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Neurodevelopmental Disorders

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Neurodevelopmental Disorders – Introduction

- Onset in developmental period
  - Typically manifest early (before grade school)
  - Personal, social, academic, occupational impairments
- Range from very specific to global impairments
  - May have symptoms of excess + deficits/delays
- **Frequently co-occur** (ASD + IDD, ADHD + SLD)

Intellectual Disability (Intellectual Development Disorder)

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Intellectual Disability – Diagnostic Criteria

A. Deficits in **intellectual functions**
   1. Confirmed by BOTH **clinical assessment + standardized intelligence testing**
   2. Reasoning, problem solving, planning, abstract thinking, judgement, academic learning, learning from experience

B. Deficits in **adaptive functioning**
   1. Results in failure to meet developmental/sociocultural standards for personal independence + social responsibility
   2. Impairs **activities of daily life**, across **multiple environments** (communication, social participation, independent living)

C. Onset of deficits **during developmental period**

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Intellectual Disability – Diagnostic Specifiers

Mild

- Conceptual Domain
  - **Preschool children**: may not be obvious
  - **School-age, adults**: difficulties in learning, academic skills, support needed
  - **Adults**: impaired abstract thinking, executive function, STM, functional use of academic skills, concrete approach

- Social Domain
  - **Communication:** conversation, language more concrete/immature
    - May be difficulties regulating emotion/behavior, difficulties noticed by peers
  - **Social cues:** immature in social interactions, difficulty accurately perceiving social cues
  - **Social judgement:** limited understanding of risk in social settings, immature for age,
    - At risk of being manipulated (gullible)

- Practical Domain
  - **Personal care:** may be able to do
  - **IADLs:** adults need supports, needs support to raise a family
  - **Recreation skills:** resemble normal, but judgement requires support
  - **Employment:** in jobs without emphasis on conceptual skill, help learning skilled vocation
  - Generally need support to make health care/legal decisions

Intellectual Disability – Diagnostic Specifiers

Moderate

- Conceptual Domain → markedly behind peers
  - **Preschool**: language, pre-academic skills develop slowly
  - **School-age**: reading, writing, math, time, money occurs slowly, markedly limited
  - **Adults**: academic skill typically at an elementary level, support needed for use of academic skills, ongoing assistance for daily life tasks, others may take over responsibility

- Social Domain
  - **Communication**: spoken language primarily, much less complex than peers
  - **Relationships:** capable with family/friends, sometimes romantic relationships in adulthood
  - **Social cues:** may not perceive or interpret accurately
  - **Social judgement:** decision making limited → requires assistance making life decisions
  - Friendships often affected, sig social + communicative support needed in work settings

- Practical Domain
  - **Personal care**: capable as adult, but may need extended period of teaching and reminders
  - **Household tasks**: participation as adult, may need extended teaching, ongoing supports
  - **Employment**: can be independent in jobs requiring limited conceptual/communication skills, need considerable support from co-workers to manage social expectations, job complexities
  - **Recreation**: can develop variety of skills, typically require additional supports

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Intellectual Disability – Diagnostic Specifiers

Severe

- Conceptual Domain → limited attainment
  - **Little understanding** of written language, concept of numbers/quantity/time/money
  - **Extensive support** from caretakes for problem-solving throughout life

- Social Domain
  - **Communication**: spoken language limited vocabulary/grammar, single words/phrases
    - Focus on here and now within everyday events, understand simple speech + gestures
  - **Relationships:** with family + familiar others, source of pleasure/help

- Practical Domain
  - **Supports for all ADLs** (meals, dressing, bathing, elimination)
  - **Supervision at all times** → cannot make responsible decisions about well-being
  - Ongoing support + assistance for household tasks, recreation, work
  - Skill acquisition involves long-term teaching, ongoing support
  - **Maladaptive behavior:** self-injury in minority

Intellectual Disability – Diagnostic Specifiers

Profound

- Conceptual Domain
  - **Only physical world** **(not symbolic processes)**
  - May use objects in goal-directed fashion, may acquire certain visuospatial skills
  - Co-occurring motor + sensory impairments may prevent functional use of objects

- Social Domain
  - **Communication**: limited understanding of symbolic communication (speech, gesture)
    - May understand some simple instructions/gestures
    - Expression through non-verbal/non-symbolic communication
  - **Relationships:** enjoyed with well-known family members, caretakes, familiar others
    - Initiates/responds to social interactions with gestures + emotional cues
  - Co-occurring sensory + physical impairments may prevent many social activities

- Practical Domain
  - **Dependent on other for all aspects** of daily physical care, health, safety
    - May be able to participate/assist in some of these activities (if no physical impairment)
  - Vocational activities: simple actions with objects, high levels of ongoing support
  - Recreational activities with support: enjoys music, movies, walks, water activities
  - Co-occurring physical + sensory impairments are frequent barriers
  - Maladaptive behavior is present in sig minority

Intellectual Disability – Diagnostic Features

- A) Deficits in general mental abilities
  - **Reasoning, problem solving, planning, abstract thinking, judgement**
    - Learning from instruction/experience, practical understanding
    - Verbal comprehension, working memory, quantitative reasoning
  - Measured with **intelligence tests**
    - Individually administered, psychometrically valid, culturally appropriate
    - Scores **2 standard deviations below mean (IQ 70±5)**
  - Test factors
    - Practice effects, "Flynn effect" (high scores due to out-of-date norms)
    - Use of brief intelligence screening tests, group tests
    - If highly discrepant subtest scores → may make overall IQ score invalid

Intellectual Disability – Diagnostic Features

- A) Deficits in general mental abilities
  - **Individual cognitive profiles** more useful (than single IQ score)
    - May identify areas of relative strength/weakness
  - IQ test scores → **approximation of conceptual functioning**
    - May be insufficient for real-life situations, mastery of practical tasks
    - IQ score >70 → may still have severe adaptive behavior problems
      - Actual functioning comparable to lower IQ

Intellectual Disability – Diagnostic Features

- B) Deficits in adaptive functioning/reasoning
  - Ability to meet standards of independence + social responsibility
    - 3 domains → **conceptual, social, practical**
    - Influenced by intellectual capacity, education, motivation, socialization, personality features, vocational opportunity, cultural experience, coexisting general medical conditions + mental disorders
  - **1) Conceptual domain**
    - Memory, language, reading, writing, math reasoning
    - Acquisition of practical knowledge
    - Problem solving, judgement in novel situations
  - **2) Social domain**
    - Awareness of others' thoughts/feelings/experiences, empathy
    - Interpersonal communication, friendship abilities, social judgement
  - **3) Practical domain**
    - Learning, self-management across life settings
    - Personal care, job, finances, recreation, behavior, tasks

Intellectual Disability – Diagnostic Features

- <u>B) Deficits in adaptive functioning/reasoning</u>
  - Assessed with both **clinical evaluation + standardized measures**
    - Educational, developmental, medical, mental health evaluations
  - **At least one domain** sufficiently impaired to require ongoing support
  - Deficits must be **directly related to intellectual impairments**

  - If standardized testing not possible → unspecified intellectual disability

Intellectual Disability – Associated Features

- Heterogeneous condition with multiple causes

- Associated difficulties
  - Social judgement, risk assessment, motivation in school/work settings
  - Self-management of behavior, emotions, interpersonal relationships
  - Lack of communication skills → may predispose to **disruptive/aggression**
  - **Gullibility** → naivete in social situation, easily led by others
    - May result in exploitation, victimization, fraud, unintentional criminal involvement, false confession, risk for physical/sexual abuse

- **Risk for suicide** if co-occurring mental disorders
  - Suicidal ideation, suicide attempts, may die from suicide
  - Increased accidental injury rates

Intellectual Disability – Prevalence

- Overall population prevalence = **1%**
  - Severe intellectual disability = **0.6%**
  - Rates vary by age

Intellectual Disability – Development & Course

- Onset in developmental period (Criterion C)
  - Specific age + characteristic features → **depend on etiology, severity**
  - If severe → delayed motor/language/social milestones within first 2 years
  - If mild → may not be identifiable until school age (academic learning)
  - Some children under age 5 may meet **global developmental delay**, then eventually intellectual disability

- If associated genetic syndrome
  - Physical phenotype → **Down syndrome**
  - Behavioral phenotype → **Lesch-Nyhan syndrome**

- If acquired form
  - Onset after illness or head trauma (during developmental period)

- If loss of previously acquired cognitive skills (severe TBI)
  - May dx BOTH intellectual disability + NCD

Intellectual Disability – Development & Course

- Generally NOT progressive
  - Periods of worsening in certain genetic disorders **(Rett syndrome)**
  - Progressive worsening in others **(San Phillippo syndrome)**

- Generally lifelong after early childhood
  - **Severity levels may change over time**
  - May be influenced by underlying medical/genetic conditions
  - Early + ongoing interventions → may improve adaptive functioning
    - Some cases improve significantly → no longer meet dx
    - Common to **delay dx** until appropriate course of intervention provided

- Older children, adults
  - Extent of support → may allow for full participation in ADLs, improve fxn
  - Distinguish stable skill acquisition vs contingent on supports

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Intellectual Disability – Risk & Prognostic Factors

Prenatal	
• Genetic syndromes • Inborn errors of metabolism • Brain malformations • Maternal disease	• Placental disease • **Environmental exposures** (alcohol, drugs, toxins, teratogens)
Perinatal	
• **Labor/delivery-related events** (leading to neonatal encephalopathy)	
Postnatal	
• Hypoxic ischemic injury • Traumatic brain injury • Infections • Demyelinating disorders • Seizures disorders	• **Social deprivation** (severe + chronic) • Toxic metabolism syndromes • Intoxications (lead, mercury)

Intellectual Disability – Culture-Related Issues

- Occurs in all races + cultures
  - Consider cultural setting during assessment

Intellectual Disability – Gender-Related Issues

- Gender ratios vary widely
  - Mild → **MALES** more likely (1.6x)
  - Severe → **MALES** more likely (1.2x)

