

## Autism Spectrum Disorder (ASD)

**Autism spectrum disorder (ASD)** is a complex collection of conditions related to brain development. ASD affects the way a person **perceives** and **socializes** with others, making it **hard to function** at school and at work.

Signs of ASD are usually visible by age 2. Some children improve as they grow older, but ASD usually lasts into adulthood. ASD is not associated with an increased risk of psychotic illness in 22q11.2DS. There is no cure for ASD, but **early intervention** can be very helpful.

### Signs of ASD

There are [many possible signs](#), and each person will likely have a **unique combination and severity**.

- **Social communication and interaction behaviors** include:
  - Not responding when called by name
  - Resists cuddling
  - Tends not to pay attention to the surroundings
  - Showing poor eye contact and lacking facial expression
  - Having very little / delayed / regressed speech
  - Reacting inappropriately in social situations
- **Patterns of behavior** include:
  - Repetitive rocking, spinning, hand flapping etc.
  - Getting very upset at changes to specific routines
  - Being very fascinated by certain details
  - Being abnormally fixated on an item or activity
  - Not engaging in make-believe play
  - Having sensory issues, e.g. hypersensitivity to sounds or textures

### Diagnosis and Treatment of ASD

Because ASD varies widely among individuals, it is difficult to diagnose. Instead of having one single diagnostic test, the healthcare provider(s) will likely ask about child's family history and check for:

- **Social interactions, communication skills, and behavior**
- **Hearing, speech, language, developmental stage**
- **Medical and neurological functions**

Treatment plans need to meet the needs of the child and may change over time. **Behavior and communication therapies, highly structured educational programs**, and **medications** may all reduce ASD symptoms and maximize the child's ability to function, develop, and learn.

### How Common is ASD?

- General population: ~2%
- 22q11.2DS: ~30% (in some studies)
- 22q11.2DupS: ~14 to 25%

ASD is more common in boys than girls.

Some children with 22q11.2DupS have restricted and repetitive behaviors but with less social communication impairments.

Those with 22q11.2DS tend to have more social communication deficit compared to restricted and repetitive interest.

### References / Resources

#### Autism Spectrum Disorder (NIMH, MD, USA)

- [Statistics](#)
- [Mental Health Info](#)

#### Autism Spectrum Disorder (Mayo Clinic, AZ, USA)

- [Symptoms & causes](#)
- [Diagnosis & treatment](#)

#### Medical Research Reports

- [Updated clinical practice recommendations for managing children with 22q11.2 deletion syndrome](#) – 2023
- [Cognitive deficits in childhood, adolescence and adulthood in 22q11.2 deletion syndrome and association with psychopathology](#) – 2020
- [22q11.2 duplication syndrome: elevated rate of autism spectrum disorder and need for medical screening](#) - 2016