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Autism Spectrum Disorder (ASD) in Children and Youth: Information for Primary Care



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Sommaire : Autism spectrum disorder (ASD) is characterized by deficits in social functioning, restricted activities and interests, typically resulting in occupational impairment. People with ASD have brains that are wired in such a way, that they have problems seeing things from other's perspective (i.e. problems with empathy and theory of mind) which can lead to severe problems interacting with other people, leading to problems at school, work and relationships. On the other hand, individuals with high functioning ASD may possess unique strengths in certain subjects, sports, or skills. Previously, this category of people received a diagnosis of “Asperger’s” or “Pervasive Developmental Disorder” (PDD NOS). Primary care providers can play a key role with early identification and connecting families to appropriate supports and interventions.

Case, Part 1

- Thomas is a 10-year-old boy, brought to see you by his parents because of complaints of bullying and stomach pains
- Since the school year started, he has been complaining of stomach pains on mornings where he has to attend school, which resolve by the weekend
- You rule out any acute medical conditions, and wonder about the bullying being a stress
- Parents report that this year, with the transition to a new school, there is a new group of peers who have been teasing and bullying him because they can see he is different
- Mother states that she has spoken to the teachers, but without success
- On the positive side, he attends school, and does well in math and science, and is strong at anything technical – he spends hours on end watching videos on the internet about engines and can tell you anything about any sort of engine
- Mother states, “He’s just different and they tease him because of that. Can you speak to the school about it? Surely there must be something you can do?”

Epidemiology

- Prevalence ~ 1 % (1/88)
- Gender: M:F – 5:1
- Average age of onset – 3.8 – 8.2 years

Etiology

- It is helpful to be familiar with some of the theories on ASD, as parents may ask "Why does my child have ASD?"
    - Baron-Cohen's Systematizing Theory
        - It has been noted anecdotally, that certain professions such as engineering and computer sciences have a disproportionately high percentage of individuals with ASD (Baron-Cohen, 2009)
        - As modern societies become increasingly dependent on technology, it has been theorized that having ASD traits are adaptive and helpful, which is one theory which might possibly explain rising rates of autism spectrum disorders (CDC, 2012).
    - Vaccine hypothesis
        - There are still some parents who may worry that vaccines cause autism, but the good news is 1) research strongly shows vaccines do not cause autism and 2) earlier evidence suggesting vaccines cause autism have in fact been shown as being fraudulent

Red Flags for Autism Spectrum Disorder (ASD)

- The following red flags suggest autism spectrum disorder in general:
    - Problems communicating verbally and/or non-verbally
    - Problems relating to others and the world around them
    - Problems with thinking and behaving flexibly

- Suspect ASD in an infant/toddler where:
    - There is a unresponsive, or lack of interest in interacting with people
    - The child focuses intently objects (or parts of people) for excessive periods of time

- Suspect ASD in a child/youth where:
    - Child has problems getting along or interacting with other people
        - Note that a high functioning ASD child may express a desire to interact with others, but interactions tend to be one-sided, and the child has troubles with two way interactions, i.e. the child will tell the other person things, but will not ask the other person about their perspective or opinion
    - Child has troubles with empathy, interpreting what others are thinking or feeling because they can’t understand social cues, such as tone of voice or facial expressions
    - Preoccupation with routines such that the individual has the same routine and same schedule every day and doesn’t like changes, such as trying new foods, or natural changes that happen with changes in teachers at school, transitions between the school year to summer holidays, and the starting of the school year, etc.

