American Academy of Pediatrics

FETAL ALCOHOL SPECTRUM DISORDERS: SCREENING, ASSESSMENT AND DIAGNOSIS

Presented by

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DEDICATED TO THE HEALTH OF ALL CHILDREN™

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Disclosure Statement

- I have no financial disclosures related to this topic.
- I will not discuss off label drugs in this talk.
- I am an FASD Champion for AAP District VII

(of course it is not a paid position☹)

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Learning Objectives

1. Describe diagnostic criteria and approaches for conditions along the continuum of FASDs, including ARND, ND-PAE, FAS, pFAS, and ARBD.
2. Distinguish major physical and neurobehavioral features for differential diagnosis of FASDs from other behavioral and genetic disorders as well as relevant comorbidities.
3. Explain the importance of screening every patient for a history of prenatal alcohol exposure at birth and during their first visit to your practice and subsequent visits as appropriate.

FASD Terminology and Acronyms

- **PAE: Prenatal Alcohol Exposure**
- **FASD: Fetal Alcohol Spectrum Disorders**
  - **U**mbrella term for a range of effects that can result from prenatal alcohol exposure (**not** a diagnosis)
  - Encompasses a group of specific disorders (more on these later), including:
- **FAS: Fetal Alcohol Syndrome**
  - The most widely known diagnosis in the spectrum
- **ARND: Alcohol Related Neurodevelopmental Disorder**
  - Used in some dx schemes for individuals without physical characteristics
- **ND-PAE: Neurobehavioral disorder associated with prenatal alcohol exposure.**
  - New category in DSM-5 Section III: Emerging Measures and Models
  - Defines more precisely the developmental and behavioral manifestations associated with PAE

The Umbrella of FASD

Fetal Alcohol Syndrome (FAS)

Partial Fetal Alcohol Syndrome (pFAS)

Alcohol Related Birth Defects (ARBD)

Alcohol Related Neurodevelopmental Disorder (ARND)

Neurobehavioral Disorder-associated with Prenatal Alcohol Exposure (ND-PAE)

DIAGNOSES RESULTING FROM IN UTERO EXPOSURE TO ALCOHOL

Fetal Alcohol Syndrome (FAS) & Partial FAS (pFAS) - Facial features, CNS dysfunction/ anomalies, possible growth deficits

ARBD - Congenital anomalies only (rare)

ARND – Neurodevelopmental/behavioral effects without cardinal dysmorphic features (common)

ND-PAE – Neurodevelopmental/behavioral effects regardless of dysmorphic features (most likely common)

Prenatal Alcohol Exposure: Relevance to Pediatric Practice

- Most common *preventable* cause of intellectual disability and behavior problems – likely seen in most practices.
- Effects can be *lifelong*.
- Effect development and function more so than other drugs or teratogens.

"Of all the substances of abuse, including cocaine, heroin, and marijuana, alcohol produces by far the most serious neurobehavioral effects in the fetus."

Institute of Medicine, 1996

- Can contribute to a range of growth deficits and structural anomalies (FASD)

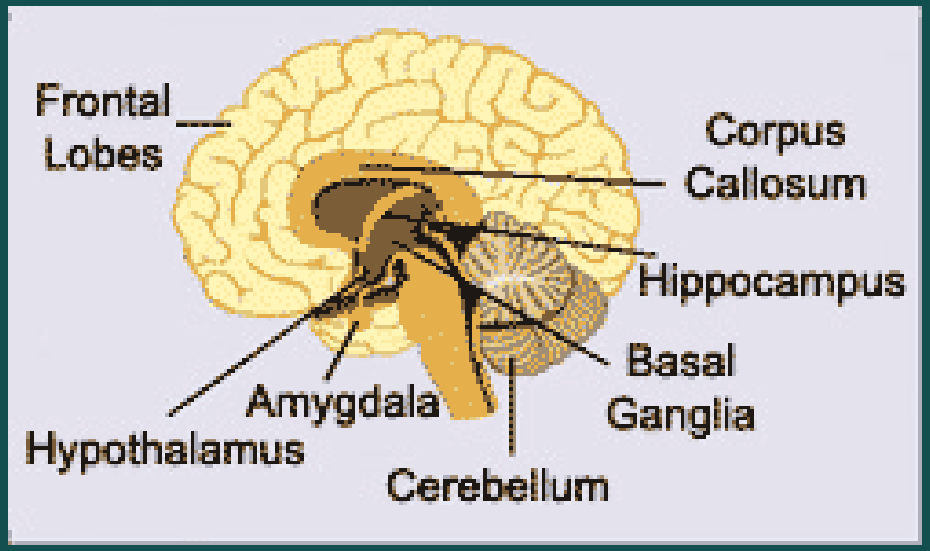
When to Consider a FASD Diagnosis?

