

Condition: Autism spectrum disorders

Inheritance:

Multifactorial

Genetic etiology:

Features of autism spectrum disorders (ASD) can be a component of many genetic and chromosomal disorders, as well as genomic copy number changes. In many cases the etiology is unknown.

Frequency:

Various estimates put frequency between 1/600 - 1/2,500.

Clinical features:

ASD is a collection of behavior disorders that begins prior to three years of age and includes impairments in social interaction, impairments in communication, and occurrence of repetitive and stereotypic behaviors. There can be associated developmental and behavioral symptoms, including abnormal sleep, unusual reactions to sound, touch, or food, and abnormal motor development. Formal diagnostic criteria have been established.

Management:

Supportive and symptomatic care, including behavioral therapies and medication.

Genetic counselling:

Empirical recurrence risks for idiopathic ASD are estimated at 4% for sibs of an affected child.