

PedsCases Podcast Scripts

This is a text version of a podcast from PedsCases.com on “Autism Spectrum Disorder.” These podcasts are designed to give medical students an overview of key topics in pediatrics. The audio versions are accessible on iTunes or at www.pedscases.com/podcasts.

Autism Spectrum Disorder

Developed by Lauren Robinson and Drs. Debra Andrews and Lyn Sonnenberg for PedsCases.com.
December 21, 2014.

Lauren: Hi everyone. I’m Lauren Robinson, a medical student at the University of

Alberta, and with me today is Dr. Lyn Sonnenberg, who is the program director for Developmental Pediatrics at the University of Alberta.

Before we start, we also wanted to acknowledge Dr. Debra Andrews, Divisional Director for Developmental Pediatrics, for her help in developing the script.

We are going to be presenting an approach to Autism Spectrum Disorder in the clinical setting. We hope that by the end of this presentation, learners will be able to *define the core features* of ASD, describe *how to screen* for ASD in the primary care setting, and know *how the diagnosis is made*. Lastly, we hope to give listeners some *tips on how to interact with patients* with ASD.

Lauren: Let’s start with the basics. Dr. Sonnenberg, *what is Autism Spectrum Disorder?*

Dr. Sonnenberg: ASD is a neurodevelopmental disorder. It’s characterized by impairments in social communication and social interaction, as well as restricted, repetitive patterns of behaviour, interests or activities.

Lauren: I’ve heard that autism is becoming more common. How common is it?

Dr. Sonnenberg: The Centre for Disease Control and Prevention’s latest numbers indicate the prevalence is 1 in 68 children, with boys affected about 4-5 times more frequently than girls. The reported increase in prevalence of autism since the 1980’s is controversial. Is there more autism or are we just better at recognizing it? There are no clear answers.

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But given how common it is, primary care practitioners must have a solid understanding of ASD because early intervention has been associated with a much-improved prognosis.

Lauren: Let's say I'm in the clinic going to see a 18 month-old boy for a well-child check-up. What should I do to address the possibility of ASD?

Dr. Sonnenberg: I think what you're getting at, is *how we screen for autism*. First, it is important to note that a positive screen does not equal a diagnosis of autism, because autism shares symptoms with other developmental disorders. A positive screen is only the first step.

In Canada, screening for *any* developmental condition, including autism, starts with developmental surveillance. This should take place universally at all well-child visits, and may include taking a history using developmental milestones or using a standardized developmental screening tool, such as the Ages and Stages questionnaire (the ASQ), the Parents' Evaluations of Developmental Status (PEDS) or, in Ontario, the Nipissing District Developmental Screen. The Canadian Paediatric Society has recommended that all children have an extended well child visit at 18 months of age and that health care providers use a standardized screener at that time.

Lauren: So what features in the developmental history would make me more worried about autism?

Dr. Sonnenberg: Simply put, failure to meet milestones in the social and communication domains. There also are some red-flags you should try to remember, which would make you want to further investigate. Do you remember what some of these are?

Lauren: Yes. No big smiles or joyful expressions by 6 months, the absence of sharing sounds, smiles or other facial expressions by 9 months, and the lack of pointing, showing, and waving by 12 months old.

In terms of communication, no words by 16 months and no two word meaningful phrases by 2 years is abnormal.

Dr. Sonnenberg: That's right. It's also important that when there is any delay, it is interpreted in the context of the child's development as a whole. For example, in a two year-old who meets the milestones of a one year-old in all developmental domains would be less specifically concerning for autism as their social-interaction capabilities are aligned with their developmental age. Of course, further work-up of global developmental delay would certainly be indicated in this child!

Lauren: Is there anything else I should be asking on history?

Dr. Sonnenberg: As for any pediatric history, it is important to start broadly and ask the caregivers if they have any concerns, developmental or otherwise. Inquiring about a family history of autism or other genetic disorders is key, as autism has high heritability. In addition, asking about feeding and a general review of systems is important, as feeding issues can be associated with autism, and development can be affected by many chronic medical conditions.

