PedsCases Podcast Scripts

This is a text version of a podcast from Pedscases.com on "**Standards of diagnostic assessment for autism spectrum disorder – CPS Podcast (Part 2 of 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.

# Standards of diagnostic assessment for autism spectrum disorder – CPS Podcast (Part 2 of ASD series)

Developed by Dr. Camila de Lima and Dr. Jessica Brian for PedsCases.com.
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## Introduction:

Hi everyone, my name is Camila de Lima and I am a pediatric resident at the Janeway Children's Hospital in Saint John's, Newfoundland. This podcast was produced by PedsCases together with the Canadian Pediatric Society. It is part two of a three-part series of podcasts on Autism Spectrum Disorder, or ASD.

Today we will be reviewing the CPS statement "Standards of diagnostic assessment for autism spectrum disorder". This podcast was developed with Dr. Jessica Brian, lead author of the statement and member of the Autism Spectrum Disorder Guidelines Task Force. For additional information and to view the complete CPS statement, please visit cps.ca. The script for this podcast can be viewed at pedscases.com.

After listening to this podcast, the learner will be able to:

1. Identify three approaches toward an ASD diagnostic assessment.
2. Learn and apply the essential components of an ASD diagnostic assessment, including key aspects on history, physical exam, use of diagnostic tools, need for further investigations, and consideration of major differential diagnoses and co-occurring conditions.
3. Identify and understand the components of a comprehensive needs assessment and whether it is required for further treatment planning.

**We will start by reviewing our case:**

Aiden is a patient previously seen by you, his pediatrician, at 18 months of age for concerns about a language delay. He presents again at two and a half years old because of some behaviour concerns. He missed his two year well-child check up and has not been back to your office since his 18 month visit. In the meantime, he attended two speech language pathology sessions as you had suggested, but the family stopped going as it was too far away from their home. You had also referred Aiden to an autism specialist at their last visit but they have not yet heard back regarding an appointment. The family will be coming back to your office next week and you are now planning the next steps in Aiden's care.

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At this point, ASD is still in your differential. You are wondering if there are any other ways to evaluate Aiden for ASD. As per the newest CPS statements, you learn there are three main approaches for such an assessment. The three approaches depend on the pediatric care provider's clinical experience, judgment, the patient's complexity of symptoms, and other family factors.

Approach 1 may be undertaken when a child's symptoms clearly indicate ASD. A sole pediatric care provider can independently diagnose ASD based on judgment and DSM 5 criteria. This can be done with or without data obtained from diagnostic assessment tools. Although efficient, this approach is often not sufficient for accessing specialized services in some Canadian jurisdictions.

Approach 2 is a shared care model. In this case, a clinician and another health care provider have joint responsibility for patient care. Here we may use information from ASD diagnostic assessment tools, as well as a consult with a health care professional with specialized knowledge to inform a diagnosis in cases where ASD is not readily apparent. These could include mild or atypical ASD presentations, a complex presentation, or symptoms suspected in a child under 2 years old.

Approach 3 is the team-based approach – in some Canadian provinces, this may be the only approach accepted for accessing specialized services, which often contributes to long wait times. Here we have a multidisciplinary team (often composed of a Developmental Pediatrician, Speech Language Pathologist, Psychologist, and Occupational Therapist, among others) where each member works independently and then they share information with one another. The team may or may not reach a diagnosis by consensus. Although this approach is efficient when it comes to intervention planning and optimizing access to supportive programs, the team-based approach may not always be required to determine a diagnosis. With the aim of reducing delays and wait times, this approach should be reserved for when diagnostic determination is more difficult. These include subtle presentations, patients with a complex medical or psychosocial background, or a presentation complicated by co-existing health concerns.

You conclude you need more information on Aiden in order to decide which approach would be best suited for his care. You know that ASD is a clinical diagnosis made through history, physical exam, direct interaction and observation for characteristic behaviors. You have some experience and extra training in ASD, so you plan on conducting an ASD diagnostic assessment at their next visit. The three key objectives of such an evaluation include:

1. Provide a definitive, categorical diagnosis of ASD – in ambiguous cases, a provisional diagnosis can be made, and the child should be monitored carefully and/or referred for further evaluation
2. Explore conditions that mimic ASD symptoms and identify comorbidities
3. Determine overall level of adaptive functioning, which will help with intervention planning. Here you should identify strengths, challenges, personal interests and priorities of your patient and/or their family.

You learn there are certain essential elements that are important in an ASD diagnostic assessment. We will go through each of these in more detail separately, but overall they include: a review of the patient's records; interviewing parents, family members, and other caregivers; an assessment for core features of ASD; a comprehensive physical exam; additional investigations; and consideration of differential diagnoses and co-occurring conditions.

