, pragmatic language
- Adaptive / Self-care skills
- Attention / Hyperactivity / Executive Functioning
- Behavioral and Emotional Functioning
- Social Communication Rating Scales
- Autism Specific Tools – Direct assessments, Rating Scales

16



Indications / When to be concerned about a co-occurring diagnosis:

- Significant impairment in learning, socialization
- Dangerous behaviors associated with aggression, injury, destructiveness
- Behaviors occur across multiple environments

Specific treatment strategies depend on:

- Severity, Frequency
- Chronologic age AND developmental level of the child
- Developmental / diagnostic profile

17

Behavior: Pharmacological Treatment

Medications can target certain symptoms: may improve physiologic regulation, emotional stability, neurocognitive processing, sleep disorders, psychiatric disorders, and can allow learning and socialization to progress

Medications do NOT teach skills, but may improve availability for learning, and may allow educational and behavioral strategies to be more successful early on

Treatment choice often depends on age of child, severity of symptoms, availability of supports and services

Requires careful consideration of risks / benefits, potential side effects

18

Pharmacological Treatment Strategy

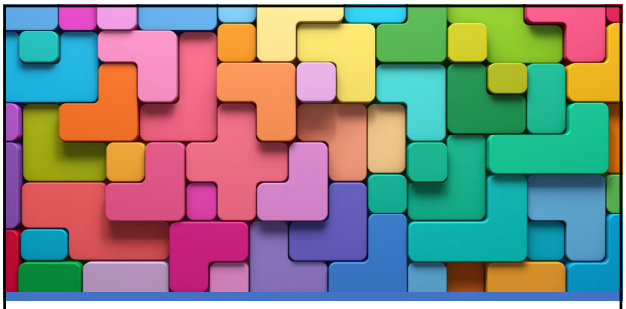
- Symptoms Medications can be used to target:
  - Impulsivity
  - Hyperactivity
  - Inattention
  - Depression
  - Mood instability (mood swings)
  - Anxiety
  - Obsessive-compulsive behavior
  - Irritability driving aggressive behavior
  - Self Injurious Behavior

19

Pharmacological Treatment Strategy

- Scientific evidence is lacking
  - Primarily case reports, expert experience
  - Most medication use is "off-label"
- More potent options → side effects more likely
- Goal: Improve function, minimize side effects
  - Start LOW, Go SLOW
  - Monitor closely
  - Avoid polypharmacy if possible

20



Autism Spectrum Disorder
in Down Syndrome

21

Background - Autism

Autism is a developmental disorder characterized by difficulty with social communication and restricted or repetitive patterns of thought and behaviors.

- Autistic Disorder → Autism Spectrum Disorder (ASD, Autism)
- Behaviorally defined disorder - DSM-5 (American Psychiatric Association): No medical "tests" to diagnose autism
- Diagnosed through a careful evaluation by specialists
  - Reliable by 18-24 months, but most diagnosed after 4 years
- Well researched condition
- Strong advocacy
- Specific evidence-based treatment strategies

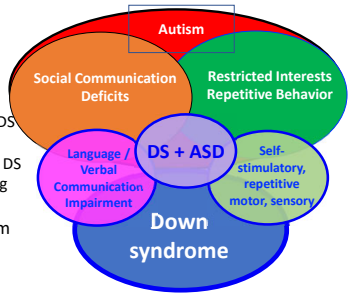
22

DS	ASD	DS+ASD
M = F 1 in 700	M >> F 1 in 36 (2.7%)	M > F 1 in 6 (4-40%)
Mild-Moderate Intellectual Disability Receptive > Expressive language Stronger Adaptive skills Stronger social relatedness	1/3 with Intellectual Disability Variable language abilities Weaker Adaptive skills Weaker social relatedness	Moderate-Severe Intellectual Disability Weaker Receptive and Expressive language Weaker Adaptive Skills Weaker social relatedness

23

Down Syndrome and Autism

- Prevalence ↑↑↑
  - 2011: 1%
  - 2022: 7-19%
- Diagnosis is complex:
  - Overlapping features in DS and ASD
  - Wide range of abilities in DS
  - Diagnostic overshadowing
- Age of Diagnosis delayed several years after symptom onset



