



**T**he original description of autism, which from a psychiatric definition relates to an individual that is 'self absorptive', was used by Leo Kanner who studied a small group of children exhibiting obsessive/repetitive behaviours and lacked emotional contact.<sup>1</sup> At a similar time Asperger described an 'autistic psychopathy' in children who had poor motor skills, obsessive behaviour and difficulties with communication. The observations of Kanner and Asperger have come to define the clinical picture of autism and Asperger syndrome. This compartmental approach to diagnosis along with aligned conditions such as pervasive developmental disorder not otherwise specified (PDD-NOS) has now shifted, and these conditions belong to what is termed an autistic spectrum disorder (ASD). This spectrum approach acknowledges that the clinical lines between each diagnostic condition may blur and overlap and that a range of abilities and disabilities may be present within the individual.

An individual with an ASD will exhibit deficits to a greater or lesser extent compared to a typically developing child in three core domains.

- A difficulty with language and communication
- Poor reciprocal social interactions
- Repetitive stereotyped behaviours and may lack creative play.

The triad of social impairment, poor language and a lack of imaginative play may be recognised in children at the age of two but owing to the differences in the severity of these signs may be overlooked and a child may not be diagnosed until later in life. The early detection of ASD in young children is important, because early intervention by speech and language therapists, occupational therapists, and the introduction of social stories to reinforce behaviour all help the child develop the areas where he/she may be lacking. The optometrist can play an important role in noticing possible behavioural differences in these young children.

ASD is a lifelong developmental condition whose prevalence is increasing owing to greater awareness of this condition rather than any single causative agent. One large study based on over 50,000 children born in the South Thames region of London and aged 9-10 years found the prevalence of ASD to be 1:116 with a male bias of 3.3:1.<sup>2</sup> Within this group, the prevalence of childhood autism defined as a child

# Autism

## Part 1 - Background to aetiology and diagnosis

In the first of a two-part series on autism and its clinical consequences, **Dr Paul Constable** takes a look at the background of the disease spectrum. **Module C16181**, one general CET point for optometrists and dispensing opticians

**TABLE 1**

### Proposed diagnostic criteria for autism spectrum disorder

Proposed classification of autism spectrum disorder DSM-5

An individual must fulfil the following three criteria:

- (1)** Clinically significant, persistent deficits in social communication and interactions, as manifest by all of the following:
  - Marked deficits in nonverbal and verbal communication used for social interaction
  - Lack of social reciprocity
  - Failure to develop and maintain peer relationships appropriate to developmental level
- (2)** Restricted, repetitive patterns of behaviour, interests, and activities, as manifested by at least TWO of the following:
  - Stereotyped motor or verbal behaviours, or unusual sensory behaviours
  - Excessive adherence to routines and ritualised patterns of behaviour
  - Restricted, fixated interests
- (3)** Symptoms must be present in early childhood (but may not become fully manifest until social demands exceed limited capacities)

years found that 75 per cent showed mild to severe social difficulties, with 25 per cent showing near normal social functioning, although most lived at home, were not employed and continued to display stereotyped repetitive patterns of behaviour. The majority of individuals with ASD will have difficulties throughout their lives; however, a small percentage may display exceptional abilities in specific areas such as mathematics, drawing or music.<sup>7</sup>

The classification of ASD is currently being revised, with the proposed revisions for the Diagnostic and Statistical Manual of Mental Disorders (DSM-5) to be published in May 2013 shown in Table 1.

