

PedsCases Podcast Scripts

This is a text version of a podcast from PedsCases.com on “**Early Detection for Autism Spectrum Disorder in Young Children – CPS Podcast (Part 1 in ASD series).**” 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.pedcases.com/podcasts.

Early Detection for Autism Spectrum Disorder in Young Children – CPS Podcast (Part 1 of ASD series)

Developed by Dr. Nehal Shata and Dr. Lonnie Zwaigenbaum for PedsCases.com.
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Introduction:

Hello, my name is Dr. Nehal Shata. I’m a Pediatric Resident at Montreal Children’s Hospital in Montreal, Quebec. This podcast was produced by PedsCases and the Canadian Paediatric Society. Today we will be discussing the CPS Position Statement “Early Detection for Autism Spectrum Disorder in Young Children.” This podcast was developed with Dr. Lonnie Zwaigenbaum, the lead author of this statement, as well as a Pediatric Associate Professor in the Department of Pediatrics at the University of Alberta.

Autism in children is a question that has led to a lot of controversial discussion. We are seeing more and more cases of Autism, otherwise known as Autism Spectrum Disorder or ASD, and the cause of this rise in ASD is debated among health care professionals everywhere. But whatever the cause of increased Autism is, the fact remains that early detection of ASD is more important than ever.

It’s possible that early detection could lead to better long-term outcomes by taking advantage of a child’s brain neuroplasticity at a younger age. This has left us with important questions about how to diagnose ASD at the earliest age possible, and today we are going to discuss how.

By the end of this podcast, the listener should be able to

1. Recognize the signs of early autism.
2. Understand how to refer a child for an early ASD diagnostic assessment.
3. Order investigations for accurate diagnosis of early Autism.

Let’s start with a case:

You are working at a family clinic. Aiden is an otherwise healthy term 18-month-old boy. At his 18-month well-baby visit with you, his family doctor, he is found to have a speech delay. He produces no intelligible words, only crying and screaming. His gross motor and fine motor development are normal: he can walk alone and steady, and he is able to scribble with a pencil. However, he is described as showing limited social responsiveness, rarely

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responding to his name and almost never engaging his parents in playtime. He is an only child, and the parents are not sure if this is normal behavior for children. Upon physical exam: there is no eye contact, and he is crying and combative through the exam.

You wonder what to do next. Now you have to ask the important question... could this be early Autism?

Let's take a moment to discuss the definition of ASD.

Autism spectrum disorder is a life-long neurodevelopmental disorder characterized by impairments in social communication, repetitive, restricted patterns of behaviour, and unusual sensory sensitivities or interests. ASD has significant impact on the lives of children and their families. And it's really common. The current estimation of ASD in Canadians from the ages of five to seventeen is 1 in every 66 kids.

That's a lot! But keep in mind that part of this high number may be because our ability to diagnose autism is better now than in the past.

Accurate ASD diagnosis is broken down into two symptom domains: The first domain is social communication impairments, and the second domain is restricted, repetitive patterns of behaviour and interests.

Let's start by looking at the diagnostic criteria for the first symptom domain: social interaction and communication. Now what does this look like, you may ask?

Well, Impairment in social interaction can look like many things. Often there is difficulty initiating or responding to social interactions. A child might seem distant and disinterested in others. Another child might seem outgoing, yet over-focused on talking about their special interests, not recognizing whether others would rather talk about something else.

Another way it can look is reduced spontaneous sharing of interests or emotions. The child might seem really excited about a favourite toy or activity, yet it seems to be a private experience, not shared with others. For example, the child might have a big smile, but not look towards another person while they're smiling.

There might be an impairment in how children communicate without words, like eye contact, talking with their hands, or directing their facial expressions. They might appear disconnected or may even use someone else's' hand to get a desired object without making eye contact.

Another diagnostic criterion in the social interaction and communication area is the failure to develop and maintain relationships. You might notice that the child has reduced, or atypical interest in their peers. They might enjoy time with the other children at daycare, but not show any interest in spending time with any one child. Or when they are older, they have trouble making friends. You might also notice that the child doesn't play pretend or engage in imaginative play with their peers. They may have a sense of imagination, but have trouble sharing control of the storyline with another child.