The first step includes reviewing records. Medical information to be reviewed consists of a patient's birth history, neonatal screen results, records of their previous routine well child visits, and any prior tests, treatments or hospitalizations. You should also review previous developmental evaluations (including ASD screening tests), hearing and vision tests, and any assessments made by other health care professionals such as a child psychologist, speech language pathologist, or occupational therapist. If available, you should also review educational records and teacher observations.

In Aiden's case, he was a term baby boy with an unremarkable birth history. His neonatal screen was negative. He has never been hospitalized. Vision and audiology tests report no abnormalities identified. You have access to the Speech Pathologist's two reports in which she states there were concerns with both his receptive and expressive language abilities. She also noted poor eye contact during her visits. From your previous notes you saw that by now he will have started daycare.

The next step is interviewing, with a focus on a family-centred approach. Even before seeing the patient in your office, you should aim to have flexible appointment scheduling times and anticipate potential barriers to access. If possible, send the family a standardized questionnaire to be completed during or ideally before the interview. Ask them to bring educational or other care provider reports to the appointment. Always consider the family's cultural and religious values and adjust your interviewing approach accordingly.

As a general rule, asking semi-structured and open-ended questions is a good way to start. This can be integrated with information from the questionnaire. You can start by reviewing the birth and medical history with the family, focusing on ASD associated difficulties such as sleep problems, unusual diets, or self-injury. If not already known, identify the original reasons for referral and explore when the first concerns emerged. A detailed development and behavioral history should be done, including whether any formal interventions have been tried.

A detailed family history should be conducted, ideally spanning at least three generations and encompassing medical as well as mental health issues. Specifically ask about a history of developmental delay, ASD, known genetic conditions, learning disorders (including ADHD), and behavioral problems. A psychosocial history should include any known family violence, trauma, substance abuse or neglect. Make sure to also ask about current family functioning, routines, what the family considers their strengths, and what resources may be available to them.

Going back to Aiden, you knew he had presented at 18 months with a language delay. Parents report that they stopped going to the Speech Pathologist as it was too far

away from their home and Aiden would only scream and cry most of the time. Over the past year, he has made some progress with his speech. He mostly uses single words or short phrases that his parents can understand, including “no”, “hot hot” for bottle, “ta ta” when he doesn’t want to be touched, and “ruff ruff rescue” when he wants the TV turned on, a phrase they recognize from his favourite show. He still does not respond to his name. His motor development has been unremarkable and parents confirm he is able to run, jump, use a spoon and draw circles. Socially, his parents think he is more interactive, often taking their hand when he needs their help with something like opening the fridge or making a toy work. He likes playing with cars and making the wheels spin. At daycare the staff have noticed that he seems to prefer playing on his own and doesn’t interact much with the other children. When Aiden has a tantrum, he will hit and break things, and both parents and daycare workers are concerned that he will hurt himself. He is described as a picky eater and doesn’t like when the foods on his plate touch. Aiden’s family history is mostly unremarkable, except for a first cousin with ADHD. There are no major concerns from a psychosocial perspective, but the family is going through financial difficulties at this time.

In the third step you should start your assessment for the core features of ASD. Inquire about patterns of behaviors and interests, keeping in mind the DSM 5 diagnostic criteria. An important part of this step involves observing and interacting with the child. An ASD diagnostic tool may be used, and we’ll talk about those next.

Firstly, it is important to note that these tools cannot and should not be used alone to diagnose ASD. Results should merely complement the diagnostic process. Secondly, training is generally required before using these tools.

Having said that, there are two types of ASD diagnostic tools – those based on direct observation and interactions of the child, and those based on parent or caregiver interviews or questionnaires. Examples of the former include the Autism Diagnostic Observation Schedule, or ADOS, and the Childhood Autism Rating Scale, or CARS. The ADOS may be used from twelve months onwards, whereas the CARS is for those aged two and above. There are many caregiver interviews and questionnaires, and they vary by age group. Examples include the Autism Diagnostic Interview-Revised, or ADI-R, for those aged two and above, and the Social Responsiveness Scale, or SRS, which has preschool and school-aged versions.

Comparatively, a recent Cochrane review found that the ADOS has the highest sensitivity and comparable specificity to the CARS and ADI-R. In some Canadian provinces, both the ADOS and the ADI-R are required to inform a diagnosis of ASD.

Let’s go back to Aiden. Luckily, you work in a large clinic where a child psychologist has been trained in administering and interpreting the ADOS. She agreed to help you evaluate Aiden during his visit. During the assessment, you saw that when Aiden gets excited, he will yell loudly and flap his hands, sometimes spinning his body around and around. He throws temper tantrums when he has to transition from one activity to another. He is unable to participate in pretend play at a doll’s birthday party.