Criteria 1 highlights the difficulties faced by a child with ASD in relating to peers and understanding emotion. A child with ASD does feel emotions of happiness and sadness, but their ability to relate to other people's feelings may be impaired. The lack of communication may be the first apparent sign of a problem with a child that fails to meet developmental targets. Communication difficulties encountered by children with ASD is another contributing factor to their ability to form relationships with their peers. A child with ASD may have a verbal age years below their chronological age. Conversely language may be well developed and the child may be hyperlexic but unable to understand the unspoken meaning of words. For example, 'John went to the store, then the bakers and finally he crossed the road to get to the post office.' Was John in the town or country? This would perplex a child with ASD because there is no direct statement about where John is. This inability to infer information and imagine is one of the characteristics of a child with ASD. This has important implications for communicating with a child on the spectrum. Language should

that fulfilled diagnostic criteria of the ADI (autism diagnostic interview)<sup>3</sup> at 4-5 years of age and met current ADOS (autism diagnostic observational schedule)<sup>4</sup> categorisation for autism and also met a clinical diagnosis for autism by trained observers was 38.9:10,000. These findings were in concordance with a similar large study based in the US that found a prevalence of 40.5:10,000.<sup>5</sup> The prevalence of a child fulfilling classification for ASD is thus ~ 1:100, while for children that fulfil the narrower definition of autism as recognised by Kanner this is ~ 3:1,000.

The mean ± SD intelligence quotient reported for ASD children was 69.4 ±24.1 in the South Thames study, which shows that children with ASD generally have poor IQ levels.<sup>2</sup> One longitudinal study based in London on children who were first seen when aged 7-8 and again when they were 23-24

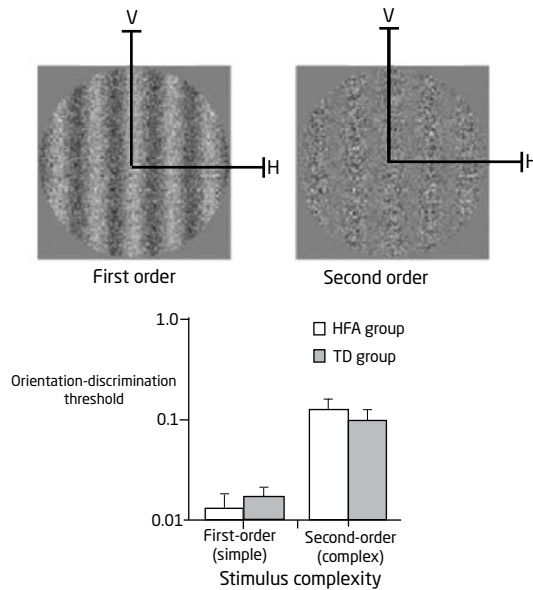
be unambiguous and with as few as words as possible. Criteria 2 emphasises that these children are likely to have stereotyped, repetitive behaviours with unusual sensory behaviour. This may be observed in the way a child will want to do the same task over and over again. They may also show hyper- or hyposensitivity, to sensory stimuli such as sound, smell or touch. Criteria 3 emphasises that these clinical signs are present in early childhood but are variable and so may not manifestly affect a child until pre-school or even later stages of development.

### Diagnosis

A child with a diagnosis of ASD will ideally have a statement of educational needs and may attend either mainstream school, with support, or a specialist school. A team of clinicians that typically includes a paediatrician, child psychologist and speech and language therapist would be involved in the initial diagnosis. Several tests that can be administered to assess whether an individual falls on the autism spectrum. The ADOS is considered more robust because the diagnosis is based on direct observation of the child rather than third-hand reports of behaviour that is used in the ADI. The ADOS is a semi-structured assessment of social interaction, communication, play, and imaginative use of materials for individuals suspected of having ASD. The ADOS can be administered to adults or children and codes the individual depending upon their responses to questions and by observing the individual at play or while engaged in an activity such as building a tower from blocks. In addition, several questionnaires are available to indicate autistic traits in children aged 4-5 such as the Social Communication Questionnaire (SCQ).<sup>8</sup> In addition to the Developmental, Dimensional and Diagnostic Interview (3di), a parental interview can also be used to assess the presence of autistic symptoms in a child.<sup>9</sup> The diagnosis of ASD relies upon a clinician's experience of seeing children with an ASD. Clinicians typically undergo peer review in the diagnostic and assessment protocols for making the clinical diagnosis.

