Case Worksheet for Learners

Case Goal

The diagnosis of autism spectrum disorder (ASD) is made on the basis of detailed information obtained from the child's caregivers, careful observation and assessment of the child, and the use of standardized tools designed to aid in the diagnosis of ASD.

Key Learning Points of This Case

1. *Be familiar with the basic principles regarding the diagnosis and epidemiology of ASD.*
   a. Describe the two domains of impairment in ASD. _____
   _____
   _____

   b. Know the currently reported estimate of the prevalence of ASD in the U.S. _____
   _____
   _____

   c. Describe the current evidence for the genetic etiology of ASD. _____
   _____
   _____

2. *Understand the diagnostic approach to evaluating a child with suspected ASD.*
   a. Identify the important elements of a comprehensive history._____
   _____
   _____

   b. Recognize the important features to assess when observing a child's behavior._____
   _____
   _____

   c. List the aspects of the physical exam that are of particular importance when evaluating for ASD._____
   _____
   _____

   d. Describe the components of a comprehensive diagnostic evaluation for ASD._____
   _____
   _____

Post Learning Exercise

1. Observe and participate in a team evaluation for autism spectrum disorder. Specific activities could include taking a history, doing a physical exam, and observing an Autism Diagnostic Observation Schedule, Second Edition (ADOS-2) exam.
2. Attend a genetics clinic and observe genetic counseling for the family of a child with a genetic cause of ASD.
3. For some, it may not be possible to observe an autism evaluation. Suggest residents:
    - Talk to a parent of a child with ASD about getting a diagnosis.
    - Interview an attending about how they introduce concerns about ASD with families.

Case Study Part I

Billy is a 3 ½ -year-old boy you are seeing for the first time in your resident practice. He was born full term following a normal pregnancy and delivery. His newborn screen and neonatal hearing test were normal. At 18 months, he underwent bilateral myringotomy tube placement following repeated bouts of otitis media. His mother also had concerns that he was not speaking any words, and his doctor said, "Let's wait and see." A follow-up appointment was scheduled for when he turned 2. At his 2-year well-child check, because of the history of global developmental delay, he was referred to early intervention (EI) services.

Three months ago, Billy began attending a specialized preschool in the local school district; there, he receives speech, physical, and occupational therapies. This is the third preschool Billy has attended. He was asked to leave his previous schools because of behavioral problems. Billy's mother reports that his current preschool teacher recommended that she take him to the pediatrician given concerns about his behavior. Since he began preschool, his teachers have reported that he is extremely hyperactive, does not follow directions, and largely ignores the children in the class.

Billy's mother reports that he has been in good health recently. She tells you that all of his milestones were delayed, particularly his language. She denies any history of developmental regression. She adds that Billy has made some nice progress since he began receiving EI services. He will now use single words like "juice" and "cookie" to make requests. At home, Billy is a "handful," but he will usually calm down when she turns on his favorite cartoon. She describes him as a sweet and loving boy, but she also shares that he will have prolonged tantrums when denied what he wants and that tantrums will often occur when they are attempting to leave their house. During these tantrums, he will frequently bang his head and bite his hand, which frightens her. For some time now she has been concerned that Billy does not like to play with his similar-aged cousins during family get-togethers, but she thought this was because he was an only child and didn't like to share. She had been eagerly awaiting his first day of preschool so that he could spend more time around other children.

When asked about family history, Billy's mother reports that her sister's 7-year-old son is in special education, but she is not sure what kind of evaluation has been done as her sister "does not like to talk about these kinds of things." She mentions that it has been hard for the family, and she finds Billy's preschool reports to be embarrassing and discouraging. She is frustrated and wants the best for Billy.

