The Diagnostic Process in British Columbia

One of the most stressful periods for parents of children with Autism Spectrum Disorder (ASD) is waiting for an assessment to establish whether a diagnosis can be made. Many wonder whether they are imagining that their child’s development is unusual, which can bring up feelings of confusion and worry as well as hope. For many families, the diagnosis of ASD comes as a relief, as well as a shock, because at last they have something concrete that they can begin to understand, in order to move forward and help their child.

While great strides have been made in British Columbia over the last decade in the area of improving the process of autism diagnosis, and providing accessible diagnosis across the province, it is still a complex process. To help families and community professionals better understand the process and the need for speedy referrals, Dr. Vikram Dua, former Medical Director of the British Columbia Autism Assessment Network and Complex Developmental Behavioural Conditions, has provided this description of the diagnostic process.

Dr. Dua is a child and adolescent psychiatrist who has assessed and treated hundreds of children and youth with ASD over the past decade. He served as the primary researcher and writer for the Ministry of Health policy paper, “Standards and Guidelines for the Diagnosis and Assessment of Young Children with Autism Spectrum Disorder.” He completed his medical school training at McMaster University in Hamilton and then went on to do a residency at Harvard Medical School. Vikram is an assistant clinical professor at the University of British Columbia and is a co-investigator at the B.C. site of the national longitudinal study of young children with ASD (“ASD Pathways”). He is a member of Advisory Committee of the Mental Health Commission of Canada and practices with the Neuropsychiatry Clinic of BC Children’s Hospital.
RED FLAGS FOR POSSIBLE AUTISM SPECTRUM DISORDER

- Delay or absence of spoken language.
- Looks through people; not aware of others.
- Not responsive to other people's facial expressions/feelings.
- Lack of pretend play; little or no imagination.
- Does not show typical interest in peers, or play near peers purposefully.
- Lack of turn taking.
- Unable to share pleasure.
- Qualitative impairment in nonverbal communication.
- Not pointing at an object to direct another person to look at it.
- Lack of gaze monitoring.
- Lack of initiation of activity or social play.
- Unusual or repetitive hand and finger mannerisms.
- Unusual reactions, or lack of reaction, to sensory stimuli.

Please note that it is not necessary for a child to demonstrate all of these “red flags” in order to have Autism Spectrum Disorder.

WHAT ARE AUTISM SPECTRUM DISORDERS?

Autism Spectrum Disorders (ASD) includes several related childhood-onset brain disorders. They are developmental-behavioral conditions that emerge early in life, generally by the time a child is 2–3 years old.

Children impacted by ASD have:

• Difficulties with social interaction both with family members and peers.
• Atypical communication (odd speech patterns, limited non-verbal communication, and challenges with conversation).
• Impaired imaginative play, and a pattern of interests and behaviors that is highly restricted, repetitive, compulsive, or unusual.

ASD includes all of the following sub-types:

• Autistic Disorder.
• PDD-NOS/Atypical Autism.
• Asperger Disorder/Syndrome.
• Rett Syndrome.
• Childhood Disintegrative Disorder.

Research has shown that although identification within the broad category of ASD can be done with great confidence, identification of sub-types within ASD is not reliable.

The term Autism Spectrum Disorder, as used in B.C. currently, is equivalent to the category of Pervasive Development Disorder (PDD) in both the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV-TR) of the American Psychiatric Association (1994) and the International Classification of Diseases (ICD-10) of the World Health Organization (1992).

Why is an accurate diagnosis of an ASD important?

A major purpose of any diagnosis is to inform treatment and intervention.

In autism there is a growing literature on interventions, but in order to apply this research to an individual child, one has to ensure that the child is similar to those in the studies. If the child is different than those described in the studies (for example, does not have autism), one cannot assume the intervention will be successful. Similarly, if a child does not have an ASD, but rather a different developmental or psychiatric condition, alternative treatments may be more effective.

There are many other reasons why an accurate diagnosis is important. Parents and caregivers benefit from an understanding of why a child has certain chal-
lenges. They want to know what caused the problem in their child. They also want to know what the future holds for the child and family. Knowing that the child has an ASD enhances understanding, alleviates potential guilt, and allows more informed planning for the future.

