Neurodevelopmental Disorders: An Overview of Autism Spectrum Disorder and Fetal Alcohol Spectrum Disorder

Ellen F. Geib, M.S.
Clinical Psychology Pre-Doctoral Intern
Track: Neurodevelopmental/Autism
University of New Mexico
Center for Development and Disability
egeib@salud.unm.edu

Disclosure

- The presenter has no financial relationship to this program.

Objectives

At the end of this presentation, participants will be able to:

1. Define and utilize the basic diagnostic vocabulary for Fetal Alcohol Spectrum Disorder (FASD) and Autism Spectrum Disorder (ASD).
2. Incorporate the step-by-step sequential process in accessing for and diagnosing FASD and ASD in your practice.
3. Provide families and other clinicians with reliable, evidence-based sources for additional information regarding FASD and ASD.

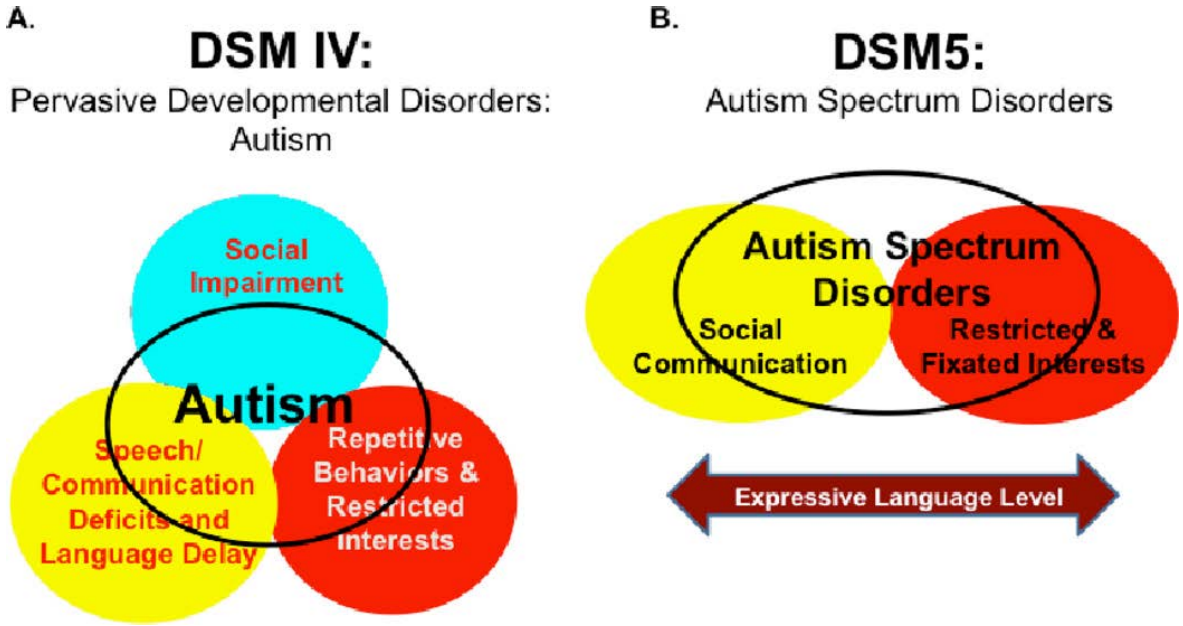
Agenda

1. Autism Spectrum Disorder (ASD)
   A. Defining Autism
   B. Differential Diagnosis of ASD
   C. Diagnostic Clinics and Additional Resources

2. Fetal Alcohol Spectrum Disorder (FASD)
   A. Defining the Spectrum of Fetal Alcohol Disorders
   B. Diagnosing FASDs
   C. Diagnostic Clinics and Additional Resources

Defining Autism

- Autism spectrum disorder (ASD) is a neurodevelopmental disorder represented on a spectrum of severity with two core impairments: social communication and restricted/repetitive behavior (DSM-5; APA, 2013) (299.0, F84)



Possible Early Red Flags for ASD

- Social communication is theorized to be the core deficit of ASD (APA, 2013)
  - Does not babble or coo by 12 months
  - Does not gesture (point, wave, grasp) by 12 months
  - Does not say single words by 16 months
  - Has any loss of any language or social skill at any age
  - No eye contact
  - Lack of social interaction
  - Dislikes physical contact

- Language delay is the most common indicator to parents that their child is not developing typically.