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Case Study Part II

At first, Billy clings to his mother, but as you continue to speak with her, he climbs down from her lap and runs around the room touching everything in sight. When you give him a few toys to play with, he briefly inspects them, smells them, and then throws them down. Billy spots a ball in your bag of toys. He points at it and says "ball" without looking at you or his mother. When you toss him the ball, he laughs and makes brief eye contact with you, but then proceeds to kick and chase the ball around the room, ignoring your efforts to engage him in play. You call his name several times and he doesn't answer. When his mother begins to undress him for the exam, he begins to scream and jump up and down while flapping his hands. She says he tends to have difficulty transitioning from one activity to another. On physical examination, Billy is at the 75th percentile for weight and height, and his head circumference is at the 98th percentile. His ears are prominent, and he has a wide nasal bridge. He has mildly hyperextensible joints. The remainder of his exam is within normal limits.

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Case Study Part III

You share your concerns about the things Billy's mother has told you and about what you have observed during the visit. You tell her that Billy shows some characteristics of a child with autism spectrum disorder. He exhibits decreased eye contact, lack of joint attention, hand flapping, resistance to transitions, and atypical exploration of objects. You recommend she take Billy for additional evaluations. Together, you agree to proceed with a referral for a multidisciplinary evaluation for the presence of autism spectrum disorder, including an audiologic assessment. You ask Billy's mother to return for a follow-up appointment to ensure the evaluation is proceeding as it should.

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Case Study Part IV

Six months later, you review the results of Billy's evaluation with his mother. His hearing evaluation, consisting of behavioral audiometry and tympanogram testing, was normal. Billy's comprehensive evaluation included assessments by a developmental-behavioral pediatrician, a psychologist, a speech-language pathologist, and an occupational therapist.

The report from the developmental-behavioral pediatrician details Billy's medical history, developmental history, current behavior, and family history, as well as findings on physical and neurological examination. According to the report, Billy meets the DSM-5 criteria for a diagnosis of ASD. He has persistent deficits in social communication and social interaction across contexts and he has two or more repetitive and restrictive behaviors. According to the report he has a severity level of 2 for social communication and a severity level of 3 for restricted and repetitive behaviors and interests.

The report also mentions that blood was drawn at that visit for a karyotype and molecular test for fragile X, but the results are still pending as Billy only saw the specialist last week. The psychologist's report reviewed Billy's performance on cognitive testing and revealed below-age-level skills in all areas, with the most pronounced weakness in the language domain.

On formal language testing, Billy's receptive and expressive skills were at the 15-to 18-month level. The occupational therapist's report described Billy's strengths and weaknesses in fine motor skills and adaptive functioning and detailed some of his sensory issues that impacted his day-to-day life. All of the clinicians who evaluated Billy thoroughly described their observations of Billy's social interactions and behaviors during their sessions. As part of the comprehensive evaluation, Billy also underwent ADOS-2 (Autism Diagnostic Observation Schedule, Second Edition) testing, which supported the diagnosis of autism. Observations included Billy's wanting to play with a wind-up toy; he threw himself on the ground when the examiner tried to take the toy away. One clinician also mentioned that when his name was called, he did not answer.

Billy's mother informs you that she took a copy of the reports to Billy's school. A meeting has been scheduled to re-assess Billy's educational plan. She has also been reading some information on autism and is looking into a parent support group in her community. Billy's mother tells you that two other families they know have sons with ASD. She asks why there seem to be so many more children diagnosed now than in the past.

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Case Study Part V - Epilogue

A few days later, you receive a call from the developmental-behavioral pediatrician who evaluated Billy. He informs you that chromosomal testing was normal, but fragile X testing revealed 383 CGG repeats, consistent with a diagnosis of fragile X syndrome. He says that he has discussed the results of the test with Billy's mother and has scheduled an appointment with a geneticist to address the issue of testing other family members and to discuss possible enrollment in clinical trials.

