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# Autism spectrum disorders

## Background

Autism spectrum disorders (ASDs) are serious neurodevelopmental disorders affecting approximately one in 160 Australians. Symptoms are apparent during the second year of life causing impairments in social interaction, communication and behaviour with restricted and stereotyped interests.

## Objective

To increase the general practitioner's awareness of the presenting symptoms of ASDs and their associated problems in children, screening for ASDs, and the assessment process, treatment options and outcomes.

## Discussion

This article discusses the five red flags that are autism alerts in young children. These red flags can enable GPs to play a key surveillance role in determining which young children might require further screening and referral for an ASD assessment. Because ASDs are lifelong, neurodevelopmental disorders and symptoms change over time. Therefore the GP has an ongoing role to support, educate and advise parents, other carers and the individual with an ASD. Treatment and pharmacological interventions are also discussed.

**Keywords:** autistic disorder; child developmental disorders, pervasive



Autism spectrum disorders (ASDs) are serious neurodevelopmental disorders affecting approximately one in 160 Australians.<sup>1</sup> In 1943, Kanner used the word 'autism' to describe children who were unable to relate to others, had delayed and disordered language, repetitive behaviours and a drive for sameness.<sup>2</sup> These three core symptoms have remained central to the diagnosis of a group of disorders referred to as 'pervasive developmental disorders' (PDDs) described in both the Diagnostic and Statistical Manual of Mental Disorders, 4th edition, text revised (DSM-IV-TR)<sup>3</sup> and the International Classification of Diseases (ICD-10).<sup>4</sup> In 1997, Wing introduced the term 'autism spectrum disorders' describing a continuum of conditions from aloof children through to 'active but odd' children who share an autistic 'triad of impairments'.<sup>5</sup> The term has since been used to describe symptoms of severity, changes that occur with development and the associated range of intellectual ability.<sup>6</sup> In line with emerging international practice, in this article the term 'autism spectrum disorders' will refer to autistic disorder, Asperger disorder and pervasive developmental disorder not otherwise specified (PDD-NOS) (atypical autism).

## Autistic disorder

Diagnosis is clear by 30–36 months. However, symptoms are apparent during the second year of life causing impairments in three main areas of functioning:

- social interaction
- communication, and
- behaviour with restricted and stereotyped interests (*Table 1*).

## Early signs of autistic disorder

Symptoms and developmental markers of autistic disorder emerge during the first 2 years of life. Developmental problems before the first birthday have been reported by parents, but the majority express concerns regarding language development and social relatedness by the age of 2 years.<sup>7,8</sup> Early developmental differences include failing to have an anticipatory posture, such as reaching out to be picked up, and absent or reduced visual attention to social



**Table 1. Core features of autistic disorder (autism)**

**Impaired social interaction**

**Progressive abnormalities in interpersonal relationships**

- Reduced responsiveness to, or interest in, people; child may appear aloof and usually have an impaired ability to relate to others
- Impaired ability in nonverbal social relating, eg. impaired use of facial expression, eye contact and difficulty with use of gestures such as waving goodbye and pointing to indicate social interest
- The ability to make friends is absent or distorted and the child is usually unable to engage in reciprocal social play with other children
- Difficulty understanding emotional expression; rarely develops age appropriate empathy. Some social relating skills may develop over time, but these skills are usually restricted or abnormal

**Delayed and disordered communication**

**Stereotyped and repetitive use of language**

- Echolalia – the repetition of words and phrases (often out of context). The child may immediately repeat words and phrases or repeat previously heard favourite phrases, such as advertising jingles or dialogue from movies
- Repetitive questioning and rituals and the creation of own words for objects and people (neologisms)
- Literal understanding of spoken language and poor understanding of sarcasm, metaphors or irony

**Difficulties with the social use of language**

- Unable to initiate or sustain a conversation
- Speaking too loudly or too softly for the context and using an unusual accent or tone

**Lack of a range of varied, spontaneous social imitative or pretend play**

- Older children may engage in what appears to be imaginative play, however it is usually the repetition of learned activities or scenes from favourite movies

**Ritualistic and stereotyped interests and behaviours**

**Preoccupations which are intense and focused**

Fascination with dinosaurs, football fixtures or weather forecasts and repeated questioning or talking in a monologue about favourite topics, even if the context is inappropriate

**Nonfunctional rituals and rigid routines**

Repetitive play – lining up, stacking or sorting objects by colour or shape; lacking imagination and social elaboration with distress if play is interrupted or the child is asked to move on to another activity

Resistance to change in routine or environment. For example, the child may become extremely distressed if there is a new teacher at school, if furniture in the house is rearranged or if the child needs to wear new clothes or shoes

The child may try to control the play of other children and rigidly apply their own inflexible version of the rules

**Repetitive motor mannerisms**

Hand flapping, finger flicking, tiptoe walking

**Preoccupation with parts of objects**

Visually attentive, eg. closely watching spinning wheels, fascination with shadows or reflections and studying collected objects such as stones or bottle-tops

stimuli, smiling in response to others, vocalisation and exploration of objects.<sup>8</sup> Regression and loss of communication and social skills are also observed in 20–40% of cases.<sup>9</sup> A recent Australian study of infants aged from 8 months found that surveillance of early signs of autism emerging by age 18 months led to a diagnosis of an ASD at 24 months.<sup>10</sup> Health professionals such as maternal and child health nurses and general practitioners can play a key surveillance role in determining which young children might require further screening and referral for an ASD assessment. The five red flags,<sup>11</sup> which are autism alerts in young children, are listed in *Table 2*.