- Developmental, cognitive, or behavioral concerns
- Complex medical concerns (e.g., cardiac)
- Growth deficits
- History of maternal alcohol or drug use
- A sibling diagnosed with a FASD
- Dysmorphic facial characteristics associated with FAS are present

Areas of the Brain Affected By Prenatal Alcohol Exposure

Frontal Lobes – impulses and judgment; controls executive function

Hypothalamus - appetite, emotions, temperature, and pain sensation



Corpus Callosum- passes information from the left brain (rules, logic) to the right brain (impulse, feelings) and vice versa.

Hippocampus – memory, learning, emotion

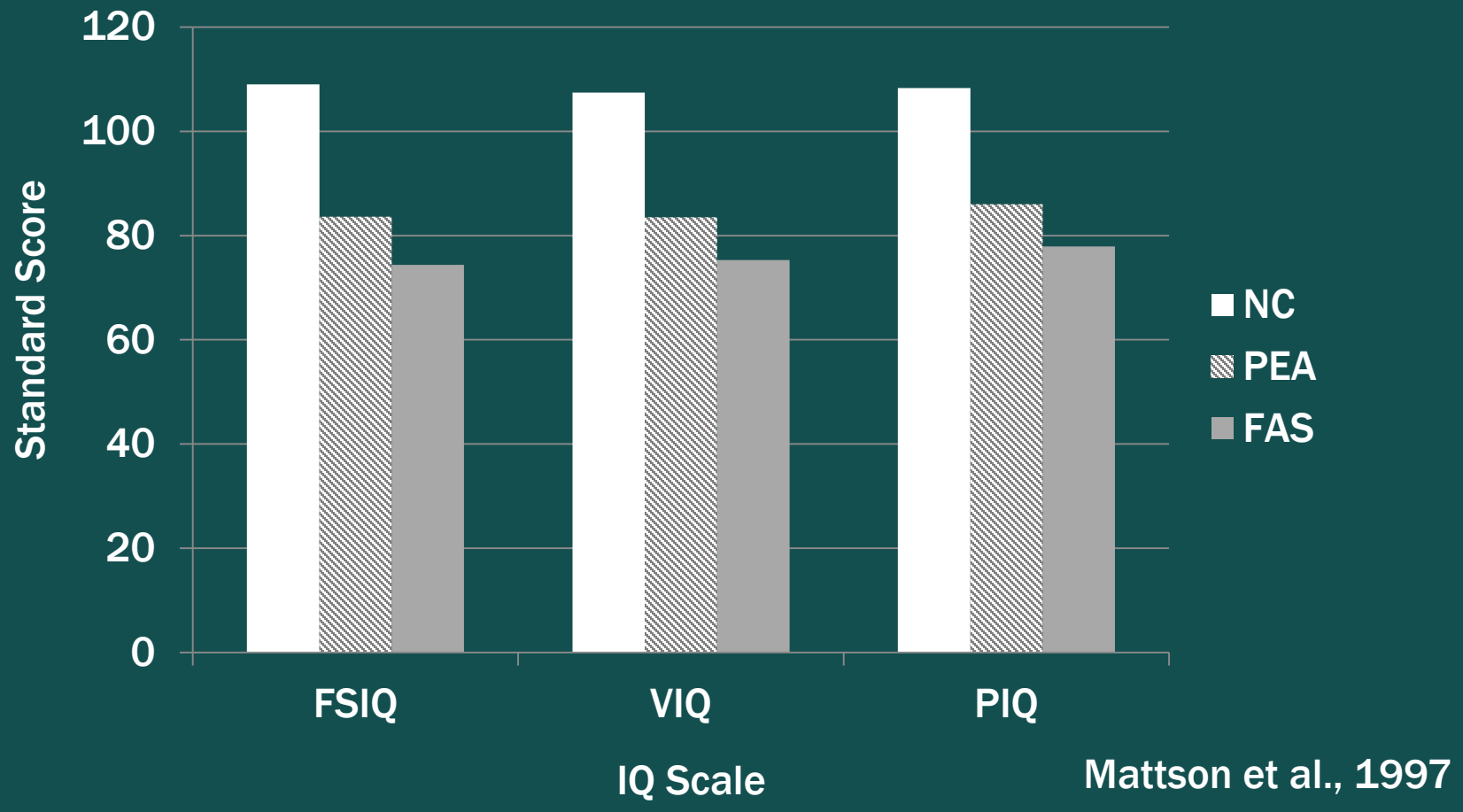
Amygdala - emotions

Cerebellum - coordination and movement

Basal Ganglia - spatial memory, switching gears, working toward goals, predicting behavioral outcomes, and the perception of time

Source: Dr. Sarah Mattson, University of San Diego

General Intellectual Performance



FASD: Relevance to Pediatric Practice *High Prevalence*

- Prevalence in a Midwestern city (May, 2014)
  - FAS: 6-9/1000 children
  - All FASD: 24-48 /1000 children (2.4% to 4.8%)
- Increased prevalence among children in child welfare (Lange, 2013)
  - FAS: 60/1000 children (6%)
  - All FASD: 169/1000 children (16.9%)

FASD: Perspectives on Prevalence

Birth defect	Prevalence
Down syndrome	1.2/1000 births
Cleft lip +/- palate	1.2/1000 births
Spina bifida	1/1000 births
Autism	12.5-14/1000*
Fetal Alcohol Syndrome (FAS)	6-9/1000*
All FASDs	24-48/1000* (May, 2014)

*per 1000 school age children

Potential Benefits of a Diagnosis

- Parental relief in the knowledge that the child's problems have a biological basis
- Facilitates access to evidence-based interventions
- Avoids unnecessary additional testing and non-specific referrals or interventions
- Discussing the cause with a biological mother may reduce recurrence risk in future offspring

Why Pediatricians Do Not Routinely Screen for FASD