Prevalence of ASD

- The U.S. Centers for Disease Control (CDC, 2014) reports the current estimate of prevalence rates of ASD as 1 in every 68 births.
- ASD is diagnosed in 1 and 42 males and 1 and 189 females.
- These rates are about 120% higher than prior 2002 estimates of 1 in 150 children according to survey estimates conducted by the CDC as part of the Autism and Developmental Disabilities Monitoring Network (Baio, 2012).

Diagnostic Challenges

- No medical test, blood test, brain scan, genetic tool to diagnose autism
- A diagnosis is based on observed behavior and standardized psychological tests assessing the “lack” of behavioral characteristics:
  - Lack of social initiations
  - Limited use of gestures
  - Inconsistent eye contact

Diagnostic Challenges

- Falsification– What else could explain these symptoms?
  - Anxiety
  - Language Delay
  - Intellectual Disability
  - ADHD
  - Trauma

Comprehensive Diagnostic Evaluation

1. **Developmental History**
2. Cognition (MSEL, DAS-II, WISC-V, WPPSI-IV)
3. Adaptive Skills (VABS-3, ABAS-3)
4. Communication (PLS-4, OWLS, CASL-2)
5. Diagnostic Instruments
    - **Autism Diagnostic Observation Schedule (ADOS-2)**
    - Autism Diagnostic Interview (ADI)

Developmental History

- Clinicians must complete a thorough review of the child's developmental history

  "Symptoms must be present in early developmental period although they may not be fully manifested until social demands exceed limited capacities" (APA, 2013)

- Symptoms must be viewed as behavioral patterns and symptom severity across time

Autism Diagnostic Observation Schedule (ADOS-2)

- ADOS-2 is a semi-structured, standardized assessment of communication, social interaction, play/imaginative use of materials, and restricted and repetitive behaviors.
- Contains 5 modules with standard activities at different developmental levels (expressive language skills) and chronological ages (toddler)

(Lord, Rutter, DiLavore, Risi, Gotham, & Bishop, 2012)

Autism Diagnostic Observation Schedule (ADOS-2)

- Consists of standard activities that provide the examiner with opportunities to observe behaviors that are directly relevant to the diagnosis of ASD.
  - Speech abnormalities
  - Reciprocal social communication
  - Use of gestures
  - Nonverbal communication
  - Social overtures
  - Restricted/Repetitive behaviors
  - Shared social enjoyment

(Lord et al., 2012)

Autism Spectrum Disorder

1. Social Communication Impairments (3)	2. Restricted/Repetitive Behaviors (≤2/4)
(A) Social-emotional reciprocity	(A) Stereotyped or repetitive motor movements or speech
(B) Nonverbal communication	(B) Insistence on sameness
(C) Development/Maintenance of Social relationships	(C) Highly restricted, fixated interests
	(D) Unusual interest in sensory aspects of the environment

APA, 2013

1. A. Social-Emotional Reciprocity

- Reduced sharing of interests, emotions, or affect
- Abnormal social approach characteristics
- Failure of back and forth conversation
- Reduced imitation of others
- Difficulty responding to complex social cues (e.g., when and how to join a conversation)

CORE = Sharing emotions with others

- Toddlers – response to name, responsiveness to social smile,
- Single Words/Phrase speech – response to name, showing toys, sharing interests, emotions, affect
- Fluent speech – back and forth conversation, sharing of interests, emotions, affect, initiating and responding to social interactions (e.g., "I had a really fun weekend…")

APA, 2013

1. B. Nonverbal Communication

- Poorly integrated verbal and nonverbal communication
- Abnormalities in eye contact and body language/body orientation
- Odd speech intonation
- Deficits in understanding and using gestures
- Lack of facial expressions
- Impaired joint attention (e.g., lack of pointing, showing, bringing objects to share interest)

CORE = Using language for the benefit of another person

- Toddlers – language for a social purposes
- Single Words/Phrase Speech – using gestures, appropriate facial expressions, eye contact
- Fluent Speech –integration of verbal and nonverbal speech

APA, 2013

1. C. Development and Maintenance of Social Relationships

- Difficulty adjusting behavior to suit social context
- Preference for solitary activities
- Difficulties making friends
- Difficulties or delay in imaginative play
- Difficulty understanding the general reciprocity of relationships with family members, coworkers, friends, etc.