24

Signs and Symptoms Reported in Children with Down syndrome + Autism

- Delayed developmental milestones, more cognitive difficulty
- Few attempts to communicate, more repetitive / inappropriate speech
- Decreased or unusual eye contact
- Less social relatedness: plays alone / socially withdrawn / less interest in others
- Unusual or repetitive play
- More difficulty with adaptive / daily living skills: toileting, feeding
- Frequent repetitive motor behaviors that are difficult to distract or redirect from
- More mood and behavioral concerns: extreme meltdowns, dysregulation, difficult to soothe
- Looking at things from strange angles, dangling, licking objects
- Sensory sensitivity: touch, haircuts, crowds, extreme food refusal
- Sleep problems
- Developmental Regression

25

Best Practices for Making ASD Diagnosis

- Diagnosis requires comprehensive developmental history AND direct assessment/observation of skills and behavior
- Clinicians must consider medical conditions, other mental health conditions, and overall developmental level
- Focus on socialization as a core feature: Development of social skills and social communication is more delayed than motor, adaptive, and cognitive functioning
- Regression of skills in early childhood may be seen
- Clinicians must be AWARE of the dual diagnosis, understand Down syndrome, autism, and how to evaluate for distinguishing features

26

Behavior and Educational Therapies are Mainstay of Treatment

- Intensive, well-coordinated, comprehensive services
- Parent training and involvement and close collaboration among parents / caregivers, educators, clinicians
- Applied Behavior Analysis (ABA) (Gold standard)
  - Many different "types" of behavioral therapy: discrete trials, naturalistic, etc.
  - Key: data driven approach
  - Addresses skill development AND reducing maladaptive behaviors
  - Social reinforcers are often helpful
  - Capitalize on strengths and interests

27

ADHD
in Down Syndrome

28

Attention Deficit Hyperactivity Disorder (ADHD)

- Symptoms and/or behaviors that have persisted ≥ 6 months in ≥ 2 settings.
- Symptoms have negatively impacted academic, social, and/or occupational functioning.
- In patients aged < 17 years, ≥ 6 symptoms are necessary
- In those aged ≥ 17 years, ≥ 5 symptoms are necessary

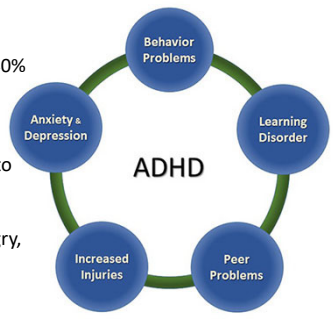
DSM 5 Diagnostic Criteria:

INATTENTION		HYPERACTIVE/ IMPULSIVE
• Fails to give close attention to details • Difficulty sustaining attention • Does not seem to listen when spoken to • Does not follow through on instructions/quickly loses focus • Difficulty organizing tasks • Avoids, dislikes, or is reluctant to engage in tasks with sustained mental effort • Loses things necessary for tasks or activities • Forgetful in daily activities	COMBINED	• Fidgets with/taps hands or feet/wiggles in seat • Leaves seat in situations when expected • Runs or climbs in situations when it's inappropriate • Unable to play or engage in leisure activities; uncomfortable being still for extended periods of time • Talks excessively • Blurts out answers before a question is finished • Difficulty waiting his/her turn • Interrupts or intrudes on others

29

ADHD: Prevalence and Associations

- ↑ Frequency of ADHD in DS
- Prevalence not certain, ?10-40%
- Must consider symptoms in relation to overall developmental level
- Symptoms shouldn't be due to
  - Specific setting
  - Mood, fear, anxiety, physiological states (hungry, tired)
  - Specific time of day



CDC, 2022

30

Evidence-Based Treatments:

- Medications
- Behavioral interventions
- Medication + Behavioral Interventions

31

Medications

- Alpha agonists, other Non-stimulant medications
  - Ex: Guanfacine*, Clonidine
    - Side effects: lowers blood pressure, drowsiness, constipation, dry mouth
- Psychostimulants:
  - Ex: Methylphenidate*, Mixed amphetamine salts
    - Side effects: Decreased appetite, insomnia, emotional problems / anxiety, agitation / irritability, tachycardia
    - ? Less well tolerated in Down syndrome
- Non-stimulants: Atomoxetine, Amantadine
  - Very little evidence, no studies in Down syndrome