- Suspect (high functioning) ASD in an adult (or parent of your patient) where:
    - There are troubles with reading social cues and seeing things from other’s perspectives, which makes it challenging for the adult to form close, intimate relationships with others
    - There may be friendships and relationships, which are primarily based on having things in common
    - The adult works in primarily a technical (e.g. engineer, information-technology), or knowledge-based field (e.g. mathematician, accountant) (as opposed to a primarily social service or human relations field such as childcare)
    - The adult has an intense preoccupation or interest in a certain area, which can be incredibly helpful for work
    - Preoccupation with routines such that the individual has the same routine and same schedule every day and doesn’t like changes, such as trying new foods, or travelling

Key features of autism spectrum disorder (ASD)

Difficulties with social communications and social interactions

- **Difficulties with communication such as non-verbal communication.** When we communicate, we use both verbal language (the words we use to express ourselves) and non-verbal language ('how' we say things, including our body language, tone of voice, facial expressions and gestures). People with autism spectrum disorder can have difficulties with non-verbal communication. For example, this may mean that they have trouble maintaining normal eye contact or understanding the gestures of other people. They may have trouble understanding social cues. As such, they may find it difficult to adjust their behaviours to various different social contexts.
- **Difficulties relating to people.** People who have autism spectrum disorder may also find that they have difficulties relating to people. This may be related to underlying difficulties with 'theory of mind', which is problems with empathy or seeing things from other's perspectives. In order to get by in the world, it helps to be able to guess or figure out how someone else is thinking or feeling. For example, if we know that a friend has had a bad day, then we can usually guess that our friend might be feeling sad or upset. However, individuals with ASD often have difficulty seeing things from someone else's perspective, which makes it much more difficult for them to deal with other people. This is why children with ASD often may appear non-empathetic or uncaring. With effective intervention, children with ASD may start to learn how to be more empathetic in situations, but it may not be a skill that comes automatically.

Behaviours or interests that are restricted and repetitive

- **Unusual play with toys and objects.** A child on the autism spectrum may play with toys, but often will play with them differently than other children do. For example, a child playing with trains may line them up over and over again or be obsessed with parts of a toy or repeatedly take apart and put a toy back together.
- **Rigid routines and/or preoccupation with routines or rituals.** Many children on the autism spectrum may have routines that they persist in doing over and over again and they may get extremely upset if they are kept from doing them. This includes placing things in a certain way or order. Because these symptoms are very similar to those seen in obsessive compulsive disorder (OCD), these children may receive an additional diagnosis of OCD by health professionals, prior to their being recognized as having an underlying autism spectrum disorder.
- **Sensory processing disorder, which is a problem processing sensory input such as sound, touch and movement.** For this reason, these children may be hypersensitive and become distressed or try to avoid sensory input such as sound, touch or movement. E.g. becoming upset with loud noises, become upset when touched, have troubles with tags on clothing or food textures. Others may be under sensitive, and in fact try to seek out sensory input such as sound, touch or movement. E.g. screaming, touching everything, or spinning. In many individuals, sensory issues can be severe and if not dealt with, can cause even more impairment than having ASD itself.
- **Difficulty with transitions such as changes in routine or one's surroundings.** Possibly due to sensory processing issues, since changes in routine/environment represent changes in sensory input, children with autism spectrum disorders may be sensitive to changes or transitions in their environment. For example, parents often report that the child has trouble shifting from one activity to another. Even the slightest changes in routine or schedules can cause problems. Parents find they need to give advance notice about activities or changes. They generally do better when their routine and environment is consistent and stable. Changes in caregivers or teachers may be particularly stressful for some children.

Individuals with Autism Spectrum Disorder may also have:

- **Difficulties with distractibility and inattention.** Individuals with ASD are often distractible and inattentive, and it is important to explore if there are any factors contributing to this. For example, perhaps the child is actually distracted from being overwhelmed from too much sound (such as the hum of the fluorescent lights), or from simply being in a large, noisy classroom. In such a case, reducing the sensory overload would thus help with the concentration. In some cases, individuals with ASD may also receive a diagnosis of Attention Deficit Disorder (ADD) or Attention Deficit Hyperactivity Disorder (ADHD), and may benefit from standard ADHD treatment (Holtmann, 2005).
- **Problems with moods such as anger or anxiety.** When we get overwhelmed, we can get angry, anxious or upset. Because of their other challenges, children with autism spectrum disorder may be at an increased risk of getting overwhelmed and upset. One way to help them is to figure out what exactly is stressing the

child, so that a plan can be put together to deal with each of the stresses. For example, typical stresses may include school (peers, teachers, school work) or home (dealing with parents, siblings).
- Delays in normal spoken language, while others may have normal spoken language but still have problems expressing their thoughts/feelings or difficulties with non-verbal communication.
- Intellectual disability (aka mental retardation), while others may actually have above average intelligence.