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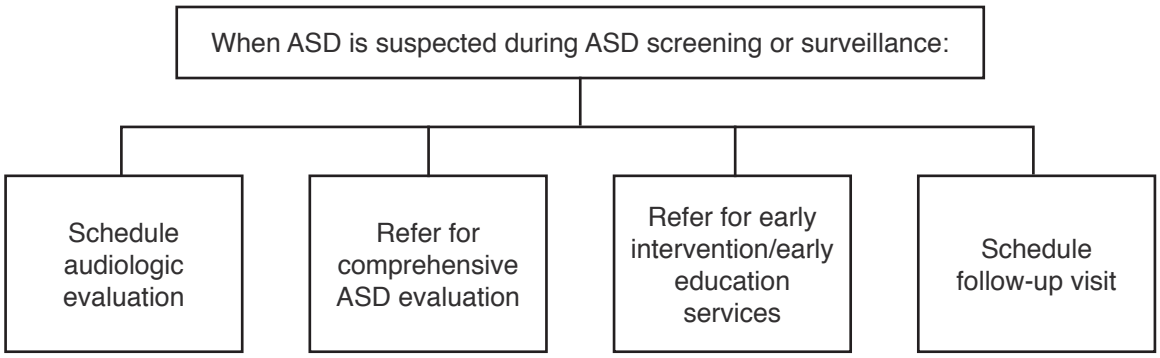
Handout I: Components of a Comprehensive Evaluation for ASD



- Detailed history from primary caregiver
- Medical history
  - Developmental history
  - Behavioral history
  - Family history
- Physical examination
  - Growth parameters, head circumference
  - General examination with special attention to skin and neurological findings
  - Inspection for any dysmorphic features
- Observation of child
  - Social interaction, response to name, joint attention
  - Play skills
  - Use of language
  - Presence of atypical behaviors or stereotypies
- Developmental/psychometric testing
- Speech/language testing
- Determination of categorical ASD diagnosis using DSM criteria and a standardized tool
  - Autism Observation Diagnostic Schedule, Second Edition (ADOS-2)
  - Autism Diagnostic Interview–Revised (ADI-R)
- Assessment of family's knowledge regarding ASD, coping skills, resources, and supports
- Medical/genetic workup as indicated by clinical picture
  - High-resolution chromosomes
  - Array CGH
  - Fragile X testing
  - Rett syndrome/MECP2 testing in girls
  - Additional tests to consider include: PTEN gene analysis, EEG, metabolic studies, neuroimaging

Macferran K, Major N, Fussell J, High P. Components of a Comprehensive Evaluation for ASD. Developed for the *Autism Case Training: A Developmental Behavioral Pediatrics Curriculum.* 2011.

Handout II: Differential and Etiologic Diagnosis of Autism Spectrum Disorder

Developmental delay/ intellectual disability	Children with intellectual disability may have "autistic features," but not meet criteria for autism spectrum disorder (ASD). Children with ASD may have intellectual disability or normal intelligence. Children with intellectual disability usually have better social and communication skills than do children with ASD with the same cognitive level.
Fetal alcohol spectrum disorders	There is an increased risk of ASD and other neurodevelopmental disorders in children exposed to alcohol in utero.
Genetic syndrome	There may or may not be a family history, depending on the specific disorder. If dysmorphic features are present, a genetic disorder should be considered. There are certain neurogenetic syndromes that tend to be associated with ASD. These include, but are not limited to: • Fragile X syndrome – intellectual disability, macrocephaly, large ears, large testicles, hypotonia, and joint hyperextensibility • Tuberous sclerosis – hypopigmented macules, central nervous system hamartomas, seizures, intellectual disability • Angelman syndrome – global developmental delay, hypotonia, wide-based ataxic gait, seizures, progressive spasticity • Rett syndrome – disorder seen primarily in girls. They have apparently normal development for the first 5 months of life and have a normal head circumference at birth. Deceleration of head growth is seen from 5 to 48 months of age resulting in microcephaly. They lose previously acquired hand skills and begin to have hand-wringing stereotypes. They often develop seizures.
Hearing Impairment	There may be a history of recurrent otitis media or fluid. Children with hearing impairments often have speech delays, but will typically use compensatory nonverbal forms of communication. They make eye contact and use facial expressions. Children with ASD may be described as having "selective hearing" (i.e., may not respond when their name is called, but are oversensitive to other noises). Children with a hearing impairment will usually be underresponsive to all noises, although this will be somewhat variable depending on the degree of hearing loss.