- Insufficient information and training
- STIGMA: Stereotypic views about mothers who might be exposing their baby to alcohol leading to prejudice, discrimination, and bias AND/OR stereotypic view about children with an FASD

Assessment Domains for Diagnosis

- Prenatal Exposure to Alcohol History
- CNS (structural, neurologic, functional)
- Growth
- Dysmorphic Facial Features

Exposure History in Context of Routine Care

- Obtaining a history of in utero exposure to alcohol before suspicion or recognition of problems is ideal
- Introduce review of family, social, and pregnancy histories as part of assessment
- Work through all potential exposures: any of parent concern, medications, environmental, recreational drugs and alcohol
- Present alcohol as commonly consumed before pregnancy is recognized and ask about that timeframe

Exposure History in Context of Routine Care

- Use open-ended questions,

  EXAMPLE: “tell me about your alcohol use in the 3 months prior to finding out you were pregnant.”

  NOT: “You didn’t drink while pregnant – Did you?”

- Ask about partner’s drinking habits

- Provide assurance that to provide the best care possible it’s important to know all of the facts about the pregnancy

AAP’s Bright Futures

- *AAP’s Bright Futures*, guidelines for health supervision of infants, children and adolescents, suggest three screening questions for the pediatric situation:
  - How often do you drink beer, wine or liquor in your household?
  - In the 3 months before you knew you were pregnant, how many times did you have 4 or more drinks in a day?
  - During your pregnancy, how many times did you have 4 or more drinks in a day?

If a positive response is obtained additional questions about amount, frequency and timing may be appropriate for diagnostic purposes.

- Suggested contact points:
  - all prenatal visits,
  - the earliest well child visits,
  - all new patient visits,
  - whenever a related concern is observed or raised.

