

# A Comprehensive Review on Autism Spectrum Disorder of Pathophysiology, Epidemiology, Causes and Treatment.

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**Abstract:** *Autism Spectrum Disorder (ASD) is a condition that affects how a child's brain develops. It changes the way they communicate, behave, and sense the world. Lately, more children seem to have autism. This review studied how often autism happens, what causes it, how it affects families and society, and how it can be diagnosed and treated. The authors say autism is a brain condition caused by both genes and surroundings, and they list things that can raise the chance of having autism. The exact cause of autism is known in only a few cases, so many possible reasons have been suggested. The earlier autism is found and help begins, the better a child does. Autism is a way to describe people who have trouble with social communication, repeat certain behaviours, have very focused interests, or react strongly to senses, starting from a young age. Today, many people with autism have a better life than 50 years ago. More can talk, read, live in the community, and some have very few symptoms as adults.*

**Keywords:** *Autism Spectrum Disorder*

## I. INTRODUCTION

In the past 50 years, autism went from being rare in children to a well-known, lifelong condition that is quite common and different for each person. Autism can cost a lot because some adults need help living. This raises health and school costs. About 1 in 700–1,000 people has autism, and about 1 in 1,000 has the usual autism symptoms. Autism mostly affects behaviour and thinking in different ways. It starts early and includes problems with talking and socializing, strong focus on certain activities, and repeating actions. Symptoms appear early in life and make it hard to interact with others and work or do daily activities. Families, teachers, and caregivers have the biggest impact on people with autism (ASD). However, doctors and other healthcare professionals also play an important role. They help by explaining how the person with ASD is doing, preparing families for upcoming changes, and connecting them with the right services and specialists when needed. IN the past, people thought autism (ASD) was caused by psychological factors. Now we know that autism happens because of differences in how the brain works and develops.

### • Brain Growth, structure and connectivity of ASD :

One of the first signs that brain development might be unusual in autism was noticing that babies and young children with autism sometimes had larger heads than usual.



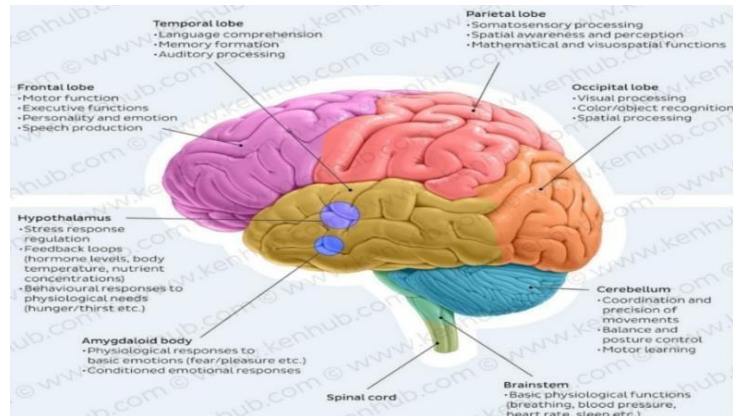


Fig 1: Brain function.

### • Epidemiology :

The World Health Organization (WHO) estimates that about 0.76% of people around the world have autism (ASD). But this number is based on information from only about 16% of the world's children. Some genetic conditions—like fragile X syndrome, tuberous sclerosis, Down syndrome, and Rett syndrome—are more likely to include autism (ASD) than in the general population. However, these genetic conditions make up only a small number of all autism cases. The CDC says that about 1.68% of 8-year-old children in the United States have autism (ASD). It might be too early to tell for sure, but the number of children with autism (ASD) in the U.S. seemed to stay the same between 2014 and 2016. Certain parts of chromosomes—especially X, 2, 3, 7, 15, 16, 17, and 22—are connected to a higher risk of autism (ASD). We know this better now because doctors use chromosome testing more often. Other things that can raise the chance of autism (ASD) are having older parents or being born early. Since we only know the exact cause of autism (ASD) in a few cases, many possible causes have been suggested and studied.

Lastly, “syndromic autism” refers to the small number of people with autism (ASD) who also have other health problems or a likely genetic cause. How common autism (ASD) seems to be can change depending on how a study is done. The results can vary based on the definitions used, who is included, and whether the cases are checked directly or just taken from records. Many people believe that autism (ASD) is increasing, but this is mostly based on official records, not on studies done in local communities. Many people believe that autism (ASD) is rising, but this is mostly based on official records, not studies done in communities.