Finally, the diagnosis of an ASD does establish eligibility for specific services and interventions designed for these children and youth. In B.C. a diagnosis of ASD establishes eligibility for a variety of government-funded services from early intervention to supports within the education system.

**Why is earliest identification of ASD important?**

Research over the last few decades has indicated that although ASDs are life-long conditions, the outcome in an individual child can be improved by treatment. There are also strong suggestions in the research that the earlier one begins such interventions, the better the long-term outcome.

As a result there has been a concerted focus on refining the processes and procedures to identify children with an ASD at the earliest time possible. Although this will undoubtedly vary depending on the particular child, some standard approaches do appear to reduce the delay between first emergence of symptoms, and formal diagnosis (and thus treatment) of challenges.

**How is ASD diagnosed?**

No two children develop alike. Identifying which young child has an ASD as opposed to a different condition is one of the greatest challenges in the field. And of course there are many unique children who do not have a developmental problem despite their unusual behaviors. Researchers and clinicians have long endeavoured to develop an accurate and reliable means of diagnosing children with ASD.

Unlike other areas of medicine, there is no blood test, brain imaging study, or other investigation that can conclusively diagnose or exclude ASD, particularly in very young children.

Fortunately, the last few decades of research has revealed a number of consistent features of ASD across children that has allowed us to be more precise in establishing whether a child is likely to have an ASD. At the same time, any diagnostic process that relies on observing a child’s behaviors has built-in risk for error. Conscientious diagnosticians will make this limitation known to parents.

The following sections provide a brief summary of the science, art, and “red tape” of autism identification and diagnosis.
THE SIGNIFICANCE OF PARENTAL CONCERNS

Parental concerns about their preschool-aged child’s communication and/or social behaviors should always be taken seriously. Many parents of children subsequently diagnosed with ASD experience first concerns about their child’s development by about 18 months of age. Parental concern about communication and social behavior in very young children is quite accurate in identifying a significant developmental challenge, whether the problem is ASD or another disorder.

Given the current awareness of ASD, however, parents of young children more frequently have concerns about unusual aspects of their child’s development. Keep in mind that no two children develop according to the same pattern. Isolated unusual features in development or behavior are not usually a red flag for ASD. Since not all unusual features represent a disorder, not all problems require professional assessment.

Parents of children with an older sibling with ASD (or other developmental or psychiatric condition) should be more vigilant about developmental problems, given the relatively high recurrence of such disorders within families. For more information on the possible role of genetics and information on genetic counselling, see Chapter 3 of this manual, “Medical Issues in Autism Treatment.”
What Should a Concerned Parent Do?

There are a host of “screens” and “checklists” available on the web and elsewhere that suggest “risks” for ASD. Parents of very young children sometimes consult such tools for reassurance, or to confirm their concerns. Unfortunately, there are no currently available screening tools that have proven accurate and reliable in predicting the presence of an ASD.

Parents of pre-schoolers may review the “Red Flags for Possible Autism Spectrum Disorder” list, on page 2 of this chapter. Other helpful insights can be found in the Autism Speaks Video Glossary available at www.autismspeaks.org/video/glossary.php.

For school-age children and youth available screening tools are even less accurate in predicting the presence of an ASD. If a parent does use a tool to review their concerns, it is critical to remember that these are only “screens” identifying children at higher risk for ASD, they are not diagnostic tools.

The desire to rule out the presence of an ASD in a young child is certainly understandable, given the emphasis on the need for the earliest intervention. However, if the child does not have a developmental disability, the impact on a family and child of going through a rigorous diagnostic process that may take many months must not be underestimated.

Whether or not a parent uses a screening tool to review their child’s behaviors, if the concerns persist, the most important action is to consult a professional. For all children and youth this would include the family doctor. In addition, for preschool children, families may consult the community public health nurse, speech and language pathology services, Infant Development Program (IDP), or Child Development Centre (CDC) programs.

It is an unfortunate reality that sometimes parental concerns about their child’s development are minimized or ignored, resulting in a delay in identification of ASD or other conditions effecting the child. If a parent has consulted professionals as suggested above and remains concerned about their child’s development, but they have not been referred for further investigation, then it is appropriate to seek another professional opinion. If a parent feels their concerns have not been adequately addressed by the primary care professional, it is reasonable to request referral to a specialist for another opinion.