### Theories of ASD

One aspect of ASD is attention to detail, which may be seen in a child that will become absorbed in one part of an object. The child with ASD may be unable to see the 'woods' but is always noticing fine details about the 'trees'. Early findings on cognitive



**Figure 1** The basis of Enhanced Perception Functioning theory is supported by superior orientation discrimination of stimuli defined by simple first order (luminance) properties compared with more complex second order (texture) defined stimuli. Reproduced with permission of Oxford University Press. HFA: high functioning autism; TD: typically developing

behaviours in ASD children found that they typically outperformed typically developing children on the Embedded Figure Task (EFT). Superiority on the EFT has formed the basis of several cognitive theories that have been developed to explain visual perception and the broader ASD phenotype. One explanation, known as enhanced perceptual functioning (EPF), posits that the central abnormality lies in the early processing of low-level visual information.<sup>10</sup> Mottron's group have demonstrated superiority in an orientation discrimination task whereby the ASD group were more adept at determining the orientation of a Gabor patch defined by luminance (first order) but worse when the Gabor was defined by texture (second order). The lower orientation discrimination thresholds in the ASD group support higher or greater perception for stimuli that are encoded by lower areas of visual cortex. This enhanced perception is not seen when the orientation was defined by texture requiring additional processing in the cortex to determine the orientation (Figure 1).

An alternative model that draws upon elements of EPF to describe ASD is 'weak central coherence' (WCC) that acknowledges that individuals with ASD have an enhanced perceptual bias. However, WCC contends that it is the way in which an individual with ASD is able to integrate the

sensory input that is impaired and this different cognitive style when dealing with sensory information results in autistic behaviours. This 'weak coherence' therefore results in an inability to create a coherent picture of the world from the constituent parts.<sup>11</sup> The altered cognitive style in ASD is evident in the manner in which ASD boys would focus on fine detail when copying a picture, and typically begin by drawing in the details before the background.<sup>12</sup> Pellicano *et al*<sup>13</sup> have demonstrated the superior ability of ASD children to locate a figure hidden in a complex shape using the childhood embedded figure test (CEFT). They found that ASD children were faster on this task (Figure 2) highlighting the atypical processing of a visual scene, by being able to detect the parts (triangle) without being distracted by the background (clock).

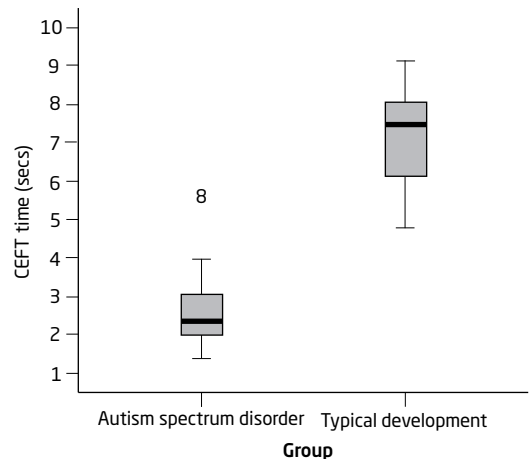
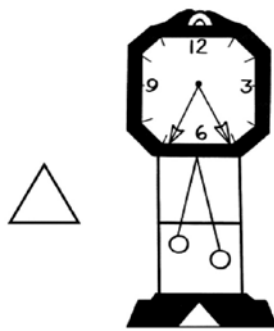
The local or fine detail over the broader, large picture perspective of the world that is observed in ASD is also present in other visual tasks, notably visual search. Faster reaction times in young children and adolescents have been replicated in several studies.<sup>14-16</sup> This superiority in visual search may be explained by enhanced perception for details of an object rather than the global 'big-picture'.<sup>17,18</sup>

One model of visual perception is the Reverse Hierarchy Theory as proposed by Hochstein and Ahissar.<sup>19</sup> In this model, there is a constant interaction between lower and higher cortical regions. At one level, orientation selective neurons detect the features of a surface and are associated with the global feed forward pathway 'the woods'. Reciprocal feed back projections to V1 then enable information to be processed regarding the texture within the boundaries, and help fill in the finer details 'the trees'. Vandenbroucke *et al*<sup>20</sup> have used this model to probe the relative balance of feed back and the feed forward connections in ASD. The authors used psychophysics and EEG recordings to study their relative contributions in a figure-ground segregation task. The overall findings supported an imbalance in favour of the feed back pathway. Therefore, in ASD individuals the imbalance would suggest that their visual style is one where the details of an object are more important than the overall shape.