So that wraps up the Diagnostic criteria for Social interaction and communication.

Now it's time to talk about the second domain of ASD diagnosis, which is about being over focused on special interests or daily routines, as well as unusual or repetitive physical movements and behaviors.

What does this look like? Well, it can look like scripted speech and behaviour. The child may repeat words or phrases that they hear from television shows or movies, and they might have habits that seem unusually repetitive, such as lining up toys or other objects. The child might also find it distressing, sometimes to the extreme, if someone else interferes with these activities. For example, taking, or even moving one of the toys.

They might show repetitive movements like body, arm, hand, or finger movements. Some common movements are spinning around, hand-flapping, or finger-flicking. Sometimes, the child might have stiff posturing of their hands or even their whole body.

Another common feature is resistance to change. The child might wear the same clothes or even the same colour every day, or they might eat the same food every day. They might show distress if the routine is changed, for example the route to preschool.

Also, the child might have restricted, fixated interests. These interests might relate to special topics like a favourite TV shows or YouTube video or certain objects. It might be that these interests are similar to that of children their age, but they can become all-encompassing like wanting to dress as their favorite Disney princess for months on end or persist past the time that their peers have long since moved on to other interests.

The child might exhibit hyper- or hypo-sensitivity to sensory input. This could include some distress or fascination with smells, sounds, textures, sights, and tastes. These sensitivities can affect their ability to participate in day-to-day activities, like a child who is so nauseated by food odours that they cannot eat close by other family members, or so upset by certain sounds that they refuse to enter a public washroom because of the sound of the hand dryer.

That wraps up the first two domains, which were: 1. Social Interaction and Communication and 2. Special Interests and Repetitive Behaviors.

These first two are the most important, and it's important to remember that according to the DSM-5 Autism Spectrum Disorder, both of these symptom domains must be fulfilled to make an accurate diagnosis of ASD.

So, let's go back to our case for a moment. Aiden fulfills both of these symptom domains.

You notice that Aiden lines up objects and flaps his hands, has a fascination for the texture of the toys, still has no single words, does not point to anything, never pretends to play, doesn't share his interests, and does not respond to his name.

But there are five other factors to consider as well.

Which brings us to a third domain of ASD diagnosis. The third domain is about when the signs and symptoms present in the child's life. Think about it this way. A child may have signs of autism during early development, but these might not be visible until they go to school, when the environment is more stressful for them. But that doesn't mean that the signs weren't there all along.

Okay. Time for the fourth domain of diagnosis. This one is about how severely the symptoms interfere with everyday functioning. Is the child functioning overall with the symptoms? That is, is the child connecting with other children and coping with the demands of daily life at home and outside of home? If the answer is no, the symptoms may qualify as criteria for an ASD diagnosis.

The fifth domain indicates the above-mentioned symptoms not fully explained by intellectual disability or global developmental delay, like ADHD or fragile x syndrome. Some children have both developmental delays and ASD, but the delays do not entirely account for the child's social challenges and behavioral symptoms.

These first five domains are enough to give us a diagnosis, but there are two final domains to consider as well. The following two domains are about further characterizing the child's diagnosis and challenges, or diagnostic qualifiers. In other words, they do not determine whether the child has Autism or not. But they are important to provide context in the diagnostic assessment.

The sixth domain indicates that ASD may occur with or without medical, genetic, neurodevelopmental, mental or behavioural disorders, or an intellectual or language impairment. Put in other words, ASD can happen alongside a second condition, making the diagnosis a bit more complicated.

The seventh, and final domain explains how to determine the severity of autism. This can help to refine the diagnosis. There are three levels of severity for ASD. Level one requires support, level two requires substantial support, and level three requires very substantial support. Keep in mind that these levels may be difficult to determine at the initial time of diagnosis with very young children.

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Back to the case. Aiden starts daycare and the schoolteacher says he doesn't associate with the other kids. While other kids are playing on the playground, Aiden sits by the fence, tapping on it repeatedly instead of engaging with others.