“If a parent feels their concerns have not been adequately addressed by the primary care professional, it is reasonable to request referral to a specialist for another opinion.”
WHAT IS THE ROLE OF PRIMARY CARE PROFESSIONALS IN THE EARLY DETECTION OF ASD?

Although parental concerns about their child’s communication or social behaviors are often predictive of a developmental problem, the absence of parental concern does not ensure normal development. As a result it is important for all primary care professionals (family doctors, public health nurses, etc.) to be vigilant for higher risk indicators of an ASD.

Earliest possible identification of ASD requires an ongoing process of general developmental surveillance of all children with specific focus on social-communication deficits. General developmental surveillance for ASD is conducted by all primary care practitioners. It can include any or all of the following components:

- Serious consideration of all parental concerns about communication, development and behavior.
- Particular attention to developmental milestones related to communication and reciprocal social interaction, two areas central to screening for ASD. See Appendix 1 of the Standards & Guidelines.
- Ongoing monitoring for the presence of “red flags” for ASD at each contact with the child and parents, including scheduled “well-child” visits.
- Administration of general developmental screening instruments.

“Parents need to go with their gut. If you feel something is ‘off’ with your child, do not just accept the first opinion you get. Some of these professionals only see your child for an hour or two. That is often not long enough to make a full evaluation of some of the nuances of ASDs. Getting a second opinion is a parent’s right, and will only benefit the child in the end. Don’t worry about ‘offending’ the professional. Your child is what is most important, and getting an accurate diagnosis and getting your child what they need is the most important thing.”

Angela Henderson
“Screening” implies a brief assessment procedure designed to identify children who are at higher risk for an ASD, and should be referred for a conclusive assessment. In the past few years there has been both much enthusiasm, and disappointment, with the promise of a specific ASD “screening” tool.

At the present time, specific ASD screening tools designed for the primary care setting, like a doctor’s office, cannot be relied on to predict the presence or absence of an ASD. However, they should be seen as a valuable means of gathering information. The lack of reliability of the screening tools now available means that screening all children for ASD is not supported. There is insufficient evidence to recommend any single test/procedure to screen for ASD.

There may be some value in primary care practitioners utilizing available screening tests for ASD with children known to be at higher risk. For example, the younger siblings of children with ASD have a recurrence risk approximately 50 times the population average).

“Failing” or “passing” on any single screening measure should not be the sole determinant of whether or not a child is referred for further assessment.

<table>
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<tr>
<th>ALTERNATIVE TO FORMAL SCREENING</th>
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<tr>
<td>Does your child …</td>
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<tr>
<td>• Not speak as well as his or her peers?</td>
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<tr>
<td>• Have poor eye contact?</td>
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<tr>
<td>• Not respond selectively to his or her name?</td>
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<tr>
<td>• Act as if he or she is in his or her own world?</td>
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<tr>
<td>• Seem to “tune others out”?</td>
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<td>• Not have a social smile that can be elicited reciprocally?</td>
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<td>• Seem unable to tell you what he or she wants, thus preferring to lead you by the hand or get desired objects on his or her own, even at risk of danger?</td>
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<tr>
<td>• Have difficulty following simple commands?</td>
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<tr>
<td>• Not bring things to you to simply “show” you?</td>
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<tr>
<td>• Not point to interesting objects to direct your attention to objects or events of interest?</td>
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<tr>
<td>• Have unusually long and severe temper tantrums?</td>
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<td>• Have repetitive, odd, or stereotypic behaviors?</td>
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<tr>
<td>• Show an unusual attachment to inanimate objects, especially hard ones (e.g., a flashlight or a chain vs. a teddy bear or a blanket)?</td>
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<tr>
<td>• Prefer to play alone?</td>
</tr>
<tr>
<td>• Demonstrate an inability to play with toys in the typical way?</td>
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<tr>
<td>• Not engage in pretend play (if older than two years)?</td>
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Appendix 3 Standards & Guidelines

What should a primary care professional do after a child/youth is identified to be at higher risk for ASD?

