



## **Tuberous Sclerosis and Autism Spectrum Disorders**

### **What is autism?**

Autism is a behaviourally defined syndrome that was first described by an American child psychiatrist called Leo Kanner in 1943. The hallmarks of autism are:

1. A particular kind of impairment in the ability to interact socially with people.
2. A particular kind of impairment in the ability to communicate using speech and/or gesture.
3. A tendency to have narrow patterns of interests and activities coupled with repetitive and obsessive behaviours and a lack of pretend or imaginative play.

These behavioural features to begin within the first three years of a child's life.

### **Variants of autism and the autistic spectrum**

Since Leo Kanner's first descriptions of the classic autistic syndrome, there has been a growing appreciation that there are a number of variants to this classic picture. Some children, for example, have clear signs 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 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, describes children who have difficulties in all three areas but fail to meet full criteria in any. Collectively, these variants of autism and classic autism are increasingly referred to as the autistic spectrum disorders.

### **The link between autism and tuberous sclerosis**

Over the last twenty years or so, it has become recognised that children with tuberous sclerosis frequently develop autism or one of the autistic spectrum disorder variants of the classic syndrome. It seems that somewhere between a quarter to a half of children with tuberous sclerosis develop an autism spectrum disorder. The rate of autism spectrum disorders in the general population is substantially lower (around 0.5 or 0.6%) so there is clearly a very substantial increase in the rate in children with tuberous sclerosis. Likewise, the rate of tuberous sclerosis in children diagnosed as suffering from an autism spectrum disorder is around 1%. Although this is a relatively low rate it is still clearly much higher than the rate of tuberous sclerosis in the general population, which is somewhere between 1 in 10,000 to 1 in 20,000 individuals. Either way, therefore, the overlap between autism spectrum disorders and tuberous sclerosis is very clear.

### **Why do children with tuberous sclerosis frequently develop an autism spectrum disorder?**

As yet there is no definitive answer to this important question, but research has thrown up some valuable leads which are beginning to form the basis of an explanation for the links. In general terms it is believed the abnormalities in brain development that occur in tuberous sclerosis sometimes interfere with the proper development of brain areas that are important in the development of social communication skills. Evidence is beginning to emerge to show that if cortical tubers (which develop in earlier stages of brain development) in children with tuberous sclerosis involve the region of the brain called the temporal lobes, then there is an increased likelihood of an autism spectrum disorder 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 on to the social communication difficulties that characterise autism spectrum disorders. It seems, however, that the presence of cortical tubers in the temporal lobes is not sufficient on its own to produce autism. Instead, it appears that when temporal lobe tubers occur in conjunction with early onset seizures often presenting as infantile spasms, then this combination of factors leads on to the much higher chance of an autism spectrum disorder. 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 give rise to both the seizures as well as autism. 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 an autism spectrum disorder in children with tuberous sclerosis?**

Some people express the view that it is enough that a child has tuberous sclerosis and yet another diagnosis such as autism is unnecessary. Although it makes sense to avoid adding diagnoses and complications, the diagnosis of an autism spectrum disorder is important for several reasons. To begin with, a diagnosis can often help parents make sense of a range of rather unusual behaviours that otherwise seem extremely puzzling. Not infrequently, parents feel that somehow they have been doing something wrong in the way in which they are raising their child, and that the difficulties that the child is having in relating, communicating or playing is because they have not been doing things correctly. It can be quite a help, therefore, just to discover that some of the unusual behaviours are part and parcel of the child's developmental problems and have little to do with the way they have been bringing the child up.

In addition, the diagnosis is important because children with autism spectrum disorders have a particular set of special needs that might be overlooked if a diagnosis isn't established. For example, they may benefit from specific

treatments for autism, or from special educational programmes designed to foster the development of children with autistic spectrum disorders. Another reason for making a diagnosis is that it can tell us something about how the future might look, and what to expect as their child grows older.

### **How is the diagnosis made?**

The diagnosis of an autism spectrum disorder is based on a report of the child's early development, detailing the way in which they have acquired skills, the areas in which they have struggled, coupled with careful observations and assessments. These evaluations need to be done by individuals who are experienced in evaluating children with complex developmental disabilities and autism spectrum disorders. The assessments may take quite some time, and it may be necessary to see the child at home or in the playgroup or nursery setting before the diagnosis can be confirmed. Ordinarily, diagnoses are made by child psychologists or psychiatrists or paediatricians, often working together as a team.

### **When is diagnosis possible?**

To some extent the answer to this depends on the child's overall level of ability. In children who are profoundly handicapped, it can sometimes be extremely difficult to make a definitive diagnosis. In general it is hard to make a confident diagnosis before the child's mental age level is at least equivalent to that of an 18 months to two year old child. In able children it might well be possible to make a diagnosis round about the age of two, whereas in the children with very significant delays in development it may not be possible until they are much older. Research is going on to try to pinpoint early markers of autism spectrum disorders in order that early treatments can be implemented.

### **What treatment is required?**

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

Special educational provision according to the particular needs of the child may also be necessary, and the exact mix and package of treatment and education rather depends on local circumstances and provision as well as the particular needs of the child.

Treatment is usually provided by a multidisciplinary team of clinical professionals including speech and language therapists, developmental psychologists, child psychiatrists and paediatricians, as well as special educationalists.

There is growing evidence to suggest that early intervention programmes may be one of the most effective current forms of treatment in children with autism, but as yet it is not known to what extent the intervention programmes of this kind are helpful for children with tuberous sclerosis. There is no special reason for thinking that they shouldn't also be helpful, but there is a need for proper evaluation of their effectiveness.

### **What will the future hold?**

Detailed knowledge about the way in which children with tuberous sclerosis and an autism spectrum disorder develop is not available, so until research has been done to answer this question, we can only be guided by knowledge of the way in which children with autism spectrum disorders in general develop. The range of outcomes here is very great. At one extreme, children can have persisting serious problems throughout childhood and into adult life. Some children with autism spectrum disorders 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, able children with Asperger's syndrome or high functioning autism 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 intellectual impairments or mental retardation. Obviously, children who have severe or profound forms of handicap are likely to have persisting difficulties. In addition, the amount of useful speech that the child acquires indicates how they will fare in the future. Lastly, the severity of the social and communication difficulties and behaviour problems is also helpful in determining what the outcome will be. The more severe the problems, the more persistent they tend to be.

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