Conversations with Mothers: Practice Compassion (NOFAS.org)

- **Mothers want their pediatrician to value and like them.** Be gentle, non-judgmental, ask, then listen to their story
- Stick to the facts, provide information and education on alcohol/drug use or dependency in matter of fact manner
- Fear of judgment is one of the main reasons women may not disclose that they drank during pregnancy.
- Remind her that you care about her child, HER and their family. Share relevant stories when possible.
- Avoid words like "FASD Kid" rather use person first language such as "child with an FASD"

Best Definition of Cognition and Behavior in FASD: ND-PAE

- DSM-5 emerging diagnosis
- Criteria describes impairment in *neurocognition, self-regulation and adaptive function*,
- Diagnosis is made in the context of confirmed prenatal alcohol exposure,
- Criteria do not require the presence of physical features

Neurobehavioral Effects

Neurocognitive deficits

- Low IQ or developmental delay
- Executive functioning deficits
- Impaired learning, memory or specific learning problems (esp. visual-spatial and math)
- Motor functioning delays for younger children

Neurobehavioral Effects

Self-regulation problems

- Self-soothing, sleep
- Difficulty managing mood
- Behavior management issues
- Attention problems (esp. shifting attention)
- Poor impulse control

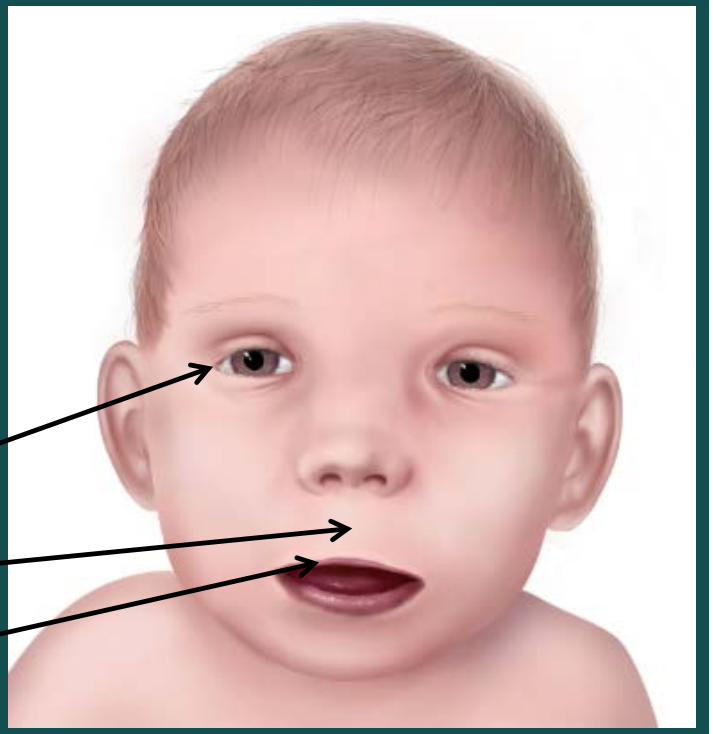
Neurobehavioral Effects

Difficulty learning/Delayed adaptive skills

- Communication deficits, especially social communications such a understanding idioms or jokes
- Problems with social skills
- Problems with self care or daily living skills
- Motor issues in younger children

Physical Effects

- Weight and/or length growth deficiency (pre or post natal)
- Abnormal brain structures (esp. small cranium, corpus callosum)
- Dysmorphic facial features:
  - Short palpebral fissures
  - Smooth philtrum
  - Thin vermillion border



FASD Diagnostic Schema Available

Currently available guidelines:

- Updated Clinical Guidelines for Diagnosing Fetal Alcohol Spectrum Disorders (Hoyme et al, *Pediatrics*, 2016)
- Canadian guidelines for diagnosis (Cook et al, *CMAJ*, 2015)
- National Task Force on FAS and FAE (2004)
- FASD 4-digit diagnostic code (Astley and Clarren, *Alcohol*, 2000)

Historically available guidelines:

- A practical clinical approach to diagnosis of fetal alcohol spectrum disorders: clarification of the 1996 Institute of Medicine criteria (Hoyme et al, Pediatrics, 2005)
- Fetal alcohol spectrum disorder: Canadian guidelines for diagnosis (Chudley et al, CMAJ) 2005
- Fetal Alcohol Syndrome (The Lancet, 1973)

The Diagnostic and Statistical Manual version 5 published by the American Psychiatric Association also proposes criteria for neurobehavioral disorder associated with prenatal alcohol exposure.