* Specifically studied in Down syndrome

32

Behavioral Interventions

- Token reward system for target behaviors
- Preferential seating near teacher
- Frequent praise and positive attention
- Clear explanations of classroom rules and expectations
- Frequent breaks, including motor breaks
- Maximize outdoor time and gross motor activity
- Shorter work periods
- Daily Report Card (DRC)

33

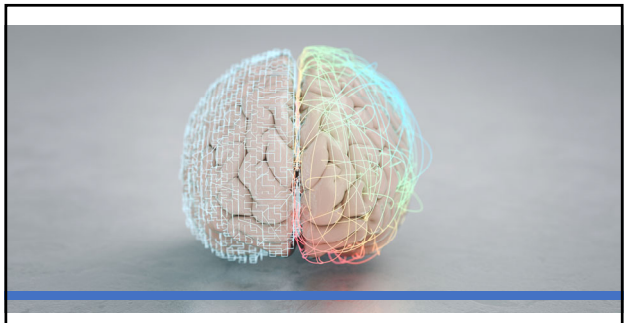
Abnormal Movements: Tics & Stereotypies

in Down Syndrome

34

	Tics	Stereotypies
Age of Onset and duration	School age (improves by adulthood)	Infancy/Toddlerhood
Pattern	Simple or complex random movements that wax/wane over time	Fixed, identical, predictable
Movement	Blinking, grimacing, shrugging, twisting(often head/face/shoulder)	Arm or hand flapping, body rocking, finger posturing, twirling, arm stiffening
Rhythm	Quick, sudden, aimless	Rhythmic
Duration	Intermittent, short, abrupt	Intermittent, repeated, prolonged
Pre-movement feelings	Yes – urge to do	?No
Trigger	Random but ↑ with excitement, stress	During periods of excitement, engrossment, stress, boredom, fatigue
Suppressibility	Self-initiated, brief	External distraction, initiation of activity
Perception	Distressing, ego-dystonic	Calming, enjoyable, ego-syntonic
Co-occurs with…	ADHD, anxiety, OCD, vocal tics	ASD
Treatment	Behavioral approaches +/- medications if interfering with function	Behavioral approaches if functionally impairing; Medication rarely effective

35



Seizure Disorders

in Down Syndrome

36

Seizures / Epilepsy

Seizure: a sudden event caused by a disruption in the brain's normal electrical activity, with nerve cells in the brain firing abnormally. May involve part of brain or whole brain. May cause changes in consciousness or awareness, abnormal movements, or other behaviors.

Epilepsy (Seizure Disorder): a condition in which someone has recurrent unprovoked seizures, due to abnormal electrical signals

37

Seizures in DS

Incidence in DS: ~5-13%
- ↑ risk compared to general population (1-2%)
- ↓ risk compared to other causes of intellectual disability (40%)

Onset at three different times in life:
- Infancy – infantile spasms
- Early adulthood – variety of seizure types
- 5th decade or later – Late Onset Myoclonic Epilepsy in DS (LOMEDS), associated with Alzheimer's disease

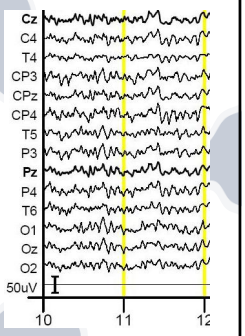
Risk factors/Etiology Variable
- Inherent genetic features
- Structural brain differences
- Altered synaptogenesis
- Membrane channel dysfunction
- Acquired injury (stroke, hemorrhage, hypoxic injury – underlying risk due to moya moya, congenital heart disease, infection)

Increased vigilance/monitoring needed

38

Diagnosis of seizures/epilepsy: EEG (electroencephalogram)

- Diagnostic test
- Measures electrical activity in the brain
- Sensors (electrodes) are attached to the head, and hooked up with wires to measure brain waves
- Looking for actual seizure activity (rarely caught unless seizures are very frequent), abnormal background rhythms, or abnormal firing of the brain that indicates increased risk of seizure



