



# AUTISM SPECTRUM DISORDER (ASD)

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# INTRODUCTION ✨

ASD is characterized by impairments in social communication/interaction and restrictive, repetitive behaviors/interests.

This disorder encompasses the **spectrum of symptomatology** formerly diagnosed as autism, Asperger's disorder, childhood disintegrative disorder, and pervasive developmental disorder.



# INTRODUCTION ✨

- Consider ASD as the diagnosis if there is a rapid deterioration of social and/or language skills **during the first 2 years of life.**
- Complete an appropriate workup, such as auditory testing, prior to diagnosing ASD.
- An extensive medical workup should be initiated **if skills are lost after the age of 2** or if more extensive losses occur (e.g., self-care, motor skills).



# EPIDEMIOLOGY ✨

- ① Recent increase in prevalence to 1% of population
- ② Ratio in males to females is 4:1
- ③ Symptoms are typically recognized between 12 and 24 months old
- ④ Caucasians have higher rates of occurrence



# ETIOLOGY



## ASD ETIOLOGY IS MULTIFACTORAL

- Prenatal neurological insults (e.g., infections, drugs (thalidomide, valproic acid))
- advanced paternal age (older than 35 years), and low birth weight.
- High comorbidity with ID
- Association with epilepsy
- Toxin exposure during pregnancy and the first 2 years of life





# ETIOLOGY



- 15% of ASD cases are associated with a known genetic mutation
- **Fragile X syndrome** is the most common known single-gene cause of ASD.
- Other genetic causes of ASD: **Down syndrome, Rett syndrome, and tuberous sclerosis.**



# TYPES OF ASD

① Autistic Disorder

② Asperger disorder

③ Pervasive developmental disorder

④ Childhood disintegrative disorder

⑤ Rett disorder

# AUTISTIC DISORDER

Also known as  
**Classic Autism** or  
referred to simply as  
**Autism**

**A neurodevelopmental condition** characterized by **significant challenges in social interaction, communication,** and the **presence of restricted and repetitive behaviors.**

Symptoms typically  
manifest **before the**  
**age of 3**

**Diagnosis** is made through **comprehensive evaluations, developmental assessments, and observations of behavior.**



# AUTISTIC DISORDER

Key Features of Autism:

## ① Social Interaction Challenges

- **Difficulty understanding and responding to social cues**, such as facial expressions and tone of voice.
  - **Limited interest in forming relationships** with peers or **engaging in social activities.**
  - **Challenges in understanding and participating in conversations.**
- 

# AUTISTIC DISORDER

Key Features of Autism:

## ② Communication Difficulties

- **Delayed speech** and language development; some may remain **nonverbal**.
- Difficulty initiating or sustaining conversations.
- Use of **echolalia** (repeating phrases or sentences) instead of spontaneous speech.



# AUTISTIC DISORDER

Key Features of Autism:

- ③ Repetitive Behaviors and Restricted Interests
- Engaging in **repetitive movements** (e.g., **hand-flapping, rocking**).
  - **Insistence on sameness and routines**, with distress when routines are disrupted.
  - **Intense focus on specific interests or topics**, often to the exclusion of other activities.
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# TREATMENT ✨

① Applied Behavior Analysis

② Speech and Language Therapy

③ Occupational Therapy

④ Individualized Education Programs (IEPs)

- While there is **no specific medication** for autism, certain medications **may be prescribed to manage specific symptoms** such as anxiety, depression, or hyperactivity.
- **Commonly used** medications include selective serotonin reuptake inhibitors (**SSRIs**) and **stimulants**.

