Assessing Autism in Primary Care

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Objectives:

1. Name assessment tools for primary care providers to screen for autism
2. Identify key features of autism
3. Use parent resource guides for families

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Primary Care

1st Contact

Role of PCP

- Early identification of autism spectrum disorder (ASD)
- Routine health maintenance
- Preventive care and care coordination for children with ASD
- Provision of support, guidance, and advocacy for families of children with ASD

CDC: Learn the Signs

"ALARM"

- Autism is prevalent (1/68)
- Listen to parents
- Act early
- Refer
- Monitor

Concerns parents may raise:

- No big smiles or warm, joyful expressions by 6 months
- No back-and-forth sharing of sounds, smiles or other facial expressions by 9 months
- No babbling or back and forth by 12 months
- No meaningful, two-word phrases (not including imitating or repeating) by 24 months
- Any loss of speech, babbling or social skills at any age

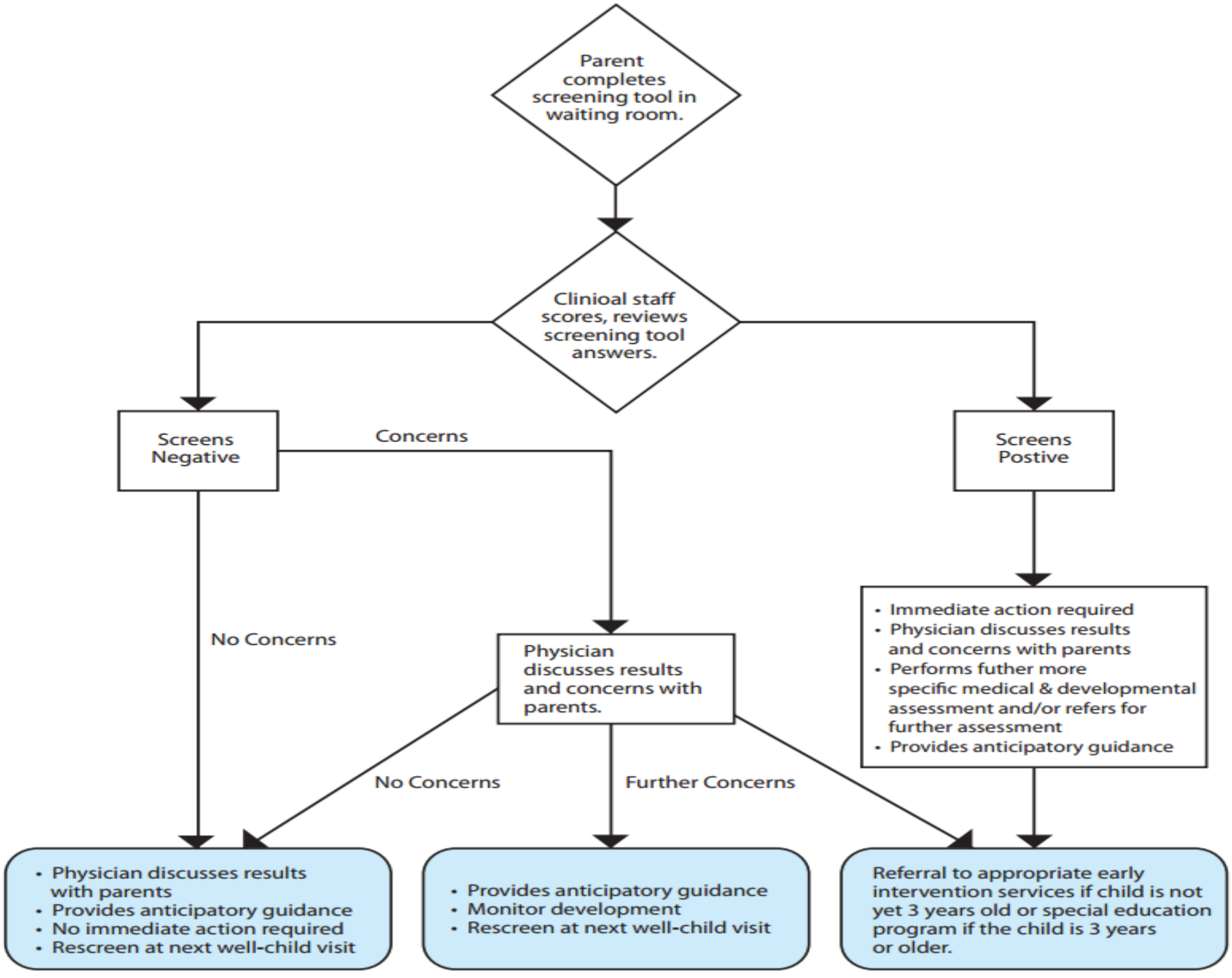
Screening for ASD

- AAP recommendations
  - Developmental Surveillance at all well child visits
  - Standard Developmental Behavioral Screening at 9, 18, and 30 (or 24) month well child visits
  - Screening for autism at 18 and 30 (or 24) month well child visits (MCHAT)