Spectrum of FASD

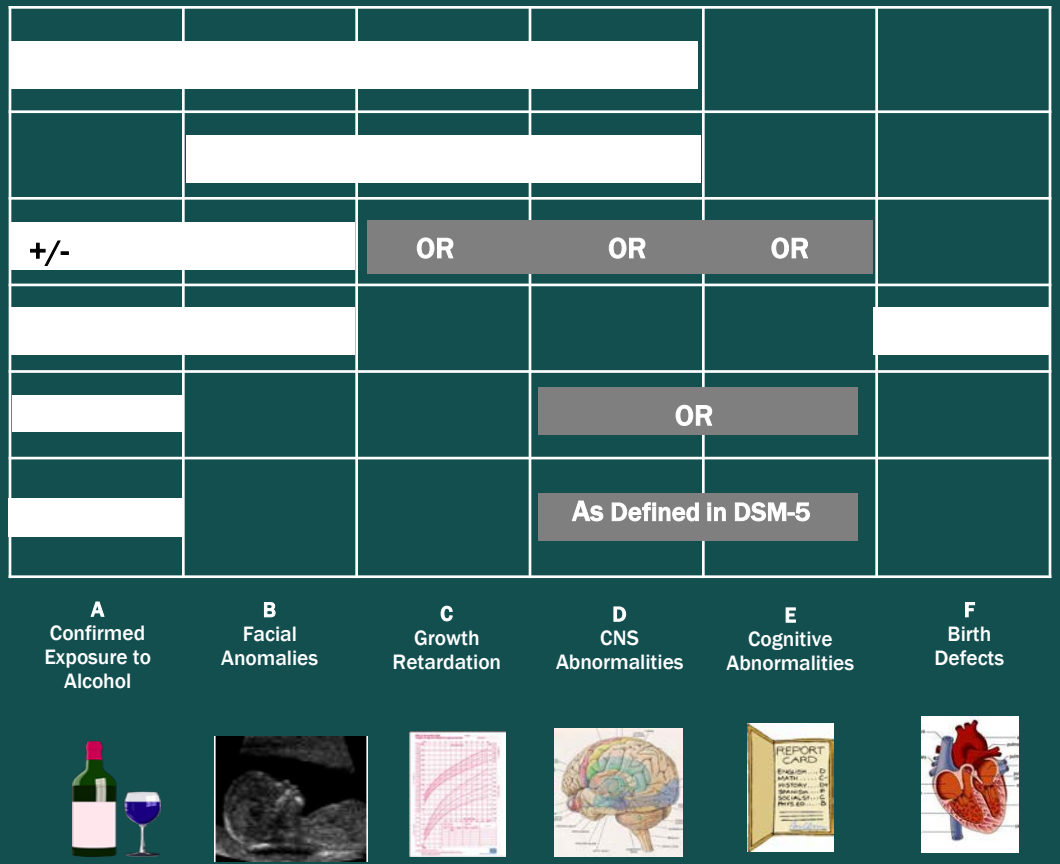
FAS
- FAS with confirmed maternal exposure
- FAS w/out confirmed maternal exposure

PFAS → Partial FAS with or w/o confirmed exposure

ARBD → Alcohol-related birth defects (ARBD)

ARND → Alcohol-related neurodevelopmental disorder (ARND)

ND-PAE → Neurobehavioral disorder associated with prenatal alcohol exposure (ND-PAE)



Adapted from *Neuroscience and Biobehavioral Reviews (2007); 31:230-238*
PEDIATRICS Vol. 106 No. 2 August 2000

Record Review & History

- Exposure History
- Birth records (weight, length, head circumference)
- Medical history/records (birth defects?, exposures?)
- Postnatal growth records
- Developmental/behavioral history
- Psychological testing, including cognitive and behavioral assessments

PATHWAY TO A DIAGNOSIS

- The AAP FASD Toolkit (www.aap.org/fasd) is a comprehensive resource for identification, diagnosis and medical home management on patients with a FASD
- Toolkit includes information on common diagnostic approaches and tools, a flow diagram for evaluation of FASDs, and guidelines for referral and diagnosis.