Another viewpoint on ASD is based upon a continuum of which autism merely represents one extreme 'male brain' perspective.<sup>21</sup> The extreme male brain theory is an alternative explanation to the behavioural characteristics of



ASD. In this model, an individual is inclined towards systemising and understanding the whole by breaking it down into component parts. Its underlying premise is that altered levels of androgens affect neural development and result in an extreme 'male brain' pattern of behaviour. Individuals with ASD often display systemising patterns of behaviour and this leaning towards detecting patterns may confer superiority in detecting fundamental features and may contribute to the ASD phenotype.



**Figure 2** ASD children are faster at finding the hidden shape (triangle) in the complex figure. The triangle is formed by the hands of the clock face. Reprinted courtesy of Elsevier from Pellicano *et al.*<sup>13</sup> CEFT: Childhood Embedded Figure Test

**Aetiology of autism**

Currently there is no single aetiological factor that can account for ASD. Several candidate genes have been linked and associated with ASD; however, the complexity of the polymorphisms and the functional roles each may play in cortical development have yet to be fully elucidated. Studies involving rodent models of ASD may provide further insights into the impact of genetic differences seen in the ASD population which may help our understanding of the ASD phenotype. Notable associations in the encoding of genes for neural development and signalling have been found, and are

assumed to play a central role in cortical development.<sup>22</sup>

Anatomical studies of post-mortem brains have also found what is described as a mini-columnopathy whereby the cortical columns in some regions of the brain are abnormally small, which may partially explain why individuals with ASD can focus in on small details owing to the greater density of cortical columns.<sup>23</sup> MRI imaging also reports

abnormal cortical development in high functioning adults that may aid in earlier diagnosis and an understanding of the altered interconnections between cortical regions.<sup>24</sup>

**Summary**

The spectrum of autism is becoming more prevalent with awareness of this condition. The individual with ASD will have lifelong difficulties

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in three core domains of language, social interaction and imagination and will require support with most daily activities. The theories surrounding ASD favour either a local enhanced perceptual model where local elements of a visual scene are processed faster. Alternately it is the linking of these local elements that is incoherent and the 'big picture' becomes difficult to comprehend. MRI and genetic data continue to find new mutations and possible functional abnormalities in the pattern of cortical development that may with the aid of animal models provide some understanding of the autistic mind. ●

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## MULTIPLE-CHOICE QUESTIONS - take part at [opticianonline.net](http://opticianonline.net)

**1** Childhood autism was originally described by?

- A Hans Asperger
- B Leo Kanner
- C Uta Frith
- D Laurent Mottron

**2** The prevalence of ASD is considered to be?

- A 1:10
- B 1:100
- C 1:1,000
- D 1:10,000

**3** Which one of the following uses direct observation of a child in the diagnosis of ASD?

- A ADI
- B 3di
- C SCQ
- D ADOS

**4** Criteria 2 of the proposed DSM-5 classification of ASD emphasises which characteristic?

- A Lack of communication
- B Clinical signs are present in early childhood
- C Stereotyped repetitive behaviours
- D Lack social reciprocity

**5** A finding that supports the EPF theory of ASD?

- A Genetic studies
- B Inferior visual search in ASD children and adults
- C Superior orientation detection for second-order stimuli
- D Superior orientation detection for first-order stimuli

**6** The reverse hierarchy model of visual processing when applied to ASD suggests that there is an imbalance in which pathway?

- A Feed forward
- B Feed back
- C Feed back and feed forward are equally affected
- D Both show weak coherence

Successful participation in this module counts as one credit towards the GOC CET scheme administered by Vantage and one towards the Association of Optometrists Ireland's scheme.

**The deadline for responses is April 21 2011**



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