American Academy of Pediatrics Council on Children With Disabilities, Section on Developmental Behavioral Pediatrics, Bright Futures Steering Committee and Medical Home Initiatives for Children With Special Needs Project Advisory Committee. Identifying infants and young children with developmental disorders in the medical home: an algorithm for developmental surveillance and screening. Pediatrics. 2006;118(1):405–420; Johnson CP, Myers SM, and American Academy of Pediatrics Council on Children With Disabilities. Identification and evaluation of children with autism spectrum disorders. Pediatrics. 2007;120(7):1183–1215.

Video Observation - Nathan & Ben

Pediatric Developmental Screening Flowchart



Screening Tools

Ages & Stages Questionnaires, Third Edition (ASQ-3) parent-centric, ages of one month to 5 ½ years

PEDS Parents Evaluation of Developmental Status: ages 0 to 8 years, 5 minutes for parents to complete, 1-2 minutes to score, multiple languages, elicits parents' concerns

M-CHAT-R

- 23 yes-no questions, 5 min
- Measures social reciprocity, language, some motor
- 18 – 30 months
- Detects ASD, language impairment, mental retardation
- Free download: https://www.m-chat.org

M-CHAT-R

“If you point at something across the room, does your child look at it?“

“Does your child play pretend or make-believe?“

“If something new happens, does your child look at your face to see how you feel about it?”

Video Observation - Joint Attention

Surveillance of ASDs

- Direct observation/interactions/“clinical probes”
  - Pointing and directing child to “look” (12–15 months)
  - Calling child’s name (12 months)
  - Asking “Where’s mommy?”
  - Eye contact, gaze referencing, pointing—evidence of joint attention
  - Observe for unusual movements (looking at things closely, out of corner of eye, repetitive movements, sensitivities)

Delayed Diagnosis

- Mean interval b/w parental concerns and seeking professional help is ~ *6 months*
- Average time between fist medical attention and diagnosis is *13 months*
- Average age of diagnosis *4 yrs*
- Earlier: significant language delay and atypical behaviors, such as hand flapping and toe walking
- Later: less severe symptoms, lack of continuity of care, hearing impairment, oversensitivity to pain, living in a rural area, Hispanic ethnicity, and lower socioeconomic status

Social/communication

Persistent deficits in social communication and social interaction across contexts, not accounted for by general developmental delays, manifested by all of the following:

- Deficits in social-emotional reciprocity
- Deficits in nonverbal communicative behaviors
- Deficits in developing and maintaining relationships appropriate to the developmental level

Restricted Interests

- Restricted, repetitive patterns of behavior, interests, or activities as manifested by at least 2 of the following:
  - Stereotyped or repetitive speech, motor movements, or use of objects
  - Excessive adherence to routines
  - Highly restricted, fixated interests that are abnormal in intensity or focus
  - Hyper- or hypo-reactivity to sensory input or unusual sensory interests

Severity levels for Autism Spectrum Disorder

- Level 1: "Requiring Support"
- Level 2: "Requiring Substantial Support"
- Level 3: "Requiring Very Substantial Support

Importance of Assessment

- Diagnosis
  - Emphasis on individual profiles, not just the diagnosis
  - However, importance of diagnosis in order to obtain services
- Access to Services
  - School: Educational Classification -- IDEA categories
  - Government Agencies: Department of Developmental Services (Formerly DMR) REGIONAL CENTER
- Treatment/Intervention
  - Assessment first step toward developing treatment goals and intervention planning

Diagnostic Tools

- Autism Diagnostic Observation Schedule (ADOS-2) – gold standard for diagnosis of autism
- Autism Behavior Checklist –57 questions completed by parent or teacher
- Autism Diagnostic Interview-Revised (ADIS)
- Childhood Autism Rating Scale - 15-item direct-observation instrument


Core Clinical Features
Associated Neurological Symptoms
Associated Physical Symptoms
Sleep Deficits
Mood
Social Communication Deficits
Intellectual Disability
Anxiety
Attention
Language Disability
Repetitive Behaviors
Hyperactivity
Tantrums
Irritability
Seizures
Self-Injury
Aggression
GI Disorders
Immune Dysfunction

What Else Can Look Like ASD?