CORE = Interest in peers

- Toddlers –what do they do when no one is demanding their attention?
- Single words/phrase speech – shared social enjoyment
- Fluent speech – ask other's thoughts, and experiences?

APA, 2013

2. A. Stereotyped or Repetitive Motor Movements, Use of Objects, or Speech

- Simple motor stereotypies (e.g., hand flapping, toe walking, finger flicking)
- Lining up toys or objects, spinning objects, flipping objects (e.g., for enjoyment and play)
- Speech echolalia (e.g., “What to go outside,” “Go outside?”)
- Idiosyncratic phrases

APA, 2013

2. B. Insistence on Sameness

- Extreme distress at small changes
- Difficulties with transition
- Rigid thinking patterns
- Need to take similar routes to destinations
- Ritualized patterns of verbal behavior
  (e.g., repetitive questioning, rigid greeting rituals)

APA, 2013

2. C. Highly Restricted, Fixed Interests

…That are abnormal in their intensity and focus

- Strong attachment to unusual objects (e.g., piece of string, drain pipes)
- Strong preoccupation with unusual objects (e.g., bus schedule)
- Excessively circumscribed and perseverative interests

APA, 2013

2. D. Sensory Processing Differences

Q = How does the child interact with his environment?

- Apparent indifference to pain, temperature
- Adverse responses to specific sounds, textures
- Excessive smelling/touching of objects
- Visual fascination with lights or movement

APA, 2013

UNM Resources and Diagnostic Clinics

- Autism Spectrum Evaluation Clinic
  - 505-272-9337
- Autism Programs Family and Provider Resource Team http://www.cdd.unm.edu/autism/portal/intex.html
  - 505-272-1852
  - 1-800-270-1861 (toll free)
- Southwest Conference on Disability http://www.cdd.unm.edu/swconf/index.html
- ADOS-2 Training Workshop http://www.cdd.unm.edu/more-events.aspx?q=Autism

Helpful Resources

- Autism Speaks www.autismspeaks.org
- Autism Speaks' New Mexico Resource Guide https://www.autismspeaks.org/resource-guide/state/NM
- Families for Early Autism Treatment (FEAT) www.feat.org
- The International Society for Autism Research (INSAR) www.autism-insar.org
- Autism Society of America www.autism-society.org
- New Mexico Autism Society www.nmautismsociety.org

Fetal Alcohol Spectrum Disorders

- Fetal Alcohol Syndrome (FAS) is a permanent birth defect syndrome caused by exposure to alcohol in utero
- Disorders across the spectrum are characterized by physical, cognitive, and behavioral deficits.
- **The term "FASD" is not a clinical diagnosis but rather represents the full range of disorders:**
  - Fetal Alcohol Syndrome (FAS)
  - Partial Fetal Alcohol Syndrome (PFAS)
  - Alcohol Related Neurodevelopmental Disorder (ARND)
  - Alcohol Related Birth Defects (ARBD)