# Diagnosis:

Based on DSM-5:

## **A. Social communication and social interaction :**

- \* Impaired social reciprocity**
- \* Deficits in nonverbal communication**
- \* Developing and maintaining relationships challenges**

## **B. Restricted, repetitive behaviors and interests**

- \* Highly restricted, fixed and peculiar interests**
- \* Inflexible adherence to rituals**
- \* Stereotyped, repetitive behaviors**
- \* Hypersensitivity or hyposensitivity or interest in sensory input**

**C. Symptoms must be present in early development but may not fully manifest until later**

**D. Symptoms must  
cause of clinically  
significant impairment  
in current functioning**

**E. Not better explained  
by intellectual  
disability or global  
developmental delay**

# Associated behavioral symptoms:

- **Disturbances in Language Development and Usage.**
- **intellectual Disability.**
- **Irritability**
- **Instability of Mood and Affect.**
- **Response to Sensory Stimuli.**
- **Hyperactivity and Inattention.**
- **Minor Infections and Gastrointestinal Symptoms.**
- **Precocious Skills.**
- **Insomnia**



## Based on DSM-5 :

**Severity is specified based on social communication impairments and restricted, repetitive patterns of behavior.**

### Severity Levels

- **Requiring very substantial support**
  - **Requiring substantial support**
    - **Requiring support**

# Severity Levels for Autism Spectrum Disorder

Severity level	Social Communication	Restricted, Repetitive Behaviors
<b>Level 1</b> <b>"Requiring support"</b>	<ul style="list-style-type: none"><li><b>*Without supports in place, deficits in social communication cause noticeable impairments.</b></li><li><b>*Difficulty initiating social interactions, and clear examples of atypical or unsuccessful responses to social overtures of others.</b></li><li><b>*May appear to have decreased interest in social interactions.</b></li><li><b>*For example, a person who is able to speak in full sentences and engages in communication but whose to-and-for conversation with other fails, and whose attempts to make friends are odd and typically unsuccessful.</b></li></ul>	<ul style="list-style-type: none"><li><b>*Inflexibility of behavior causes significant interference with functioning in one or more contexts.</b></li><li><b>* Difficulty switching between activities.</b></li><li><b>*Problems organisation and planning hamper independence</b></li></ul>

# Severity Levels for Autism Spectrum Disorder

Severity level	Social Communication	Restricted, Repetitive Behaviors
<p><b>Level 2</b> <b>“Requiring substantial support”</b></p>	<p><b>*Marked deficits in verbal and nonverbal social communication skills; social impairments apparent even with supports in place;</b></p> <p><b>*limited initiation of social interactions; and reduced or abnormal responses to social overtures from others.</b></p> <p><b>*For example, a person who speaks simple sentences, whose interaction is limited to narrow special</b></p>	<p><b>*Inflexibility of behavior, difficulty coping with change, or other restricted/behaviours appear frequently enough to be obvious to the casual observer and interfere with functioning in a variety of contexts.</b></p> <p><b>*Distress and/or difficulty changing focus or action</b></p>

# Severity Levels for Autism Spectrum Disorder

<b>Severity level</b>	<b>Social Communication</b>	<b>Restricted, Repetitive Behaviors</b>
<b>Level 3 "Requiring very substantial support"</b>	<p><b>*Severe deficits in verbal and nonverbal social communication skills cause severe impairments in functioning.</b></p> <p><b>* very limited initiation of social interactions, and minimal response to social overtures from others.</b></p> <p><b>*For example, a person with few words of intelligible speech who rarely initiates interaction, and when he or she does, makes unusual approaches to meet needs only and responds to only</b></p>	<p><b>*Inflexibility of behavior, extreme difficulty coping with change, or other restricted/repetitive behaviors markedly interfere with functioning in all spheres.</b></p> <p><b>* Great distress/difficulty changing focus or action</b></p>

BASED ON DSM-5 :

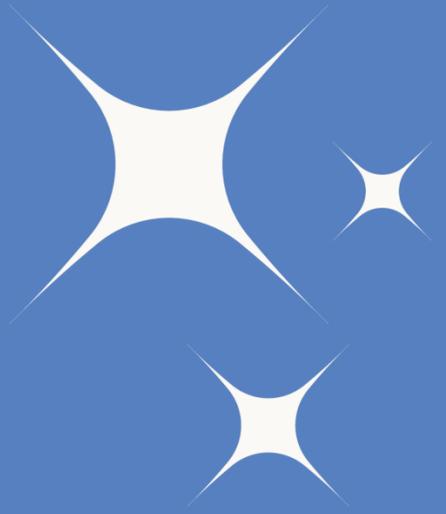
• Specify If

- **With or without accompanying intellectual impairment**
- **With or without accompanying language impairment**
- **Associated with a known genetic or medical condition/environmental factor**
- **Associated with a neurodevelopmental, mental, or behavioral problem**
- **With catatonia (refer to catatonia criteria)**