39

Infantile Spasms

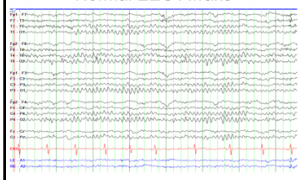
- The most frequent epilepsy syndrome in Down syndrome, prevalence ~5%
- Sudden, brief jerks of the neck, trunk, extremities
  - Last 1-2 seconds
  - Often occur in clusters with relaxation between each spasm
  - Often occurs with sleep-wake transitions

West Syndrome: TRIAD of symptoms:
1. Infantile Spasms
2. Hypsarrhythmia on EEG
3. Developmental regression or arrest

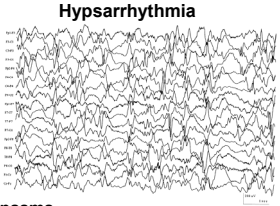
- Treatment
  - Must stop the spasms AND improve the EEG pattern
  - Start immediately!
  - ACTH (adrenal corticotropin hormone given via IM injection) or steroids, 1-2 months+
  - Frequent EEG + close clinical monitoring needed
  - Other options (2nd line):
    - Vigabatrin
    - Ketogenic diet
    - Other anti-seizure medications - topiramate

40

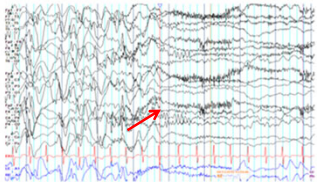
Normal EEG Awake



Hypsarrhythmia



Infantile Spasms



Sudden calming of electrical activity immediately after a spasm

41

Outcomes

- Good outcomes in terms of seizures/epilepsy
  - More research needed on developmental outcomes
- **The key is to diagnose and treat early!**
- Delayed treatment has been associated with:
  - Longer time for eliminating spasms
  - Decreased likelihood of full developmental recovery
  - Increase in autistic features
  - Persistence of seizures

42

Not all funny movements or behaviors are spasms or seizures!

Other Considerations:

- Pain/Colic/Reflux - digestive irritability, "Sandifer Syndrome"
  - Arching with feeds, Timing may be different
- Exaggerated startle reflex
  - Moro reflex: neonatal reflex up to 4-5 months of age
- Shuddering Attacks
  - Infancy - movements resembling shivering and straining without impaired consciousness or epileptiform EEG, resolves by toddlerhood
- Repetitive mannerisms / Stereotypic movement disorder
- Sleep myoclonus
  - Jerks when transitioning into and out of sleep
- Other medical diagnoses
  - Spasmas Nutans – nystagmus, head bobbing and head tilt
  - Paroxysmal Tonic Upgaze – eye deviation upward

Key Factors Leading To The Diagnosis:

- Age: first year of life
- Description of the movements , clustering, timing
- Developmental Concerns
- EEG pattern of hypsarrhythmia

43

Beyond Infantile Spasms

- Age 5 years through young adulthood
  - Emergence of other seizures types
    - Focal onset epilepsy with impaired awareness (Video)
  - Unclear if higher incidence compared to general population; probably higher likelihood in children with DS+ASD
  - Treatment approaches same as general population
- Late Adult-onset (40s+)
  - Unprovoked myoclonic (jerky movements) and generalized tonic-clonic seizures
  - Initial good response to treatment, but can progress to refractory myoclonus
  - Dementia usually proceeds first seizure, but not always
- Any time - Provoked seizures
  - Electrolytes, medication withdrawal, etc.
- Adults - Psychogenic Nonepileptic Seizures
  - No EEG correlate

44

Sleep
in Down Syndrome

45

Sleep and Neurodevelopment

Sleep problems reported in ~30-50% of children with DS

- Obstructive sleep apnea (OSA)
- Frequent insomnia
- Decreased sleep efficiency
- More fragmented sleep patterns
- More abnormal sleep arousals

46

Sleep disordered breathing

Very common (>50%)

Risk factors

- Inherent craniofacial differences / midface hypoplasia
- Relative macroglossia
- Upper airway anatomic differences and laryngomalacia
- Hypotonia / Decreased pharyngeal tone
- Obesity

Presentation

- Snoring
- Apneas
- Night-time awakenings
- Unusual sleep positions
- Daytime behavior changes – irritability, inattention

Many young children are asymptomatic, or symptoms are not obvious to parents

47

Why does detecting OSA matter?

