

Autism: What you need to know

By Victoria Crosbie, MD, FRCPC

Few disorders are more confusing than autism. Stephen's behavior (see case at right) illustrates many of the symptoms seen within the autistic spectrum.

The stereotype of the withdrawn, mute child with self-absorbed, repetitive actions does not accurately reflect the spectrum of symptoms seen in clinical practice today.¹

Individuals with autism have one of a group of developmental disorders of brain function. These disorders have a broad range of behavioral consequences with varying severity. They are referred to as the Pervasive Developmental Disorders (PDD) in the fourth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV).²

Within the spectrum of PDD, there exists several more specific diagnoses (Table 1):

- Autistic disorder,
- Asperger's syndrome,
- Childhood disintegrative disorder,
- Rett syndrome, and
- Pervasive Developmental Disorder, Not Otherwise Specified (PDD, NOS).

The terms autism, autistic, and autistic spectrum disorder will be used to refer to the broad range of abnormalities characterizing this group of disorders.

Case



Stephen is an independent and self-sufficient three-year-old. He spends long periods of time involved in self-absorbed solitary play. When excited, he flaps his hands, speaks in gibberish, and paces incessantly. While he possesses an extensive vocabulary, it is rarely used for anything besides protesting and requesting. He quotes movie scripts verbatim, becoming upset if interrupted. Sensitivities to loud noises, crowds, and certain textures have been noted. Imaginary play skills are just emerging, as is a growing interest in other children.

Autism

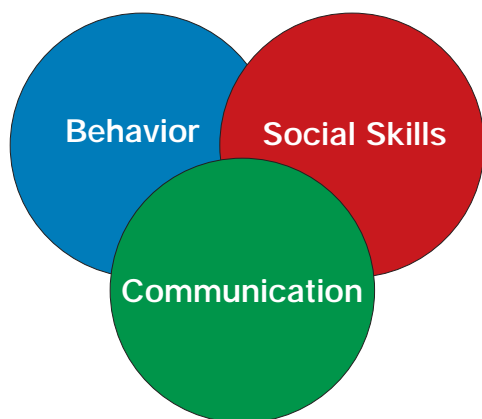
A diagnosis is usually suspected in toddlers, predominantly male, between 18 months and three years of age, when health-care providers have noted delayed language development, lack of typical social interest/curiosity, and/or a pattern of language and social regression.

Symptoms of Autism

The main symptoms of autism are deficits in social interaction, reciprocal verbal and non-verbal communication, and limited range of interests and activities. Symptoms are attributed to three main domains: behavior, social skills, and communication. (Figure 1)

Figure 1

Domains of Autism



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It is the degree, severity, and deviance from typical developmental patterns that leads one to suspect a possible autistic spectrum disorder.

Within each domain examined, many specific typical developmental milestones are sought. Communication deficits tend to fall into broad categories of phonology (speech sounds), prosody (rhythm and melody of speech), syntax (grammar and word order), semantics (vocabulary and meaning of language), and pragmatics (communicative and conversational use of language). Input from an experienced speech-language pathologist is critical.

Behavior refers to developmental patterns of play development, attention span, and activities of interest. An autistic child's play includes the manipulating, or lining up of toys without awareness of the toys intended use. Lack of pretend or imaginary play before the expected age of two years is typical. The observation of play is a sensitive method of detecting traits of autism.³ Several observation schedules rely heavily on the observation of play. The social skill deficits have a broad range of presentation. Aloofness, no response to the name being called, and lack of enjoyment in social interaction are all noted. Lack of non-verbal communication strategies (*i.e.*, pointing/gestures) are seen early in presentation.

Prevalence

Autism was once thought to be rare, with occurrences of four in 10,000 children.^{4,5} In 1998, a Nova Scotia survey of 20,800 children aged six to 14 years reported a prevalence of one autistic child per 1,000.⁶ More recent estimates have quoted figures as high as three to five cases of autism per 1,000 children. The Centres for Disease Control (CDC) are currently sponsoring a prevalence study in Brick, New Jersey.

It is this apparent growing prevalence of autism that has led many to talk of an autism epidemic. Whether the increase is actual, or due to a broadening of diagnostic criteria and heightened awareness is debated among experts. Regardless, diagnostic centres, intervention services, and education agencies are struggling to meet the demands placed upon them.

Prognosis

For individuals with autistic disorders, figures of 70% to 75% meeting the criteria for mental retardation have been widely published. While some older individuals are noted to have areas of great skill, in general, the outlook for independent adult living has been poor.

Many of the children diagnosed today are making rapid gains in all areas of function. Long-term outcomes of this generation of patient cohort need to be followed if accurate predictions of prognosis are to be made.

Causes of Autism

The underlying mechanism of autism has yet to be defined. Undoubtedly, an interplay of genetics and environment is at work. Many defined biologic causes exist, none of them unique to autism.

Prenatal factors include: intrauterine rubella, tuberous sclerosis, Cornelia de Lange syndrome, chromosomal abnormalities, as well as brain malformations, such as hydrocephalus.

A variety of medical conditions ranging from infantile spasms to herpes encephalitis have been associated with autism.⁶

A mere 10% to 30% of cases of autism are felt to be attributable to known factors.

Growing evidence highlights the importance of genetics. Studies of monozygotic and same sex dizygotic twins have shown concordance rates of 90%, and 5% to 10% respectively for autism spectrum diagnosis.⁷ Siblings of children with autism have an 8% to 10% recurrence risk.