### 1. Prevalence:

Estimates of how common autism is vary depending on how studies are done, including how autism is defined, who is studied, and how cases are identified. The Global Burden of Disease study looked at all available data and estimated that in 2010 about 52 million people worldwide had autism, which is about 1 in 132 people. Some people think autism is increasing, but studies show it hasn't really changed over time. Surveys of children and adults confirm this, and it's not less common in older people. Autism is more common in boys than girls, about 4 boys for every 1 girl.

### 2. Environmental factors:

Some things in the environment may raise the chance of autism, like older parents and problems at birth, especially if the baby gets too little oxygen. These factors don't cause autism by themselves, but they might play a part. Other things that may increase autism risk are long labor, C-section or assisted delivery, early water breaking, and fertility treatments. They might affect risk through gene changes, inflammation, or low oxygen.



• **Mechanisms/pathophysiology**

People have suggested many ideas to explain autism. Some focus on social skills, like understanding others' thoughts, and others focus on thinking skills, like paying attention, planning, or seeing the big picture. Watching babies from before birth or birth until they are 2–3 years old helps researchers see how autism and related behaviors first appear. Brain studies show small but widespread differences in people with autism. Autism likely comes from many interacting factors, where the brain, behavior, and genes all affect each other, rather than from just one single cause.

**1. Genetics:**

Genes play a big role in autism. Over 100 gene changes have been linked to it, some very small and some bigger. Changes that affect important parts of DNA are most connected to autism, and these rare changes can greatly increase the risk. Autism is mostly linked to rare new gene changes, while adult mental health problems usually involve many common genes with small effects. Some genes can influence very different brain conditions, but we don't fully understand how yet. Rare gene changes teach us a lot about how autism works. One rare mutation can cause more risk than many common gene changes in other disorders. But studying just one mutation doesn't make it easy to help most people with autism.

**2. Molecular pathophysiology**

Over the past 10 years, scientists have studied rare single-gene forms of autism, which are less than 10% of cases, and also looked at rare new gene changes in regular autism. Autism risk genes usually affect many brain areas and functions as the brain develops. So, changing one risk gene in an animal or lab model might show some effects, but it doesn't always explain how it causes social difficulties in humans. Being able to focus future experiments on specific regions, cell types, and stages of development linked to disease risk will help scientists better understand the exact mechanisms involved. Since gene therapy has worked and been approved for some early brain disorders, like spinal muscular atrophy type 1, scientists now think it might also be possible to target single genes that strongly contribute to autism, whether the cause is known or not.

• **Cause :**

1. ASD is a condition that affects the brain. Both genes and the environment can play a role. Genes can increase the chance of having ASD.
2. Brothers and sisters of someone with ASD are more likely to have it. Identical twins have an even higher chance, but not always.
3. Studies of many genes have helped us find genes linked to ASD. Looking at many genes helps us find those related to ASD.
4. Knowing what these genes do can explain how ASD works. Looking at many genes helps us find those related to ASD. Knowing what these genes do can explain how ASD works.
5. A small study of young children's brains showed that the layers in the outer part of the brain were often disrupted.
6. This suggests problems with how brain cells develop and form these layers. Children with ASD can have bigger brains and extra fluid around the brain.
7. Few studies exist, but they show that children with ASD can have differences in several brain areas, including the limbic system, cerebellum, and the front and side parts of the brain.
8. Children born early have a higher chance of having ASD and other brain development problems.
9. A study found that babies with low birth weight, born early, low health scores at birth, C- section, or bleeding in the womb were more likely to have autism.
10. There is no evidence that vaccines or mercury cause ASD, even though.