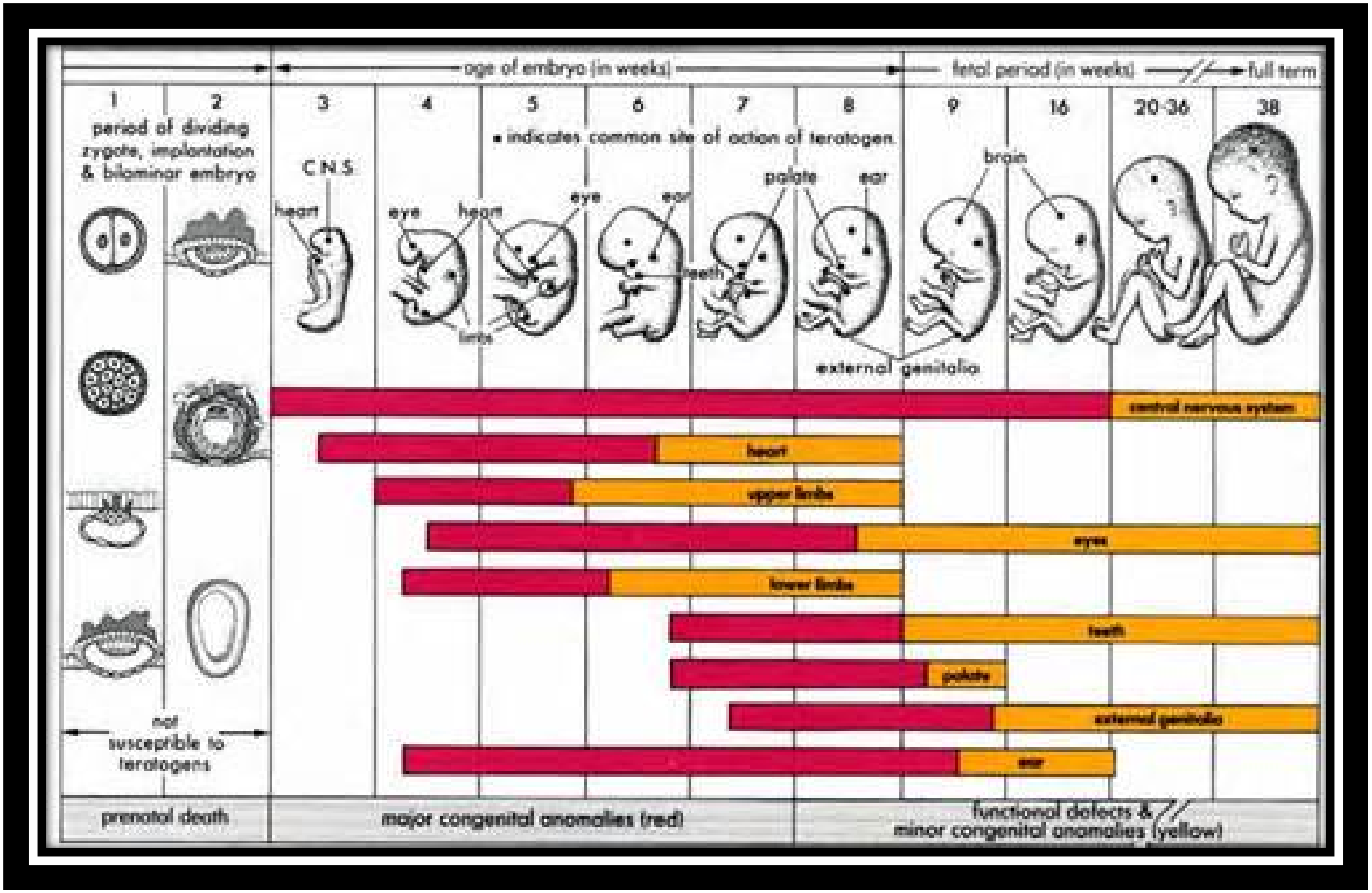
Defining FASD

- DSM-5 – 315.8 (F88) - Neurodevelopmental disorder associated with prenatal alcohol exposure
- ICD-10 – Q86.0 – Fetal Alcohol Syndrome
- IOM Categories (Hoyme et al., 2016):
  - Fetal Alcohol Syndrome (FAS)
  - Partial Fetal Alcohol Syndrome (PFAS)
  - Alcohol Related Neurodevelopmental Disorder (ARND)
  - Alcohol Related Birth Defects (ARBD)

Alcohol in Pregnancy

- Safe Amount
  - **There is NO "safe" level of alcohol use during pregnancy at any time using any form of alcohol (American Academy of Pediatrics, 2015)**
  - Larger amounts of alcohol and binge drinking appear to increase risk of adverse outcomes
- Timing of Alcohol Use
  - Alcohol exposure appears to be the most harmful during the first 3 months of pregnancy although severe effects are also possible later in pregnancy
  - **It is never too late for a mother to stop drinking**