Neurodevelopmental Impacts!

- Declines in verbal IQ
- Poor cognitive flexibility
- Worse visual-perceptual skills
- Increased disruptive and behavioral symptoms
- Poorer overall developmental milestone acquisition
- Worsened school performance
- Mood issues

48

Why does OSA matter?

Detectable

- Screen for symptoms
- Screening sleep study by age 4 years for all

Treatable

- **Surgery**
  - Adenotonsillectomy
  - Lingual tonsillectomy
  - Drug-induced sleep endoscopy (DISE) → supraglottoplasty, epiglottopexy
  - Craniofacial surgery evaluation
- **Medications**
  - Flonase/Singulair (Allergic Rhinitis) – may be less effective
  - Management of Reflux and Asthma
- **CPAP: Continuous Positive Airway Pressure**
- **Other**
  - Weight loss
  - Dental approaches/appliances
  - Hypoglossal nerve stimulator

49

Beyond OSA: other sleep issues

Arousals/Parasomnias

- Unusual and undesirable physical events/behaviors or experiences that disrupt sleep
- Sleepwalking, night terrors, excessive nightmares, confusional arousals
- More commonly endorsed by parents of children with DS
- Most are benign in nature, most don't need treatment
- Differentiate from nocturnal frontal lobe epilepsy
- Anticipatory guidance and education on sleep hygiene

Sleep-related movement disorders

- Rhythmic movements of the legs, limbs, or jaw during sleep
- Periodic limb movement disorder, restless leg syndrome, teeth grinding
- More common in DS
- Association with iron deficiency → evaluate Ferritin level, iron supplementation to goal of Ferritin >50

Behavioral sleep disorders

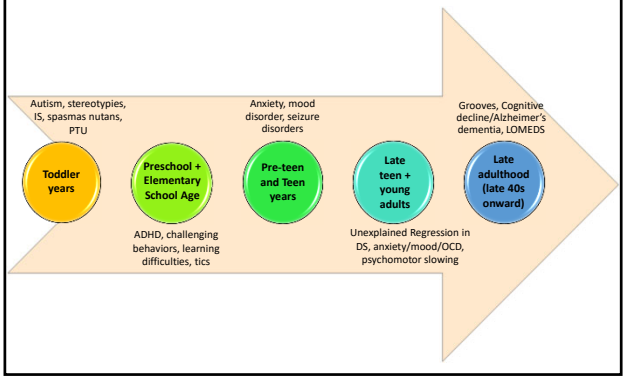
- Insomnia due to challenges with sleep initiation or sleep maintenance
- Associated with ↑daytime behavior issues and parental stress
- Programs on self-soothing, sleep-hygiene
- Behavioral strategies (planned ignoring, fading adult support, bedtime pass)
- Medications can help, more for initiation than maintenance
  - Antihistamines, alpha-agonists, trazodone, gabapentin, quetiapine

50

Mental Health
in Down syndrome

51

Progression of concerns over time



52

Anxiety

Onset	• Late childhood, adolescence
Symptoms	• Excessive worry, increasing situational anxiety, more difficulty with transitions, avoidance behaviors, hypervigilance, increased rigidity and ritualistic behavior • Physical symptoms (pacing, skin-picking, nailbiting, stomachaches, etc.)
Forms	• Generalized anxiety, specific phobias, selective mutism, panic disorders
Assessment	• Parent completed standardized questionnaires • Child/young-adult completed questionnaires

53

Obsessive Compulsive Disorder

- Obsessions = Recurrent, unwanted, preoccupying thoughts
- Compulsions = Repetitive behaviors one feels compelled to perform
- Interfere with daily functioning
- Common symptoms
  - Excessive inflexibility with routines, repeated ordering, arranging, checking, touching, hoarding, handwashing/maintaining cleanliness
  - Challenging to assess obsessional component
- Differentiate from routinized, compulsive-like behaviors
  - More common in DS than other forms of ID
  - Common in younger children
  - Adult "Grooves"

54

The Groove

- Patterns and routines that individuals with DS tend to want to follow
- Function?
  - Help organize/manage unpredictability of life
  - Cope with stress
  - Exert control over environment
  - Express preferences
- Challenging if individual gets stuck in pattern
- If pervasive, frequent, impairing function → OCD?