- Male preponderance
  - May be due to sex-linked factors, male vulnerability to brain insult

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Intellectual Disability – Diagnostic Markers

- Comprehensive evaluation
  - **Intellectual capacity + adaptive functioning**
  - **Genetic + nongenetic etiologies**
  - **Associated medical conditions** (cerebral palsy, seizure disorder)
  - **Co-occurring mental, emotional, behavioral disorders**

- May include
  - Basic pre-/perinatal medical history
  - **3 generation family pedigree**
  - Physical examination
  - Genetic evaluation (karyotype, chromosomal microarray, specific syndrome testing)
  - Metabolic screening
  - Neuroimaging

Intellectual Disability – Differential Diagnosis

- Major/mild neurocognitive disorders
  - May co-exist (Down syndrome developing Alzheimer's, IDD + TBI)
- Communication disorders, specific learning disorder
  - No deficits in intellectual or adaptive behavior
  - May co-occur → can dx BOTH
- Autism spectrum disorder
  - IDD common among ASD → assessment may be complicated
  - Reassessment across developmental period
  - **IQ scores in ASD may be unstable** (esp early childhood)

Intellectual Disability – Comorbidity

- Comorbidities frequent
  - **Mental disorders, cerebral palsy, epilepsy** → 3-4x higher (vs gen pop)
  - IDD influences prognosis + outcome of comorbidities
  - May need to modify assessment
    - Communication disorders, ASD, motor/sensory disorders

- Most common comorbidities
  - ADHD, depressive disorders (MDD can occur throughout range of severity)
  - Bipolar disorder, anxiety disorders
  - Autism spectrum disorder, stereotypic movement disorder (self-injury)
  - Impulse-control disorders
  - Major NCD

Global Developmental Delay

Global Developmental Delay

- **Age <5** (clinical severity cannot be reliably assessed)
  - Fails to meet expected **developmental milestones** in several areas
  - **Unable to undergo** systematic assessment
  - **Too young to participate** in standardized testing

- **Requires RE-assessment** after period of time

Unspecified Intellectual Disability

Unspecified Intellectual Disability

- **Over age 5**
  - Assessment of degree of intellectual disability **difficult/impossible**
    - Locally available procedures
    - Sensory/physical impairments (blindness, prelingual deafness)
    - Locomotor disability
    - Severe problem behaviors
    - Co-occurring mental disorder

- Only used in **EXCEPTIONAL circumstances**
- **Requires reassessment** after period of time

Communication Disorders

Communication Disorders - Preamble

- Speech
  - Expressive production of sounds
  - Articulation, fluency, voice, resonance quality
- Language
  - Form, function, Use of conventional system of symbols (spoken word, sign language, written words, pictures)
  - In a rule-governed manner for communication
- Communication
  - Verbal/non-verbal behavior (intentional or unintentional)
  - That influences behavior, ideas, attitudes of another individual
- Assessment must take into account culture + language context

Language Disorder

Language Disorder – Diagnostic Criteria

A. Difficulties in **acquisition + use of language** across modalities, due to deficits in **comprehension or production**
   1. Reduced **vocabulary** (word knowledge + use)
   2. Limited **sentence structure** (grammar, morphology)
   3. Impairments in **discourse** (ability to explain, describe)

B. Substantially + quantifiably **below expected for age,** resulting in **functional limitations**

C. Onset in **early developmental period**

D. Not due to sensory/motor impairment, AMC/ANC, or IDD/GDD

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Language Disorder – Diagnostic Features (1)

- Deficits evident in **spoken, written, sign** communication
  - **Expressive ability** → production of vocal, gesture, verbal signals
  - **Receptive ability** → receiving + comprehending language messages
  - May differ in severity → assess both modalities

- Usually affects **vocabulary + grammar** → limit discourse
  - Likely delayed first words/phrases
  - Vocabulary → smaller size, less varied
  - Sentences → shorter, less complex, grammatical errors, esp past tense

- Deficits in **comprehension** → often underestimated
  - May be able to **infer meaning** through context
  - Word-finding problems, impoverish verbal definitions
  - Poor understanding of synonyms, multiple meaning, word play

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Language Disorder – Diagnostic Features (2)

- Difficulties remembering new words/sentences
  - **Following instructions** of increasing length
  - **Rehearsing** strings of verbal info (phone numbers, shopping list)
  - Remember **novel sound sequences** (learning new words)

- Difficulties with discourse
  - Providing **adequate info** about key events
  - Narrating a **coherent story**

- Below expected for age → interferes with function
  - Academics, occupation, communication, socialization
  - Based on history, direct observation in different contexts
  - Standardized tests of language ability

Language Disorder – Associated Features (1)

- Family history of language disorder often present
- May be adept at accommodating their limited language
  - May **appear shy/reticent** to talk
  - May prefer to communicate **only with family members/familiar people**
- May co-occur with **speech sound disorder**

Language Disorder – Development & Course

- Onset
  - During **early developmental period**
    - Sig variation in acquisition → NOT predictive of later outcomes
  - **By age 4** → more stable differences → HIGHLY predictive of outcomes
    - If dx from age 4 → likely to be stable over time, persist into adulthood
    - Particular profile of language may change over course

Language Disorder – Risk & Prognostic Factors

- **Receptive language impairments**
  - **POORER prognosis** (vs expressive)
  - More **resistant to treatment**
  - Often difficulties with **reading comprehension**

- Genetic & Physiological
  - **HIGHLY heritable**
  - Family members more likely to have history of language impairment
    - Family hx of speech or language disorders often present

Language Disorder – Differential Diagnosis

- Normal variations in language
  - May be difficult to distinguish before age 4
- History or other sensory impairment
  - When language deficits are **in excess**
- Intellectual disability
  - Language delay often **presenting feature**
  - Can dx both if language deficit **in excess** of intellectual limitations
- Neurological disorders
  - Epilepsy, acquired aphasia, Landau-Kleffner syndrome
- Language regression/loss
  - Before age 3 → may be sign of **autism** or specific neurological condition **(Landau-Kleffner syndrome)**
  - After age 3 → may be sx of **seizures/epilepsy**

Language Disorder – Comorbidity

- Strongly assoc with other neurodevelopment disorder
  - **Specific learning disorder** (literacy, numeracy)
  - **ADHD**
  - **Autism spectrum disorder**
  - **Developmental coordination disorder**
  - **Social (pragmatic) communication disorder**

Speech Sound Disorder

Speech Sound Disorder – Diagnostic Criteria

A. Difficulty with **speech sound production**, interferes with **intelligibility of speech**, or prevents **verbal communication**

B. Causes limitations in **effective communication + function**

C. Onset in **early developmental period**

D. Not due to congenital/acquired conditions
   - Cerebral palsy, cleft palate, deafness, hearing loss, TBI, other AMC/ANC

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Speech Sound Disorder – Diagnostic Features

- Speech sound production
  - Requires clear articulation of **phonemes** (individual sounds → words)
  - Requires both **phonological knowledge + ability to coordinate**
    - Movements of **articulators** (jaw, tongue, lips)
    - With **breathing + vocalizing** for speech

- Speech sound disorder = different underlying mechanisms
  - **Phonological disorder, articulation disorder**
  - Below expected for age/developmental stage
  - NOT result of physical/structural/neurological/hearing impairment
  - ***"Verbal dyspraxia"*** = speech production problems

- In typically developing children
  - Age 2 → 50% understandable
  - Age 4 → overall intelligible

Speech Sound Disorder – Associated Features

- Language disorder
  - Esp **expressive deficits** → co-occur with speech sound disorder
  - **Positive family hx** of speech/language disorder common

- If inability to rapidly coordinate articulators
  - May have hx of delay/incoordination in related skills
    - Chewing, maintaining mouth closure, blowing nose
  - Other impaired areas of motor coordination (developmental coordination)

- Genetic conditions
  - **Down syndrome**
  - **22q deletion**
  - **FoxP2 gene mutation**

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Speech Sound Disorder – Development & Course

- Articulation of speech sounds follows developmental pattern
  - Reflected in age norms of standardized tests
  - Typically developing children may shorten words/syllables while learning
    - Progression to mastering speech sound production mostly by age 3
  - If disorder → **continue immature phonological simplification**
  - **By age 7** → most speech sounds + word pronunciation accurate
  - **"Late eight"** = *l, r, s, z, th, ch, dzh, zh (*misarticulation normal up to age 8)

- **Lisping** → misarticulating sibilants
  - Common → may involved frontal/lateral pattern of airstream direction
  - Assoc with abnormal tongue-thrust swallowing pattern

- Most children respond well to tx, improve with time (not lifelong)
  - Disorder may not be lifelong
  - If language disorder ALSO present → poorer prognosis
    - May be assoc with specific learning disorders

Speech Sound Disorder – Differential Diagnosis

- Normal variations in speech
- Hearing or other sensory impairment
  - Hearing impairment, sensory deficit, speech-motor deficit
  - If speech deficit in excess → can dx speech sound disorder
- Structural deficits
- Dysarthria
  - Motor disorder (cerebral palsy), neurological signs, distinct voice features
  - May be difficult to differentiate in age <3
  - Or if minimal general body motor involvement (Worster-Drought syndrome)
- Selective mutism
  - Limited use of speech (may be sx of selective mutism or anxiety disorder)
  - May develop with speech disorder (due to embarrassment)
  - Many exhibit normal speech in “safe settings”

Childhood-Onset Fluency Disorder (Stuttering)

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COFD (Stuttering) – Diagnostic Criteria

A. Disturbances in **normal fluency + time patterning of speech,** inappropriate for age + language skills, persistent (1+ of 7 sx):
   1. **Sound + syllable repetitions**
   2. **Sound prolongations** (consonants + vowels)
   3. **Broken words** (pauses within a word)
   4. **Audible/silent blocking** (filled/unfilled pauses in speech)
   5. **Circumlocutions** (word substitutions to avoid problematic words)
   6. **Excess of physical tension** (producing words)
   7. **Monosyllabic whole-word repetitions** (I-I-I-I see him)