Strengths of autism spectrum disorder (ASD)

The following is a list of classic strengths that people with ASD are felt to have:

- Being very good at following rules and laws, as well as trying to understand things according to rules and laws (Baron-Cohen, 2003). Their "analytical" or logical reasoning skills may be helpful in fields such as accounting, engineering and computers.
- Other individuals with ASD have interests across other fields that include arts, music, drama, and social sciences.
- Being able to focus their attention for long periods of time, even on tasks which others may find boring or mundane.
- Being able to stay objective, and not be as affected by 'peer pressure' or others opinions (e.g. children on the autism spectrum are far less influenced by peer pressure such as with clothing choices or trying drugs.)
- Being visual. Many may have exceptional (even "photographic") visual memory (i.e. memory for things that they have seen). These strong visualization skills (the ability to think in pictures) may help in engineering, design and other visual fields.
- Being auditory and verbal. Rather than being visual however, individuals with Autism Spectrum Disorder may have exceptional auditory memory (i.e. memory for things that they have heard.) Strong verbal skills in fact, can help in future professionals such as being a writer, editor, tour guide, or lecturer...
- Show great depth of knowledge in areas of interest, which can help them become experts in their fields of interest.

History / Interviewing Questions:

- Ask parents about child's functioning in 2 core areas:
    - Social relatedness
        - Example: "Does your child seem to have problems knowing how to interact with people? How about with other family members? With other children at school / day-care / at the park?"
        - Example: "Does your child have difficulty paying attention to you? Do you feel like your child ignores you?"
    - Repetitive behaviour
        - Example: "Does your child do certain things repeatedly?" (clarify if these are purposeful or purposeless movements).
        - Example: "What sorts of things does your child enjoy / is your child interested in?" (look for variety vs. fixed interests)

More...

Screening

- The American Academy of Pediatrics (AAP) recommends the following screening schedule:
    - 9-months – broad developmental screen
    - 18-mos. – broad developmental screening plus autism-specific screening
    - 24-mos. – broad developmental screening plus autism-specific screening

- Factors to increase your index of suspicion: 1) preterm birth, 2) low birth weight, 3) family history of ASD in a sibling or parent.

Diagnosis

ASD is a condition characterized by:

A. Deficits in multiple contexts with

- Social communication,
- Social interaction

B. Restricted, repetitive patterns of behavior, interests and/or activities

More...

Changes from DSM-IV-TR to DSM-5:

The Diagnostic and Statistical Manual of Mental Disorders, 5th edition (DSM-5) contains one disorder for the entire spectrum of autistic symptoms: Autism Spectrum Disorder (ASD) with two domains:

- Social Communication (combination of the first two DSM-IV-TR domains)
- Repetitive Behaviour

More...

Differential Diagnosis (DDx)

- Rett's syndrome: Rare form of autism that affects only girls. Characterized by normal early growth and development, following by slowing of development, loss of purposeful use of hands, slowed brain and head growth, problems with walking, seizures and intellectual disability.
- Non-verbal learning disorder (NVLD): Problems with non-verbal communication and social skills. Has a distinct psychological profile with significant verbal/non-verbal split, whereby verbal strengths are significantly stronger than non-verbal strengths.
- Selective mutism: Child who is able to communicate verbally at home with family, but has significant problems talking with those at school or daycare.
- Language disorder and social (pragmatic) communication disorder: New diagnosis in DSM-5 that recognizes those with significant problems with verbal and non-verbal communication for social purposes, which causes impaired communication, social function and relationships.
- Intellectual disability (intellectual developmental disorder) without ASD
- Stereotypic movement disorder: Motor disorder characterized by repetitive, nonfunctional motor behavior (e.g. hand waving or head banging) that causes marked impairment.
- ADHD with severe impulsivity that causes social skills problems
- Schizophrenia with sufficient positive/negative symptoms