Handout II: Differential and Etiologic Diagnosis of Autism Spectrum Disorder

Mental health disorders	This is a broad category of differential diagnoses with variable symptomatology depending on the specific diagnosis. a. Obsessive compulsive disorder (OCD) – The obsessive thoughts and repetitive actions seen in OCD can appear very similar to the ritualistic behaviors and motor stereotypes seen in ASD. b. Anxiety – Children who have problems with anxiety may be hesitant to interact with others. They may have difficulties with transitions. Children with anxiety are still socially related and have appropriate social insight. c. Depression – Depression in children can present in a variety of ways. Children may be withdrawn and isolate themselves. They may have a blunted affect and avoid eye contact. d. Attention deficit-hyperactivity disorder (ADHD) – Children with ADHD may have impairments in their social skills due to their hyperactivity and impulsivity. They may have difficulty sustaining a conversation because of inattention. Children with ASD often have problems with hyperactivity, impulsivity, and inattention. e. Oppositional defiant disorder (ODD)/behavior problems – The behavior problems seen in children with ODD are usually intentional. Most children will have temper tantrums at some point. Children with ASD are more likely to have tantrums associated with transitions or "for no apparent reason." f. Tourette syndrome – Tics seen in Tourette syndrome may appear similar to motor stereotypes associated with ASD. Children with Tourette syndrome will usually not have the social or communication impairments seen with ASD. However, there may be some social isolation due to embarrassment or peer avoidance.
Psychosocial (e.g., neglect)	Children who have a history of significant abuse or neglect may be withdrawn and hesitant to interact with others. They may also have regression of skills, such as loss of language, and behavior problems.
Sensory problems	Children with ASD often have sensory issues such as being hypersensitive to loud noises or avoiding certain food textures. A child that has sensory impairments but is not on the autism spectrum will not have the core features of ASD (impairments in social communication, etc.).
Speech/language disorder	Children with speech/language disorders will compensate with nonverbal forms of communication such as pointing and gestures. They lack severe social deficits, although there may be some social impairment due to the communication difficulties.

Suggested Citation: Macferran K, Major N, Fussel J, High P. Differential and Etiologic Diagnosis of Autism Spectrum Disorder. Developed for the Autism Case Training: A Developmental-Behavioral Pediatrics Curriculum. 2011.

Handout III: AAP Screening Guidelines

Surveillance and Screening Algorithm: Autism Spectrum Disorders (ASDs)