55

Anxiety management

Behavioral strategies – Building a skill set
- Routine/structure, visual supports, social stories
- Sensory strategies (weighted blankets, headphones, etc.)
- Relaxation strategies (yoga, meditation, deep breaths, etc.)

Modified cognitive-behavioral therapies
- Modified exposure + response prevention therapy (ERP)

Pharmacologic intervention
- SSRIs (higher dose for OCD)
- SNRIs
- Buspirone
- Augmentation with antipsychotic medications

56

Depression

Symptoms	• Sad/low mood often not self-reported or may be hard to detect • Changes in vital functions: sleep, appetite, movement/activity level, self-care • Social withdrawal, decreased interest in preferred activities
Rule out medical causes	• OSA, hearing/vision, pain – MSK, constipation, hypothyroidism, celiac
Onset	• Psychosocial stressor – may be perceived as minor to others
Treatment	• Prevent/plan for triggering events • Supportive counseling • Pharmacologic treatment - SSRIs

57

Psychosis, etc.

Psychosis	• Hallucinations, delusions, paranaoia • Far less common than other mood-based issues • Likely no more common than general population
Things that are NOT psychosis:	• Self-talk and self-answering (sometimes with different voices/tones) • Self-assurance, cognitive processing • If negative → think about mood/self-esteem • Shouts/screeches (vocal tics?) • Symptoms modeled by others/learned and imitated • Loftry, seemingly grandiose hopes/dresms

58

Unexplained Regression in Down syndrome

Down Syndrome Disintegrative Disorder

Onset
- Adolescence, young adulthood
- Decline may be precipitous or gradual

Differentiate from:
- Autism with regression (ages ~2-6 yrs)
- Alzheimer's disease (late 40s+ yrs)

Symptoms:
- Loss of previously acquired skills
  - Language – whispering, mute
  - ADLs – feeding, dressing, toileting
- Withdrawal/reduced social engagement and family participation
- Sleep dysfunction/insomnia
- Personality changes, mood lability
- New movements, tics
- May look "autistic" and meet criteria for catatonia

59

Regression

Often occurs in context of psychosocial stressors

May occur in setting of underlying mood disorder

? Autoimmune/autoinflammatory process

Must evaluate for contributing medical causes

Extensive workup is recommended though is often unrevealing for a definitive cause
- Brain MRI, EEG, Lumbar puncture. Workup for autoimmune encephalitides
- Medical workup – workup for Hashimoto's including thyroid function and antibodies, celiac screening, sleep apnea evaluation, etc.

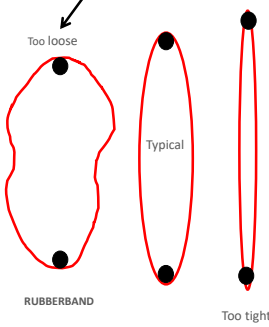
Treatment
- Catatonia approach → high dose benzodiazepines, ECT
- Other psychopharmacology → SSRI, neuroleptics
- Immunotherapy → steroids, IVIG, JAK inhibitors?
  - NIH clinical trial

