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Asperger Syndrome Fact Sheet

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What is Asperger syndrome?

Asperger syndrome (AS) is a developmental disorder that is characterized by: ¹

limited interests or an unusual preoccupation with a particular subject to the exclusion of other activities

- repetitive routines or rituals
- peculiarities in speech and language, such as speaking in an overly formal manner or in a monotone, or taking figures of speech literally
- socially and emotionally inappropriate behavior and the inability to interact successfully with peers
- problems with non-verbal communication, including the restricted use of gestures, limited or inappropriate facial expressions, or a peculiar, stiff gaze
- clumsy and uncoordinated motor movements

AS is an autism spectrum disorder (ASD), one of a distinct group of neurological conditions characterized by a greater or lesser degree of impairment in language and communication skills, as well as repetitive or restrictive patterns of thought and behavior. Other ASDs include: classic autism, Rett syndrome, childhood disintegrative disorder, and pervasive developmental disorder not otherwise specified (usually referred to as PDD-NOS).

Parents usually sense there is something unusual about a child with AS by the time of his or her third birthday, and some children may exhibit symptoms as early as infancy. Unlike children with autism, children with AS retain their early language skills. Motor development delays – crawling or walking late, clumsiness – are sometimes the first indicator of the disorder.

The incidence of AS is not well established, but experts in population studies conservatively estimate that two out of every 10,000 children have the disorder. Boys are three to four times more likely than girls to have AS.

Studies of children with AS suggest that their problems with socialization and communication continue into adulthood. Some of these children develop additional psychiatric symptoms and disorders in adolescence and adulthood.

Although diagnosed mainly in children, AS is being increasingly diagnosed in adults who seek medical help for mental health conditions such as depression, obsessive-compulsive disorder (OCD), and attention deficit hyperactivity disorder (ADHD). No studies have yet been conducted to determine the incidence of AS in adult populations.

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Why is it called Asperger syndrome?

In 1944, an Austrian pediatrician named Hans Asperger observed four children in his practice who had difficulty integrating socially. Although their intelligence appeared normal, the children lacked nonverbal communication skills, failed to demonstrate empathy with their peers, and were physically clumsy. Their way of speaking was either disjointed or overly formal, and their all-absorbing interest in a single topic dominated their conversations. Dr. Asperger called the condition "autistic psychopathy" and described it as a personality disorder primarily marked by social isolation.

Asperger's observations, published in German, were not widely known until 1981, when an English doctor named Lorna Wing published a series of case studies of children showing similar symptoms, which she called "Asperger's" syndrome. Wing's writings were widely published and popularized. AS became a distinct disease and diagnosis in 1992, when it was included in the tenth published edition of the World Health Organization's diagnostic manual, *International Classification of Diseases* (ICD-10), and in 1995 it was added to the *Diagnostic and Statistical Manual of Mental Disorders* (DSM-IV), the American Psychiatric Association's diagnostic reference book.

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What are some common signs or symptoms?

The most distinguishing symptom of AS is a child's obsessive interest in a single object or topic to the exclusion of any other. Some

children with AS have become experts on vacuum cleaners, makes and models of cars, even objects as odd as deep fat fryers. Children with AS want to know everything about their topic of interest and their conversations with others will be about little else. Their expertise, high level of vocabulary, and formal speech patterns make them seem like little professors.

Children with AS will gather enormous amounts of factual information about their favorite subject and will talk incessantly about it, but the conversation may seem like a random collection of facts or statistics, with no point or conclusion.

Their speech may be marked by a lack of rhythm, an odd inflection, or a monotone pitch. Children with AS often lack the ability to modulate the volume of their voice to match their surroundings. For example, they will have to be reminded to talk softly every time they enter a library or a movie theatre.

Unlike the severe withdrawal from the rest of the world that is characteristic of autism, children with AS are isolated because of their poor social skills and narrow interests. In fact, they may approach other people, but make normal conversation impossible by inappropriate or eccentric behavior, or by wanting only to talk about their singular interest.

Children with AS usually have a history of developmental delays in motor skills such as pedaling a bike, catching a ball, or climbing outdoor play equipment. They are often awkward and poorly coordinated with a walk that can appear either stilted or bouncy.

Many children with AS are highly active in early childhood, and then develop anxiety or depression in young adulthood. Other conditions that often co-exist with AS are ADHD, tic disorders (such as Tourette syndrome), depression, anxiety disorders, and OCD.