When a child/youth of any age is identified as being at higher risk for an ASD, referral for further specialized ASD evaluation through the British Columbia Autism Assessment Network (BCAAN) must be considered.

**BCAAN accepts referrals from all community physicians, and does not require a specialist (such as a pediatrician or psychiatrist) to assess the child/youth prior to accepting the referral.**

For children/youth where an ASD is suspected, in addition to initiating referral to BCAAN, the primary care professional should concurrently arrange/facilitate the following:

- Confirmation of hearing status through public health services.
- Depending on the age of the child/youth, referral to appropriate community-based services (e.g., the Infant Development Program (IDP), developmental preschools, Child Development Centres, school special education services, child and youth mental health).
- Every child with a language delay/disorder should have a community speech-language evaluation (through public health if preschool-aged, and through the school district if school-aged).
- Every school-aged child with academic challenges should be assessed for intellectual abilities and/or learning disabilities by the school district psychologist.
- Every child with a developmental delay and pica should have lead screening. (Pica is the persistent eating of non-food items, which puts children at increased risk to exposure to environmental toxins such as lead.)

**What is the BCAAN Process for Assessing a Child or Youth with ASD?**

The topic of diagnostic assessment of children and youth for possible ASD (and for that matter all developmental and behavioral conditions) is a complicated, technical, and debated subject. Most of this is not relevant to the typical parent or caregiver, other than to understand that B.C. has adopted perhaps the most evidence-based and comprehensive approach to assessing and diagnosing ASD of anywhere in the world. BCAAN endeavours to ensure that all children and youth in B.C. obtain equitable assessment, and one that represents the state-of-the-art in the field.
The core of a BCAAN ASD assessment is termed the Clinical Diagnostic Assessment (CDA). The CDA is conducted by a professional called the Qualified Specialist (QS)—who can be a psychiatrist, pediatrician or registered doctoral-level psychologist (in some situations a component of the assessment may be completed by another clinician, but results are ultimately integrated by the QS).

Amongst other things, the CDA includes a detailed history, clinical estimate of developmental status, and depending on the particular child may include a pediatric and/or psychiatric assessment.

The cornerstones of the CDA are the use of standardized ASD diagnostic tools—the Autism Diagnostic Interview – ADI-R (which solicits detailed developmental history), and the Autism Diagnostic Observation Schedule—ADOS (which provides a standardized direct assessment of the child/youth). It is important to understand that although the tools are enormously informative and provide various scores, there is no specific test or instrument that either confirms or excludes ASD as a diagnosis.

Ultimately the QS integrates the data from the ADI-R and ADOS, together with information from other reports and testing to arrive at a final diagnosis based on clinical judgment and criteria in the DSM or ICD.

Multidisciplinary Assessments

Many children and youth referred to BCAAN will receive additional assessments beyond the CDA. These assessments can have a variety of functions, including assisting with the differential diagnosis of ASD, identifying potential “causes” of the ASD, as well as accurately describing co-existing functional impairments (in intellectual development, communication, motor or sensory function, or mental health). Assessment by different disciplines provides for a comprehensive picture of the individual child’s functional skills.

In addition to the CDA most children who are under the age of 6 years will have an assessment of their intellectual development (by a psychologist), communication (by a speech and language pathologist), and medical examination (by a pediatrician).

In a number of situations some of these have already been completed in the community prior to the BCAAN assessment and do not need to be repeated.

Depending on the child’s profile some children may have assessment of their motor or sensory function (by an occupational therapist and/or physiotherapist) or mental health (by a child psychiatrist).
Children and youth over 6 years of age may not require multidisciplinary assessments beyond the CDA in order to establish whether or not an ASD is present. In general, it is the expectation that multidisciplinary assessments, if indicated to provide a comprehensive developmental formulation, will be done in the community (for example through the school districts or mental health teams) either before or after the BCAAN assessment.

Roles and Functions in the BCAAN Multidisciplinary Team

- **Case Manager:** This role is filled by different professionals, including nurses and social workers. The primary role of the Case Manager is to gather necessary information prior to the BCAAN assessment, to support the family before, during and after the assessment, and to ensure the child/family makes appropriate connections to services agencies after completion of the assessment.