So, you order a genetic test and a hearing test, and both came back normal. The mother says that it seems like he lives in his own bubble. She's tired and she doesn't know what to do. You tell her that you will be with her in every single step and then you provide her with local resources to help families with ASD.

Keep in mind to always be sensitive to distress related to developmental concerns and potential impacts on family life. Be mindful of family vulnerability, fears for the future, and financial pressures as they work through the assessment process.

She asks you if you think that Aiden is autistic. You tell her that Autism has not been ruled out, but that you'd like to find out more. Then the mother wants to know what the risk factors are.

Which brings us to our next section:

What are the etiology and risk factors of ASD?

The etiology of ASD is not completely understood, though recent findings suggest an interplay among genetic, epigenetic, and environmental factors.

Let's take a look at table two. Table two is about environmental risk factors for ASD, like genetics, prenatal and postnatal.

The risk factors associated with genetics are as follows. The major one, and the one most people know about, is Male sex. Over 75% of autism cases are found in males, although numbers of females are climbing.

Another factor is having a sibling with ASD, or even a more distant relative, like a cousin, with ASD or related developmental disorder, like severe language delay.

Prenatal exposures modestly associated with ASD are as follows: parental age of 35 years and up, maternal obesity, diabetes, hypertension, in utero exposure to valproate, pesticide, or traffic-related air pollution, maternal infections like rubella, or close spacing of pregnancies.

Other risk factors are: low birth weight and extreme prematurity.

You explain to Aiden's parents what they can expect during all the stages of the assessment process. You give them space for questions, and you counsel them and provide resources where they can find more information on their own, as well as contact information for a local family support group.

As you already know, Aiden had a normal vision and hearing screening, which was considered the first round of screening, but you are still concerned about his failure to meet milestones in the social and communication domains. At present, he has no single words, an absence of compensatory gestures, a lack of pretend play, and limited joint attention like initiating, responding or sharing of interests. The fact that Aiden shows more than one diagnostic criteria in these cases is enough to move on to a more intensive, ASD-focused round of screening.

Let's talk about the importance of screening tools. At this stage, these generally include standardized assessments including questionnaires and interviews to gather more information to inform your clinical impression and referral decisions.

In Aiden's case, you will need an autism-specific screening tool. This can be a questionnaire such as the Modified Checklist for Autism in Toddlers, which is for children age 16-30 months.

So you administer the questionnaire to Aiden's parents. Aiden's mother responds 'no' to eight of the 20 questions, in which eight or more 'no' responses are considered high-risk. These results tell you that no interview is needed to move to next steps.

After a positive screen like this, you refer Aiden immediately for local early intervention services.

For Aiden, you determine that the services necessary are speech-language therapy, occupational therapy, and targeted preschool support.

In younger cases than Aiden, there is infant development available. As a general rule, A CPS statement is recommended, enhancing developmental assessment and parental education during the 18-month well-baby visit—a critical time when signs or symptoms of ASD often emerge. The Rourke Baby Record may be used to chart global development, physical examination data, immunization, nutrition, and other milestones.

Always keep in mind: 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.

After multiple meetings with you and multidisciplinary team, Aiden receives a confirmed diagnosis of Autism and the parents receive a training session on how to take care of him and the team provided the parents with all available resources and tools that will help provide him with help and support. At the end, Aiden's parents tell you that they are grateful for your help and resources, and feel prepared to move forward with Aiden's support and development plan.

Now let's sum up the key points.

Remember, our learning objectives were:

1. Recognize the signs of early autism.
2. Understand how to refer a child for an early ASD diagnostic assessment
3. How to order investigations for accurate diagnosis of early Autism.

First, Autism is a common neurodevelopmental disorder marked by two main types of symptoms— which are broken up into two categories. The first is impairments in social

communication and social interaction, and the second is restricted, repetitive patterns of behaviour, interests or activities.

Remember, screening includes universal developmental surveillance, and in individuals at higher risk, autism-specific screening tools like the M-CHAT.

And finally, you helped the child's family access intervention before a definitive diagnosis, 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.

I hope you found this useful and thank you for listening.