**Associated features**

Associated features include unusual and restricted diet, sleep disturbance, difficulty regulating emotions and self injurious behaviour. Sensory and perceptual abnormalities are also common, including sensitivity to sound and smell, lack of response to pain, and preoccupation with visual or tactile stimulation. These features are not specific to children with autism and may occur in association with intellectual disability.

**Intellectual ability**

The majority of children with autistic disorder have an intellectual disability. Approximately 50% have severe intellectual disability and 30% mild to moderate disability. The remaining 20% have intellectual abilities in the normal range and are referred to as having ‘high functioning autism’ (HFA). Cognitive assessment usually reveals a scatter of abilities with more difficulty in verbal and language skills and better performance in visual motor activities.

**Asperger disorder**

Children with Asperger disorder (AD) are differentiated from children with autistic disorder because they do not have a delayed receptive and expressive language development or cognitive development. In common with children with autistic disorder, children with AD have clinically significant impairment in their social interactions and social communication and restricted, repetitive and stereotype patterns of behaviour and interests. They may not come to clinical attention until they are at preschool or primary school when their social difficulties and rigid, odd



and repetitive behaviours become more noticeable and problematic. The key factor differentiating AD from HFA is language development, as those with HFA have delayed and disordered language.

Despite clear differentiating diagnostic criteria, confusion and debate continues regarding whether or not AD constitutes a separate disorder. A proposed fifth revision of the DSM will specify only ASD, manifest as delays and abnormalities in social interactions and the presence of rigid and repetitive behaviours. Asperger disorder will no longer be specified as a separate disorder and the nature of any language disorder and profile of intellectual abilities will need to be separately described.<sup>12</sup> This debate highlights the continuing necessity to describe the full range of symptoms, developmental features, language ability and profile of cognitive skills in order to plan an appropriate management program and provide a baseline to monitor outcome.

## Screening and assessment

General practitioners can use several instruments to screen for an ASD. These include:

- the modified checklist for autism in toddlers (M-CHAT), a parent-completed checklist screening for autism for children aged 16–30 months;<sup>13</sup> and
- the developmental behaviour checklist (DBC), a parent-completed questionnaire of emotional and behavioural problems that includes an autism screening algorithm for children aged 4–18 years<sup>14</sup> and also younger children aged 18–48 months.<sup>15</sup>

A positive screen for autism is not diagnostic, but indicates that referral to a paediatrician, child psychiatrist or autism assessment team is necessary.

Multidisciplinary assessment of development/cognition, language, play skills and sensory sensitivities contribute essential information to help with planning appropriate management and early intervention. As part of the Australian Federal Government's 'Helping Children with Autism Early Intervention Funding Program', specialist Medicare ASD diagnosis numbers and psychology and allied health Medicare assessment item numbers are available.<sup>16</sup> This funding initiative assists families and carers of children aged 0–6 years diagnosed with an ASD and provides \$12 000 funding. The eligibility criteria documentation requires a copy of a definitive statement of diagnosis of a pervasive developmental disorder as classified by the DSM-IV (ie. autistic disorder, Asperger disorder, PDD-NOS, Rett disorder or childhood disintegrative disorder). The diagnosis cannot be suggestive, indicative or provisional.

The definitive diagnosis must be made by a paediatrician, psychiatrist or a multidisciplinary team including a psychologist, speech pathologist and occupational therapist.

## After the diagnosis – the role of the GP and treatment

Autism spectrum disorders are life-time neurodevelopmental disorders and symptoms change over time. The GP has an ongoing role to support, educate and advise parents, other carers and the individual

with an ASD. There are no cures and best practice treatment comprises interventions tailored to help the individual with an ASD to adapt as effectively as possible to their environment.<sup>17</sup>

Because of the serious and chronic nature of ASDs parents are understandably prey to claims of scientifically unsubstantiated and usually expensive treatment. There is emerging evidence that a multimodal program of early intervention tailored to address the profile of symptoms and abilities of each child is more likely to promote development, improve behaviour and reduce stress experienced by the child and their family.<sup>18–20</sup> For example, communication and social skills can be enhanced by the use of visual prompts such as picture scripts. Timetables and specific social skills can be taught using social behaviour scripts, and for higher functioning children, role play, video modelling and social stories. Common elements of an effective early intervention program for children with an ASD are listed in *Table 3*.

