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Autism Spectrum Disorder (ASD): Information for Primary Care

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Summary: Autism spectrum disorder (ASD) is characterized by deficits in social functioning restricted activities and interests, typically resulting in occupational impairment. Primary care providers can play a key role with early identification and connecting families to appropriate supports and interventions.

Epidemiology

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

Etiology

- Certain professions such as engineering and computer sciences have a disproportionately high percentage of individuals with ASD (Baron-Cohen, 2009), and has been portrayed in popular culture, such as the comic strip Dilbert, and the television show Big Bang Theory
- Furthermore, the risk of having a child with ASD increases greatly when both parents are engineers
- The empathizing-systemizing (E-S) theory suggests that people may be classified along two dimensions:
    - Empathizing (E), which is a patient's ability to identify and understand the thoughts/feelings of others and to respond appropriate to them.
    - Systemizing (S), which is an indication of their strength of interest in systems. A system is anything which follows rules, e.g. mechanical systems, natural systems, abstract systems, collectible systems.
- Individuals with ASD classically typically struggle with empathy, yet have great strengths in systemizing
- In increasingly technological societies, the theory is that having systemizing traits is adaptive, which may explain apparent increased rates of autism spectrum disorders (CDC, 2012).

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
    - Thinking and behaving flexibly

- Suspect ASD in an infant 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 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

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

Screening Tools

- The following are freely available online tools can be used for autism-specific screening purposes
- For toddlers
  - M-Chat
    - Parent rating scale, takes 5-10 minutes
    - Online version https://www.m-chat.org/mchat.php
  - Communication and Symbolic Behaviour Scales Devlopmental Profile Infant/Toddler Checklist (CSBS-DP-IT Checklist)
    - Parent rating scale, takes 5-10 minutes
    - License: The Checklist is copyrighted (Wetherby & Prizant, 2001) but remains free for use and can be downloaded from the Internet and freely photocopied or duplicated by other methods.
    - PDF versions available http://firstwords.fsu.edu/index.php/early-identification-of-communication-delays/26-csbschecklist
- For children/youth
  - Autism Spectrum Quotient
    - Self-rated, requires 5-10 minutes
    - Online version: http://aq.server8.org/
    - Online version (alternate link): http://www.autismresearchcentre.com/arc_tests
- For adults
  - Autism Spectrum Quotient
    - Self-rated, requires 5-10 minutes
    - Online version: http://aq.server8.org/
    - Online version (alternate link): http://www.autismresearchcentre.com/arc_tests
  - RAADS-14 Screening
    - Self-rated, 14-item questionnaire requiring 5-10 minutes
    - PDF version online http://www.biomedcentral.com/content/supplementary/2040-2392-4-49-S1.pdf

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

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

In the previous version, DSM-IV-TR, the disorder was previously divided into:

- Autistic Disorder
- Asperger's Disorder
- Rett's Syndrome
    - Removed in DSM-5, as it was considered an atypical inclusion. ASD symptoms are usually brief in early childhood in patients with Rett's. An organic etiology for Rett's has been identified; ASD and its predecessors have been defined by symptoms of behaviour, not etiologies)
    - Nonetheless, in DSM-5, Rett's Syndrome patients who have autistic symptoms may be diagnosed with ASD along with the specifier "Associated with a known medical or genetic condition or environmental factor".
- Childhood Disintegrative Disorder
- Pervasive Developmental Disorder Not Otherwise Specified (PDD-NOS)

In DSM-5, the remaining DSM-IV-TR disorders (Asperger's, Childhood Disintegrative and PDD NOS), are combined along with Autistic Disorder into **Autism Spectrum Disorder** in the DSM-5.

As for the diagnostic criteria, DSM-IV-TR had three domains of deficits:

- Social Function
- Communication
- Repetitive Behaviour

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 disability: 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
    - ADHD: Under DSM-IV it was not possible by definition to have comorbid ADHD, but under DSM-5, it is now recognized that patients with ASD can indeed have comorbid ADHD.
    - Developmental coordination disorder
    - 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 DSM-5 diagnosis, sensory processing issues are common in ASD such as sensory hypersensitivities (e.g. auditory, visual, tactile)

When to Refer

- If you suspect that your patient in primary care may have autism spectrum disorder (ASD), then the next step will be referral to a professional or agency that can make the diagnosis
- Other indications for referral
    - 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, where the diagnosis is typically done by a psychologist, psychiatrist, or developmental paediatrician
    - 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
            - Monitoring is required for betabolic adverse events, such as weight gain and dyslipidemia
    - 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 charts

Medication charts 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	Up to 60 mg daily	? maximum Ritalin dosage for adults
Adderall XR	Dextroamphetamine salts	Stimulant	Children 3 – 5 Initial daily dosage is 2.5 mg; increased by 2.5 mg weekly intervals until optimum response Children age6	Start with 20 mg daily

Vyvanse	Lisdexamfetamine	Stimulant	Not indicated for	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 adults	Dosage for children
Catapres	Clonidine	Alpha adrenergic agent		
Tenex	Guanfacine	Alpha adrenergic agent		

Other management 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.

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About this Document

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