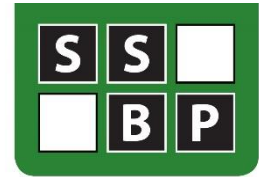


SSBP Syndrome Sheets



Autism Spectrum Disorder

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, 2013; International Classification of Disorders [ICD] 11; World Health Organisation, 2018) 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. In addition, both classifications 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, or genetic condition or environmental factor, or is associated with another neurodevelopmental, psychiatric or behavioural disorder. 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.

Associated conditions

There is a significant association between autism and a wide range of other developmental and genetic disorders including Tuberous Sclerosis and Fragile X (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 problems, the most common being sensory impairments, autoimmune disorders, and obesity, gastrointestinal, and sleep disorders (Rydzewska et al., 2021). The overall prevalence of epilepsy is around 12% (Liu et al., 2022) with rates being highest (at around 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 disorders vary widely from study to study, a recent meta-analysis, based on cases diagnosed via clinical interview, reported an overall prevalence rate of 60% (Lugo Marin 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), (Arnett et al., 2019). 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 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 (Ma et al., 2021).

Environmental risk factors

Recent research has highlighted the impact of gene-environment interactions and a number of potential environmental risks has been identified (Hertz-Picciotto et al., 2018). These include high maternal and paternal age; maternal health factors such as obesity or drugs 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. However, there is no evidence that MMR or other vaccines are a cause of autism.

Prevalence

Prevalence estimates of autism vary, both across and within countries. The most recent systematic review update, based on 71 studies (Zeidan et al., 2021), 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 other studies now suggest that the apparent gender bias may be at least partly due to the fact that formal diagnostic criteria may fail to identify some autistic girls and women (Driver & Chester, 2021).

Physical phenotype

There is no distinct physical phenotype although minor physical anomalies and dysmorphic features are common. There are also increased rates of chronic and acute medical problems across the life span (Bishop-Fitzpatrick & Rubenstein, 2019). Imaging studies have so far failed to identify any neurological anomalies that are either consistently associated with, or unique to autism (Hashem et al., 2020).

Life expectancy/natural history

An increased risk of premature mortality in autism, especially among individuals of lower IQ, has been reported in a number of studies and is associated with a range of disorders of the nervous, circulatory, respiratory and digestive systems. Among autistic adults of average or above intellectual ability, premature mortality is significantly associated with suicide, particularly among females (Hirvikoski, et al., 2020). Epilepsy is one of the most common causes of early death in individuals of low IQ (Hirvikoski, et al., 2016)

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 remaining “minimally verbal”. Although intellectual disability was once thought to be a common feature of autism, more recent research indicates that 60%-70% of autistic people are of at least average intellectual ability (Zeidan et al., 2020).

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 IQ and severity of social and communication impairments, and the adequacy of educational, occupational and other support systems (Howlin, 2021; Lord et al., 2022).

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 help 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; Lord et al., 2022).

Websites: There are numerous national and international websites offering information and support for individuals, families and professionals e.g.:

www.nas.org.uk; www.autistica.org.uk; <https://www.autismspeaks.org/>

There are also many websites designed specifically for autistic people: e.g.

info@SPARKforAutism.org; iancommunity.org/cs/adults

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