Comorbidity

- Comorbid conditions are very common with ASD, as 70% will have at least one disorder; 40% will have two or more disorders
- Psychiatric conditions such as
    - Attention deficit hyperactivity disorder (ADHD): Under DSM-5, it is recognized that patients with ASD can indeed have comorbid ADHD.
    - Developmental coordination disorder (DCD)
    - Anxiety disorders
    - Depressive disorders
    - Eating disorders (avoidant/restrictive type)
- General medical conditions
    - Seizure disorders

    - Sleep disorders
    - Constipation
- Genetic:
    - 10% will have a genetic disorder
    - Down syndrome
    - Fragile X syndrome
    - Tuberous sclerosis
- Other
    - Though not formally recognized as a separate DSM-5 diagnosis, sensory processing issues are common in ASD such as sensory hypersensitivities (e.g. auditory, visual, tactile)

Physical Exam

- Physical exam is primarily targeted towards ruling out other etiologies.
    - General observations:
        - Look for dysmorphic features which may suggest genetic syndrome
    - Weight, height, head circumference
        - Slowing of the normal rate of head growth may indicate Rett's syndrome
    - Neurological exam
    - Hearing screen
    - Dermatologic: Consider Wood’s lamp to rule out tuberous sclerosis

Laboratory Testing

- Based on history and physical exam, laboratory testing can be acquired to further rule out other etiologies.
    - Genetic testing such as G-banded karyotype, fragile X testing, chromosomal microarray
    - EEG if history of seizure or history of significant regression in social or communication functioning.
    - Neuroimaging

Ancillary Testing

- Reasons for ancillary testing:
    - Identify treatable causes of symptoms
    - Rule out other conditions with similar symptoms
    - Guide future planning (e.g. educational needs)
- Testing can include assessments of:
    - Vision
    - Hearing
    - Speech Language Pathology
    - Occupational Therapy (i.e. sensorimotor testing)
    - Neuropsychological testing (include verbal skills, non-verbal skills, adaptive skills and overall function assessments)
    - Sleep assessment

When and Where to Refer for Assessment and Diagnosis, i.e. Child/Youth Not Yet Diagnosed

When to refer

- If you suspect autism spectrum disorder (ASD)

Where to refer

- Agency or Centre specializing in ASD
- Developmental Pediatrician, Neurologist, Psychiatrist
- Psychologist

When and Where to Refer for Ongoing Treatment, i.e. Child Already Has an ASD Diagnosis

When to refer:

- If support from other disciplines (e.g. OT, SLP) is required
- If symptoms (or behaviours) are poorly controlled despite behavioural techniques and medications
- If patient has co-morbid mental illness with an unclear picture (e.g. ASD with ADHD, ASD with Depression, etc.)

Where to refer:

- Local autism and developmental agencies
- Typical interventions include:
    - Behavioural
        - Types of behavioural therapies
            - Applied Behavioural Analysis (ABA): Uses Pavlonian concepts of reinforcement to increase desired behaviour and decrease undesired behavior
            - Early Intensive Behavioural Intervention (EIBI): This is an intensive sub-type of ABA using discreet trial training initially, then progressing to complex skills.
        - Most specialized autism-spectrum programs will have ABA therapists
    - Communication
        - Speech language pathologist (SLP) can help the patient with developing an individualized plan to help with any language and communication difficulties
        - Interventions depend on the individual situation
        - Until communication ability improves, alternate communication modalities can be employed. Some examples include:
            - For patients who are non-verbal, the use of communication boards and teaching sign language can be helpful
            - For patients who have troubles with daily routines, the use of visual supports and picture exchange systems can help
            - For patients who are verbal, but who may have problems with non-verbal communication, there are specific interventions to help
    - Occupational therapy (OT)
        - Occupational therapy can help with sensory issues, as well as daily routines (e.g. using strategies such as visual supports)
        - Most specialized autism spectrum services will have OT
- Mental health professionals in private practice such as a psychiatrist, psychologist, paediatrician
- Support programs such as self-help, mutual aid services for autism, e.g. Autism Society