Alcohol in Pregnancy



Prevalence of Exposure to Alcohol

- FAS is the leading known cause of preventable intellectual disabilities/developmental disabilities
- About 13% of all babies are exposed to alcohol to some degree while in utero
- FASD occurs in about 10 to 25.2 per 1,000 live births (May et al., 2014)
- FAS occurs in about 3 per 1,000 live births (Stratton et al., 1996)

Diagnostic Challenges

- **Individuals with prenatal alcohol exposure can present with a wide range of outcomes because of the timing, frequency, and quantity of alcohol exposure**
- It is difficult to determine a diagnosis:
  - History of alcohol use by the birth mother may not be known
  - Stigma of maternal alcohol use leads to under-reporting
- FAS can still be diagnosed without confirmation of alcohol exposure if all the symptoms are present

Comprehensive Diagnostic Evaluation

A multidisciplinary team is best practice

1. Developmental History
2. Physical Exam
   - Neurological “Hard” and “Soft” Impairments
   - Birth length/weight, stature, weight, head circumference
3. Standardized psychological assessment
4. Diagnostic instruments
   - **IOM Diagnostic Algorithm (Hoyme et al., 2016)**
   - 4-Digit Diagnostic Code (Astley, 2004)

FASD Diagnostic Algorithm (Hoyme et al., 2016)

1.

2.

3.

4.