- **Qualified Specialist:** Can be a psychiatrist, pediatrician or registered psychologist, and is ultimately responsible for arriving at a diagnostic conclusion. The Qualified Specialist completes the Clinical Diagnostic Assessment and integrates the results from other reports and assessments.

- **Pediatrician:** When not functioning as a Qualified Specialist, a pediatrician may examine a child for possible associated medical problems or causes of the child’s condition. In general this involves taking a history with the parents/caregivers and detailed physical examination of the child for evidence of atypical physical features. In some cases the pediatrician may suggest investigations such as blood tests or other studies.

“A whole year after our initial concerns began, we got the diagnosis. This is ridiculous. I can’t help but think how much better my son may have been and how much extra help he could have received. Everyone says ‘early intervention is key.’ Well, we tried to get it for him and no one would listen. I think a major problem is that people, including physicians, are not aware of the Autism Spectrum. People think autism, and they think of the worst-case scenario. They don’t realize that people with autism can make eye contact and speak and do all sorts of things.”

Laura Costas
• **Psychologist:** When not functioning as a Qualified Specialist, a registered doctoral-level psychologist assesses the intellectual, adaptive, and learning profile of a child/youth. This involves using a number of standardized tests with the child/youth, as well as interviewing the parents about the child’s functioning. This assessment establishes whether there is an associated intellectual disability or learning disorder. The psychology report also provides guidance with regards to education and learning.

• **Psychiatrist:** When not functioning as a Qualified Specialist, the psychiatrist evaluates for the presence of psychiatric conditions which can frequently both mimic and accompany ASD. This involves obtaining a history from the caregivers, use of questionnaires and checklists, and mental status examination of the child/youth. This assessment establishes whether there is an associated psychiatric disorder, and provides recommendations for treatment.

• **Speech Language Pathologist (SLP):** The SLP assesses the child/youth for communication difficulties. Using a number of standardized tests, as well as examination of oral structures, this assessment establishes whether the child/youth has a language and/or speech disorder. The SLP report provides recommendations for intervention of communication problems.

• **Occupational Therapist (OT)/Physiotherapist (PT):** The OT assesses fine motor skills and sensory function. The physiotherapist assesses gross motor function. The results for these assessments establish whether the child/youth has a motor coordination or sensory integration disorder.

**What if there is Disagreement with the BCAAN Diagnostic Conclusions?**

Although the procedures and tools used by BCAAN professionals represent the state-of-the-art, as with other areas of medicine, on occasion team members, families, or community providers may not all agree on whether or not the child/youth has an ASD. In order to promote the most accurate diagnostic formulations and to meaningfully address questions that can arise from the community or family, BCAAN has established standard processes for regional BCAAN teams to request a second opinion assessment. Each case is considered individually, and although not all children are re-assessed, the primary objective is to provide families with the best information to guide supporting their child’s development.
A CONSISTENT DEFINITION OF AUTISM SPECTRUM DISORDER ACROSS ALL GOVERNMENT MINISTRIES

In British Columbia, the Ministries of Children and Family Development, Health, and Education have all adopted a consistent definition of Autism Spectrum Disorder as defined by the Standards & Guidelines. This means that a BCAAN diagnosis of Autism Spectrum Disorder (ASD) is accepted by the Ministry of Education as entitling a student to services.

The British Columbia Autism Assessment Network

In 2002, the B.C. Autism Assessment Network (BCAAN) was established by the Ministry of Health. BCAAN’s mandate is to assess children and youth under 19 years of age who are referred with a query of ASD. It is the only program in Canada, and perhaps North America that consistently provides state-of-the-art, comprehensive assessments for all children and youth within a large jurisdiction.

BCAAN provides approximately 1000 assessments annually, dispersed across the five health regions of the province. Each health region is responsible for providing diagnostic and assessment services to children and youth in their region. The Provincial Autism Resource Centre (PARC) based at Sunny Hill Health Centre for Children currently serves as the regional team for Vancouver Coastal Health Authority, as well as providing assessment for complicated cases from across B.C. Roughly 50% of the children referred for diagnosis are found to have an Autism Spectrum Disorder.