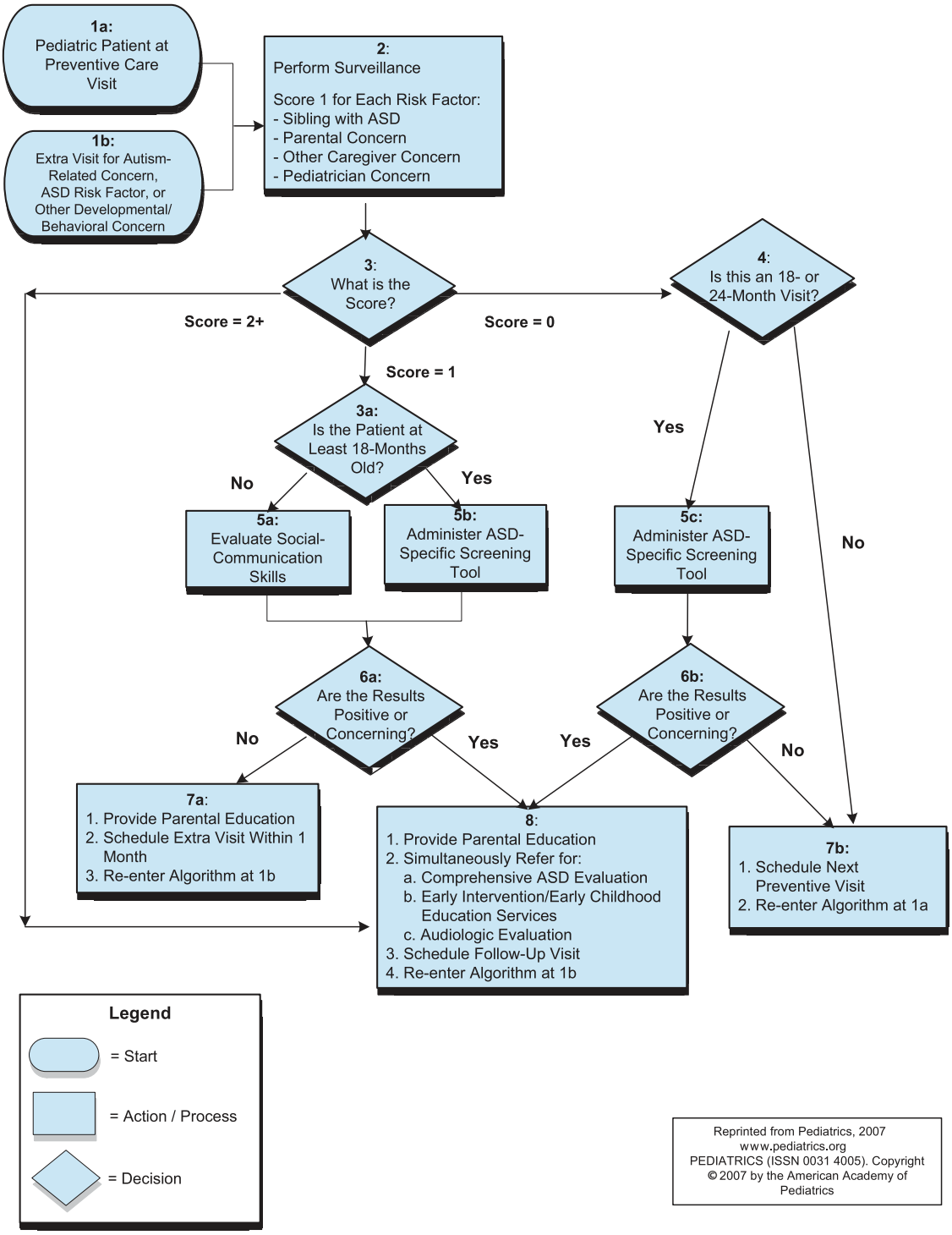


FIGURE 1
Surveillance and screening algorithm: ASDs.

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Handout III: AAP Screening Guidelines

Surveillance and Screening Algorithm: Autism Spectrum Disorders (ASDs)

1a: Pediatric Patient at Preventive Care Visit

1a - Developmental concerns, including those about social skill deficits, should be included as one of several health topics addressed at each pediatric preventive care visit through the first 5 years of life. *(Go to step 2)*

1b: Extra Visit for Autism-Related Concern, ASD Risk Factor, or Other Developmental/ Behavioral Concern

1b – At the parents' request, or when a concern is identified in a previous visit, a child may be scheduled for a "problem-targeted" clinic visit because of concerns about ASD. Parent concerns may be based on observed behaviors, social or language deficits, issues raised by other caregivers, or heightened anxiety produced by ASD coverage in the media. *(Go to step 2)*

2:
Perform Surveillance

Score 1 for Each Risk Factor:
- Sibling with ASD
- Parental Concern
- Other Caregiver Concern
- Pediatrician Concern

2 - Developmental surveillance is a flexible, longitudinal, continuous, and cumulative process whereby health care professionals identify children who may have developmental problems. There are 5 components of developmental surveillance: eliciting and attending to the parents' concerns about their child's development, documenting and maintaining a developmental history, making accurate observations of the child, identifying the risk and protective factors, and maintaining an accurate record and documenting the process and findings. The concerns of parents, other caregivers, and pediatricians all should be included in determining whether surveillance suggests that the child may be at risk of an ASD. In addition, younger siblings of children with an ASD should also be considered at risk, because they are 10 times more likely to develop symptoms of an ASD than children without a sibling with an ASD. Scoring risk factors will help determine the next steps. *(Go to step 3)*

For more information on developmental surveillance, see "Identifying Infants and Young Children With Developmental Disorders in the Medical Home: An Algorithm for Developmental Surveillance and Screening" (*Pediatrics* 2006;118:405-420).