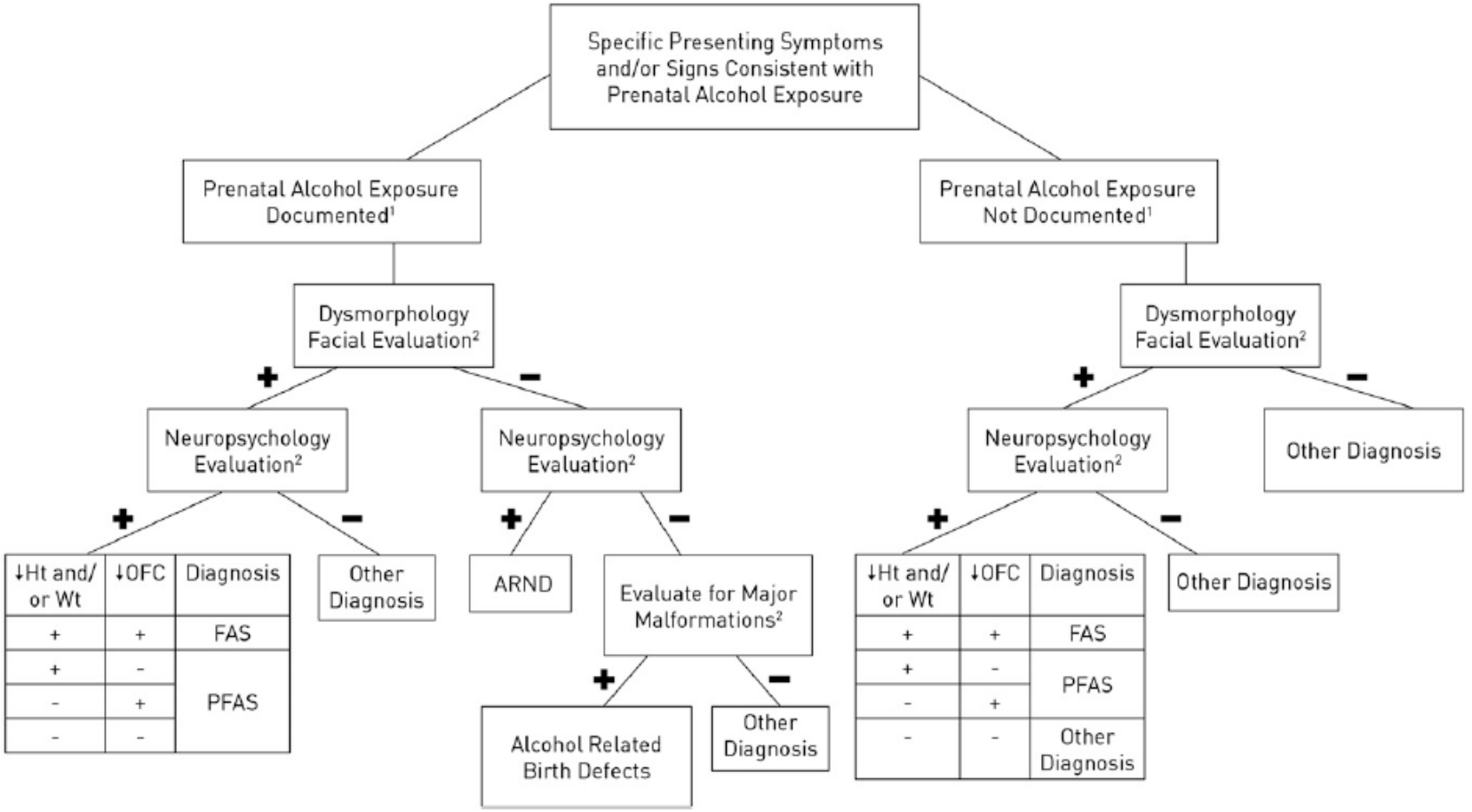


FIGURE 1
FASD diagnostic algorithm. See text for complete discussion. A positive dysmorphology facial evaluation requires 2 of the 3 cardinal facial features of FASD (short palpebral fissures, smooth philtrum, and this vermilion border of the upper lip). Cutoffs for neuropsychological testing are −1.5 SD. Cutoffs for stature, weight, and head circumference are at the 10th percentile.

Developmental History to Determine Prenatal Alcohol Exposure

Biological mother report or reliable collateral source:

1. ≥ 6 drinks/week for >2 weeks of pregnancy
2. ≥ 3 drinks per occasion on ≥2 occasions during pregnancy
3. Documentation of alcohol-related social or legal problems during pregnancy
4. Documentation of intoxication during pregnancy by blood, breath, or urine
5. Positive testing with established alcohol-exposure biomarkers during pregnancy
6. Increased prenatal risk associated with drinking during pregnancy as assessed by a validated screening tool
   (e.g., T-ACE, AUDIT)

FASD Diagnostic Algorithm (Hoyme et al., 2016)