B. Causes **anxiety about speaking** or **limits function**

C. Onset in **early developmental period**

D. Not due to speech-motor or sensory deficit, dysfluency assoc with neurological insult or AMC/AMD

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COFD (Stuttering) – Diagnostic Features

- Abnormal fluency + time patterning of speech
  - Frequent repetitions/prolongations of sounds/syllables
  - Other speech dysfluencies

- Interfering (academic, occupational, social)
  - Extent varies by situation → **more severe if special pressure**
  - Often **absent** during oral reading, singing, talking to pets/inanimate objects

COFD (Stuttering) – Associated Features

- Fearful anticipation of the problem may develop
  - May use **linguistic mechanisms** to avoid dysfluencies
    - Altering rate of speech, avoiding certain words/sounds
    - Avoiding certain speech situations (phone, public speaking)
  - Dysfluency exacerbated by stress + anxiety

- May be accompanied by **motor movements**
  - Eye blinks, tics, tremors of lip/face, jerking of head
  - Breathing movements, fist clenching

- Range of language abilities
  - Unclear relationship between fluency disorder + language abilities

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COFD (Stuttering) – Development & Course

- Onset
  - Ranges from age 2-7 → **80-90% by age 6**
  - **Typically gradual** (can be insidious or more sudden)
    - Repetition of initial consonants, first words, long words
    - Child may not be aware of dysfluencies
      - Progressive → eventually occurs on most meaningful words/phrases
  - As child becomes more aware → may develop avoidance behaviors
    - Avoiding public speaking, use of short/simple utterances

- Course
  - **65-85% recover** from dysfluency
  - **Severity at age 8** → predicts recover vs persistence

COFD (Stuttering) – Risk & Prognostic Factors

- Genetic & Physiological
  - Risk among **first-degree biological relatives = 3x higher** (vs gen pop)

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COFD (Stuttering) – Functional Consequences

- Stress + anxiety
  - May exacerbate dysfluencies
  - May impair social functioning

COFD (Stuttering) – Differential Diagnosis

- Sensory deficits
  - Hearing impairment, sensory deficits, speech-motor deficits
  - If speech dysfluencies in excess → can make dx
- Normal speech dysfluencies
  - If worsens with age → can make dx
- Medication side effects
- Adult-onset dysfluencies
  - Assoc with specific neurological insults, AMC, AMD (not DSM dx)
- Tourette's disorder
  - Vocal tics, repetitive vocalizations

Social (Pragmatic) Communication Disorder

Social (Pragmatic) Communication Disorder – Diagnostic Criteria

A. Difficulties in social verbal/nonverbal communication (need all 4/4):
   1. Deficits using communication for **social purposes** (greeting, sharing)
   2. Inability to **change/adapt** communication to **context/listener**
   3. Difficulty following **conversation conventions**
   4. Difficulty understanding **what is not explicitly stated**

B. Functional limitations

C. Onset in **early developmental period**

D. Not due to AMC/ANC, low abilities in grammar/word structure, ASD, IDD, GDD, AMD

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Social (Pragmatic) Communication Disorder – Diagnostic Features

- Primary difficulty with pragmatics
  - **Social use** of language + communication (verbal/non-verbal)
  - Understanding + following **social rules in naturalistic contexts**
  - **Changing language** according to needs of listener/situation
  - **Following rules** for conversations and storytelling

- Not better explained by low abilities in domains of structural language or cognitive ability

Social (Pragmatic) Communication Disorder – Associated Features

- Most common associated feature = **LANGUAGE IMPAIRMENT**
  - Hx delay in reaching language milestones
  - Structural language problems (language disorder)

- May **avoid social interactions**

- More likely to have:
  - **ADHD**
  - **Specific learning disorders**
  - **Behavior problems**

Social (Pragmatic) Communication Disorder – Development & Course

- Diagnosis **RARE younger than age 4**
  - By age 4-5, most children should posses **adequate speech/language**
  - To permit identification of specific deficits in social communication
- Milder forms → may not be apparent until **early adolescence**
  - More complex language + social interactions
- Variable outcomes
  - Some have **persisting difficulties** into adulthood
  - Some children **improve substantially** over time
    - But early pragmatic deficits may cause **lasting impairments**
    - Social relationships, behavior, skill acquisition, written expression

Social (Pragmatic) Communication Disorder – Development & Course

- Genetic & Physiological
  - Increased risk IF family history of:
    - **Autism spectrum disorder**
    - **Communication disorders**
    - **Specific learning disorders**

Social (Pragmatic) Communication Disorder – Differential Diagnosis

- Autism spectrum disorder
  - ASD → **restricted/repetitive patterns** of behavior/interests/activities
    - May only occur in early developmental period

- ADHD

- Social anxiety disorder
  - Overlapping sx → differentiate **timing of symptom onset**
  - S(P)CD → **NEVER** had effective social communication
  - SAD → social communication skills developed appropriately
    - But not utilized because of anxiety

- Intellectual disability
  - If social communication deficits in excess → can make dx

Unspecified Communication Disorder

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Unspecified Communication Disorder

- Does not meet any full criteria
- Clinician choose NOT to specific reason

Autism Spectrum Disorder

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Autism Spectrum Disorder – Diagnostic Criteria

A. Deficits in social communication + social interaction
   1. Deficits in **social-emotional reciprocity**
   2. Deficits in **non-verbal communicative behaviors**
   3. Deficits in **relationships (developing, maintaining, understanding)**

B. Restricted/repetitive behaviors/interests/activities (2+/4):
   1. **Stereotyped/repetitive** motor movements, use of objects, speech
   2. **Insistence on sameness**, inflexible routines, ritualized behavior
   3. **Highly restricted, fixated interests** with abnormal intensity/focus
   4. **Hyper/hypo-reactivity to sensory input** or unusual interest

C. Onset in early development
D. Sig functional impairment
E. Not better explained by IDD, GDD (may co-occur)

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Autism Spectrum Disorder – Diagnostic Specifiers

- *Specify current severity*
  - For Criteria A & B (separately)

- *Specify if:*
  - **With or without accompanying intellectual impairment**
    - Separate estimates of verbal/non-verbal skills
  - **With or without accompanying language impairment**
    - Separate evaluations of receptive + expressive language skills
  - **Associated with known medical or genetic condition or environmental factor**
    - Epilepsy, syndromes (Rett, Fragile X, Down), VPA, FASD, very LBW
  - **Associated with another neurodevelopmental, mental or behavioral disorder**
  - **With catatonia**

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Autism Spectrum Disorder – Diagnostic Specifiers

	Social Communication	Restricted/Repetitive Behaviors
Level 1 "support"	• Noticeable impairments without supports • **Difficulty initiating interactions** • Atypical/unsuccessful responses • May appear disinterested • Can speak **full sentences** • Conversations fail • Odd/unsuccessful attempts to make friends	• Interference with function in some contexts • **Difficulty switching activities** • Hampers independence • Difficulty organizing + planning
Level 2 "substantial support"	• Marked deficits • Apparent impairments even with supports • Decr/abnormal responses • **Simple sentences** • Limited interaction, to narrow special interests • Markedly odd nonverbal communication	• Difficulty coping with change • **Obvious to casual observer** • Interferes with function in variety of contexts • Distress/difficulty changing focus/action
Level 3 "very substantial support"	• Severe deficits cause severe impairments • Very limited initiation of social interactions • Minimal response • **Few words of intelligible speech** • Unusual approaches to meet needs only • Responds only to very direct approaches	• **Extreme difficulty coping with change** • Marked interference with function in all areas

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Autism Spectrum Disorder – Diagnostic Features

- Core diagnostic features evident in **developmental period**
  - Intervention, compensation, current supports → **may mask difficulties**
  - Multiple sources, clinician observations, caregiver history, self-report
  - **Standardized behavioral diagnostic** tools with good psychometrics
    - Caregiver interviews, questionnaires, clinical observation measures

- Manifestation vary greatly (severity, developmental level, age)
  - ASD now encompasses:
    - Early infantile autism, childhood autism, **Kanner's autism,** high-functioning autism, atypical autism, **Asperger's disorder,** pervasive developmental disorder NOS, and childhood disintegrative disorder

- Language deficits varies
  - Speech → **complete lack, poor comprehension,** **echoed speech**
    - May be stilted + overly literal language
  - Even if formal language skills intact → reciprocal communication impaired

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Autism Spectrum Disorder – Diagnostic Features

- **A1) Social-emotional reciprocity deficits**
  - Ability to engage with others, share thoughts/feelings
  - Clearly evident in young children with ASD, may show:
    - **Minimal initiation** of social interaction
    - **No sharing of emotions**
    - **Reduced imitation** of others' behavior

- If language exists → often one-sided
  - **Lacks social reciprocity**
  - **Language used to request/label** (vs comment, share, converse)

- In adults without intellectual disabilities/language delays
  - Difficulty **processing/responding to complex social cues**
    - Joining a conversation, what not to say
  - May develop **compensation strategies**
    - But still struggle in **novel/unsupported situations**
    - Suffer from **effort + anxiety of consciously calculating** what is socially intuitive for most individuals

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Autism Spectrum Disorder – Diagnostic Features

- **A2) Non-verbal communication deficits**
  - **Eye contact** → absent, reduced, atypical (vs cultural norm)
  - **Gestures** → limited repertoire, fail to use spontaneously
  - **Facial expression, body orientation** → odd, wooden, exaggerate
  - **Speech intonation**
  - **Overall poor integration for social communication**

- Early feature = **IMPAIRED JOINT ATTENTION**
  - Failure to follow someone's pointing or eye gaze

Autism Spectrum Disorder – Diagnostic Features

- **A3) Deficits in relationships (develop, maintain, understand)**
  - **Social interest** → absent, reduced, atypical
    - Rejection of others, passivity
    - Inappropriate approaches (may appear aggressive, disruptive)

- Evident in young children:
  - Lack of **shared social play + imagination**
  - Insistent on **playing by very fixed rules**