- Global developmental delay/intellectual disability
- Social (pragmatic) communication disorder
- Developmental language disorder
- Language-based learning disability
- Hearing impairment
- Landau-Kleffner syndrome
- Rett disorder Severe early deprivation/reactive attachment disorder
- Attention-deficit/hyperactivity disorder (ADHD) plus anxiety
- Nonverbal learning disability
- Anxiety plus language delay (with/without sensory issues)
- Cognitive delay plus anxiety
- Obsessive-compulsive disorder

- http://www.cdc.gov/ncbddd/actearly/act.html

Epidemiology

- "True" increase or "epidemic?"
- Increased awareness, broader diagnostic criteria, diagnostic substitution…
- Probably a little of both…

Etiology

- Unknown
  - Initially described as a disorder of parenting/“refrigerator mothers”
- Neurobiologic disorder
- Gene/environment interaction

Further work-up

- Labs: Chromosomal microarray (CMA) and DNA analysis for fragile X
- Hearing Test
- Metabolic testing, lead, Neuroimaging, EEG only if clinically indicated

Treatment: Getting Started

- School IEP/504
- Speech/Language Therapy: social stories
- Occupational Therapy
  - Sensory Integration
- Behavior Management
- Early Intervention/Pre-School/School-Age

Resources for parents

- Tool Kit https://www.autismspeaks.org/family-services/tool-kits
- U of Washington Autism Center for parents: "My Next Steps: A Parent's Guide to Understanding Autism." http://depts.washington.edu/uwautism/resources/autism-resource-dvd.html
  - Government Resources: www.nichd.nih.gov/autism; www.nimh.nih.gov/publicat/autism.pdf
  - Community Resources: e.g., Autism Spectrum Resource Center (www.ct-asrc.org)
- Behavioral therapy: http://www.ucdmc.ucdavis.edu/ddcenter/adept.html

Common Comorbid Disorders

- Eczema
- Allergies
- Asthma
- Ear and respiratory infections
- Gastrointestinal problems
- Severe headaches/migraines
- Seizures

Common Comorbid Disorders

- ADHD
- Anxiety
- Increased risk of schizophrenia and bipolar disorder

Common sources of pain and discomfort include:

- Headache l Earache
- Toothache l Sore Throat
- Sensory hyper-responsitivity: hyperacusis, tactile defensiveness, sensitivity to light
- Covering ears with hands
- Teeth grinding

Comorbid Illness?

- Constant eating/drinking/ swallowing
- Facial grimacing, wincing, tics (frequent clearing of throat,
- Mouthing behaviors, chewing on clothes
- Tapping behavior: finger tapping on throat
- Sobbing ‘for no reason at all/ vocal expressions of moaning, groaning, sighing, whining
- Agitation: pacing, jumping up and down*
- Blinking, sudden screaming, spinning and fixed look
- Repetitive rocking or other new repetitive movement
- Walking on toes
- Heightened anxiety and/or avoidance behaviors
- Increase in self-injury

Treatment: Medical

- Regular follow-up recommended
  - Monitoring, screening of common medical issues
    - Sleep issues, GI issues, nutritional concerns
  - Medication management
- Medications
- Alternative medical treatments

Alternative Treatments

- Diet
  - Gluten and casein free diet
- Vitamins
  - B6, Mg, B12, Omega-3 fatty acids, Zn
- Yeast treatments
- Manipulation
- Secretin, intravenous immune globulin (IVIG), steroids, antivirals, chelation

Autism and Vaccines

- Currently no evidence to support a relationship between vaccines and autism
- Theories continue to change
- Recent retraction of Lancet article
- Families still “caught in the middle” between research and media

Maintenance Treatment PCP

- A "practice" visit to familiarize the patient with the office setting, staff, and routine
- Having the family use a "social story" or visual topic board reviewing the expected parts of the visit, to help the child understand what to expect
- Allowing enough time to take the history and talk with the parent or caregiver before the examination
- Allowing the child to manipulate instruments and materials
- Keeping instructions and language simple, not abstract
- Using visual cues and supports
- Having family and/or familiar staff available
- Minimizing portions of the examination that might be overwhelming or overstimulating

Prognosis

- Previous studies summarized:
  - 10–15% with good outcomes
  - 15–25% with fair outcomes
  - 15–25% with poor outcomes
  - 30–50% with very poor outcomes
- Few current longitudinal studies exist
- Issues of diagnostic “shift,” diagnostic stability

Better Prognosis with

- Presence of joint attention
- Functional play skills
- Higher cognitive abilities
- Decreased severity of ASD symptoms
- Early identification
- Involvement in intervention
- A move toward inclusion with typical peers

Poorer Prognosis with

- Lack of joint attention by four years of age
- Lack of functional speech by five years of age
- IQ <70
- Seizures or other comorbid medical or neurodevelopmental conditions
- Severe ASD symptoms

Summary

- “ALARM”
  - Autism is prevalent (1/68)
  - Listen to parents
  - Act early (surveillance and screening)
  - Refer
  - Monitor

Resources

- Autism Society of America – www.autism-society.org
- Autism Speaks – 100 Day Kit – www.autismspeaks.org
- Centers for Disease Control and Prevention – www.cdc.gov
- American Academy of Pediatrics – www.aap.org