11. A large study In Denmark showed that MMR vaccines do not cause ASD.

**• Symptoms**

1. Social challenges: Difficulty understanding or interacting with other people.
2. Communication problems: Trouble talking, understanding language, or using gestures.
3. Repetitive behaviors: Doing the same actions or routines over and over.
4. Sensory differences: Being very sensitive or not sensitive enough to sights, sounds, touch, or other senses.
5. Social difficulties: Trouble making friends or keeping them, Limited eye contact or facial expressions, Difficulty understanding other people's feelings or social cues
6. Communication challenges: Delayed speech or language development, Trouble starting or keeping conversations, Using unusual tone of voice, gestures, or body language.
7. Repetitive behaviors and routines: Repeating words, phrases, or actions, Needing things to stay the same or following strict routines, Strong interest in specific topics.
8. Other possible signs: Difficulty adapting to changes, Strong reactions to minor changes in environment, Unusual ways of playing with toys.

**• Diagnosis:**

Autism is diagnosed by looking at behavior. Even though people with autism are different, doctors can spot main signs in social skills, talking, and repetitive or unusual sensory behaviors. Sure! Here's a simpler version of your passage:

o Third Phase: Confirming the Diagnosis

In this phase, doctors decide for sure if the person has autism and what type it is. To do this:

1. They look at what specialists and parents have observed about the person.
2. They do the necessary tests to check behaviours and skills.
3. They compare all the information to the DSM-5 rules to make the final diagnosis

**1. Diagnostic criteria:**

Asperger's is now called ASD because doctors didn't always agree on the diagnosis. The focus should be on studying the differences in people with autism using science, not just using unclear labels like Asperger's or PDD-NOS. In DSM-5, unusual reactions to senses (too sensitive, not sensitive enough, or seeking certain feelings) are counted as part of repetitive behaviours in autism.

**2. Diagnosis and screening in children :**

There are many autism tests, but only a few have been checked carefully. The best tests, ADI-R and ADOS-2, work well but are not always easy to use everywhere. To diagnose autism, doctors ask parents about the child's early development and watch how the child behaves with parents and strangers. Some people with autism form groups, and their symptoms can change over time. This might be due to cultural differences, girls being tougher or protected, or tests not catching autism in girls well enough.

**3. Diagnosis and screening in adults**

There isn't much research on how to diagnose autism in adults, and few tests have been checked carefully. Doctors usually use child-based methods to diagnose adults, using childhood history, but the ADOS Module 4 test also works well for adults. Adults being tested for autism may also have other mental health problems, so tests must tell the difference between them and autism.

It's usually harder to diagnose autism in adults than in children because some autism signs can look like anxiety or ADHD.