Lauren: What about the physical exam? Are there any typical physical findings in children with Autism?

Dr. Sonnenberg: In many children, no. Sometimes head circumference may be increased, especially in comparison to body proportions, in the toddler years, particularly if it started out in the normal range. There is also a subgroup of children with Autism who have medical syndromes, such as fragile X, which may have findings on physical exam. Eye contact and hand flapping can also be noted, so a thorough physical exam is important, looking especially for dysmorphology and minor anomalies.

Lauren: Let's say during developmental surveillance this 18 month-old boy had not been pointing to objects to get his parents' attention, and spent hours everyday sitting spinning the wheels of his favorite toy car. In terms of language, he was able to say three words.

He seemed to be aligned with his peers in terms of motor development, but never seemed very interested in the other toddlers at daycare. His physical exam was normal.

Dr. Sonnenberg: Now you are suspicious of Autism. In this case, it would be appropriate to move onto the second level of screening. Indications for secondary screening are: parental concern, having a sibling with autism, and having social or communication delays during developmental surveillance. Secondary screening involves using an autism-specific screening tool, such as the Modified Checklist for Autism in Toddlers Revised with Follow-up (or the M-CHAT-R/F). It's a questionnaire administered by a health practitioner to parents for children age 16-30 months.

Lauren: Okay, so say I decide to use the M-CHAT R/F, and I score it and it's positive. What should I do next?

Dr. Sonnenberg: After a positive screen, the next step is referral to a multidisciplinary team for diagnostic assessment. The team varies depending on local resources but usually includes a physician, (usually a developmental pediatrician, child psychiatrist, or pediatric neurologist), psychologist, speech language pathologist, nurse, social worker, and/or occupational therapist. These are all individuals with expertise in the diagnosis of autism and other forms of developmental delay.

Lauren: I've heard that can take a long time. Are there any lab tests or other investigations I could do while waiting for the child to see the multidisciplinary team?

Dr. Sonnenberg: You could start to work up the language delay component. Do you remember the other common causes of speech-language delay in a toddler?

Lauren: It can be part of global delay, or isolated speech-language delay, or sometimes a motor problem. And hearing loss—we should check hearing.

Dr. Sonnenberg: Right. Audiology to check hearing is top priority, and a community speech language pathologist is able to characterize the type of language impairments and start therapy.

Genetic testing depends on what your lab can offer. Most can provide a karyotype and fragile X testing. Microarray or comparative genetic hybridization or CGH analysis may be used to examine for microdeletions and duplications that may or may not already be associated with autism. More and more labs have this testing available, and it's becoming the genetic test of choice in children with no physical findings that suggest a syndrome. If you think there is a syndrome, a geneticist can best direct the labwork for specific conditions.

Testing beyond this is dependent on history and physical findings and may include metabolic testing, neuroimaging and EEG. Epilepsy is more prevalent in children with autism; however, EEG is only indicated if there is suspected or witnessed seizure activity.

Neuroimaging may be indicated if there is evidence of tuberous sclerosis (for example, if there are hypopigmented skin lesions) or other neurologic findings.

Lauren: So it sounds like investigations are highly dependent on history and physical exam, and it is important not to send these children for tons of tests without reason.

Dr. Sonnenberg: Exactly! Although the cause of autism is not known, most cases seem to be polygenetic. Very rarely is there ever one gene, metabolic abnormality, brain lesion, or environmental influence leading to autism.

Lauren: That makes sense. So once children see the multidisciplinary team, how is the diagnosis made?

Dr. Sonnenberg: It is likely that the team will use several tools, such as the Autism Diagnostic Inventory Revised (or ADI-R) for the autism history and the Autism Diagnostic Observation Schedule (or ADOS) for a standardized, semi-structured observation of the child assessing socialization, communication, and play skills.

The diagnostician will use the results from these tools, the results of the assessments of other multi-disciplinary team members, and their own clinical judgment in accordance with the Diagnostic Statistical Manual of Mental Disorders, Fifth Edition.