# Diagnosis:



- **Consider ASD as the diagnosis if there is a rapid deterioration of social and/or language skills during the first 2 years of life.**
- **Complete an appropriate workup such as auditory testing, prior to diagnosing ASD**
- **An extensive medical workup needs to be initiated if skills are lost after age 2, or more extensive losses occur (self-care, motor skills)**



# ASPERGER'S SYNDROME



## 2 ASPERGER'S SYNDROME



Asperger's syndrome is a neurodevelopmental disorder that falls under the autism spectrum disorders (ASD).

Characterized by impaired **social** and **communication skills** as well as **repetitive or restrictive thinking and behavior**.

People diagnosed with Asperger's syndrome typically have **high intelligence** and **no speech delays**. However, they tend to play, learn, speak, and act differently from others

### **DSM-5 Criteria for Autism Spectrum Disorder (ASD):**

Asperger's is no longer a separate diagnosis; it falls under ASD – usually Level 1: requiring support, with no intellectual or language impairment.



## 2 ASPERGER'S SYNDROME



A. Persistent deficits in social communication and social interaction across multiple contexts, as manifested by **all three of the following**:

1. Deficits in social-emotional reciprocity, such as:

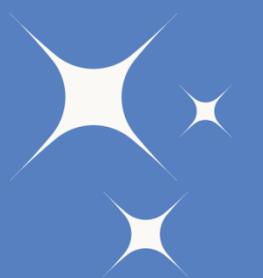
- Abnormal social approach
- Failure of normal back-and-forth conversation
- Reduced sharing of interests, emotions, or affect
- Failure to initiate or respond to social interactions

2. Deficits in nonverbal communicative behaviors used for social interaction, such as:

- Poorly integrated verbal and nonverbal communication
- Abnormal eye contact or body language
- Deficits in understanding and using gestures
- Lack of facial expressions

3. Deficits in developing, maintaining, and understanding relationships, such as:

- Difficulty adjusting behavior to suit social contexts
- Difficulty sharing imaginative play or making friends
- Apparent absence of interest in peers



## 2 ASPERGER'S SYNDROME



**B.** Restricted, repetitive patterns of behavior, interests, or activities, as manifested by **at least two of the following:**

1. Stereotyped or repetitive motor movements, use of objects, or speech.
  - (e.g., hand flapping, lining up toys, echolalia, idiosyncratic phrases)
2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of behavior.
  - (e.g., extreme distress at small changes, rigid thinking, need for the same route or food)
3. Highly restricted, fixated interests that are abnormal in intensity or focus.
  - (e.g., strong attachment to unusual objects, excessive interest in specific topics)
4. Hyper- or hypo-reactivity to sensory input or unusual interest in sensory aspects.
  - (e.g., indifference to pain/temperature, adverse response to specific sounds/textures, fascination with lights or movement)

**C.** Symptoms must be **present in the early developmental period.**

**D.** Symptoms cause **clinically significant impairment.**

**E.** Disturbances **are not** better explained by **intellectual disability (ID)** or **global developmental delay.**

## 2 ASPERGER'S SYNDROME



- **Onset:**

**usually before two years** with long term duration, And is often diagnosed belatedly

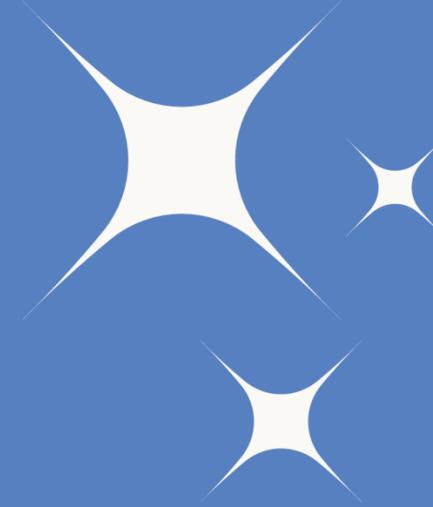
- **Causes & Diagnosis:**

Experts don't know exactly what causes ASD. But they believe it's a combination of **genetics and environmental** factors . **Diagnosed clinically**, often in childhood, using developmental history and observation.