3: What is the Score?

3 - Scoring risk factors:

- If the child does not have a sibling with an ASD and there are no concerns from the parents, other caregivers, or pediatrician: Score=0 *(Go to step 4)*
- If the child has only 1 risk factor, either a sibling with ASD or the concern of a parent, caregiver, or pediatrician: Score=1 *(Go to step 3a)*
- If the child has 2 or more risk factors: Score=2+ *(Go to step 8)*

3a: Is the Patient at Least 18-Months Old?

3a –

- If the child's age is <18 months, *Go to step 5a*
- If the child's age is ≥18 months, *Go to step 5b*

4: Is this an 18- or 24-Month Visit?

4 – In the absence of established risk factors and parental/provider concerns (score=0), a level-1 ASD-specific tool should be administered at the 18- and 24-month visits. *(Go to step 5c)* If this is not an 18- or 24-month visit, *(Go to step 7b)*.

Note: In the AAP policy, "Identifying Infants and Young Children With Developmental Disorders in the Medical Home: An Algorithm for Developmental Surveillance and Screening", a general developmental screen is recommended at the 9-, 18-, and 24-or 30-month visits and an ASD screening is recommended at the 18-month visit. This clinical report also recommends an ASD screening at the 24-month visit to identify children who may regress after 18 months of age.

5a: Evaluate Social-Communication Skills

5a - If the child's age is <18 months, the pediatrician should use a tool that specifically addresses the clinical characteristics of ASDs, such as those that target social-communication skills. *(Go to step 6a)*

5b: Administer ASD-Specific Screening Tool

5b - If the child's age is ≥18 months, the pediatrician should use an ASD-specific screening tool. *(Go to step 6a)*

5c: Administer ASD-Specific Screening Tool

5c – For all children ages 18 or 24 months (regardless of risk factors), the pediatrician should use an ASD-specific screening tool. *(Go to step 6b)*

AAP-recommended strategies for using ASD screening tools: *"Autism: Caring for Children with Autism Spectrum Disorders: A Resource Toolkit for Clinicians" (in press)**

6a: Are the Results Positive or Concerning?

6a – When the result of the screening is *negative*, *Go to step 7a*

When the result of the screening is *positive*, *Go to step 8*

6b: Are the Results Positive or Concerning?

6b – When the result of the ASD screening (at 18- and 24-month visits) is *negative, Go to step 7b*

When the result of the ASD screening (at 18- and 24-month visits) is *positive*, *Go to step 8*

7a:
1. Provide Parental Education
2. Schedule Extra Visit Within 1 Month
3. Re-enter Algorithm at 1b

7a – If the child demonstrates risk but has a negative screening result, information about ASDs should be provided to parents. The pediatrician should schedule an extra visit within 1 month to address any residual ASD concerns or additional developmental/ behavioral concerns after a negative screening result. The child will then re-enter the algorithm at 1b. A "wait-and-see" approach is discouraged. If the only risk factor is a sibling with an ASD, the pediatrician should maintain a higher index of suspicion and address ASD symptoms at each preventive care visit, but an early follow-up within 1 month is not necessary unless a parental concern subsequently arises.

7b:
1. Schedule Next Preventive Visit
2. Re-enter Algorithm at 1a

7b – If this is not an 18- or 24-month visit, or when the result of the ASD screening is *negative*, the pediatrician can inform the parents and schedule the next routine preventive visit. The child will then re-enter the algorithm at 1a.

8:
1. Provide Parental Education
2. Simultaneously Refer for:
   a. Comprehensive ASD Evaluation
   b. Early Intervention/Early Childhood Education Services
   c. Audiologic Evaluation
3. Schedule Follow-up Visit
4. Re-enter Algorithm at 1b

8 – If the screening result is *positive* for possible ASD in step 6a or 6b, the pediatrician should provide peer reviewed and/or consensus-developed ASD materials. Because a positive screening result does not determine a diagnosis of ASD, the child should be referred for a comprehensive ASD evaluation, to early intervention/early childhood education services (depending on child's age), and an audiologic evaluation. A categorical diagnosis is not needed to access intervention services. These programs often provide evaluations and other services even before a medical evaluation is complete. A referral to intervention services or school also is indicated when other developmental/behavioral concerns exist, even though the ASD screening result is negative. The child should be scheduled for a follow-up visit and will then re-enter the algorithm at 1b. All communication between the referral sources and the pediatrician should be coordinated.