- Older individuals:
  - May struggle to understand **appropriate behavior by contexts**
  - May prefer **solitary activities**
  - May prefer to interact with **much younger or older people**

- Often desire to establish friendships
  - But **incomplete/unrealistic idea** of what friendship entails
  - One-sided, based solely on shared special interests

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Autism Spectrum Disorder – Diagnostic Features

- **B1) Stereotyped, repetitive behaviors**
  - **Simple motor stereotypies** → hand flapping, finger flicking
  - **Repetitive use of objects** → spinning coins, lining up toys
  - **Repetitive speech** → echolalia, parroting words, "you" referring to self
  - **Stereotyped speech** → use of words/phrases, prosodic patterns

- **B2) Excessive adherence to routines, restricted patterns**
  - **Resistance to change** → distress at small changes
    - Insistence on adherence to rules, rigid thinking
  - **Ritualized patterns of verbal/non-verbal behavior**
    - Repetitive questioning, pacing a perimeter

- **B3) Highly restricted, fixated interests**
  - Tend to be with **abnormal intensity/focus**
  - Eg. preoccupation with pan, vacuum cleaners, writing out timetables

Autism Spectrum Disorder – Diagnostic Features

- **B4) Hyper/hyporeactivity to sensory input**
  - Extreme responses to **specific sounds/textures**
  - Excessive **smelling/touching** of objects
  - Fascination with **lights/spinning objects**
  - Apparent **indifference to pain, heat, cold**
  - Extreme reaction/rituals **involving food** (taste, smell, texture, appearance)
    - May have excessive **food restrictions**
    - May be a **presenting feature** of ASD

- Many adults without intellectual/language disabilities
  - Learn to **suppress repetitive behavior in public**
  - Special interests may be source of **pleasure/motivation**
    - May provide avenues for **education + employment**
  - Sx must have been clearly present during childhood (if not current)

Autism Spectrum Disorder – Associated Features

- Intellectual ± language impairment
  - Even if average/high intelligence → may have **uneven profile of abilities**
  - Often **large gap** between intellectual + adaptive functional skills

- Motor deficits → often present
  - **Odd gait, clumsiness,** abnormal motor signs (walking on tiptoes)

- Self-injury → may occur (head banging, wrist biting)

- Disruptive/challenging behavior → **MORE COMMON in autism**
  - (vs other disorders, vs IDD)

- Anxiety, depression → adolescents + adults prone

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Autism Spectrum Disorder – Associated Features

- Catatonia
  - Some **"catatonia-like"** → slowing, freezing
  - Some **"full catatonia"** → mutism, posturing, grimacing, waxy flexibility
  - Greatest risk during **adolescent years**

Autism Spectrum Disorder – Prevalence

- Prevalence = **1%** (US + non-US)
  - **Similar in child + adult samples**
  - Unclear if true increase in frequency
    - Or reflects expansion of diagnostic criteria (DSM-IV to DSM5)

Autism Spectrum Disorder – Development & Course (1)

- Onset
  - Sx typically recognized during **2nd year of life** (age 12-24 months)
    - May be seen **earlier if severe developmental delay**
    - May be seen **later if more subtle sx** (more rare)

- Behavioral features → first evident in early childhood
  - **Lack of interest in social interaction** in first year of life

- May experience developmental plateaus or regression
  - Early developmental delays
  - **Loss of social/language skills** (rapid/gradual) → often in first 2 years
    - May be "red flag" (losses rare in other disorders)
    - If loss of non-social skills or after age 2 → unusual, medical work-up
      - Loss of self-care, toileting, motor skills **(Rett syndrome)**

Autism Spectrum Disorder – Development & Course (2)

- First symptoms of autism
  - **Delayed language** development, **unusual communication** patterns
  - **Lack of social interest**, unusual social interaction, **odd play patterns**
  - (rule out deafness)

- During second year
  - **Odd/repetitive behaviors + absence of typical play** → more apparent
  - Many typically developing children have strong preference, enjoy repetition
    - May be difficult to distinguish during preschool years
    - Based on type, frequency, intensity

- Some will be first diagnosed in adulthood
  - May be prompted by autism in child, breakdown of relations at work/home
  - Need developmental history (no good social/communication skills as child)

Autism Spectrum Disorder – Development & Course (3)

- NOT a degenerative disorder
  - Symptoms most marked in **early childhood + early school years**
    - Developmental gains typical in later childhood
    - Typical for **learning + compensation to continue throughout life**
  - **MOST improve behaviorally** during adolescence (minority deteriorate)
  - Little known about ASD in old age

- Minority live/work independently in adulthood
  - Those who do → **superior language/language abilities**
    - Able to find niche that matches special interests/skills
    - May remain **socially naïve/vulnerable**
    - Difficulty **organizing practical demands** without support
    - Prone to **anxiety + depression**
  - **Compensation strategies + coping mechanisms** → to mask difficulties
    - May suffer from **stress + effort** to maintain socially acceptable facade

Autism Spectrum Disorder – Risk & Prognostic Factors

- Best established prognostic risk factors for individual outcome
  - **Associated intellectual disability**
  - **Associated language impairment**
    - **Functional language by age 5** is good prognostic sign
  - **Additional mental health problems**
  - **Epilepsy** → greater intellectual/verbal disability

- Environmental (non-specific)
  - Advanced **parental age**, **low birth weight**, fetal exposure to **valproate**

- Genetic & Physiological
  - Heritability = **37 – 90%** (based on twin concordance studies)
  - **15% of cases** → **known genetic mutation** (but not fully penetrant)
    - Rest of cases are polygenic risk

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Autism Spectrum Disorder – Culture-Related Issues

- Marked impairment compared to cultural norms
  - Cultural + socioeconomic factors **may affect age at recognition/dx**
  - US → later/underdiagnosis in African American children

Autism Spectrum Disorder – Gender-Related Issues

- More commonly diagnosed in **MALES (4x)**

- **FEMALES** with ASD
  - More likely to have **accompanying IDD**
  - May suggest underdiagnosis of females without IDD, language delays

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Autism Spectrum Disorder – Functional Consequences

- Young children
  - Social/communication deficits → may **hamper learning**
  - Insistence on routines → may affect **eating, sleeping, routine care**

- Adaptive skills typically below measured IQ
  - **Academic achievement affected** (even if above-average intelligence)
    - Difficulty planning, organizing, coping with change
  - During adulthood → **difficulty establishing independence, employment**
    - Continuing rigidity, difficulty with novelty
    - Poor psychosocial functioning

- Functional consequences in old age unknown
  - Social isolation, communication problems (decr help-seeking)

Autism Spectrum Disorder – Differential Diagnosis

- Rett Syndrome
  - **Regressive phase (age 1-4)** → disruption of social interaction
    - Young girls may appear to meet ASD criteria
  - After this period → **most improve social communication skills**
    - No longer concern about ASD
  - Only consider if ALL diagnostic criteria of ASD met

- Selective mutism
  - **Early development not disturbed**
  - Appropriate communication skills in certain contexts
  - Social reciprocity not impaired, no restricted/repetitive patterns of behavior

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Autism Spectrum Disorder – Differential Diagnosis

- Language disorders, social (pragmatic) communication disorder
  - May be communication problems, secondary social difficulties
  - SLD → no abnormal non-verbal communication, **no restricted/repetitive patterns of behavior**
  - S(P)CD → **no restricted/repetitive patterns of behavior**

- Intellectual disability without ASD
  - May be difficult to distinguish in very young children
  - **No apparent discrepancy** between all intellectual skills
    - Additional ASD dx if social communication impairment in excess to developmental of nonverbal skills (fine motor, etc.)

- Stereotypic movement disorder
  - May dx BOTH if stereotypies cause **self-injury** + **become focus of tx**

Autism Spectrum Disorder – Differential Diagnosis

- ADHD
  - Both have abnormal attention, hyperactivity
  - Additional ADHD dx if symptoms in excess (can dx BOTH)

- Schizophrenia
  - Childhood onset schizophrenia → usually develops after period of **normal or near normal development**
  - **Prodromal state** (social impairment, atypical interests)
  - **Hallucinations, delusions** → not features of ASD
    - Consider concrete interpretations of questions

Autism Spectrum Disorder – Comorbidity

- Frequent associations
  - **Intellectual impairment, structural language disorder** (specifiers)
  - **Psychiatric symptoms** (not meeting disorder criteria)
    - **70% have 1** comorbid mental disorder
    - **40% have 2+** comorbid mental disorders
    - Can diagnose ASD with ADHD, DCD, anxiety, depressive disorders
    - If non-verbal/language deficits, may notice changes in sleep, eating, challenging behaviors → evaluation for anxiety or depression

- Specific learning difficulties, DCD → COMMON
- Avoidant-restrictive food intake disorder → frequent presenting
  - Extreme/narrow food preferences may persist

- Medical conditions
  - **Epilepsy, sleep problems, constipation**

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Attention-Deficit/Hyperactivity Disorder

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ADHD – Diagnostic Criteria (*at a glance*)

A. Inattention and/or hyperactivity/impulsivity
   1. **Inattention for >6 mo**, (6+/9)
   2. **Hyperactivity/impulsivity for >6 mos**, (6+/9)
   3. Inconsistent with developmental level, negative impact on activities

B. Symptoms present **before age 12**

C. Symptoms present in **2+ settings**

D. Interferes with function

E. Not exclusively during psychosis, not better explained by AMD

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ADHD – Diagnostic Criteria

- A1) Inattention, >6 mos (6+/9):
  - (If age ≥17 → need 5+/9)
  1. Not attentive to **details**, careless mistakes
  2. Difficulty sustaining **attention**
  3. Does not seem to **listen**
  4. Does not follow through on **instructions**, fails to finish work
  5. Difficulty **organizing** tasks/activities
  6. **Avoids**/dislikes/reluctant with tasks requiring sustained mental effort
  7. **Loses** necessary things
  8. Easily **distracted**
  9. **Forgetful** in daily activities