The fourth step of your assessment involves a comprehensive physical exam and deciding which additional investigations are necessary, if any. As always, you should start with anthropometric measures and vital signs. Look specifically for macrocephaly since 20% of those with ASD have this finding. Assess for any dysmorphic features or congenital anomalies that might point towards a syndrome or known genetic condition. Some disorders that have been associated with ASD are Tuberous Sclerosis, Fragile X syndrome, Rett Syndrome, Neurofibromatosis, Angelman, or Trisomy 21. As such, in addition to your cardiorespiratory and abdominal exams, you will also want to do a full neurological exam and a skin exam looking for cutaneous stigmata.

In terms of investigations, you should start with hearing and vision assessments and further referrals to audiology, optometry, or ophthalmology as needed. Further investigations are recommended only if clinically indicated. Suspicion of seizures should prompt an EEG. If microcephaly, seizures or abnormalities on neurological exam are present, an MRI can be considered. A chromosomal microarray should be offered for any children with a developmental disability, dysmorphic features, or congenital anomalies. Metabolic testing can be considered in the context of symptoms such as cyclic vomiting, developmental regression, or seizures. Blood lead levels should be reserved for when the child lives in a high-risk environment, exhibits pica symptoms, or has a developmental delay. Fragile X testing can also be considered given its overlapping behavioral symptoms with ASD, especially if there are typical dysmorphic features present on exam. These would include, for example: long ears, elongated face, prominent forehead or macroorchidism.

The fifth step is to consider the differential diagnoses and comorbidities of ASD. There are a number of genetic, neurodevelopmental, behavioral, mental, neurological and medical conditions whose symptoms may overlap with or mimic those present in ASD. Some of them we have already mentioned, but the list is quite extensive. They include but are not limited to ADHD, global developmental delay or intellectual disability, anxiety or depressive disorders, oppositional defiant disorder, cerebral palsy, and epilepsy. For a more complete list of conditions, please refer to Table two of our statement.

Step six would involve establishing the diagnosis, using the DSM 5 criteria along with clinical judgment to differentiate ASD from other developmental disorders. You should check out part one of our podcast series if you would like to review the diagnostic criteria for ASD.

In Aiden's case, his physical examination was all within normal limits. He has already had audiology and optometry assessments, both of which were within normal limits. He had high scores on the ADOS and parents reported significant concerns with his language development, play, and difficulties with transitions. You consider possible comorbidities and the differential diagnoses but believe the yield of pursuing further investigations would be too low in his case. Based on your history, physical exam, diagnostic tools and clinical judgment, you feel strongly that Aiden meets the diagnostic criteria for ASD based on the DSM 5 criteria. Given that you have consulted with the clinical psychologist in order to complete the ADOS, you have operated under the shared care model to determine this diagnosis.

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**What if the diagnosis wasn't as clear?**

In a case where the diagnosis is unclear, you can consider the following steps: gather information from other sources, observe the child in a different setting (home, day care), obtain a second opinion from a specialized ASD team, or conduct a repeat assessment after a certain period of time, for example after initiation of therapy or school entry. In cases where the child is very young and symptoms are more subtle, it is important to book a timely follow-up appointment or referral for further assessment.

**Once a diagnosis of ASD is made, how should you communicate the findings to Aiden's family? And what are the next steps for a pediatrician after a child has been diagnosed with ASD?**

Ideally, communicating your assessment findings should be done in a face to face appointment with parents and the child (if age appropriate). If not possible, a telephone or video conference should be arranged. Lone parents should be encouraged to bring a support person, and interpreters should be present if necessary.

Communicating a diagnosis of ASD entails a sensitive and supportive discussion. You can start by clearly stating, for example, that "Aiden meets DSM 5 criteria for a diagnosis of ASD". Give the family time to process the information, and then aim to go through a summary of what led to the referral and diagnosis of ASD. Describe how presenting symptoms or behaviors meet the DSM 5 criteria, and explain the diagnostic tool used, if any. Ask the family *what* questions they might have, as opposed to *if* they have questions. You should provide information both verbally and as part of a comprehensive written report, with language suitable for a lay audience.

You should also discuss any co-occurring conditions that have been identified or require further investigation, explaining the rationale behind each.

If the initial evaluation did not include a comprehensive needs assessment for treatment and intervention planning, arranging for this should be part of the clinician's role. This needs assessment may evaluate a series of domains including: cognitive/academic functioning; speech, language, communication skills; sensory and motor functioning, sensory sensitivities; adaptive functioning (self help skills); behavioural and emotional functioning; as well as physical health and nutrition.

