

For the Primer, visit doi:10.1038/s41572-019-0138-4

➔ Autism spectrum disorder — or autism — is a neurodevelopmental disorder with a typical onset early in childhood. Manifestations of autism vary in severity between individuals and include impairments in communication and interaction with others, sensory anomalies, repetitive movements and can include intellectual disability.

EPIDEMIOLOGY

Autism is more common in males than in females, with estimated sex ratios ranging from 2:1 to 5:1. Prevalence estimates vary depending on the methodology used in the study, with a prevalence of 52 million individuals reported in the 2010 Global Burden of Disease Study. After accounting for methodological differences between studies, there is no clear evidence that the prevalence of autism is increasing over time.

! Environmental factors associated with autism include increased parental age, neonatal hypoxia and preterm birth. There is no link between vaccination and autism.

DIAGNOSIS

Manifestations of autism occur gradually during early childhood, starting at ~2 years of age. Diagnosis comprises a detailed developmental history of the child (often obtained from their parent or parents), together with observing the child interact with familiar and with unfamiliar adults. Many adults with autism have not received a formal diagnosis; methods used for diagnosis typically rely on childhood developmental data.

Rx MANAGEMENT

Management of autism varies depending on the age of the individual

Interventions in young children focus on improving the core manifestations of autism and can include parent–therapist sessions that encourage communication and interaction between the parent and child

In school-aged children and adolescents, social skills training and therapies that target co-occurring emotional and behavioural problems can be beneficial

Interventions in adults aim to promote independence, such as support with finding and maintaining employment as well as addressing co-occurring mental health issues

! Most children with autism who reside in low-income or middle-income countries will never meet an autism expert

QUALITY OF LIFE

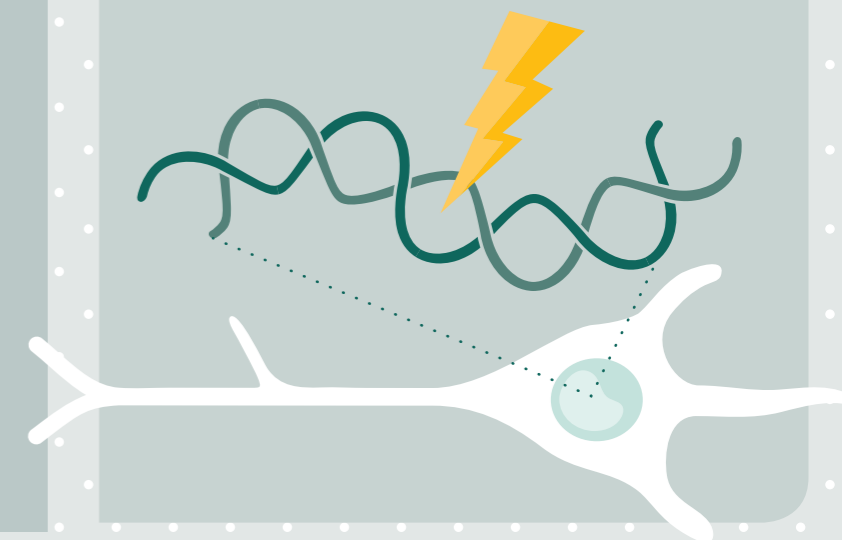
Studies evaluating quality of life (QOL) in people with autism have suggested that both subjective and objective QOL are reduced in this population. Higher QOL is associated with, for example, higher levels of language development, IQ and adaptive

! Therapies that target co-occurring disorders include cognitive behavioural therapy for anxiety or pharmacological treatments for hyperactivity

behaviour scores, as well as less-severe manifestations and fewer challenging behaviours.

MECHANISMS

More than 100 genes or genomic regions have been associated with autism, many of which have a role in glutamatergic neurons during development. These genes encode proteins that have a role in synaptic structure and function (such as receptors, ion channels, cell adhesion molecules, phosphatases and kinases) or chromatin modification. MRI and electroencephalography can be used to assess how the brain develops in people with autism. Although further research is required to confirm findings from these studies, alterations in the cortex, overall brain volume, the temporal and middle gyri, the amygdala and the insula have been demonstrated in children with autism.



OUTLOOK

With further research, MRI or electroencephalography studies could potentially provide detailed insights into the neural mechanisms of autism. In addition, these techniques could be used to identify subgroups of people with autism and similar neuronal alterations that account for their clinical manifestations. Eventually, these techniques could be used for diagnostic purposes or even to guide therapeutic intervention.