ADHD – Diagnostic Criteria

- A2) Hyperactivity/impulsivity, >6 mos (6+/9):
  - (If age ≥17 → need 5+/9)
  1. Often **fidgets** with hands/feet, squirms in seat
  2. Often **leaves seat** when expected to remain seated
  3. Often **runs about** when inappropriate
  4. Often unable to play **quietly**, engage in leisure activities quietly
  5. Often **"on the go".** "driven by a motor"
  6. Often **talks excessively**
  7. Often **blurts out** answer, before question completely
  8. Often has **difficulty waiting their turn**
  9. Often **interrupts/intrudes** on others

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ADHD – Diagnostic Specifiers

- *Specify whether:*
  - **Combined presentation**
  - **Predominantly inattentive presentation**
  - **Predominantly hyperactive/impulsive presentation**

- *Specify if:*
  - **In partial remission**

- *Specify current severity:*
  - **Mild:** few sx in excess of those required, only minor impairments
  - **Moderate:** between mild + severe
  - **Severe:** many sx in excess of those required, marked impairment

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ADHD – Diagnostic Features

- Inattention
  - Wandering off task, lacking persistence, difficulty sustaining focus
  - Being disorganized
  - **NOT due to defiance or lack of comprehension**

- Hyperactivity
  - Excessive motor activity, excessive fidgeting/tapping/talking

- Impulsivity
  - Hasty actions, without forethought, high potential for harm
  - Desire immediate rewards or inability to delay gratification
  - Social intrusiveness
  - Important decisions without considering of long-term consequences

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ADHD – Diagnostic Features

- Begins in childhood
  - Several sx must be **present before age 12 (subclinical)**
  - Adult recall of childhood sx tends to be unreliable → need collateral

- Must be present in multiple settings
  - Sx may vary depending on context
  - May have minimal sx if:
    - Receiving **frequent rewards**
    - **Closer supervision**
    - Novel settings
    - **Especially interesting activities**
    - Consistent external stimulation
    - Interacting in one-on-one situations

ADHD – Associated Features

- Mild delays in language, motor, social development
  - Often co-occur with ADHD (but not specific)
  - May have **low frustration tolerance, irritability, mood lability**
  - **Academic/work performance** often impaired
    - Even without specific learning disorder
  - Inattention associated with underlying **cognitive processes**
    - May notice problems on tests of attention, executive function, memory (tests are **not** diagnostic)

- By early adulthood → **incr risk of suicide attempt**
  - Esp if comorbid with mood, conduct, substance use disorders

- No diagnostic biological marker
  - EEG → incr slow waves
  - MRI → decr total brain volume, ?delayed post-to-anterior cortical maturation

- If known genetic cause (Fragile X, 22q11) → can still dx ADHD

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ADHD – Development & Course

- May be difficult to distinguish from normal behavior before age 4

Toddler	• May observe excessive motor activity as toddler
Preschool	• Mainly hyperactivity
Elementary school	• Inattention more prominent • When **ADHD usually identified**
Early adolescence	• Relatively stable course • Some have worsened course (antisocial behaviors)
Adolescence	• Motor hyperactivity less common • May be fidgetiness, inner feeling of jitteriness, restless, impatience
Adulthood	• Inattention, restlessness, impulsivity persist • Hyperactivity diminished • Substantial proportion still relatively impaired

ADHD – Risk & Prognostic Factors

- Temperamental (predisposing, not specific to ADHD)
  - Decr **behavioral inhibition**, decr effortful control/constraint
  - **Negative emotionality**
  - Incr **novelty seeking**

- Environmental
  - **Very low birth weight** (<1500 grams) → 2-3x risk for ADHD
    - Most children with LBW DO NOT develop ADHD
  - Correlation with smoking during pregnancy → common genetic risk
  - Minority of cases may be related to diet
  - May have hx of **child abuse, neglect, multiple foster placements**
  - **In utero exposures** (correlations, unknown if causal)
    - Neurotoxins (lead), infections (encephalitis), alcohol exposure

ADHD – Risk & Prognostic Factors

- Genetic & Physiological
  - **Substantial heritability**, some specific genes correlated (not causal)
    - Incr risk among 1º biological relatives with ADHD
  - Possible influences on ADHD sx
    - Visual/hearing impairments, metabolic abnormalities, sleep disorders, nutritional deficiencies, epilepsy
  - **NO specific physical features**
    - May have incr rates of **minor physical anomalies**
      - Hypertelorism, highly arched palate, low-set ears
  - May have subtle motor delays, neurological soft signs

- Course modifiers
  - **Family interactions patterns** in early childhood
    - May influence its course (unlikely to cause ADHD)
    - Or contribute to secondary development of **conduct problems**

ADHD – Culture-Related Issues

- Differences in ADHD prevalence rates across regions
  - Mainly attributable to difference diagnostic practices
  - May be cultural variation in attitudes
    - Lower rates in African American, Latinos (vs white)

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ADHD – Gender-Related Issues

- More frequent in **MALES**
  - Children → **MALES (2x)**
  - Adults → **MALES (1.6x)**
  - Females more likely to present primarily with **inattentive** features

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ADHD – Functional Consequences

- Inattention → academic deficits, school problems, peer neglect
- Hyperactivity/impulsivity → peer rejection, accidental injury

- Children
  - Decr school performance, academic attainment, reduced intellectual scores
  - More likely to develop **conduct disorder** in adolescence
    - More likely to develop **antisocial PD** in adulthood
    - Consequently incr likelihood for **SUD + incarceration**

- Adults
  - Poorer **occupational performance**, attainment, attendance
  - Higher probability of unemployment, incr interpersonal conflict

- Social impairment
  - Difficulty with sustained efforts → **interpreted by others as laziness**, irresponsibility, failure to cooperate
  - Family relationships → discord, negative reactions
  - Peer relationships → peer rejection, neglect, teasing

- More likely to be **injured, traffic accidents/violations, obesity**

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ADHD – Differential Diagnosis

- Oppositional defiant disorder
  - Resist work/school because they **resist conforming to others' demands**
  - **Negativity, hostility, defiance**
  - ADHD may develop secondary oppositional attitudes

- Intermittent explosive disorder
  - Share high levels of impulsive behavior
  - IED → serious aggression towards others, no attention difficulties
    - Rare in childhood, but both may be dx together

- Other neurodevelopmental disorders
  - Incr motor activity vs repetitive motor behavior
    - Stereotypic movement disorder, autism → fixed, repetitive
    - Tourette's disorder → freq multiple tics
    - ADHD → fidgetiness (not tics)

ADHD – Differential Diagnosis

- Specific learning disorder
  - May appear inattentive → due to frustration, lack of interest, inability
    - Not impairing outside of academic work

- Intellectual disability
  - ADHD sx common if placed in setting **in**appropriate to intelligence
    - Not present during non-academic tasks
  - Can dx BOTH, if ADHD sx are in excess of mental age

- Autism spectrum disorder
  - Both exhibit inattention, social dysfunction, challenging behavior
  - ASD → social disengagement, isolation, indifference to social cues
    - May have tantrums due to intolerance of change
  - ADHD → tantrums due to impulsivity, poor self-control

ADHD – Differential Diagnosis

- Reactive attachment disorder
  - May show social disinhibition → not full ADHD syndrome
  - Lack of enduring relationships

- Anxiety disorders → inattention due to worry, rumination
- Depressive disorders → poor concentration (only during MDE)
- Bipolar disorder
  - May have incr activity, poor concentration, incr impulsivity **(EPISODIC)**
  - With accompanying euphoria, grandiosity, mood lability, etc.
    - ADHD → may have mood lability within day
  - **Bipolar rare in pre-adolescents** (even with severe irritability/anger)
  - ADHD common if excessive anger/irritability

ADHD – Differential Diagnosis

- Disruptive mood dysregulation disorder
  - Pervasive irritability, frustration intolerance
  - Not impulsiveness, disorganized attention
  - Most children with DMDD → also meet ADHD (dx BOTH)

- Substance use disorders
  - Need clear evidence of ADHD before substance misuse

- Personality disorders
  - In adolescents/adults → ddx borderline, narcissistic, other PDs
    - Disorganization, social intrusiveness, emotional dysregulation, cognitive dysregulation
  - ADHD → no fear of abandonment, self-injury, extreme ambivalence

ADHD – Differential Diagnosis

- Psychotic disorders

- Medication-induced sx of ADHD
  - Bronchodilators, isoniazid, neuroleptics (akathisia), thyroid-replacement
  - Dx as other specified or unspecified other (or unknown) substance-related disorder

- Neurocognitive disorders
  - Early major/mild NCD → not assoc with ADHD
    - May present with similar clinical features
    - LATE ONSET

ADHD – Comorbidity

- Comorbidities frequent
  - **ODD** → co-occurs in 50% of ADHD-C, 25% of ADHD-I
  - **Conduct disorder** →co-occurs 25% of ADHD-C

  - Most DMDD have ADHD (less ADHD have DMDD)
  - Specific learning disorder commonly co-occurs with ADHD

  - Anxiety disorders, MDD → occur in minority of ADHD (more than gen pop)
  - IED → minority of adults with ADHD (more than gen pop)
  - SUD → minority of adults with ADHD (more than gen pop)

- Personality disorders, antisocial PD
- OCD, tic disorders, autism spectrum disorder

Other Specified ADHD

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Other Specified ADHD

- Does not meet any full criteria
- Clinician chooses to specify reason

- ADHD with insufficient inattention symptoms

Unspecified ADHD

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Unspecified ADHD

- Does not meet any full criteria
- Clinician chooses NOT to specify reason

Specific Learning Disorder

Specific Learning Disorder – Diagnostic Criteria

A. <u>Difficulties learning + using academic skills, >6 months (1+/6):</u>
   1. Inaccurate, slow, effortful **word reading**
   2. Difficulty understanding **meaning of what is read**
   3. Difficulties with **spelling**
   4. Difficulties with **written expression**
   5. Difficulties mastering **numbers** (sense, facts, calculation)
   6. Difficulties with **mathematical reasoning**