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What causes AS? Is it genetic?

Twin and family studies suggest there is a genetic vulnerability to AS and the other ASDs, but a specific gene for AS hasn't been identified. It is likely that multiple genes cause AS, since the symptoms and the severity of symptoms vary so widely among individuals.

Researchers recently identified an association between certain behavioral traits (the insistence on strict routines and repetitive behavior) in a group of children with autism and a specific gene – GABRB3. Another study discovered a strong association between autism and the mutation of a gene the researchers call ENGRAILED 2. Additional evidence for the link between inheritable genetic mutations and AS is seen in the higher incidence of family members of children with an ASD who have similar behavioral symptoms, but in a more limited form. For example they may have mild social, language, or reading problems.

Current research points to structural abnormalities in the brain as a cause of AS. These abnormalities impact neural circuits that control thought and behavior. Researchers think that gene/environment interactions cause some genes to turn on or turn off, or turn on too much or too little in the wrong places, and this interferes with the normal migration and wiring of embryonic brain cells during early development.

Researchers at the University of California, supported in part by the National Institutes of Health, have proposed the disorder stems from abnormal changes that happen during critical stages of fetal development. Defects in the genes that control and regulate normal brain growth create abnormal growth patterns, which cause overgrowth in some brain structures and reduced growth, or excessive cell loss, in others.

Using advanced brain imaging techniques, scientists have revealed structural and functional differences in specific brain regions between the brains of normal versus AS children. One study found a lack of activity in the frontal lobe of AS children when asked to respond to tasks that required them to use their judgment. Another found differences in brain activity when children were asked to respond to facial expressions. Other methods of investigating brain function have revealed abnormal levels of particular proteins in the brains of adults with AS, which correlate with obsessive and repetitive behaviors.

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How is it diagnosed?

The diagnosis of AS is complicated by the lack of a standardized diagnostic screen or schedule. In fact, because there are several screening instruments in current use, each with different criteria, the same child could receive different diagnoses, depending on the screening tool the doctor uses.

To further complicate the issue, some doctors believe that AS is not a separate and distinct disorder. Instead, they call it high-functioning autism (HFA), and view it as being on the mild end of the ASD spectrum with symptoms that differ -- only in degree -- from classic autism. Some clinicians use the two diagnoses, AS or HFA, interchangeably. This makes gathering data about the incidence of AS difficult, since some children will be diagnosed with HFA instead of AS, and vice versa.

Most doctors rely on the presence of a core group of behaviors to alert them to the possibility of a diagnosis of AS. These are:

- abnormal eye contact
- aloofness
- the failure to turn when called by name
- the failure to use gestures to point or show
- a lack of interactive play
- a lack of interest in peers

Some of these behaviors may be apparent in the first few months of a child's life, or they may appear later. Problems in at least one of the areas of communication and socialization or repetitive, restricted behavior must be present before the age of 3.

The diagnosis of AS is a two-stage process. The first stage begins with developmental screening during a "well-child" check-up with a family doctor or pediatrician. The second stage is a comprehensive team evaluation to either rule in or rule out AS. This team generally includes a psychologist, neurologist, psychiatrist, speech therapist, and additional professionals who have expertise in diagnosing children with AS.

The comprehensive evaluation includes neurologic and genetic assessment, with in-depth cognitive and language testing to establish IQ and evaluate psychomotor function, verbal and non-verbal strengths and weaknesses, style of learning, and independent living skills.

An assessment of communication strengths and weaknesses includes evaluating non-verbal forms of communication (gaze and gestures); the use of non-literal language (metaphor, irony, absurdities, and humor); patterns of inflection, stress and volume

modulation; pragmatics (turn-taking and sensitivity to verbal cues); and the content, clarity, and coherence of conversation. The physician will look at the testing results and combine them with the child's developmental history and current symptoms to make a diagnosis.

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Are there treatments available?

The ideal treatment for AS coordinates therapies that address the three core symptoms of the disorder: poor communication skills, obsessive or repetitive routines, and physical clumsiness. There is no single best treatment package for all children with AS, but most professionals agree that the earlier the intervention, the better.