Medications

- There are no medications that "cure" the core symptoms of ASD, such as problems with theory of mind, perspective taking and social relationships
- However, medications are often used with certain target symptoms that have not responded to other interventions such as behavioural interventions
    - For aggression, low frustration tolerance, irritability, hyperactivity, stereotypy

        - Atypicals such as
            - Risperidone
            - Aripiprazole
    - For repetitive behaviours
        - Historically, SSRIs such as Fluoxetine have been used
        - However, a recent review in 2005 (Young et al., 2015) suggests that selective serotonin reuptake inhibitors are not effective in improving repetitive behaviors in children with ASD, and frequently cause activating adverse events
    - For hyperactivity, inattention
        - Stimulant medications for attention such as Methylphenidate (Ritalin), Dextroamphetamine (Dexedrine), or others
        - Atomoxetine (Strattera)
    - For hyperactivity, irritability
        - Alpha 2 agonists such as
            - Clonidine (Catapres)
            - Guanfacine (Intuniv)
    - For anxiety / depressive symptoms
        - SSRIs such as Fluoxetine (Prozac)
    - For insomnia
        - Sedating antipsychotic at bedtime, e.g. Risperidone, Clonidine
        - Melatonin
        - Trazodone

Medication Tables

Courtesy of the Department of Human Services of the State of Oregon, USA, and was retrieved on May 5, 2014. http://www.dhs.state.or.us/caf/safety_model/procedure_manual/appendices/ch4-app/4-14.pdf

Medication Chart for Atypicals

Brand name	Generic Name	Class	Dosage for children	Dosage for adults
Risperdal	Risperidone	Atypical	0.25-1 mg daily	4-16 mg daily
Abilify	Aripiprazole	Atypical	5-10 mg daily	10-30 mg daily
Zyprexa	Olanzapine	Atypical	2.5-5 mg daily	5-20 mg daily
Seroquel	Quetiapine	Atypical	50-300 mg daily	300-600 mg daily

Medication chart for SSRIs

Brand name	Generic Name	Class	Dosage for children	Dosage for adults
Prozac	Fluoxetine	SSRI	5-10 mg daily	20-60 mg daily

Medication chart for ADHD medications

Brand name	Generic Name	Class	Dosage for children	Dosage for adults
Concerta	Methylphenidate	Stimulant	1 mg/kg/daily Max 72 mg daily	1 mg/kg/daily Max 72 mg daily

Ritalin	Methylphenidate	Stimulant	Dosage 1 mg/kg/daily Start at 5 mg breakfast and lunch; Up to 60 mg daily	Start at 5 mg twice daily (breakfast and lunch); increase by 5-10 mg daily at weekly intervals; max 60 mg total daily
Adderall XR	Dextroamphetamine salts	Stimulant	Children age 6-12: Start 10 mg morning; increase by 5-10 mg daily at weekly intervals; max 30 mg daily Youth aged 13-17: Start at 10 mg daily; increase by 10 mg daily; max 30 mg daily	Start with 20 mg mornings, which is also the target dosage; max 50 mg daily though studies show > 20 mg daily shows no added benefit.
Vyvanse	Lisdexamfetamine	Stimulant	Not indicated for under 6 Age 6-12: Start at 20-30 mg daily; titrate up at 10 mg at weekly intervals; max 50 mg daily if necessary	Patient age 6+: Starting with 30 mg od AM in patients ages Adult: 10 mg or 20 mg at weekly intervals up to max dose of 70 mg/day.
Strattera	Atomoxetine	Non-stimulant	Up to 1.4 mg/kg/daily	Up to 1.4 mg/kg/daily