B. Substantially **below expected for age**, interferes with **function**

C. Began during **school-age years** (may fully manifest later)

D. Not better explained by AMD, AMC, ANC, psychosocial factors

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Specific Learning Disorder – Diagnostic Specifiers (1)

- *Specify if (include all impaired domains + subskills):*
  - **With impairment in reading**
    - Word reading accuracy
    - Reading rate or fluency
    - Reading comprehension
    - *Dyslexia* → problems with word recognition, decoding, spelling
  - **With impairment in written expression**
    - Spelling accuracy
    - Grammar + punctuation accuracy
    - Clarity or organization of written expression
  - **With impairment in mathematics**
    - Number sense
    - Memorization of arithmetic facts
    - Accurate or fluent calculation
    - Accurate math reasoning
    - *Dyscalculia* → problems with processing numerical information, learning arithmetic facts, performing calculations

Specific Learning Disorder – Diagnostic Specifiers (2)

- *Specify current severity:*
  - **Mild:** some difficulties in 1-2 academic domains
    - Able to compensate or function well with supports
  - **Moderate:** marked difficulties in 1+ academic domains
    - Unlikely to become proficient without intensive + specialized teaching
    - Some support required for part of day to complete activities
  - **Severe:** severe difficulties in several academic domains
    - Unlikely to learn skills without intensive + specialized teaching ongoing
    - May not be able to complete activities efficiently, even with supports

Specific Learning Disorder – Diagnostic Features

- SLD = neurodevelopmental disorder
  - **Biological basis** for cognitive abnormalities, assoc with behavioral signs
    - Genetic, epigenetic, environmental factors
    - Affect brain's ability to perceive/process verbal/non-verbal information

- A) Difficulties learning key academic skills
  - Onset during **formal school** (developmental period)
  - Academic skills have to be **taught + learned explicitly** (vs talk/walk)
    - **Reading, spelling, writing, mathematics**
  - SLD disrupts normal pattern of learning academic skills
    - Not just due to lack of learning opportunity or inadequate instruction
    - May affect other subjects (history, science, social studies)
  - **Dyslexia** → one of the MOST COMMON manifestations
    - Difficulty mapping letters with sounds, to read printed words

- Persistent learning difficulties → >6 months, despite extra help

Specific Learning Disorder – Diagnostic Features

- A1) Word reading *(aka dyslexia)*
  - Single word difficulty, frequently guessing words, difficulty sounding out
- A2) Understanding meaning of what is read
  - May read accurately, but not understand sequence, relationship, inferences or deeper meaning
- A3) Spelling
  - May add/omit/substitute vowels or consonants
- A4) Written expression
  - Frequent grammatical/punctuation errors, poor paragraph organization, written expression of ideas lacks clarity
- A5) Numbers (sense, facts, calculating)
  - Poor understanding of magnitude + relationships, counts on fingers, add single-digit numbers (instead of recalling math fact), gets lost in arithmetic computation, may switch procedures
- A6) Mathematical reasoning
  - Severe difficulty applying math concepts, facts, procedures to solve quantitative problems

Specific Learning Disorder – Diagnostic Features

- Assessment of learning difficulties
  - Observable, probed through clinical interview, rating scales,
  - Cumulative school reports, educational/psychological assessments
  - Portfolios of evaluated work, curriculum-based measures

- B) Performance well below average for age
  - Low academic achievement for age
    - OR average achievement with significant supports
    - In adults → avoidance of activities requiring academic skills
  - Requires **psychometric evidence** (no natural cut-off)
    - At least 1.5 standard deviations below (<78, 7th percentile)
    - Lower thresholds (1.0 – 2.5 SD) may used with clinical judgement

- C) Apparent in early school years (may fully manifest later)

Specific Learning Disorder – Diagnostic Features

- <u>"Specific" learning disorders</u>
  - Not due to **intellectual disabilities** (IDD, GDD)
    - Otherwise demonstrate normal levels of functioning (IQ >70)
    - "Unexpected academic underachievement"
    - May occur in intellectually "gifted"
      - Adequate academic function with compensatory strategies/effort
  - Not due to **general external factors**
    - Economic/environmental disadvantage, chronic absenteeism
  - Not due to **neurological, motor, vision/hearing disorders**
    - If present, assessments should account
  - May be restricted to one academic skill/domain

- <u>Can only be diagnosed AFTER formal education starts</u>
  - Any point afterwards (children, adolescents, adults)
  - **Typically persists into adulthood**

Specific Learning Disorder – Associated Features

- Frequently preceded by delays in preschool years
  - Attention, language, motor skills → may persist or co-occur with SLD
- Uneven profile of abilities → common
  - May have poor reading/writing, but high drawing/design/visuospatial
  - Typically have poor performance on tests of **cognitive processing**
    - Cognitive deficits found in other neurodevelopmental disorders
    - Therefore assessment of cognitive processing deficits NOT required
- **Increased risk for suicidal ideation + attempts** (across ages)
- No known biological markers that are useful for diagnosis
  - Circumscribed alterations in cognitive processing, brain structure, function
  - Genetic differences at group level

Specific Learning Disorder – Prevalence

- Among school-age children = **5 – 15%**
  - Across different languages + cultures

- Prevalence in adults = **4%**

Specific Learning Disorder – Development & Course (1)

- Onset, recognition, diagnosis → usually **elementary school**
  - When required to learn to read, spell, write, math
  - **Developmental delays common** in early childhood, before school
    - Language, rhyming, counting, fine motor
  - May have behavioral **manifestations** (reluctance to engage, oppositional)

- Course → **lifelong,** persists into adulthood, but variable
  - Depends on environment, support systems, intervention
  - Symptoms may change with age → persisting or shifting difficulties

- Preschool children
  - Lack of interest in games with language sounds (repetition, rhyming)
  - Trouble learning nursery rhymes, learning to count
  - Frequent baby talk, mispronounce words
  - Difficulty remember names of letters, numbers, days of week
  - May fail to recognize letters in own names

Specific Learning Disorder – Development & Course (2)

Preschool	Kindergarten
• Low interest in **games with language** sounds (repetition, rhyme) • Trouble learning **nursery rhymes** • Frequent **baby talk**, **mispronounce words** • Difficulty remember **names of letters**, **numbers, days of week** • Difficulty **learning to count** • May fail to recognize **letters in own names**	• Unable to **recognize/write letters**, **own name** • May use **invented spelling** • Trouble splitting spoken **words into syllables** • Trouble recognizing **words that rhyme** • Trouble **connecting letters with their sounds** • Trouble recognizing **phonemes**

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Specific Learning Disorder – Development & Course (3)

Elementary School
• Difficulty with **letter-sound correspondence**, **fluent word decoding**, **spelling, math facts** • **Reading aloud** difficulties • Difficulty understanding **magnitude of numbers (spoken or written)**

Primary (Grades 1-3)	Middle (Grades 4-6)
• Difficulty with **phonemes, reading one-syllable words**, common **irregularly spelled words** • **Reading errors** in connecting sounds + letters • **Sequencing numbers + letters** • **Number facts** • **Arithmetic procedures** (adding, subtracting) • May **avoid math**	• Mispronounce **long, multisyllable words** • Confuse **words that sound alike** • Difficulty remembering **dates, names, phone numbers** • Difficulty **completing homework or tests on time** • Poor **reading comprehension** • Trouble **reading prepositions** • **Poor spelling + written work** • May **avoid reading aloud**

Specific Learning Disorder – Development & Course (4)

Adolescents	Adults
• May have **mastered word decoding** • **Reading still slow, effortful** • Difficulties in **reading comprehension, written expression** • **Poor mastery of math facts**, problem solving • May **avoid activities demanding** reading or arithmetic	• **Spelling mistakes** • **Reading slow, effortful** • Trouble **pronouncing multisyllable words** • Need to **reread material to understand** • Difficulty **making inferences** from written or numerical information • May **avoid activities demanding** reading or arithmetic • May use **alternative approaches** (audio)

Specific Learning Disorder – Development & Course (5)

- Alternative clinical expression
  - **Circumscribed learning difficulties**, persistent across lifespan
    - E.g. basic number sense, or word identification/spelling

- Avoidance in demanding activities → COMMON (all ages)
  - May have episodes of **severe anxiety**, panic attacks, somatic complaints
    - Common across lifespan
    - Accompany both circumscribed + broader expression of SLD

Specific Learning Disorder – Risk & Prognostic Factors

- Environmental
  - Incr risk for SLD → **prematurity, very LBW, prenatal nicotine exposure**

- Genetic & Physiological
  - **Aggregates in families** → esp if reading, spelling, math affected
    - RR of SLD increased in 1^{o} relatives with same SLD (vs without)
      - **Reading = 4-8x, Math = 5-10x**
    - Family hx of dyslexia + parental literacy skills
      - Predicts literacy problems or SLD in offspring
  - **High heritability** for most manifestations of learning abilities/disabilities
    - Reading in **alphabetic + non-alphabetic languages**
    - High covariation between various learning difficulties

Specific Learning Disorder – Risk & Prognostic Factors

- Course modifiers
  - **Inattentive behavior** (preschool age) → predictor of:
    - Later difficulties in reading + math (not necessarily SLD)
    - Non-response to effective academic interventions
  - **Delays in speech, language, cognitive processing** (preschool age)
    - Predicts later SLD in reading + written expression
  - **ADHD comorbidity** → predictive of worse mental health outcome
  - **Evidence-based interventions** → may improve/ameliorate difficulties
    - Systematic, intensive, individualized instruction
    - May mitigate poor outcomes

Specific Learning Disorder – Culture-Related Issues

- Occurs across languages, culture, race, socioeconomic
  - May vary by spoken/written symbol systems, cultural, educational practice
    - English → inaccurate, slow reading of single words
    - Other languages → just slow reading (but accurate)
      - Spanish, German → more direct mapping between sounds + letters
      - Chinese, Japanese → non-alphabetic languages
    - Limited proficiency in English vs SLD

- Risk factors for SLD in English-language learners
  - Family hx of SLD or language delay in **native language**
  - Family hx of learning difficulties in English, failure to catch up to peers