AAP information for parents about ASDs includes: "*Is Your One-Year-Old Communicating with You?**" and *"Understanding Autism Spectrum Disorders.**"

*Available at www.aap.org

Handout IV: DSM-5 ASD Checklist

_____ **A.** Persistent deficits in social communication and social interaction across multiple contexts, as manifested by the following, currently or by history

- ☐ A1. Deficits in social-emotional reciprocity, ranging, for example, from abnormal social approach and failure of normal back- and- forth conversation; to reduced sharing of interests, emotions, or affect; to failure to initiate or respond to social interactions.
- ☐ A2. Deficits in nonverbal communicative behaviors used for social interaction, ranging, for example, from poorly integrated verbal and nonverbal communication; to abnormalities in eye contact and body language or deficits in understanding and use of gestures; to a total lack of facial expressions and nonverbal communication.
- ☐ A3. Deficits in developing, maintaining, and understanding relationships, ranging, for example, from difficulties adjusting behavior to suit various social contexts; to difficulties in sharing imaginative play or in making friends; to absences of interest in peers.

_____ **B.** Restricted, repetitive patterns of behavior, interests, or activities as manifested by <u>at least 2 of 4 symptoms</u> currently or by history

- ☐ B1. Stereotyped or repetitive motor movements, use of objects, or speech (e.g., simple motor stereotypies, lining up toys or flipping objects, echolalia, idiosyncratic phrases).
- ☐ B2. Insistence on sameness, inflexible adherence to routines, or ritualized patterns of verbal or nonverbal behavior (e.g. extreme distress at small changes, difficulties with transitions, rigid thinking patterns, greeting rituals, need to take same route or eat same food everyday).
- ☐ B3. Highly restricted, fixated interests that are abnormal in intensity or focus (e.g., strong attachment to or preoccupation with unusual objects, excessively circumscribed or preservative interest).
- ☐ B4. Hyper- or hyporeactivity to sensory input or unusual interest in sensory aspects of the environment (e.g., apparent indifference to pain/temperature, adverse response to specific sounds or textures, excessive smelling or touching of objects, visual fascination with lights or movement)

_____ **C.** Symptoms must be present in the early developmental periods (but may not become fully manifest until social demands exceed limited capacities, or may be masked by learned strategies in later life)

_____ **D.** Symptoms cause clinically significant impairment in social, occupational, or other important areas of current functioning. (minimum = level 1)

_____ Social Communication Severity Level (1, 2, or 3)

_____ Restricted Repetitive Behavior Severity Level (1, 2, or 3)

_____ **E.** These disturbances are not better explained by intellectual disability (intellectual development disorder) or global developmental delay.

Patient meets criteria for ASD
(criteria A-E satisfied)

Severity Level for ASD	Social Communication	Restricted Interests & Repetitive Behaviors
Level 3 'Requiring very substantial support'	Severe deficits in verbal and nonverbal social communication skills cause severe impairments in functioning, very limited initiation of social interactions, and minimal response to social overtures from others. For example, a person with few words of intelligible speech who rarely initiates interaction and, when he or she does, makes unusual approaches to meet needs only and responds to only very direct social approaches.	Inflexibility of behavior, extreme difficulty coping with change, or other restricted/repetitive behaviors markedly interfere with functioning inall spheres. Great distress/difficulty changing focus or action.
Level 2 'Requiring substantial support'	Marked deficits in verbal and nonverbal social communication skills; social impairments apparent even with supports in place; limited initiation of social interactions; and reduced or abnormal response to social overtures from others. For example, a person who speaks simple sentences, whose interaction is limited to narrow special interest, andwho has markedly odd nonverbal communication.	Inflexibility of behavior, difficulty coping with change, or other restricted/ repetitive behaviors appear frequently enough to be obvious to the casual observer and interfere with functioning in a variety of contexts. Distress and/or difficulty changing focus or action
Level 1 'Requiring support'	Without supports in place, deficits in social communication cause noticeable impairments. Difficulty initiating social interactions, and clear examples of atypical or unsuccessful response to social overtures of others. May appear to have decreased interest in social interactions. For example, a person who is able to speak in full sentences and engages in communication but whose to-and-fro conversation with others fails, and whose attempts to make friends are odd and typically unsuccessful	Inflexibility of behavior causes significant interference with functioning in one or more contexts. Difficulty switching between activities. Problems of organization and planning hamper independence.