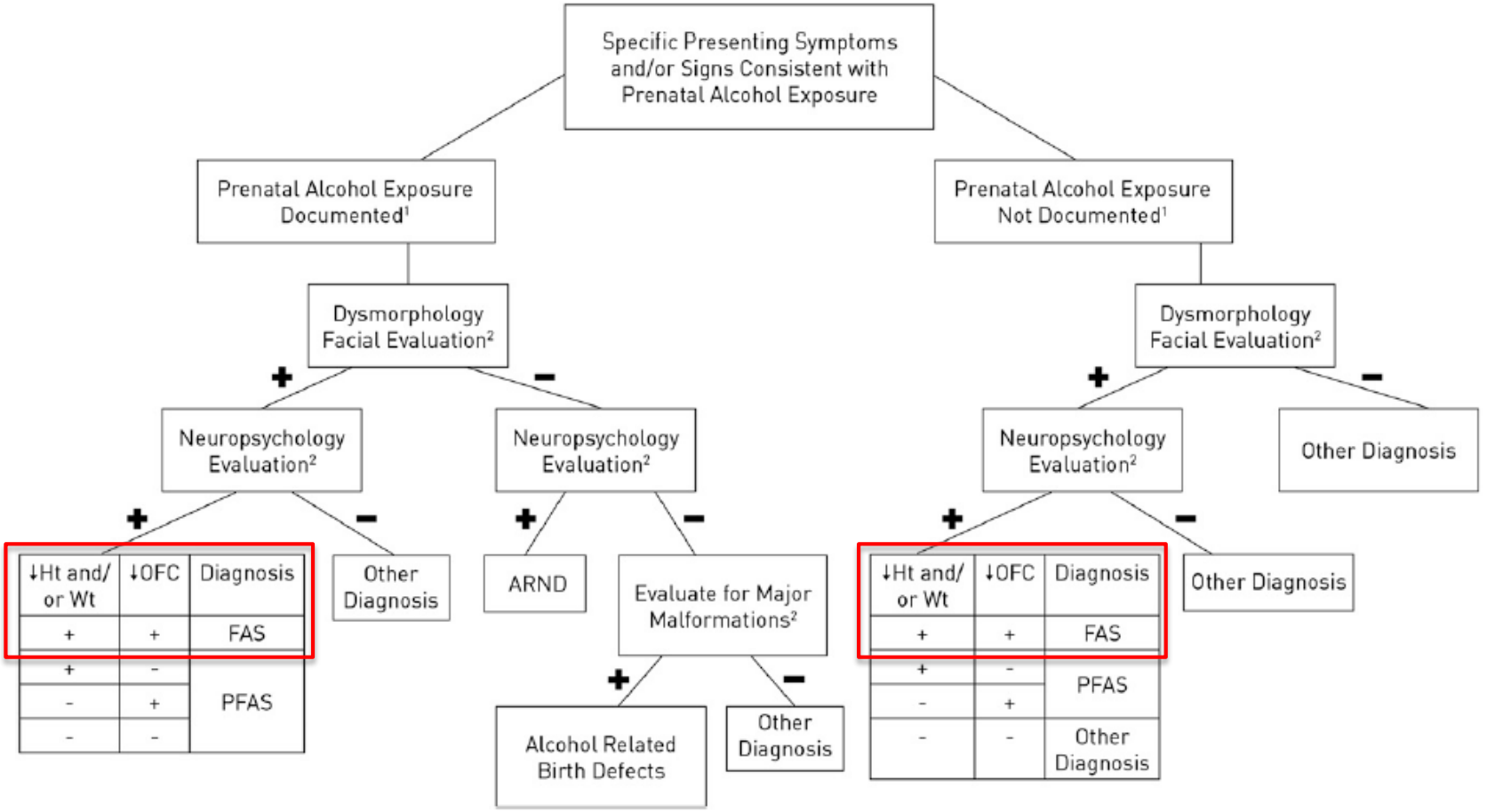


FIGURE 1
FASD diagnostic algorithm. See text for complete discussion. A positive dysmorphology facial evaluation requires 2 of the 3 cardinal facial features of FASD (short palpebral fissures, smooth philtrum, and this vermilion border of the upper lip). Cutoffs for neuropsychological testing are −1.5 SD. Cutoffs for stature, weight, and head circumference are at the 10th percentile.

Fetal Alcohol Syndrome (FAS)

1. Facial abnormalities (>2 of the following, all 3 if alcohol exposure is not documented)
   - Short palpebral fissures (<10th percentile)
   - Thin vermilion border of the upper lip
   - Smooth philtrum
2. Prenatal and/or postnatal growth deficiency
   - Birth length/weight, growth deficiency
3. Deficient Brain growth/abnormalities
   - Head circumference, brain anomalies, seizures
4. Neurobehavioral impairment (>1.5 SD)
   - Global impairment, OR
   - Cognitive or Behavioral deficit in at least 1 domains (>1.5 SD), or Developmental Delay (children <3)