Specific Learning Disorder – Gender-Related Issues

- SLD more common in **MALES** **(2-3x)**
  - Independent of ascertainment bias, definitional/measurement variation, language, race, SES

Specific Learning Disorder – Functional Consequences

- Can have negative functional consequences across lifespan
  - Lower academic attainment
  - Higher rates of high school dropout
  - Lower rates of post-secondary education
  - High levels of psychological distress
  - Poorer overall mental health
  - Higher rates of unemployment/under-employment
  - Lower incomes

- Risk factors for poor mental health outcomes (incl suicidality)
  - **School dropout**
  - **Co-occurring depressive sx**

- Protective factors for better mental health outcomes
  - High levels of social/emotional support

Specific Learning Disorder – Differential Diagnosis

- Normal variations in academic attainment
  - Distinguish SLD vs external factors
- Intellectual disability
  - SLD in presence of normal intellectual functioning (IQ >70)
  - Can dx both if learning difficulties in excess of those usually associated with ID
- Learning difficulties due to neurological/sensory disorders
- Neurocognitive disorders
- ADHD → can dx both
  - Learning (SLD) vs performing (ADHD) academic skills
- Psychotic disorders
  - Often see a rapid decline in functional domains

Specific Learning Disorder – Comorbidity

- Commonly co-occurs with other disorders
  - Neurodevelopmental
    - ADHD, autism spectrum disorder
    - Communication disorders, developmental coordination disorder
  - Other mental disorders
    - Anxiety, depressive, bipolar disorders
  - May make **testing more difficult**

Developmental Coordination Disorder

DCD – Diagnostic Criteria

A. Impaired acquisition + execution of **coordinated motor skills**, substantially below expected age + opportunity
   - Manifested as clumsiness, slowness, inaccuracy

B. Interferes with function

C. Onset in **early developmental period**

D. Not better explained by IDD, visual impairment or ANC

DSM5

DCD – Diagnostic Features

- Manifestation varies with age
  - **Young children** → may or may not achieve typical **motor milestones**
    - May be delayed in stairs, pedaling, buttoning, puzzles, zippers
    - Even if skill achieved, execution may be slow or less precise
  - **Older children, adults** → skills in **education, work affected**
    - Puzzles, building models, playing ball games
    - Handwriting (legibility, speed), typing, driving, self-care skills

- Onset in early developmental period
  - Typically NOT dx before **age 5**
    - Considerable variation in acquisition in early childhood
    - Other causes of motor delay may not have fully manifested

- Not discrete subtypes → may be mainly gross or fine motor
  - Also known as *childhood dyspraxia, specific developmental disorder of motor function, clumsy child syndrome*

DCD – Associated Features

- <u>May have additional motor activity</u>
  - **Choreiform movements** (of unsupported limbs) or **mirror movements**
  - Considered "overflow" movements, usually suppressed
  - *Neurodevelopmental immaturities, neurological soft signs* (vs abnormality)
  - Unclear role in dx

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DCD – Prevalence

- Prevalence in age 5-11 = **5-6%**
  - Age 7 → 1.8% dx with SEVERE DCD, 3% with PROBABLE DCD

- More commonly affects **MALES (2-7x)**

DCD – Development & Course

- Onset → in **early childhood**
  - First signs → delayed motor milestones, attempting certain motor tasks
    - Holding knife + fork, buttoning clothes, playing ball games
  - **Middle childhood** → puzzles, building models, handwriting
    - Organizing (when motor sequencing + coordination required)

- Course → variable, but stable
  - **50-70% continue into adolescence**
  - **Early adulthood** → difficulty learning complex/automatic motor skills
    - Driving, using tools
    - Inability to take notes, handwrite quickly (may affect work)
  - Comorbidities have additional impact on presentation, course, outcome

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DCD – Risk & Prognostic Factors

- Environmental
  - **Prenatal alcohol exposure**
  - **Preterm, LBW**

- Genetic & physiological
  - Impairments in underlying neurodevelopmental processes
    - Esp **visual-motor perception, visuospatial mentalizing**
    - Affects **rapid motoric adjustments** in more complex movements
    - ? Cerebellar dysfunction
  - Consistent **co-occurrence in twins** → only in SEVERE cases
    - Otherwise shared genetic effect only proposed

- Course modifiers
  - **ADHD + DCD** → more impairment (than ADHD without DCD)

DCD – Culture-Related Issues

- Occurs across cultures, races, SES
  - Consider whether appropriate opportunities to learn/practice activities

DCD – Functional Consequences

- Impaired function in activities of daily living
  - Poor **self-esteem + sense of self-worth**
  - Emotional/behavioral problems
  - Impaired **academic achievement**
  - Decr physical activity
    - Decr participation in team play/sports
    - Poor physical fitness
    - Obesity

- Impairment increases with comorbidities

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DCD – Differential Diagnosis

- Motor impairments due to AMC
  - **Visual impairment**, **neuromuscular disorders**
  - Specific **neurological disorders**
    - Cerebral palsy, progressive cerebellar lesions
  - **Joint hypermobility syndrome** → hyperextensible joints, pain
- Intellectual disability
  - Can dx both if motor difficulties in excess of expected with IDD
- ADHD → can dx both
  - Whether clumsiness due to distractibility + impulsiveness (vs DCD)
- Autism spectrum disorder → can dx both
  - May just be uninterested in tasks requiring complex coordination skills

DCD – Comorbidity

- MOST COMMON = **ADHD (50% co-occurrence)**
  - Just inattention problems also common

- Common comorbidities
  - Speech + language disorder
  - Specific learning disorder (esp reading + writing)
  - Autism spectrum disorder
  - Disruptive + emotional behavior problems
  - Joint hypermobility syndrome

- Comorbidity clusters
  - Severe reading disorders, fine motor problems, handwriting problems
  - Impaired movement control, motor planning

Stereotypic Movement Disorder

Stereotypic Movement Disorder – Diagnostic Criteria

A. **Repetitive, driven, purposeless motor behavior**
   - Hand shaking/waving, body rocking, head banging, self-biting, hitting self

B. Interferes with function, may result in self-injury

C. Onset in **early developmental period**

D. Not due to substance, ANC, AMD

Stereotypic Movement Disorder – Diagnostic Specifiers

- *Specify if:*
  - **With self-injurious behavior** (or requiring preventative measures)
  - **Without self-injurious behavior**

- *Specify if:*
  - **Associated with a known medical or genetic condition, neurodevelopmental disorder or environmental factor**
    - (Lesch-Nyhan syndrome, intellectual disability, intrauterine alcohol exposure)

- *Specify if:*
  - **Mild:** easily suppressed (by sensory stimulus/distraction)
  - **Moderate:** requires explicit protective measures + behavioral modification
  - **Severe:** requires continuous monitoring + protective measures to prevent serious injury

Stereotypic Movement Disorder – Diagnostic Features (1)

- A) Repetitive, seemingly driving, purposeless motor behavior
  - **Often rhythmical** → movements of head, hands, body
    - **No obvious adaptive function**
    - May or may not respond to efforts to stop them
  - In typically developing children → repetitive movement may be stopped
    - Suppressed when attention directed to child, or distracted
  - In neurodevelopmental disorders → movement typically LESS responsive
    - May try to **self-restrain** (sitting on hands, use protective device)

- Behaviors vary → individual "signature" patterns
  - **Non-self-injurious** → body rocking, bilateral hand flapping/rotating,
    - Flicking/fluttering fingers in front of face, arm flapping, head nodding
  - **Self-injurious** → head banging, face slapping, biting (hands, lips, etc)
    - Eye poking (esp in visual impairment)
  - May be combination of multiple movements

Stereotypic Movement Disorder – Diagnostic Features (2)

- Frequency varies
  - May occur **many times during day** → seconds to minutes
    - May have **weeks between episodes**
  - Vary in context → engrossed in activity, excited, stressed, fatigued, bored
  - "Apparently" purposeless → but may **reduce anxiety** (external stressors)

- Presence of stereotypic movements
  - May indicate **undetected neurodevelopmental problem** (esp age 1-3)

Stereotypic Movement Disorder – Prevalence

- Simple stereotypic movements
  - Common in young typically developing children (e.g. rocking)
  - Complex stereotypical movements → **much less common (3-4%)**

- If IDD → **4 – 16% engage in stereotypy + self-injury**
  - Greater risk if severe IDD
  - If IDD living in residential facilities → **10 – 15% have SMD + self-injury**

Stereotypic Movement Disorder – Development & Course

- Stereotypic movements → typically begin within first 3 years
  - Simple → **common in infancy, part of motor mastery**
  - Complex → **80% before age 2**
    - 12% between age 2-3
    - 8% after age 3
  - In most typically developing → **resolve over time, can be suppressed**

- Onset of complex motor stereotypies
  - **In infancy, or later in developmental period**

- If IDD → stereotyped, self-injurious behaviors **may persist**
  - Pattern of self-injury may change

Stereotypic Movement Disorder – Risk & Prognostic Factors

- Environmental
  - **Social isolation** → risk factor for self-stimulation
    - May progress to stereotypic movements with repetitive self-injury
  - **Environmental stress** may be a trigger
  - **Fear** may incr frequency of stereotypic behavior

- Genetic & physiological
  - **Lower cognitive functioning** → greater risk, poorer response to tx
  - **Mod-severe IDD** → more frequent stereotypic movements
    - Syndromes (Rett syndrome), environmental (insufficient stimulation)
  - **Neurogenetic syndromes** → behavioral phenotype
    - Lesch-Nyhan syndrome (dystonic movements, self-mutilation)
    - Rett syndrome, Cornelia de Lange syndrome (hand-mouth stereotypy)
  - **Painful medical conditions**
    - Middle ear infections, dental problems, GERD

Stereotypic Movement Disorder – Culture-Related Issues

- Occurs in all races + cultures
  - Cultural attitudes may result in delayed diagnosis

Stereotypic Movement Disorder – Differential Diagnosis (1)