Medication chart for Impulsivity

Brand name	**Generic Name**	**Class**	**Dosage for children**	Dosage for adults
Catapres	Clonidine	Alpha adrenergic agent	Pediatric Dosage: 0.05mg QHS, increased to maximum dosage of 0.35 mg daily in divided doses.	
Tenex	Guanfacine	Alpha adrenergic agent	Initially 0.5 mg at bedtime titrated to maximum dosage of 3 mg daily in divided doses	

Medication Monitoring
CAMESA Guidelines for medication monitoring
www.camesa.org

Other Issues

- Inform the school regarding the diagnosis of autism spectrum disorder (ASD)
    - The school can implement a variety of accommodations, modifications and supports, depending on the specific situation
    - Types of interventions / supports may range from
        - In-class supports: Student remains in a regular classroom, with accommodations/modifications
        - Specialized placement: Certain schools may offer a special classroom specifically for students with ASD.
    - Testing interventions may include:
        - Psychoeducational, or neuropsychological testing to help identify individual educational needs

- Allied health disciplines such as OT or PT
    - With the help of the inter-disciplinary team, an individualized education plan can

be developed

- Specialized services can aid in delivering Structured Educational Models – education delivery methods that have shown efficacy in autism. Some examples include:
    - Early Start Denver Model
    - Treatment and Education of Autism and related Communication handicapped Children program (TEACCH program)

- If the diagnosis has been officially made, consider filling out forms for financial supports such as:
    - Disability Tax Credit from Revenue Canada
    - More information about provincial supports is available from the corresponding provincial autism society and local treatment agencies

- Help connect family with self-help and other community supports for autism spectrum disorders
    - There are many provincial, and local autism support organizations that can provide inavaluable support and information

- Ask about alternative/complementary treatments
    - There are several alternative / complementary therapies that exist, often endorsed by popular media figures, but most treatments have no proved benefit
    - Treatments that have been shown specifically not to work:
        - IV infusion of secretin
        - Oral vitamin B6 and Magnesium
        - Gluten-free, Casein-free diet
        - Omega-3 fatty acids
        - Oral human immunoglobulin
    - Treatments with greater potential risk:
        - Chelation
        - Contaminants in “natural” compounds
    - Although complementary treatments may not be directly harmful, they can still impose a burden on the family by diverting limited financial or other resources from a family

- Provide ongoing follow-up and case management as necessary
    - Although primary care physicians will most likely not be involved in delivering specialized services, primary care physicians nonetheless are involved over the life span of the patient
    - Primary focus changes over the life span
    - In very young children, the primary issues are those of diagnosis and identification of treatment programs
    - In school-aged children, behavioural and psycho-pharmacological issues predominate
    - In adolescents, the focus changes to vocational / prevocational training and planning for independence / self-sufficiency
    - It is also helpful to be aware that service utilization by patients with ASD and their families can often be sporadic in nature.
    - Therefore, long periods of limited follow-up do not mean completion of management.

Case, Part 2

- Thomas is a 10-year-old boy, brought to see you by his parents because of complaints of bullying and stomach pains
- Because of his bullying, you ask more about social skills, and parents tell you that he has had longstanding problems related to other people

- At the core, he appears to have troubles with seeing things from other's perspectives (i.e. empathy)
- You wonder about ASD and you refer him to a local specialist to provide further assessment and possible diagnosis; in addition, you also inform the school of your suspicions
- A few months later, his mother reports that indeed, he has received a diagnosis of ASD, and interestingly enough, mother is now wondering if the father (who works as a software programmer) may also have some ASD traits as well
- The school has involved their 'ASD Team' and has come up with a new individualized education plan (IEP) that takes into account his issues, especially social skills issues
- They are on the waitlist for local publicly funded autism services and although he is still struggling, and mother tells you that now at least she has hope…
- Mother thanks you for pointing them in the right direction, and for taking her concerns seriously…

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About this Document

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