First described in 1943 as a syndrome impacting behavior, autism is typically diagnosed within the first three years of a child’s life. The three areas evaluated to reach a diagnosis of autism are:

1. An impairment in the ability to interact socially with people; often demonstrating a lack of eye contact and disinterest in physical contact such as hugging or hand-holding;
2. An impairment in the ability to communicate using speech and/or gestures; and
3. A tendency to have narrow patterns of interests and activities coupled with repetitive and obsessive behaviors, and a lack of pretend or imaginative play; often children with autism find it necessary to have rigid and structured routines.

There are a wide range of variants and degree of demonstrated behavior such that autism is often defined as “autistic spectrum disorder” or ASD. Some children have clear signs of ASD in two of the main areas required for diagnosis, but have less obvious features in the third. In these instances, individuals are said to have an atypical form of autism. When the intellectual abilities are normal, early language development is not significantly delayed and speech is well developed, then individuals may meet criteria for another variant called Asperger’s syndrome. A third variant, termed pervasive developmental disorder (not otherwise specified) or PDD, describes individuals who have difficulties in all three areas but fail to meet full criteria in any of the areas.

What Is the Link Between Autism Spectrum Disorder and Tuberous Sclerosis Complex?

Over the years, it has become recognized that between one-fourth and one-half of all children with tuberous sclerosis complex (TSC) develop ASD. The rate of ASD in the general population is substantially lower (around 0.5 or 0.6% of the total population), so there is clearly a very substantial increase in the rate of ASD in children with TSC. Likewise, the rate of TSC in children diagnosed with ASD is around 1 percent. Although this is a relatively low rate it is still clearly much higher than the rate of TSC in the general population, which is somewhere between 1 in 6,000 individuals. Either way, the overlap between ASD and TSC is clear.

ASD is usually diagnosed in young children between the ages of 2 and 4, but in individuals with TSC, the diagnosis of ASD may go unrecognized or be delayed due to other developmental disabilities. The importance of an accurate diagnosis of ASD for individuals with TSC is so that the individual can receive appropriate educational services and life-long support, as needed.
Why Do Individuals with TSC Frequently Develop ASD?
Current research does not definitely answer the questions related to the increase of ASD in individuals with TSC. However, some important leads are beginning to form the basis of an explanation for the link. In general, it is believed the abnormalities in brain development that occur in TSC sometimes interfere with the proper development of brain areas that are important for the development of social communication skills (the ability to appropriately interact with other individuals).

Evidence is beginning to emerge that shows that if cortical tubers (which develop in earlier stages of brain development) in individuals with TSC involve the region of the brain called the temporal lobes, then there is an increased likelihood of an ASD developing. The temporal lobes are important for processing auditory information, especially speech sounds as well as information about faces and facial expressions. Interference with the development of these key skills may then lead to the social communication difficulties that characterize ASD.

It seems, however, that the presence of cortical tubers in the temporal lobes is not sufficient on its own to produce ASD. Instead, it appears that when temporal lobe tubers occur in conjunction with the onset of seizures at a young age, often presenting as infantile spasms, then this combination of factors leads to the much higher chance of ASD. Although the link with early onset epilepsy and infantile spasms raises the possibility that the seizures may play a role in interfering with normal development of brain systems important in social communication, it is possible that the link with early seizures instead reflects the presence of cortical tubers and related structural abnormalities in key locations in the brain. These structural abnormalities may give rise to both the seizures as well as ASD. Further research to try to determine which of these two explanations is correct is required, especially as it has such important implications for treatment.

Is It Important to Diagnose ASD in Individuals with TSC?
Some people express the view that it is enough that an individual has TSC, so another diagnosis such as ASD is unnecessary. Although it makes sense to avoid adding diagnoses and labels, the diagnosis of an ASD is important for several reasons. A diagnosis can often help parents make sense of a range of rather unusual behaviors that otherwise seem extremely puzzling. Often, parents feel that somehow they have been doing something wrong in how they are parenting their child, and that the difficulties that the child is having in relating to others, communicating or playing is somehow the parents’ fault. It can be quite helpful for parents to discover that some of the unusual behaviors their child may be demonstrating are part of the developmental delays a child may be experiencing related to the autistic process.

In addition, the diagnosis is important because children with ASD often benefit from early intervention services that support improvement in speech, language and behaviors. Early intervention services are available for very young children and their families. These services include physical therapy, speech therapy and occupational therapy. Early intervention services work with not only the child with ASD, but also the parents and siblings. The goal of early services is to foster the development of children with ASD.
How Is the Diagnosis Made?
The diagnosis of an ASD is based on a report of the child’s early development, detailing the way in which he or she acquired skills and the areas in which he or she has struggled, coupled with careful observations and assessments. These evaluations need to be performed by individuals who are experienced in evaluating individuals with complex developmental disabilities and ASD. The assessments are lengthy, and it may be necessary for the evaluator to see the child at home or in the playgroup or nursery setting, often referred to as a functional contextual assessment, before the diagnosis can be confirmed. The diagnosis of ASD is made through a team evaluation, including reports from therapists, pediatricians, teachers, parents and psychologists.