- Normal development
  - Simple stereotypic movements → **common in infancy, early childhood**
    - May occur in transition from sleep to awake
    - Usually **resolves with age**
  - Complex stereotypies → LESS common
    - Can usually be **suppressed by distraction or sensory stimulation**
  - Rarely affects daily routine or cause distress

- Autism spectrum disorder
  - Stereotypies may be presenting sx of ASD
  - Also deficits of social communication/reciprocity, rigid behaviors/interests
  - **Can dx both** if self-injury or if stereotypies sufficiently severe

Stereotypic Movement Disorder – Differential Diagnosis (2)

Tics	Stereotypies
• Later onset (age 5-7)	• Earlier onset (before age 3)
• Variable in presentation	• Consistent, fixed pattern
• Eyes, face, head, shoulders	• Arms, hands, entire body
• Brief, rapid, random, fluctuating	• Fixed, rhythmic, prolonged
• Both reduced by distraction	• Both reduced by distraction

Stereotypic Movement Disorder – Differential Diagnosis (3)

- OC&R disorders
  - OCD → rules that must be applied rigidly (vs purposeless)
  - Trichotillomania, excoriation disorder → **onset around puberty**
    - Not apparently purposeless, not patterned/rhythmical

- Other neurological and medical conditions
  - Habits, mannerisms, paroxysmal dyskinesias, benign hereditary chorea
  - Features suggestive of other disorders
    - Myoclonus, dystonia, tics, chorea
  - **Tardive dyskinesia** → chronic neuroleptic use
    - Oral/facial dyskinesia, irregular trunk/limb movements
    - Do not result in self-injury
  - **Amphetamine intoxication/abuse** → skin picking/scratching
  - Neurological disorders assoc with repetitive choreoathetoid movements

Stereotypic Movement Disorder – Comorbidity

- SMD may occur as PRIMARY or SECONDARY disorder
  - Common manifestation of some neurogenetic disorders
    - **Lesch-Nyhan syndrome**
    - **Rett syndrome**
    - **Fragile X syndrome**
    - **Cornelia de Lange syndrome**
    - **Smith-Magenis syndrome**
  - Both should be diagnosed

Tic Disorders

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1) Tourette's Disorder – Diagnostic Criteria

A. **Both multiple motor + vocal tics**, not necessarily concurrently

B. **Duration >1 year** since first tic, may wax/wane in frequency

C. Onset **before age 18**

D. Not due to substance or AMC

DSM5

2) Persistent (Chronic) Motor or Vocal Tic Disorder

A. Single or multiple **motor or vocal tics**, not both

B. **Duration >1 year** since first tic, may wax/wane in frequency

C. Onset **before age 18**

D. Not due to substance or AMC

E. **Never met criteria for Tourette's disorder**

- *Specify if:*
  - **With motor tics only**
  - **With vocal tics only**

3) Provisional Tic Disorder

A. Single or multiple **motor and/or vocal tics**

B. **Duration LESS than 1 year** since first tic

C. Onset **before age 18**

D. Not due to substance or AMC

E. **Never met criteria for Tourette's disorder** or **persistent (chronic) motor or vocal tic disorder**

DSM5

Tic Disorders – Diagnostic Features (1)

- 4 diagnostic categories → **hierarchical order**
  - Tourette's' disorder
  - Persistent (chronic) motor or vocal tic disorders
  - Provisional tic disorder
  - Other specified/unspecified tic disorders

- Motor or vocal tics
  - **Sudden, rapid, non-rhythmic**
  - **Recurs in characteristic fashion** at any point in time
    - But may have various tic sx over time
  - Can include almost any muscle group or vocalization
    - Common → **eye-blinking, throat-clearing**
  - Experienced as **involuntary** → can be voluntarily suppressed (time varies)

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Tic Disorders – Diagnostic Features (2)

Simple Motor Tics	Simple Vocal Tics
• **Short duration (milliseconds)** • Eye-blinking • Shoulder shrugging • Extremity extension	• **From contraction of diaphragm or oropharyngeal muscles** • Throat-clearing • Sniffing • Grunting

Complex Motor Tics	Complex Vocal Tics
• **Longer duration (seconds)** • Often **combination** of simple tics • **Simultaneous** head turning + should shrugging • May appear purposeful • **Copropraxia** (sexual, obscene) • **Echopraxia** (imitation)	• **Coprolalia** (obscenities, slurs, but lacks prosody of speech) • **Echolalia** (repeating last heard) • **Palilalia** (repeating own sounds)

Tic Disorders – Diagnostic Features (3)

- A) Presence of tics
  - Tourette's (both motor + vocal), Persistent (either motor or vocal, not both)

- B) 1-year duration (If less than 1 year → provisional tic disorder)
  - May wax/wane in severity, may have **tic-free periods (weeks-months)**

- C) Onset of tics before age 18
  - Typically begin in **prepubertal period** (average onset **age 4 – 6**)
    - Incidence decreases in teen years
    - Exceedingly rare onset in adulthood → usually drug exposure (cocaine)
  - But not uncommon to present for **initial assessment as adol/adult**

- D) Not better explained
- E) Hierarchical exclusion

Tic Disorders – Prevalence

- Common in childhood → usually **transient**

- Tourette's disorder → **0.3-0.8%** school-aged children
  - More commonly affects **MALES (2-4x)**
  - Lower rates among African Americans, Hispanic Americans

Tic Disorders – Development & Course

- Onset → typically **age 4 – 6**
  - Peak severity **age 10 – 12**
    - **Severity DECLINES** during **adolescence, adulthood**
    - Small % have persistently severe/worsening sx into adulthood

- Course
  - **Manifests similarly** in all age groups, across lifespan
    - May wax/wane, may change affected muscle group/vocalization
  - With age, may report **premonitory urge**
    - Somatic sensation that precedes tic → relieved with expression of tic
    - May not feel complete involuntary → **some ability to resist**
  - May need to perform tic in specific way/number → **"just right"**

- Development of co-occurring conditions change with age
  - Prepubertal → ADHD, OCD, separation anxiety
  - Adolescents, adults → new onset MDD, bipolar, SUD

Tic Disorders – Risk & Prognostic Factors

- Temperamental
  - Tics worsened by → **anxiety, excitement, exhaustion, stressful events**
  - Tics improved by → **calm, focused activities**
    - Engaged in schoolwork/work tasks (vs relaxing at home)

- Environmental
  - **Observe gesture/sound** → may repeat → misperceived as purposeful
    - May be problematic with authority figures

- Genetic & physiological
  - Risk alleles for Tourette's disorder
  - Rare genetic variants in families with tic disorders
  - Factors assoc with worse tic severity
    - **Older paternal age, maternal smoking during pregnancy**
    - **Obstetrical complications, LBW**

Tic Disorders – Culture-Related Issues

- Similar clinical characteristics, course, etiology
  - Race, ethnicity, culture may impact perception, help-seeking, management

Tic Disorders – Gender-Related Issues

- More commonly affects → **MALES**
- NO gender differences in **type of tic, age of onset, course**
- Women with persistent tic disorders
  - More likely to experience **anxiety + depression**

Tic Disorders – Functional Consequences

- Depends on tic severity
  - Mild-mod → many **no distress/impairment, unaware**
  - More severe → more impairment, but **some may still function well**
  - **Comorbidities** can have greater impact on function (ADHD, OCD)

- Impairment from tics less common
  - Social isolation, interpersonal conflict, peer victimization
  - Inability to go to work/school
  - Lower quality of life, substantial psychological distress

- Rare complication of Tourette's
  - **Physical, orthopedic, neurological injury**

Tic Disorders – Differential Diagnosis (1)

- Abnormal movements accompanying AMC or SMD
  - **Motor stereotypies** (involuntary, rhythmic, repetitive, predictable)
    - Earlier onset, prolonged duration, fixed form
    - Exacerbated when **engrossed in activities**
    - **No premonitory urge**
  - **Chorea** (rapid, random, continual, abrupt, irregular, unpredictable)
    - Usually bilateral, affects all parts of body
    - Timing, direction, distribution varies moment to moment
    - Usually worsen during attempted voluntary action
  - **Dystonia** (simultaneous, sustained contracture of agonist/antagonist)
    - Results in distorted posture/movement
    - Often triggered by attempts at voluntary movements
    - NOT seen during sleep

Tic Disorders – Differential Diagnosis (2)

- Substance-induced + paroxysmal dyskinesias
  - Usually dystonic or choreoathetoid movements
  - Precipitated by voluntary movement or exertion

- Myoclonus
  - Sudden, unidirectional movement → NON-rhythmic
  - Worsen by movement, may occur during SLEEP
  - **Rapidity, lack of suppressibility, no premonitory urge** (vs tics)

- OC&R disorders
  - OCD → cognitive-based drive, particular fashion, symmetry, "just right"
  - More goal directed + complex (vs tics)

Tic Disorders – Comorbidity

- Common comorbidities → ADHD, OCD

- Comorbid OCD
  - More aggressive symmetry + order sx
  - Poorer response to pharmacotherapy (with SSRIs)

- Comorbid ADHD
  - Disruptive behavior, social immaturity, learning difficulties
  - May interfere with academic progress, interpersonal relationships
  - Greater impairment than from tic disorder

- Other comorbidities
  - Other movement disorders
  - Other mental disorder → depressive, bipolar, substance use

Other Specified Tic Disorder

Other Specified Tic Disorder

- Does not meet any full criteria
- Clinician chooses to specific reason

- With onset after age 18 years

Unspecified Tic Disorder

Unspecified Tic Disorder

- Does not meet any full criteria
- Clinician chooses NOT to specific reason

Other Specified Neurodevelopmental Disorder

Other Specified Neurodevelopmental Disorder

- Does not meet full criteria
- Clinician chooses to specify reason

- Neurodevelopmental disorder associated with prenatal alcohol exposure

Unspecified Neurodevelopmental Disorder

Unspecified Neurodevelopmental Disorder

- Does not meet full criteria
- Clinician chooses NOT to specify reason