What can you tell me about the diagnosis of Autism in the DSM-V?

Lauren: Well, the DSM-V replaced the DSM-IV TR in 2013 and changed multiple aspects of the diagnostic criteria of autism. For example, the diagnostic categories of Pervasive Developmental Disorder, Pervasive Developmental Disorder not otherwise specified, and Asperger's Disorder were removed from the DSM-V. Now, the term Autism Spectrum Disorder is used for everyone meeting the criteria. That's a lot less confusing.

In the DSM-V, Autism Spectrum Disorder (ASD) has *two* symptom domains that must be fulfilled. First, there must be persistent deficits in reciprocal social communication and social interactions across multiple contexts. Second, there must be restricted, repetitive patterns of behavior, interests or activities.

Symptoms must be present from an early developmental age, but may not fully manifest until social demands exceed limited capabilities. This opens the diagnosis up to older children, adolescents, and adults. In addition, these impairments must affect everyday functioning, so you need to check adaptive behaviour skills. Lastly, limitations may not be better accounted for by a global developmental delay or intellectual impairment, and if delay is present, social impairments are more than what you'd expect for developmental age—which means we have to know what their overall developmental level is.

Dr. Sonnenberg: That's right. And different children may present with different severity levels within the two main symptom groups, so autism can include everything from a very developmentally impaired non-verbal child with severe repetitive behaviour to a child who can talk in full sentences but who is very awkward socially, plays by himself and seems stuck on his favourite topic—the kind of person who talks *at you* instead of *with you*. It's always important to remember the “spectrum” in Autism Spectrum Disorder, even though we often shorten the name to Autism.

I'll add one thing about diagnosis of ASD. Comorbidities are very common and include intellectual impairment, ADHD, and developmental coordination disorder or DCD. In adolescents and adults with autism, anxiety and depression are common. So you can see why a thorough assessment by a team is needed—but the team needs primary care providers to think of autism and recognize its early signs, to get things started.

Lauren: So once we have a diagnosis, what are the treatments available?

Dr. Sonnenberg: Early, intensive educational and behavioral therapy has been associated with improved language and social skills. To get a sense what is meant by “intensive”, most interventions are than 25 hours per week.

Lauren: That's a lot of intervention! Aren't there any medications that help with autism?

Dr. Sonnenberg: No pharmacotherapy has been identified that improves the core symptoms of autism; however, medications are sometimes prescribed for co-morbid ADHD or disruptive behavior, such as intensely repetitive actions, aggression, and self-injurious behaviors. Psychopharmacotherapy may also be used to treat co-morbid depression or anxiety in later years.

Lauren: As we are nearing the end of the interview, do you have any *tips for us when it comes to interacting with patients with autism?*

Dr. Sonnenberg: Sure. First and foremost, remember that children with autism can still have underlying medical conditions. For example, constipation and gastroesophageal reflux are common problems that are often overlooked. Trust the parents when they say something is wrong or different. Secondly, approach the child slowly and speak in a quiet voice as children with autism may have sensory differences and can be set off by louder noises or bright lights. Again, ask the parent how to best approach their child as each child with Autism is different. They really do know their child best.

Lauren: Thank-you so much, Dr. Sonnenberg, for sharing your expertise in the area. Before we finish, let's quickly summarize our main learning objectives:

- Autism is a common neurodevelopmental disorder marked by two main types of symptoms—1) impairments in social communication and social interaction and 2) restricted, repetitive patterns of behaviour, interests or activities.
- Screening includes universal developmental surveillance, and then in individuals at higher risk, autism-specific screening tools like the M-CHAT.
- Definitive diagnosis is made after assessment by a multidisciplinary team, but don't wait for the team to get things going because the earlier the intervention, the better chance the child will benefit from intervention! You can start the process by doing a good history and physical, ordering audiology testing and referring the child to a speech language pathologist as soon as possible.

That concludes our presentation! Thanks for listening to PedsCases Podcasts.

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