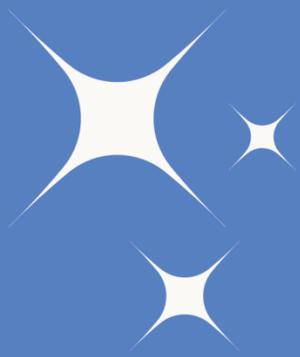
- **Management:**

- Behavioral and educational interventions: social skills training, structured learning environments. Failure of normal back-and-forth conversation
- Speech and language therapy: for pragmatic (social) communication skills.
- Occupational therapy: for motor or sensory challenges.
- Counseling / CBT: for anxiety, depression, or emotional regulation.
- Medication: not for Asperger's, but for associated conditions (ADHD, anxiety, depression, irritability).





# RETT'S SYNDROME



### 3 RETT'S SYNDROME

Rett syndrome is a **neurodevelopmental disorder** almost exclusively **affecting girls**. It is characterized by a period of normal early development followed by **loss of acquired skills** and the emergence of **stereotypic hand movements**.

- **Genetics:**

Usually caused by mutations in the **MECP2 gene on Xq28**.  
**Most cases are sporadic**, though rare familial cases exist.

- **Epidemiology:**

Primarily **affects females** (males with MECP2 mutations often die in infancy).  
Prevalence: ~1 in 10,000–15,000 female births.

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### 3 RETT'S SYNDROME



#### DSM-5 Diagnostic Criteria for Rett Syndrome:

Rett syndrome is not classified under Autism Spectrum Disorder even though some features overlap (social withdrawal, communication difficulties), due to its known etiology.

According to DSM-5, **all of the following criteria must be met:**

- A period of apparently normal development for the **first 6–18 months of life**.
- **Loss of purposeful hand skills** (e.g., hand-wringing, washing, clapping) and the emergence of **stereotypic hand movements**.
- **Loss of acquired spoken language**.
- **Gait abnormalities:** impaired, unsteady, or absent.
- **Deceleration of head growth** (microcephaly develops after early normal growth).

The disorder is excluded if the clinical features are better explained by another medical or neurological condition.



### 3 RETT'S SYNDROME



- **Stages and clinical feature of Rett Syndrome:**

Stage 1: Early Onset (6–18 months)

- Subtle developmental slowing.
- Mild loss of interest in surroundings.
- Slight delays in gross motor skills (e.g., sitting, crawling).
- Sometimes misdiagnosed as autism or developmental delay.
- **Head growth may begin to slow.**

Stage 2: Rapid Destructive Stage (1–4 years)

- **Loss of purposeful hand skills** (grasping, holding objects).
- **Stereotypic hand movements emerge:** wringing, clapping, washing.
- Loss of spoken language.
- Motor abnormalities appear: unsteady gait, hypotonia, ataxia.
- Social interaction may decline temporarily.

This is often when diagnosis becomes obvious.



### 3 RETT'S SYNDROME



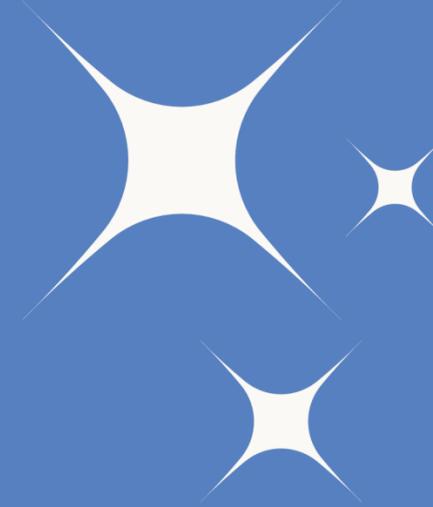
Stage 3: **Plateau or Pseudostationary Stage** (2–10 years, sometimes longer)

- Stabilization of regression; some skills may improve.
- Hand stereotypies persist but may become less severe.
- **Seizures often develop (up to 70–90% of patients).**
- Behavioral abnormalities: anxiety, irritability, autistic-like features.
- Children are generally alert and socially engaged, despite motor limitations.