Standards and Guidelines for the Assessment and Diagnosis of Young Children with ASD in British Columbia

In 2002, the B.C. government brought together a group of B.C. professionals with expertise in autism. They were given the job of developing a set of standardized procedures for early identification, diagnosis and assessment of young children with ASD. The result was the Standards and Guidelines for the Assessment and Diagnosis of Young Children with Autism Spectrum Disorder in British Columbia (S&G), which established the essential content and process of diagnostic assessment. The complete guidelines can be found at www.healthservices.gov.bc.ca/cpa/publications/asd_standards_0318.pdf.

Regardless of where in B.C. a child is diagnosed, all BCAAN assessments must meet the Standards & Guidelines for the Assessment and Diagnosis of Young Children with Autism Spectrum Disorder in British Columbia.

In 2006, BCAAN adopted policy revisions to the S&G that included extension of the methods and procedures to children over 6 years of age, refinement of assessment procedures, and formalization of training and competency requirements of BCAAN diagnosticians.
Suggested Resources for Parents

There are many books and websites that have useful insights on the diagnostic process for parents and for the professionals who support them.

Books


Websites

- The British Columbia Autism Assessment Network website can be found at [www.phsa.ca/HealthPro/Autism/default.htm](http://www.phsa.ca/HealthPro/Autism/default.htm).

- The Autism Speaks Video Glossary. For a remarkable resource made available in 2008 see the new ASD Video Glossary available on the Autism Speaks website at [www.autismspeaks.org/video/glossary.php](http://www.autismspeaks.org/video/glossary.php). Using over a hundred videos of children with ASD, this web-based tool allows parents and professionals to gain a better understanding of the subtle differences between children who are typically developing as compared to those who are demonstrating the “red flags” associated with ASD.

- For the answers to frequently asked questions about Autism, see [www.actcommunity.net/AI/FAQ.htm](http://www.actcommunity.net/AI/FAQ.htm).

ADVOCATING FOR A TIMELY DIAGNOSIS — A MESSAGE FROM ACT

Considering a Private Diagnosis?

Some parents are taking the route of paying for a private diagnosis because they are concerned about waiting for a BCAAN diagnosis. At ACT we advise parents that a BCAAN diagnosis is preferable, but we understand parental frustrations with waiting for a diagnosis. A private diagnosis is an option but take precautions, as assessments by a speech pathologist and a psychologist can cost between $2,000 and $3,000 in total. Below are some points to consider.

Make your decision based on full information.

First check on the BCAAN waiting lists in your region—don’t depend on second hand or out-of-date information! Ask the BCAAN Regional Referral Coordinator how long it will be for an appointment. Then ask the private diagnostician when you can expect to have all the components of a diagnosis from them, including the Speech & Language and Psychology Assessments. A list of BCAAN Regional Referral Coordinators is available at www.phsa.ca/AgenciesServices/Services/Autism/autism-assessment-programs.htm or you may call ACT – Autism Community Training for more information. ACT does have the names of private diagnosticians to share with families, but we cannot guarantee that their work will be accepted by the Ministry of Children and Family Development.

The Standards & Guidelines apply to private diagnosticians, too.

A number of parents have complained to ACT that the diagnosis that they have received privately has not been accepted by government agencies. Make sure that your diagnostician has received the necessary training in these diagnostic tools before you begin the diagnostic process.

Don’t make the assumption that private is better.

BCAAN has quality assurance processes which private practitioners are not part of—although some private practitioners also work part-time for BCAAN.

Advocate for a Timely Diagnosis for all B.C. Children

The BCAAN model is the envy of many jurisdictions across North America, but many families feel there is a need to advocate together to make timely diagnosis available for all children with suspected ASD in B.C. Currently BCAAN has an annual limit of 1000 children for whom they are funded to provide a diagnosis. However, as the number of children being referred for diagnosis continues to climb, so do the waiting times for diagnosis when the funding has been exhausted. Parents who are waiting longer than 3-6 months for a diagnosis may wish to consider contacting the ACT office for more information on raising their concerns about delays.

Ensuring that the referral for your child has been received by BCAAN

There have been instances when a referral from a family doctor or pediatrician has not been received by BCAAN. It is always a good idea to check with the Regional Referral Coordinator that your doctor’s referral has been received and that an appointment for your child is in the works. We recommend that you make note of the date of the person you spoke with and their name.