There are several assessments that are used to reach a diagnosis of ASD; the most commonly used is called the Autism Diagnostic Observation Schedule (ADOS). This assessment should be performed by someone familiar with ASD who is trained to utilize the ADOS.

When Is Diagnosis Possible?
To some extent the answer to this depends on the individual’s overall level of ability. In individuals who have the most severe cognitive disabilities, it can sometimes be extremely difficult to make a definitive diagnosis. In general it is hard to make a confident diagnosis before the individual’s cognitive age level is at least equivalent to that of an 18-month to 2-year-old child. In less affected individuals, it might well be possible to make a diagnosis around the age of 2, whereas in the individuals with very significant delays in development it may not be possible until they are much older. Research is continuing to identify the early markers of ASD so early treatments can be implemented.

What Treatment Is Suggested?
Treatment options vary based on the individual’s age and ability. The focus of the treatment is often targeted at strengthening skills in individual areas of difficulty. Special education provisions and accommodations are incorporated in a child’s individual education plan (IEP). This often includes the individual working with a multidisciplinary team of clinical professionals that provide several different services, including speech and language therapists, developmental and child psychologists and pediatricians.

According to the Autism Society of America, treatment approaches include:

- Applied Behavioral Analysis (ABA) and Discrete Trial Training
- Treatment of Autistic and Related Communication Handicapped Children (TEACCH)
- Picture Exchange Communication System (PECS)

**ABA and Discrete Trial Training** are often used interchangeably. These methods include intense repetitive, structured tasks in which good behavior is rewarded and undesirable behavior is ignored. It is time intensive and focuses on changing current behaviors and does not prepare individuals to respond in new situations. Some individuals with TSC who have ASD have significantly benefited from ABA programs.

**TEACCH** (Treatment of Autistic and Related Communication Handicapped Children) was developed at the University of North Carolina. TEACCH focuses on adapting the environment...
to the individual with ASD instead of trying to make the individual adapt to the environment. This is achieved through high structure, organizational charts and schedules. While many favor this approach, some feel that it is too structured and makes the individual too dependent on charts and other organizational tools.

**PECS** (Picture Exchange Communication System) is used to encourage communication. By using pictures, an individual can point to or hand an object to someone to demonstrate what he or she wants.

Options vary and the treatment program needs to be tailored to the individual’s age and ability. Treatment is targeted at fostering skills in the three main areas of difficulty — social and communication skills and the development of imaginative play. In addition, treatment aims to ensure that the repetitive or obsessive behaviors do not become too marked or prominent and do not interfere with family life. Lastly, the treatment aims to help parents foster their child’s development and support them during the early, often very demanding, years.

There is growing evidence to suggest that early intervention programs may be one of the most effective current forms of treatment in individuals with ASD, but it is not yet known to what extent the intervention programs of this kind are helpful for children with TSC. Research is needed to evaluate effectiveness of these programs for individuals with TSC who have ASD.

**What Will the Future Hold?**
Detailed knowledge about the way individuals with TSC and ASD develop is currently being gained through studies in the U.S. and the U.K, so for now we can only be guided by the development of individuals with ASD who do not have TSC. The range of outcomes here is very great. At one extreme, individuals can have persisting serious problems throughout childhood and into adult life. Some individuals with ASD are prone to self-injury, particularly if they get upset or frustrated when their routines or activities are interrupted, or if they get frustrated over their communication difficulties.

At the other extreme, individuals with Asperger Syndrome or high-functioning ASD can largely outgrow their difficulties and lead an independent or semi-independent life in adulthood. The outcome is to some extent related to the severity of the associated cognitive impairments or a demonstrated level of cognitive disabilities. Individuals who have severe or profound forms of cognitive ability are likely to have persisting difficulties. In addition, the amount of useful speech that the individual acquires indicates how they will fare in the future. Lastly, the severity of the social and communication difficulties and behavior problems is also helpful in determining what the outcome will be. The more severe the problems, the more persistent they tend to be.

**Additional Resources**
Autism Today
[www.autismtoday.com](http://www.autismtoday.com)

Autism PDD Resources
[www.autism-pdd.net](http://www.autism-pdd.net)
Autism Resources
www.autism-resources.com

Center for the Study of Autism
www.autismwebsite.com

National Institutes of Health Autism Research Network
www.autismresearchnetwork.org/AN/

Autism Society of America
7910 Woodmont Avenue, Suite 300
Bethesda, MD 20814
Phone: 301-657-0881 or 1-800-328-8476
www.autism-society.org

Autism Information Center
Centers for Disease Control and Prevention
Phone: 1-800-CDC-INFO
Email: cdcinfo@cdc.gov
www.cdc.gov/ncbddd/dd/ddautism.htm

Autism Speaks
1 East 33rd Street, 4th Floor
New York, NY 10016
Phone: (212) 252-8584
Fax: (212) 252-8676
E-mail: contactus@autismspeaks.org
www.autismspeaks.org/index2.php

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