This may involve advocating for further assessment, which is in turn influenced by the approach used for an ASD diagnostic evaluation. For approach one, where diagnosis is identified by a sole pediatric care provider, a referral to an interdisciplinary team or to multiple professionals may be required. For approach two, involving a second care partner, the latter may be able to provide additional information for intervention planning such as insight into cognitive functioning or language abilities. In Aiden's case, for example, the psychologist who completed his ADOS may be able to conduct a cognitive assessment to determine his baseline learning strengths and needs. Further referrals may only be needed to understand additional areas of functioning, like sensory or motor, if indicated. For approach three, additional assessments may not be necessary since the child is evaluated

by a multidisciplinary team and therefore an assessment for baseline intervention planning is likely to occur at the same time. In any of these approaches, individualized treatment planning may be conducted by the child's educational or treatment team.

When disclosing the next steps of management with the family, this includes going over the patient's current functioning level and the recommended supports to be put in place. Once again, these may consist of referrals for services, general intervention recommendations, or any additional assessments. Regardless of meeting diagnostic criteria, developmental concerns should be addressed with referrals for services or further assessments addressing the concerns in question.

The last step of the family meeting consists of providing resources for parents and/or other family members. They should be given information about ASD, support and parent advocacy groups, and funding opportunities. A follow up plan should be clearly stated.

After familiarizing yourself with the available resources, you schedule a follow up appointment with Aiden and his family to disclose his diagnosis and next steps. You explain the importance of a multilevel approach to care when it comes to ASD, and that a follow-up appointment has been made with the psychologist for a cognitive assessment. You provide them with handouts regarding ASD as well as local parental support groups. You suggest a referral to Social Work in order to explore funding and local service opportunities, and the family agrees. You also provide the family with the necessary information to gain access to applied behaviour analytic (ABA)-based services in the community. The family inquires about how to overcome his picky eating habits, and you explain they will likely explore that as part of the ABA program. If not, you will consider a referral to an occupational therapist in the future. The family is happy with the plan.

Based on this experience, you decide you will also pursue further training in using and interpreting ASD diagnostic tools.

**We will finish by discussing some final considerations in a diagnostic evaluation of ASD.**

Firstly, the importance of confirming or ruling out ASD as early as possible cannot be overstated. Although a definitive diagnosis for ASD is possible in children under two, this can be challenging given that symptoms may be subtle or less distinguishable from other delays, or even typical development! Those with a provisional ASD diagnosis will need a timely follow up evaluation or referral for further assessment since symptoms can substantially change during development, and many jurisdictions will not provide ASD-focused services without a confirmed diagnosis.

Secondly, clinicians should be aware that girls are probably under-diagnosed for ASD. Boys receive a diagnosis of ASD four times more frequently than girls, which may be due, at least in part, to gendered differences in symptom presentation. Evidence also suggests that girls may be better at camouflaging symptoms and use compensatory strategies to overcome social and communication difficulties.

Thirdly, we know that children from racial or ethnic minorities are diagnosed later than their peers in the general population. It's unclear whether this disparity is attributable to limited access to services, interpretation of symptoms by families, health-care system related factors, or a combination of reasons. Cultural factors may affect help-seeking behaviors and impact aspects of care.

Fourthly, children living in rural/remote communities are diagnosed with ASD later than peers in urban settings. Shared care between a local provider and an ASD specialist can be facilitated through telehealth and by travel clinics on a seasonal schedule.

This brings us to the end of this PedsCases podcast on "Standards of diagnostic assessment for autism spectrum disorder". Let's review a summary of our key points:

1. There are three main approaches for assessment and evaluation of ASD, depending on the pediatric care provider's clinical experience and judgment, the patient's complexity of symptoms, and family factors. These include a diagnosis made by a sole pediatric care provider, a shared care model, or a team model.
2. Formal diagnostic assessment tools such as the ADOS may complement a comprehensive ASD diagnostic assessment but should not be used alone.
3. A comprehensive ASD assessment should include a review of the patient's records; interviewing parents, family members, and other caregivers; an assessment for core features of ASD; a comprehensive physical exam; additional investigations; and consideration of differential diagnoses and co-occurring conditions.
4. Investigations should almost always include hearing assessments; further investigations should be tailored to your history and physical exam.
5. After diagnosis, a baseline needs assessment may be necessary in order to evaluate a series of developmental domains in your patient, depending on the approach taken. This may require further evaluation, referrals, or advocacy from the primary care provider. Specific, individualized treatment planning may be conducted by the child's educational or treatment team.
6. Regardless of meeting diagnostic criteria, developmental concerns should be addressed with referrals for services or further assessments addressing the concerns in question.

Thanks for listening! As you can see, a pediatrician or primary care provider plays a vital role when it comes to diagnosing and supporting a child with ASD. This also includes providing routine care, managing co-occurring medical conditions, and coordinating care. For more information on condition management and follow-up care of ASD, tune in to part three of our ASD podcast series.

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