Several candidate areas of chromosomes are being studied in the human genome. The short arms of chromosomes 1, 9, and 16 and long arms of chromosomes 2, 7, and 13 are some of the more promising areas.

Environmental factors have received much attention. The most widely discussed being the measles-mumps-rubella (MMR) vaccine after publication of a British study in 1998.⁸



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Table 1

Characteristics of Pervasive Developmental Disorders

Autistic disorder (classic autism)

- Presence of ≥ 6 of 12 potential deficits involving all three behavioral domains that define the autistic spectrum:
 - ≥ 2 deficits in sociability, empathy, and insight into other persons' feelings and agendas.
 - ≥ 1 deficit in communication language and imagination.
 - ≥ 1 deficit in behavioral and cognitive flexibility.
- Detectable before the age of three years.
- Diagnosis not excluded by the level of cognitive competence or the existence of other handicaps.

Asperger's disorder

- Troublesome social ineptness, lack of insight.
- Behavioral inflexibility with a narrow range of interests.
- No delay in the emergence of speech.

Pervasive developmental disorder, not otherwise specified

- Applies to less severely affected children who do not meet criteria for either autistic disorder or Asperger's disorder.

Disintegrative disorder

- Early development, entirely normal, including speech.
- Severe regression between the ages of two and 10 years, affecting language, sociability, cognition, and competence in skills of daily life.

Rett's syndrome

- Severe global regression in infant girls (rarely in boys), resulting in lifelong severe mental retardation, lack of language and purposeful hand use, and other neurologic deficits.
- Mutation on the long arm of the X chromosome.

Both American and British public health authorities have studied the possible causative role of MMR in detail. To date, conclusions have stated, "there is no conclusive evidence that any vaccine can cause autism."

Pathology studies have been undertaken to study the brains of individuals with autism.⁹ A paucity of Purkinje cells and granular cells in parts of the cerebellar cortex and smaller, more tightly packed cells in the amygdala and hippocampus, have been noted. These findings suggest prenatal dysgenesis. Details of current hypotheses in embryology are eloquently described in "The Early Origins of Autism" by Dr. Patricia Rodier.¹⁰

Although the exact cause of autism has not yet been found, it is clear that quality of parenting does not play a causative role.

Diagnosis

While diagnosis tends to fall to a variety of specialists and sub-specialists, symptom recognition occurs in the family physician's office.

An accurate developmental history, with focus upon patterns of behavior, communication, and social development typical of autism, is crucial.

When specific medical conditions (*i.e.*, infantile spasms, chromosomal abnormalities) are suspected, the appropriate investigations should be done. Little useful data is

provided by routine metabolic workup, chromosome analysis, electroencephalogram (EEG) studies, and diagnostic imaging procedures. Standard measures of cognition are often conducted and performance is rarely age-appropriate given the underlying language and behavioral disorder.

Ultimately, the clinician's high index of suspicion and attentiveness to the care providers' concerns are most important. Autism must be approached in the logical, stepwise manner like all other medical diagnoses. Only after the appropriate observations, investigations, and referrals have been obtained, can a diagnosis of autism be excluded and the family reassured.

Intervention

Currently, no drug or treatment has been shown to cure autism. The world of autism intervention contains an array of therapies and treatments, from both traditional and complementary medicine.

Drug therapies can be used to target troublesome behaviors that interfere with intervention programs and adaptive functioning. Hyperactivity, anxiety, and aggression have all been successfully "treated" with a variety of pharmaceuticals.

The most important intervention appears to be early, intensive education, targeting both behavioral and communication disorders. Education strategies employ techniques that are highly structured, individualized, and delivered in high teacher to student ratios.

This intervention is usually put in place for 20 hours to 40 hours per week. Understandably, cost is an issue. Several provinces now have provincially funded programs.



Autism

Occupational therapy, speech-language therapy and physical therapy should be used to address specific areas of need.

Many theories have received attention. Currently, there are several popular theories dealing with autism:

- Gluten/casein sensitivity;
- Fungal overgrowth;
- Mercury intoxicification;
- Specific vitamin/mineral deficiencies;
- Sub-clinical food allergies.

None of these theories/treatments have bodies of rigorous scientific data to support them. Supportive evidence is primarily anecdotal. Web sites devoted to autism give much space to their practical implementation however.

A cautious, yet open-minded, approach needs to be taken. Involvement of a skilled and knowledgeable dietician is important whenever elimination diets or vitamin/mineral supplementation is entertained. Given the current lack of definitive etiology, most families are willing to explore any avenue of potential benefit. An empathetic, informed care provider is invaluable to support families in these difficult decisions.

Knowledge of provincial programs and funding policies is critical. Links with tertiary referral services for diagnosis confirmation, and ongoing advocacy, are important for families and primary health-care providers.

Conclusion

What was once a rare disorder now rivals cystic fibrosis and juvenile diabetes in prevalence. Diagnostic centers, intervention services, and educational agencies are scrambling to meet the demands placed upon them.

As waiting lists grow, so does the stress and frustration of the families involved. The family

physician is in a unique position to support families through the physical and mental health, marital, and financial crises encountered while trying to love, nurture, and provide for a child with autism.

Collaborative relationships between primary-care providers, diagnostic centres, and intervention services need to evolve as the struggle to meet the needs of the ever-increasing numbers of children with autism continues. [CME](#)

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Suggested Readings

1. Rapin I: Current Concepts: Autism. *New Engl J Med* 1997; 337(2):97-103.
2. www.nichd.nih.gov/default/htm (search term: Autism).
3. <http://www.cdc.gov/nip/vacsafe/concerns/autism/>.