

The early detection of autism in clinical practice

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Autism is the prototypical form of a spectrum of related, complex neurodevelopmental disorders referred to as autistic spectrum disorders (ASDs; also known as the pervasive developmental disorders; PDDs). In addition to autistic disorder, the ASDs include Asperger syndrome, which is distinguished by relatively strong cognitive and language skills, and PDD-not otherwise specified, a less severe variant of autism (1,2). Like other complex neurodevelopmental disorders (eg, schizophrenia), ASD is thought to be the final common pathway of multiple etiological (largely genetic) and neuropathological mechanisms (3), thus, complicating the search for universal, autism-specific biological markers. In the absence of a biological marker, ASD is defined behaviourally, by impairments in three functional domains: socialization; communication; and a lack of behavioural flexibility, with reliance on routines (1,2).

Autism is among the most prevalent of the severe disorders of development. Health practitioners throughout Canada and elsewhere are currently experiencing a staggering increase in the numbers of children coming to attention. Prevalence is now estimated at one to two per 1000 for autism, and six to seven per 1000 for the entire spectrum of autistic disorders (4,5). While the issue of whether there is a real increase in ASD remains controversial (6,7), the incremental increase over time is generally attributed to the gradual broadening in how we define autism/ASD, and our increased awareness and detection of its diverse manifestations, particularly in the more cognitively or linguistically capable child (4). The fact remains that individuals with ASD are among the most difficult and costly to treat (8,9). The impact on families and their quality of life is enormous and sometimes devastating (10,11).

Although prognosis for individuals with autism has historically been poor (12,13), recent evidence points to improved outcomes with early intensive behavioural intervention (14-16). These interventions focus on the structured and systematic teaching of targeted communication and other functional skills, using empirically derived positive behavioural principles. Evidence of treatment effectiveness, together with the increasing availability throughout Canada of publicly funded early autism-specific intervention programs, serves to underscore the critical importance of concerted efforts aimed at the earlier detection of ASDs

(hereafter autism). At present, autism is typically not identified before four years of age, often more than two years after parents first seek medical attention (17). Initial contacts usually result in misplaced reassurance, followed by a series of professional consultations before a definitive diagnosis of autism is made. The long delay frustrates parents and postpones appropriate, autism-specific intervention.

In an attempt to optimize early access to best practices intervention, a panel of cross-disciplinary experts and parent representatives have recently issued practice guidelines for early community-based detection of autism (18). These guidelines have been adopted by the American Academies of Pediatrics (19), Neurology (18) and Child and Adolescent Psychiatry (20), and after minor revisions, endorsed by several jurisdictions in Canada and the United States. Recommendations are that practitioners adopt a two-stage early detection strategy: monitoring for signs of autism within the context of routine developmental surveillance; and in cases of concern, followed by the use of an early autism screening instrument to identify children at highest risk. Because there does not yet exist an ideal screening instrument for community use, practice guidelines are aimed at identifying infants/toddlers at high risk for autism, who can then be targeted for more comprehensive developmental assessment (18).

Our purpose in this paper is to elaborate on this two-stage strategy, with a particular focus on its implementation. In this context, we outline what is currently known about the early signs of autism and available screening instruments, and address concerns regarding false positive and equivocal cases. We end by commenting on the thoughtful essays of parents that accompany this paper.

CLINICAL MONITORING FOR EARLY SIGNS OF AUTISM

As a first step toward earlier detection, guidelines call for the monitoring of early signs of autism during regular health visits as an adjunct to ongoing surveillance or monitoring of development. Evidence on the early signs of autism derives largely from parents' retrospective reports (21,22) and home videos (23,24) of children later diagnosed with autism, and is thus limited by its retrospective and restricted nature (25). However, remarkable consistency exists between these

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TABLE 1
Early signs of autism

Atypical social-emotional engagement/connectedness and play, notably a lack of:

- Social smiling (specifically in response to the smiles of others)
- Shared affect/interest in social games such as peek-a-boo (in absence of physical contact such as tickling)
- Interest in other children (eg, ignores/avoids or does not imitate their play)
- Orienting to own name being called
- Interest in toys, or plays with them in unusual ways (eg, lining up, spinning or opening/closing a part rather than using toy as a whole)
- Pretend play (eg, feeding a stuffed animal or having a tea party)

Delays/abnormalities in language and non-verbal communication, notably a lack of:

- Following others eye gaze to a nearby object/person
- Shared/joint attention (eg, eye gaze with another person and an object/event of interest)
- Communicative gestures such as using eye gaze to request something, pointing at a distant object of interest (eg, an airplane), or waving good-bye
- Social/reciprocal babbling, as in conversing with others
- Meaningful language (eg, delayed, odd first words or unusually repetitive)