Stage 4: **Late Motor Deterioration Stage** (Adolescence and beyond)

- **Severe motor problems:** spasticity, rigidity, decreased mobility.
- **Many become wheelchair-bound.**
- Hand skills may partially improve but remain limited.
- Communication abilities: limited, often nonverbal.
- Cognitive function is relatively preserved compared to motor skills.





CHILDHOOD  
DISINTEGRATIVE  
DISORDER (CDD)



# 4

## CHILDHOOD DISINTEGRATIVE DISORDER (CDD)



Childhood Disintegrative Disorder (CDD), also known as **Heller syndrome**, is a rare neurodevelopmental disorder characterized by **normal development for at least two years, followed by severe regression** in multiple areas, including **language, social skills, motor skills, and adaptive behaviors**.

- **Epidemiology**

Incidence: Approximately 1.7 per 100,000 children.

Gender: More common in males than females.

- **Diagnosis**

Based on the DSM-5, CDD is now considered part of the broader category of Autism Spectrum Disorder (ASD). The diagnosis is made when there is:

- **Normal development for at least 2 years.**
- **Significant loss of skills in at least two areas.**
- Absence of other medical or neurological conditions that could explain the regression.



# 4

## CHILDHOOD DISINTEGRATIVE DISORDER (CDD)



### Clinical Features:

- Normal Development: Achieves typical milestones in language, motor, social, and adaptive domains during the first 2 years.
- Regression: After the initial period, there is a marked loss of previously acquired skills in at least two of the following areas:
  - Language: Loss of both expressive and receptive language.
  - Social Skills: Withdrawal from social interactions, loss of interest in peers.
  - Motor Skills: Loss of coordination, muscle tone abnormalities.
  - Adaptive Behavior: Loss of self-care skills, such as toileting and feeding.
- Behavioral Changes: Development of stereotypic behaviors, irritability, and other behavioral disturbances.



# 4

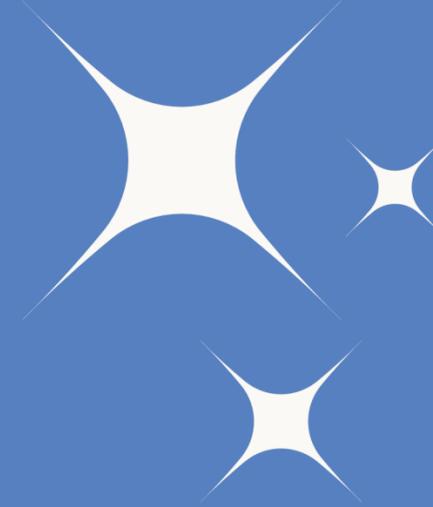
## CHILDHOOD DISINTEGRATIVE DISORDER (CDD)



### Management:

- Therapies:
  - Behavioral Therapy: To address social and communication deficits.
  - Speech and Occupational Therapy: To improve language and motor skills.
  - Antiepileptic Medications: If seizures are present.
  - Psychiatric Medications: To manage irritability and other behavioral issues.
- Educational Support: Tailored programs to meet the child's developmental needs.
- **No Cure:** Treatment is supportive and symptomatic.





PERVASIVE  
DEVELOPMENTAL  
DISORDER (PDD)



## 5 PERVASIVE DEVELOPMENTAL DISORDER (PDD) ✨

Pervasive Developmental Disorder (PDD) is an outdated term that has been largely replaced by Autism Spectrum Disorder (ASD), encompassing a range of developmental delays affecting social and communication skills.

This disorder has historically been used to **describe individuals who did not fully meet the specific criteria for autism. Used as "catch-all" for atypical or milder presentations.**

### Management

- Behavioral interventions.
- Speech and occupational therapy.
- Pharmacotherapy: For irritability, hyperactivity, or comorbid psychiatric conditions
- Educational support tailored to individual needs.

Q AND A

THANK

YOU

