SSBP Syndrome Sheets

Autism Spectrum Disorder (Autism)

Classification

Although there are some slight differences between the two main diagnostic classification systems for autism (Diagnostic and Statistical Manual [DSM]-5; American Psychiatric Association, 2022; International Classification of Disorders [ICD] 11; World Health Organisation, 2019/21) both require evidence (currently or by history) of difficulties in two core domains: (i) the ability to initiate and sustain reciprocal social interaction and social communication, and (ii) a range of restricted, repetitive and inflexible patterns of behaviour and interests. Both classifications also include hyper- or hypo-reactivity to sensory input and/or unusual interests in sensory stimuli. Diagnostic ascertainment should specify if autism is accompanied by additional intellectual or language impairments; is associated with a known medical, genetic or environmental factor, or associated with another neurodevelopmental, emotional or behavioural condition. To meet diagnostic criteria, symptoms must be sufficiently severe to cause impairment in personal, family, social, educational, occupational, or other important areas of functioning. DSM-5 also incorporates overall severity ratings ('requiring very substantial support"; "requiring substantial support" and "requiring support"). Symptoms must have been present in early development although they may not become apparent until social demands exceed the individual's capabilities; symptom severity may also vary according to social, educational, or other contexts. Sub-categories of autism that were previously included in DSM-IV/ICD10 (e.g. Asperger Disorder, Autistic Disorder, Pervasive Developmental Disorder NOS) are no longer specified. (See also Hirota and King, 2023 for a general review).

Associated conditions

There is a significant association between autism and a wide range of other developmental and genetic disorders including Fragile X syndrome, Tuberous Sclerosis Complex, Angelman syndrome and Neurofibromatosis Type 1 (Lubbers et al., 2024; Pan et al., 2021). The comorbidity between autism and ADHD, both at a genetic and symptom level, is particularly high (Rong et al., 2021; Thapar & Rutter, 2021). There are links, too, with conditions such as maternal rubella, cytomegalovirus and phenylketonuria, although the phenotype in these cases tends to be atypical. Autistic people have a significantly increased risk of physical health problems, the most common being sensory impairments, autoimmune disorders, and obesity, gastrointestinal, and sleep disorders (Rydzewska et al., 2021). Rates of functional neurological disorders are high (Vickers et al., 2025); the overall prevalence of epilepsy is around 12% (Liu et al., 2022) with rates being highest (20%-30%) in autistic individuals with intellectual disability. Mental health problems, especially related to anxiety and depression, are also extremely common. Although estimated rates of mental health conditions vary widely from study to study, a meta-analysis, based on cases diagnosed via clinical interview, reported an overall prevalence rate of 60% (Lai et al., 2019)

Genetics

Overall heritability estimates for autism vary somewhat but median rates are around 80%. Family

genetic studies indicate significantly increased rates of autism in siblings (around 20%); the "Broader Autism Phenotype" (i.e. having problems related to social, language and/or cognitive functioning) is also estimated to occur in up to 20% of first-degree family members (Thapar & Rutter, 2021) However, there is wide genetic heterogeneity, with multiple modes of inheritance including high rates of de novo mutations and a wide range of possible rare and common copy number variations (e.g. submicroscopic chromosomal deletions or substitutions; Yasuda et al., 2023). Diverse clinical phenotypes and limited sample sizes add to the challenges of identifying the specific genes involved and currently only around 10% to 15% of cases of autism appear to be associated with a known, specific, genetic mutation Moreover, as research into the genetics of autism has progressed, the shared genetic influences between autism and other conditions, especially ADHD, has become increasingly clear (Zhou et al., 2023).

Environmental risk factors

Recent research has highlighted the impact of gene-environment interactions and identified a number of potential environmental risks (Williams et al., 2022). These include high maternal and paternal age; maternal health factors such as obesity, diabetes, or medications taken during pregnancy (e.g. thalidomide, SSRI's and Valproate); immune system abnormalities; pre or peri-natal perturbations, and pre-natal exposure to pollutants and pesticides. There is no evidence that MMR or other vaccines are a cause of autism (Gabis et al., 2022).

Prevalence

Prevalence estimates of autism vary widely across the world (from 0.02% in China to 3.66% in Sweden; Salari et al., 2022) and are affected many factors including cohort size, geographic location, and diagnostic ascertainment methods. A systematic review update, based on 71 studies (Zeidan et al., 2022), reported ranges from 1.09/10,000 to 436.0/10,000, with a median prevalence of 100/10,000 (i.e.1%). The median percentage of autism cases with co-occurring intellectual disability was 33.0%. The median male-to-female ratio was 4.2, although, increasingly, research suggests that the apparent gender bias may be at least partly due to the lower sensitivity of current diagnostic criteria for identifying autism in girls and women (Lockwood et al., 2021).

Physical phenotype

There is no distinct physical phenotype although minor physical anomalies and dysmorphic features are common and there is an increased risk of chronic and acute medical problems across the life span (Liu et al., 2023). Compared to the general population, autistic people are more likely to suffer from neurological abnormalities including epilepsy, macrocephaly, hydrocephalus, cerebral palsy, migraine/headache, and congenital abnormalities of the nervous system (Pan et al., 2020). However, to date, imaging studies have failed to identify any neurological anomalies that are either consistently associated with, or unique to autism.

Life expectancy/natural history

An increased risk of premature mortality in autism, especially among females and individuals of lower IQ, has been highlighted in a number of systematic reviews and is associated with a range of disorders of the nervous, circulatory, respiratory and digestive systems (Forsyth et al., 2023). Among autistic adults of average or above intellectual ability, suicide is a significant cause of premature mortality, particularly among females (Santomauro et al., 2024). Epilepsy is one of the most common causes of early death in individuals of low IQ (Forsyth et al., 2023).

Behavioural and cognitive characteristics

Difficulties in reciprocal social communication and the presence of ritualistic and stereotyped behaviours/interests are core characteristics of autism. The onset of spoken language is often delayed and around 30% of individuals are described as having "minimal" speech. Although intellectual disability was once thought to be a common feature of autism, more recent research indicates that up to 60% of autistic people are of at least average intellectual ability (Katusic et al., 2021).

Outcomes and intervention

Longitudinal studies indicate that many individuals, especially those who do not have additional intellectual disabilities, show significant improvements in core autism symptoms and behavioural difficulties with age. However, prognosis is affected by many individual and environmental factors, including severity of cognitive, social and communication impairments, and the adequacy of educational, occupational and other support systems (Howlin, 2021; 2025; Lord et al., 2022; Tafolla et al., 2025).

Autism is a highly heterogeneous condition and interventions must be tailored to individual and family needs. For very young children, approaches with a focus on social communication are recommended. For older children, support to enhance learning and social inclusion in school is required. Many adults need assistance to develop self-help and independence skills, and to maintain good mental health. The provision of programmes to ensure access to college, employment, and independent living is also crucial. There are no drugs that can be used to treat autism per se, but access to adequate medical care is needed to reduce the impact of co-occurring physical and mental health problems (Fuentes et al., 2021;Howlin, 2025; Lord et al., 2022).

Useful websites/associations for more information

There are numerous national and international websites offering information and support for individuals, families and professionals e.g.:
https://www.autism.org.uk/
https://www.autismspeaks.org/
https://www.autistica.org.uk/
https://sparkforautism.org/portal/page/contact-us/

There are also many websites designed specifically for autistic people: e.g.
https://www.iancommunity.org
https://autisticadvocacy.org/
https://awnnetwork.org/

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