Regression/loss of early words and/or social-emotional engagement/connectedness

Other atypical behaviours

- Lack of active visual exploration of environment
- Tends to stare or visually fixate on certain objects/patterns (eg, lights)
- Tends to be under- and/or over-reactive to sounds or other forms of sensory stimulation (eg, seeks out certain stimuli in an unusual way, or covers ears to loud sounds)

sources and emerging prospective data from our longitudinal study of high-risk infants (all with a sibling with autism), some of whom have developed autism (25). Thus, as part of routine developmental surveillance, primary care physicians are advised to assess for the early social-communicative, play and behavioural signs of autism outlined in Table 1. These signs can be detected by questioning parents and through direct observation in the office. For example, it is possible to check for whether:

- the infant/toddler smiles in response to your smiles and is able to follow your gaze or point to a picture on the wall;
- he/she engages in active back-and-forth looking that includes looking at you (versus blank or random looking and/or staring at particular objects);
- when you step outside the child's direct view, he/she will orient to his/her name called.

Taking time to play peek-a-boo with a blanket, to pretend to feed a stuffed animal or to see whether the child engages with you as you blow bubbles may give more confidence in having had sufficient interaction to judge the typicality of the child's social communication. Also, ask the

mother whether she has any concerns about her child's social or communicative development: whether, for example, the child looks at her when nursing/feeding from a bottle, and whether the child engages in interactive play. Special care is required with first time mothers who may have no easy reference point for typical behaviour.

Signs of autism should be monitored from at least 12 months of age, as some cases can be detected by 18 months and signs may be evident even earlier. Special attention should be paid to development between 18 and 24 months of age, a period during which many (but not all) cases of autism are detectable. There is also a small subgroup in whom initial signs of autism may not be apparent until after the second year, thus emphasizing the importance of ongoing surveillance rather than screening at a single point in time. We emphasize that *careful consideration should always be given to parent's concerns*, which are typically legitimate. With the possible exception of regression (which parents should be questioned about), no single sign is diagnostic, nor does the presence/absence of any one or even more of the signs outlined in Table 1 preclude the possibility of autism. Rather, the focus should be on the profile or pattern of behaviours, and on their relative frequency compared to what is age or developmentally typical. Even in children with a general developmental delay, most of the social-communicative behaviours outlined in Table 1 would be expected to be present by a developmental age of 12 months. For example, a profile of concern is one in which an 18-month-old, with or without language, smiles but not reciprocally, has fleeting eye contact and rarely uses eye gaze to communicate, points to nearby but not distant objects and usually orients to name only after being called repeatedly.

USE OF EARLY AUTISM SCREENING INSTRUMENTS

As outlined above, once a child has been observed to have 'autistic' signs (Stage 1), recommendations are that a screening instrument be used to identify those at highest risk (Stage 2) (18). However, it bears emphasizing that existing early autism screening instruments preclude any simple decision rule (25). All are at various stages of testing, and none has been validated for community use. While efforts aimed at the earlier detection of autism cannot await an ideal screen, we recommend that existing screening instruments be used only to assist in the assessment of risk, not as a basis for decision making; ultimately, the decision is a clinical one. One such instrument, the Modified Checklist for Autism in Toddlers (26), is designed for use at two years of age, and is completed by parents, thus making it feasible for clinical practice. Screening at two years has been shown to identify many children with autism/ASD. However, strong empirical grounds exist for assuming that significant numbers are still missed (27), thus emphasizing the importance of ongoing surveillance or monitoring of screen-negative children. Of the false-positive cases identified at two-year follow-up, all but a negligible few had some other (non-autistic) developmental problem requiring attention (28).

The main goal is a timely referral of children at high risk for autism to specialized diagnostic or treatment services, being cognizant of the fact that long waiting lists exist for these services in most regions across the country. Rather than postponing treatment until a final diagnosis is made, we recommend immediate referral to generic early intervention/developmental/speech and language services, as well as ongoing clinical monitoring for autism. Our experience, as well as that of others, is that virtually all children initially suspected of having ASD have some form of developmental delay, whether autism/ASD or not, and that they should have the opportunity as soon as possible to benefit from whatever intervention services are available in their community. Ultimately, efforts aimed at the earlier detection and treatment of children with autism and related developmental disorders serve a critically important purpose: to optimize the developmental trajectories and long-term outcomes of the many children affected, and in so doing, improve the quality of life of all family members.

Finally, early detection, diagnosis and treatment of autism provide the strong foundation sought by virtually every parent we have met. But the reality is that this

foundation forms only the beginning of a long process of teaching the many skills that all children need. The accompanying articles, written by parents, are powerful testimonies to this fact. Barbara Oland reminds us that we must never deny parents hope, for as she so beautifully expresses, every child can learn and grow, and deserves the opportunity to be and feel valued for who they are and what they have achieved. Penny Gill, whose child is older, is dedicated to the same principles; she eloquently and unabashedly argues that to achieve these goals teachers need to be trained in positive, evidence-based methods of teaching children with autism. We are grateful for their rich contributions to this series.

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