American Psychiatric Association. Pervasive developmental disorders. In: Diagnostic and Statistical Manual of Mental Disorders. 5th ed.-text revision (DSM-5). Washington, DC: American Psychiatric Association; 2013.

Facts About Fragile X Syndrome

What is fragile X syndrome?

Fragile X syndrome (FXS) is the most common known cause of intellectual disability (formerly referred to as mental retardation) that can be inherited, that is passed from parent to child. It is estimated that FXS affects about 1 in 4,000 boys and 1 in 6,000 to 8,000 girls. Both boys and girls can have FXS, but girls usually are more mildly affected.

What causes FXS?

The cause of FXS is genetic. FXS occurs when there is a change in a gene on the X chromosome called FMR1. The FMR1 gene makes a protein needed for normal brain development. In FXS, the FMR1 gene does not work properly. The protein is not made, and the brain does not develop as it should. The lack of this protein causes FXS. Other Fragile X-associated Disorders (FXDs) can be present in the extended family, even if not currently evident. Talk with a genetic counselor for more information.

What are some signs of FXS?

Children with FXS might:

- Sit up, crawl, or walk later than other children
- Have trouble with learning and solving problems
- Learn to talk later, or have trouble speaking
- Become very anxious in crowds and new situations
- Be sensitive about someone touching them
- Bite or flap their hands
- Have trouble making eye contact
- Have a short attention span
- Be in constant motion and unable to sit still
- Have seizures

Some children with FXS have certain physical features such as:

- A large head
- A long face
- Prominent ears, chin, and forehead
- Flexible joints
- Flat feet
- Macroorchidism (enlarged testicles in males; more obvious after puberty)

These physical features tend to become more noticeable as the child gets older.

What conditions are common among children with FXS?

Children with FXS might have learning disabilities, speech and language delays, and behavioral problems such as attention-deficit/hyperactivity disorder (ADHD) and anxiety. Some boys can develop aggressive behavior. Depression can also occur. Boys with FXS usually have a mild to severe intellectual disability. Many girls with FXS have normal intelligence. Others have some degree of intellectual disability, with or without learning disabilities. Autism spectrum disorder (ASD) occurs more often among children with FXS.

What can I do if I think my child has FXS?

Talk with your child's doctor or nurse. If you or your doctor thinks there could be a problem, the doctor can order a blood test for FXS or refer you to a developmental specialist or geneticist, or both. Also, contact your local early intervention agency (for children younger than 3 years of age) or public school (for children 3 years of age or older) to find out if your child qualifies for intervention services. To find out whom to call in your area, contact the Center for Parent Information and Resources at **www.parentcenterhub.org/find-your-center/**.

In addition, CDC has links to information for families at **www.cdc.gov/fragilex**.

Additional resources include the National Fragile X Foundation **(www.fragilex.org)** and the FRAXA Research Foundation **(www.FRAXA.org)**. CDC also supports the efforts of the Fragile X Clinical & Research Consortium **(www.FXCRC.org)** which can be reached through the National Fragile X Foundation.

While there is no cure for fragile X syndrome, therapies and interventions can improve the lives of those affected and of their families. It is **very** important to begin these therapies and

Interventions as early as possible to help your child reach his or her full potential. Acting early can make a real difference!

1-800-CDC-INFO | www.cdc.gov/fragilex

National Center on Birth Defects and Developmental Disabilities
Division of Human Development and Disability

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