60

Motor and Musculoskeletal Issues in Down Syndrome

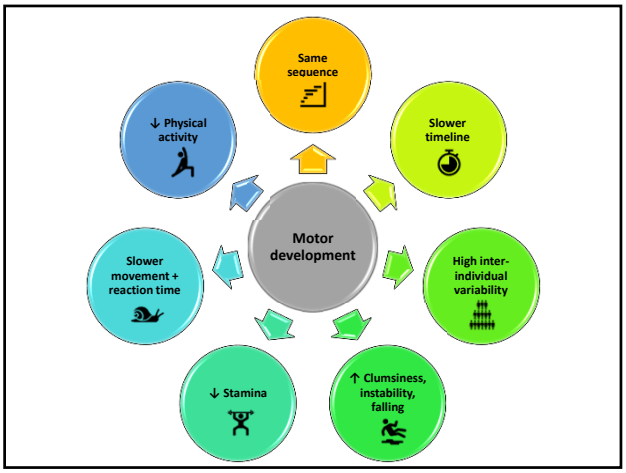
61

Hypotonia



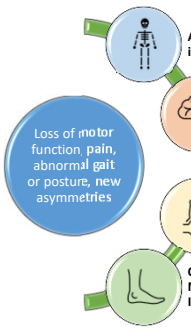
- Decreased resistance to passive muscle stretch or movement
  - Subjective component of physical examination
  - Separate from strength, but often associated with mild generalized weakness
- Contributes to
  - Hyperextension and instability at the joints
  - Gross motor delays
- Common findings
  - Delayed gross motor milestones
  - Over-pronation and flat feet (pes planus)
  - Difficulty with postural control
  - Breathing difficulties
  - Feeding problems, excessive drooling, and articulation difficulties
  - Constipation
  - Easy fatiguing

62



63

When to seek help…



Atlanto-axial instability

- The joint between the upper spine and base of the skull is unstable due to ligamentous laxity
- Symptoms: Gait changes, new-onset torticollis or head tilt, neck pain, and decreasing mobility overall
- Screen for symptoms throughout childhood with any concerns warranting further evaluation via cervical x-ray and spinal MRI
- Treated with monitoring vs. surgery, anesthesia precautions, activity limitations

Moyamoya

- Abnormal vasculature in the brain (puff of smoke) due to upstream carotid abnormality → leads to ischemic or hemorrhagic strokes or transient ischemic attacks
- Symptoms: seizures, headaches, or sudden or intermittent focal neurologic deficits such as new onset weakness or paralysis
- Requires immediate neurological and neurosurgical evaluation

DS-arthropathy

- Joint inflammation/arthritis that can lead to permanent damage
- Often involves small joints of hands/wrists with onset in teen years, usually involves multiple joints
- Presents with joint swelling, pain, often delayed diagnosis
- Different clinical and lab findings compared to other classic forms of juvenile arthritis
- Requires referral to rheumatology, disease-modifying medications involving immunosuppression

Other Musculoskeletal Issues

- Subluxation of hips
- Scoliosis
- SCFE (Slipped Capital Femoral Epiphysis)
- Worsening pes planus

Involvement of orthopedics, neurosurgery, rheumatology, physiatry, physical therapy, and orthotic services can be essential for appropriate management of these conditions that impact motor function in DS.

64

Conclusion

DS is characterized by a specific developmental, cognitive, and learning profile, though we are only beginning to understand the complex neurobiology underlying this profile.

Individuals with DS are at higher risk for many medical conditions that may impact development and functioning, underscoring the importance of awareness, screening, and appropriate treatment.

It is important to be aware of possible neurologic conditions in DS, such as hypotonia and motor impairments, epilepsy, and abnormal movements. These conditions have different clinical presentations, trajectories over time, and management approaches.

Co-occurring neurodevelopmental and mental health conditions are relatively common in DS, may sometimes be misattributed to manifestations of ID alone, and are important to recognize because they call for different management strategies that can improve outcomes and enhance supports for the individual.

65

Resources

ADHD

- Attention Problems in DS
- Finding a therapist:
  - https://www.abct.org
  - https://www.psychologytoday.com/us/therapists
  - https://www.findatherapist.com
- Resources, Advocacy, Education:
  - https://chadd.org
- Daily Report Cards:
  - https://www.additudemag.com/daily-report-card-to-improve-adhd-classroom-behavior/
  - http://www.healthyinfo.com/consumers/ho/ADD.daily.report.card.pdf
- Books:
  - Taking Charge of ADHD: The Complete Authoritative Guide for Parents by Russell A. Barkley, PhD

Autism:

- https://downsyndromecenter.libsyn.com/80-autism-and-down-syndrome-with-dr-nicole-baumer
- Down Syndrome Center of Western Pennsylvania Podcast #80 - Autism and Down Syndrome (with Dr. Nicole Baumer)

66