## Behavioural therapy

There is some evidence that daily intensive behavioural therapy may have positive benefit, particularly with cognitive skills, but there is considerable variability in outcome and this intervention is not effective for some children with an ASD.<sup>21,22</sup> Sensory integration training, based on the theory that functional performance deficits are due to problems with processing sensory information, is widely promoted but does

**Table 2. Early developmental surveillance. Red flags for an ASD<sup>13</sup>**

- Does not babble or coo by 12 months of age
- Does not gesture (point, wave, grasp) by 12 months of age
- Does not say single words by 16 months of age
- Does not say two-word phrases on his or her own (rather than just repeating what someone says to him or her) by 24 months of age
- Has any loss of any language or social skill at any age

**Table 3. Common elements of effective early intervention programs for children with ASDs**

- An autism specific curriculum focusing on communication, attention to task, the development of social, play, self help and motor skills and the training and modification of behaviour
- Supportive and aid assisted environments that are structured and predictable and which help manage emotional and behavioural problems such as anxiety, rituals and resistance to change
- A comprehensive support plan for children in transition, eg. from preschool to primary school
- The inclusion of parents as collaborative partners in the planning and implementation of interventions
- Education and skills training for parents, access to parent support groups and the provision of respite care services and family support



not currently have sufficient evidence to support its use as a primary intervention method in ASDs. Specific sensory integration interventions such as the use of weighted vests and auditory integration training have been shown in empirical studies to be ineffective or even lead to deterioration in some children.<sup>23</sup>

## Diet therapy

There is no empirical evidence that diet or other mineral and/or vitamin supplements are effective treatment. If a child has a lactose intolerance or gluten enteropathy, treatment with an appropriate diet is likely to lead to some improvement in behaviour and relief of discomfort.

## Medication

There is no specific medication for the treatment of autism. Medication may have a role in the treatment of associated emotional and behavioural problems such as anxiety and depression. Anxiety is a common comorbid condition in individuals of all ages with an ASD. Depressive illness becomes more prevalent in adolescents with an ASD, perhaps in response to the development of insight into their difficulties, increased educational and social pressure and because of a potential increased genetic vulnerability in those with a family history of depression.<sup>24</sup>

Management of anxiety and depression includes altering the environment to reduce stress and anxiety, creating the experience of successful achievement at school, psychological treatments such as cognitive behavioural therapy modified to take account of the child's cognitive abilities, and the use of the selective serotonin reuptake inhibitor (SSRI) fluoxetine in some cases.

Risperidone in low doses has been shown to be effective in the treatment of disruptive, aggressive and self injurious behaviour in children with an ASD<sup>25</sup> but should only be initiated by a specialist paediatrician or child psychiatrist because of potentially serious side effects such as dystonic reactions, weight gain and risk of development of a metabolic disorder. Increased risk of epilepsy is reported, particularly early in childhood or again in early adolescence, and is associated with deterioration in the child's emotional and behavioural adjustment when poorly treated and uncontrolled.<sup>24</sup>

Approximately 20% of children with an ASD suffer from severe symptoms of inattention, impulsiveness and hyperactivity. Stimulant medication and other drugs used for the treatment of attention deficit hyperactivity disorder (ADHD) might be prescribed, but these are usually not as effective and are more likely to cause troublesome side effects than in the general population.<sup>26</sup> A sedative antihistamine or melatonin might help manage persistent problems with sleeping. Regular review of medication is necessary to respond to the development of any side effects and treatment response should be followed using a systematic behavioural record.

## Summary of important points

- Autism spectrum disorders affect approximately one in 160 Australians.
- Symptoms are apparent during the second year of life causing

impairments in social interaction, communication and behaviour with restricted and stereotyped interests.

- GPs can play a key surveillance role in determining which children might require further screening and referral for an ASD assessment.
- Red flags for autism are: does not babble or coo by 12 months of age; does not gesture by 12 months; does not say single words by 16 months; has any loss of any language or social skill at any age; and does not say two-word phrases on his or her own by 24 months of age.
- There is emerging evidence that a multimodal program of early intervention, including parent education, tailored to address the profile of symptoms and abilities of each child is more likely to promote development, improve behaviour and reduce stress experienced by the child and their family.
- Autism spectrum disorders are also associated with other mental health problems such as anxiety, depression and ADHD, which need to be the focus of targeted management.
- The GP has an ongoing role to support, educate and advise parents, other carers and the individual with an ASD.

## Resource

Factsheets on ASDs, including M-ChAT, the Developmental Behaviour Checklist, and early signs are available at [www.med.monash.edu.au/spppm/research/devpsych/actnow](http://www.med.monash.edu.au/spppm/research/devpsych/actnow).

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Conflict of interest: none declared.

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