Potential Differential & Comorbid Diagnoses

Behavioral disorders examples

- ADHD
- Intellectual disability
- Early Trauma
- Conduct disorder/Oppositional defiant disorder
- Parenting issues

Potential Differential & Comorbid Diagnoses

Genetic and growth disorders examples

- Williams syndrome
- Dubowitz syndrome
- Fetal valproate syndrome
- Maternal PKU fetal effects
- Nutritional insufficiency – growth
- Prenatal smoking - growth

Delivering the Diagnosis

- Approach should be similar to delivering any diagnoses that may have significant impact on a child's future.
- Sensitivity to the possibility of stigma is critical, particularly when the biological mother is involved.
- Explanation of how the diagnosis leads to treatment options and better parent-child interactions is helpful.
- Only the FAS diagnosis can be made without confirmed in utero exposure to alcohol – for other dx's revisit as appropriate

Diagnosis Established
What now?

- Counsel re: Natural history risk of neurodevelopmental manifestations over time, need to intervene to avoid secondary conditions.
- Medical Home: http://www.aafp.org/practice-management/transformation/pcmh.html
- AAP toolkit: www.aap.org/fasd

Referrals

- For diagnosis: If uncertain whether findings satisfy criteria, depending on available resources:
  - FASD diagnostic clinic
  - Genetics and dysmorphology clinic
  - Neurodevelopmental/behavioral pediatrician
  - Neuropsychologist or behavioral psychologists for ND-PAE or ARND
- For treatment/care needs and care planning or management:
  - Neuropsychologist, clinical psychologist, school psychologist, early intervention
  - SpL, OTR or PT as indicated
  - Social work
  - Medical specialists as indicated (e.g., Otorhinolaryngologist to address frequent ear infections)

Interprofessional Care for Patients with FASD

Health Care is Changing

- Move towards outcomes-based, value-based and ***team-based*** care.
- Reimbursement with the Medicare Access & CHIP Reauthorization Act of 2015 (MACRA) will support values-based care and reimburse ***team-based*** care.

More information on potential team members is available in the AAP FASD Toolkit

Interprofessional Care

- Create and develop a team from different specialties to collaborate in planning & care.
- Establish a common goal
- Care should be interdependent, complimentary, coordinated.
- The patient and family should be considered team members if practicable.

Resources

- American Academy of Pediatrics (AAP): www.aap.org/fasd
- Centers for Disease Control and Prevention (CDC): www.cdc.gov/ncbddd/fasd
- National Organization on Fetal Alcohol Syndrome: www.nofas.org
- National Institute on Alcohol Abuse and Alcoholism: www.niaaa.nih.gov

Take Homes

- FASD are more common than recognized. You most likely have children in your practice with a FASD,
- Obtaining history of prenatal exposure to alcohol is good practice and should be routine for all patients,
- Children will most likely present with neurodevelopmental/neurobehavioral problems,
- But a comprehensive physical and behavior assessment is best to establish the appropriate diagnosis and care plan

Review

- The primary facial dysmorphic features associated with FAS are:
  - Short palpebral fissures, smooth philtrum, thin upper lip.
- The diagnosis of ND-PAE (DSM-5) requires confirmed alcohol exposure and
  - Neurocognitive impairment, self-regulation impairment and deficits in adaptive functioning.
- Traditional behavioral therapies may not work best for children with an FASD.

Review

- The following approaches are recommended for children diagnosed with an FASD:
  - Regularly scheduled follow-up in the medical home
  - Evaluation by a psychologist to assess neurocognitive functioning, self-regulation, and adaptive functioning
  - Medication management for co-occurring conditions if necessary

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Questions?