An effective treatment program builds on the child's interests, offers a predictable schedule, teaches tasks as a series of simple steps, actively engages the child's attention in highly structured activities, and provides regular reinforcement of behavior. This kind of program generally includes:

- social skills training, a form of group therapy that teaches children with AS the skills they need to interact more successfully with other children
- cognitive behavioral therapy, a type of "talk" therapy that can help the more explosive or anxious children to manage their emotions better and cut back on obsessive interests and repetitive routines
- medication, for co-existing conditions such as depression and anxiety
- occupational or physical therapy, for children with sensory integration problems or poor motor coordination
- specialized speech/language therapy, to help children who have trouble with the pragmatics of speech – the give and take of normal conversation
- parent training and support, to teach parents behavioral techniques to use at home

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Do children with AS get better? What happens when they become adults?

With effective treatment, children with AS can learn to cope with their disabilities, but they may still find social situations and personal relationships challenging. Many adults with AS are able to work successfully in mainstream jobs, although they may continue to need encouragement and moral support to maintain an independent life.

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What research is being done?

The National Institute of Neurological Disorders and Stroke (NINDS) is one of the federal government's leading supporters of biomedical research on brain and nervous system disorders. The NINDS conducts research in its laboratories at the National Institutes of Health in Bethesda, Maryland, and also awards grants to support research at universities and other facilities.

Many of the Institutes at the NIH, including the NINDS, are sponsoring research to understand what causes AS and how it can be effectively treated. One study is using functional magnetic resonance imaging (fMRI) to show how abnormalities in particular areas of the brain cause changes in brain function that result in the symptoms of AS and other ASDs. A large-scale study is comparing neuropsychological and psychiatric assessments of children with possible diagnoses of AS or HFA to those of their parents and siblings to see if there are patterns of symptoms that link AS and HFA to specific neuropsychological profiles. A clinical trial is testing the effectiveness of an anti-depressant in individuals with AS and HFA who exhibit high levels of obsessive/ritualistic behavior. Other investigators are conducting a long-range study to collect and analyze DNA samples from a large group of children with AS and HFA and their families to identify genes and genetic interactions that are linked to AS and HFA.

¹Adapted from the *Diagnostic and Statistical Manual of Mental Disorders IV* and the *International Classification of Diseases - 10*

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Where can I get more information?

For more information on neurological disorders or research programs funded by the National Institute of Neurological Disorders and Stroke, contact the Institute's Brain Resources and Information Network (BRAIN) at:

BRAIN
P.O. Box 5801
Bethesda, MD 20824
(800) 352-9424
<http://www.ninds.nih.gov>

Information also is available from the following organizations:

MAAP Services for Autism, Asperger's, and PDD

P.O. Box 524
Crown Point, IN 46308
chart@netnitco.net
<http://www.maapservices.org>
Tel: 219-662-1311
Fax: 219-662-0638

Autism Network International (ANI)

P.O. Box 35448
Syracuse, NY 13235-5448
jisincla@mailbox.syr.edu
<http://ani.autistics.org>

Autism Society of America

7910 Woodmont Ave.
Suite 300
Bethesda, MD 20814-3067
<http://www.autism-society.org>

Autism Research Institute (ARI)

4182 Adams Avenue
San Diego, CA 92116
<http://www.autismresearchinstitute.com>
Tel: 619-281-7165

National Institute of Mental Health (NIMH)
National Institutes of Health, DHHS
6001 Executive Blvd. Rm. 8184, MSC 9663
Bethesda, MD 20892-9663
nimhinfo@nih.gov
<http://www.nimh.nih.gov>
Tel: 301-443-4513/301-443-8431 (TTY)
866-615-NIMH (-6464)
Fax: 301-443-4279

**National Institute on Deafness and Other Communication Disorders
Information Clearinghouse**
1 Communication Avenue
Bethesda, MD 20892-3456
nidcdinfo@nidcd.nih.gov
<http://www.nidcd.nih.gov>
Tel: 800-241-1044 800-241-1055 (TTD/TTY)

**National Institute of Child Health and Human
Development (NICHD)**
National Institutes of Health, DHHS
31 Center Drive, Rm. 2A32 MSC 2425
Bethesda, MD 20892-2425
<http://www.nichd.nih.gov>
Tel: 301-496-5133
Fax: 301-496-7101

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Prepared by:
Office of Communications and Public Liaison
National Institute of Neurological Disorders and Stroke
National Institutes of Health
Bethesda, MD 20892

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