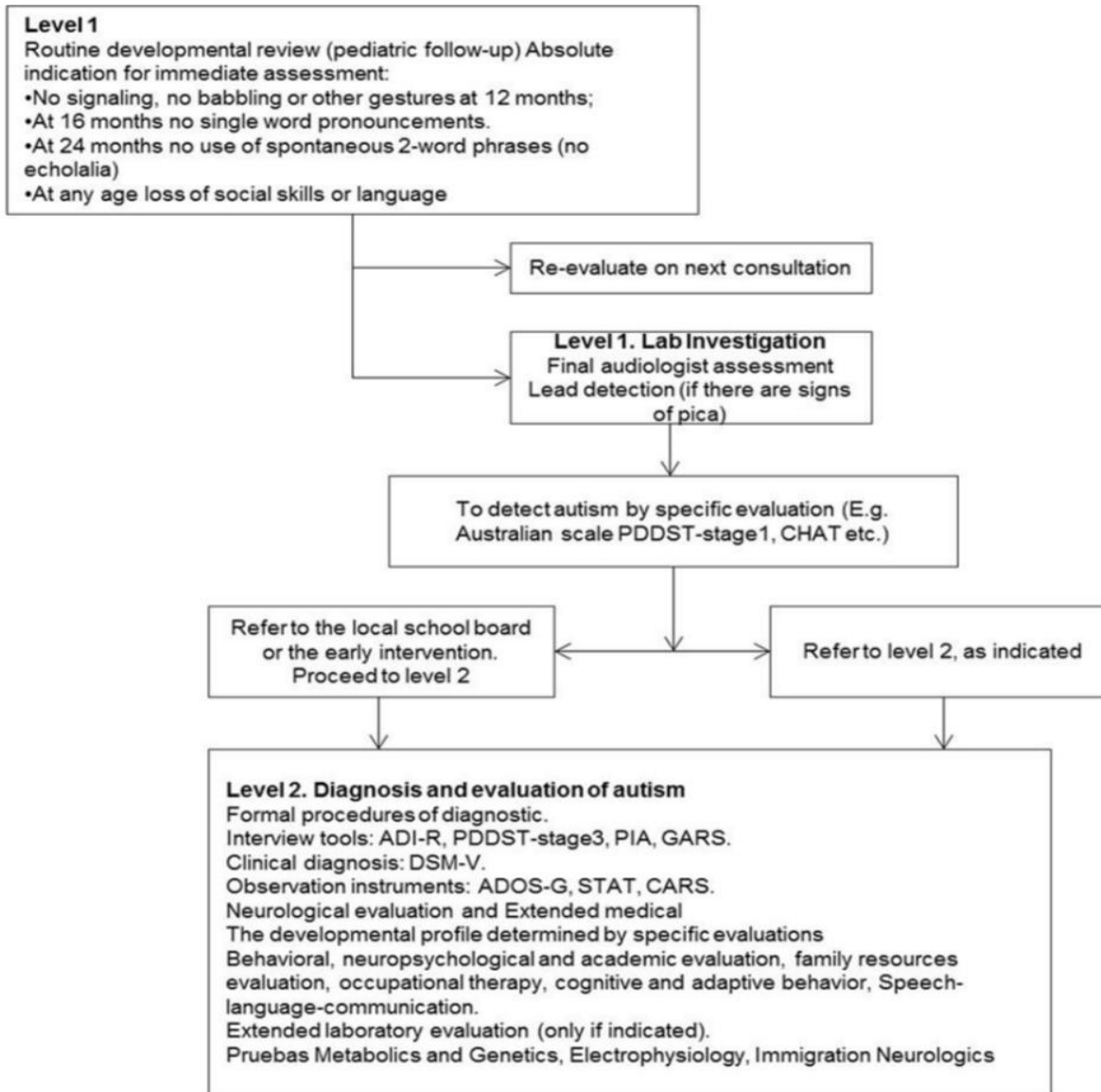


Fig 2 : levels of diagnosis for autism.

**• Treatment**

The help children and adults with ASD get is very different in different places. Studies show that parents with less education often have a harder time getting special support for their child. In some parts of Europe, even cheap or free services were not available until a year after diagnosis.

**1. Early parent-mediated interventions:**

These treatments are easy to use, low cost, and can be at home or in a clinic. They help young children at risk, even if they don't develop ASD. Early parent-led programs have been shown to help, with benefits lasting into later childhood.



## **2. Naturalistic behavioural developmental interventions**

Parent-led programs mainly help with social skills, while one-on-one programs can help with language, thinking, and daily life skills. Kids may benefit from both, but we don't yet know which works best. Sensory treatments are used a lot in North America but less elsewhere. They can help young children improve movement and sensory skills for a short time.

## **3. Behavioural and social treatments for school-age children, adolescents, and adult:**

Many programs use CBT to help children with ASD. Group sessions for kids and parents can reduce anxiety. They usually last 3 months and help about half of the children, while very few improve without it. It's harder to help with depression. There are few studies on behavior treatments for adults with ASD, except for anxiety and depression. More research is needed on life skills, anger control, self-help, and health. Helping teens and young adults get jobs in normal community programs is also important.

## **4. Pharmacology**

Children with ASD who also have epilepsy or mental health problems should usually be treated like other children. Because we don't know how well medicines work for them, safer options like therapy or counseling are preferred.

## **5. Medication:**

A health care provider may prescribe medication to treat specific symptoms, including:

- Aggression
- Anxiety and depression
- Attention
- Hyperactivity
- Irritability
- Repetitive behavior
- Self-injurious behavior

## **II. CONCLUSION**

Life for people with autism is much better today than it was 50 years ago. More adults with autism can now talk, read, drive, finish school, and live on their own, even though the way autism is diagnosed and the range of abilities have changed over time. Caregivers can feel hopeful because life for most people with autism is better now and will keep improving. We hope future research helps people with autism who still struggle and finds ways to help them be more included and independent. Both research and government actions can help bring about these positive changes. By working with families, schools, and community programs, doctors and therapists can help children and adults with autism by giving them helpful information, support, and hope.

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