Cardinal Facial Features of FAS

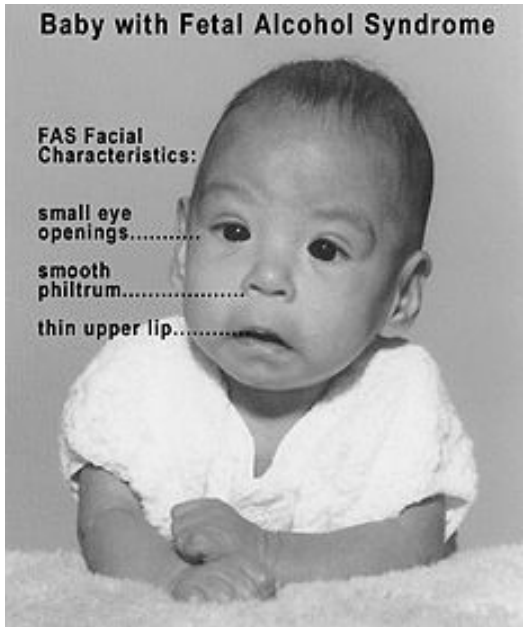


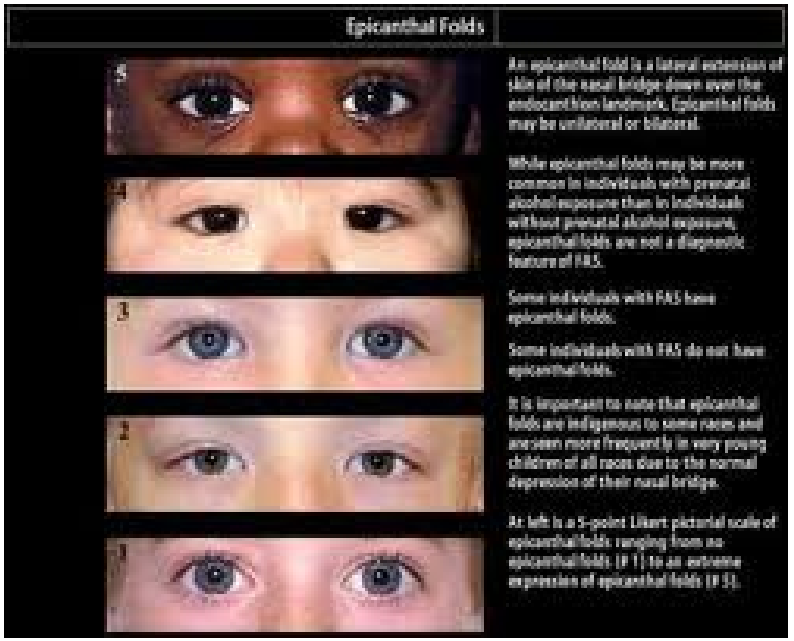
FIGURE 2
Typical child with FAS. The 3 cardinal facial features are evident: short palpebral fissures, smooth philtrum, and relatively thin vermilion border of the upper lip. Midface hypoplasia is also apparent.

(Hoyme et al., 2016)

Facial Diagnostic Guides (Astley, 2004)

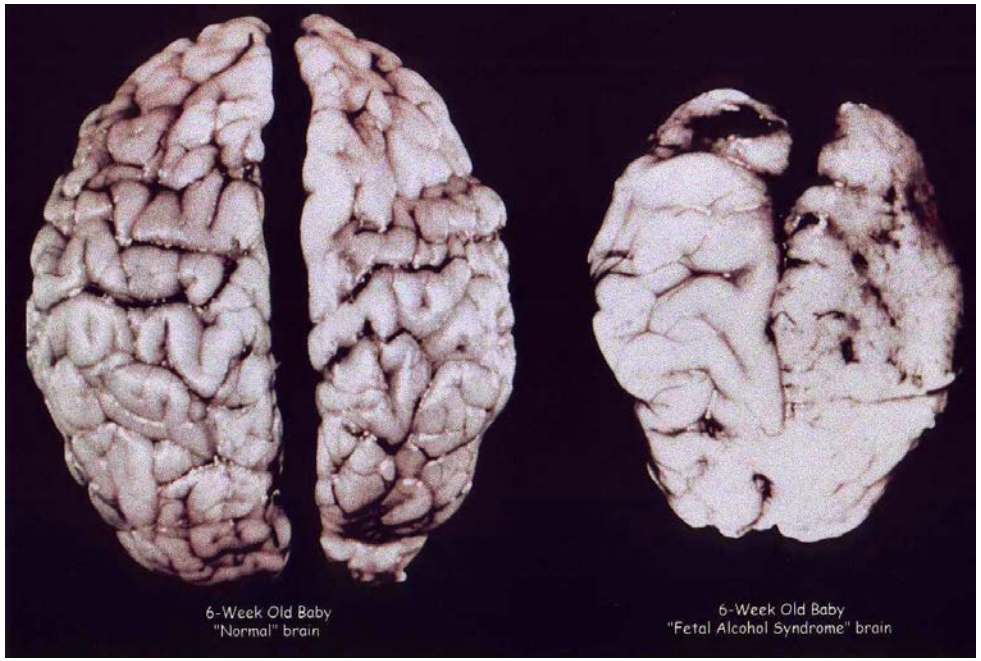
Philtrum and Upper Lip

Epicanthal Eye Folds

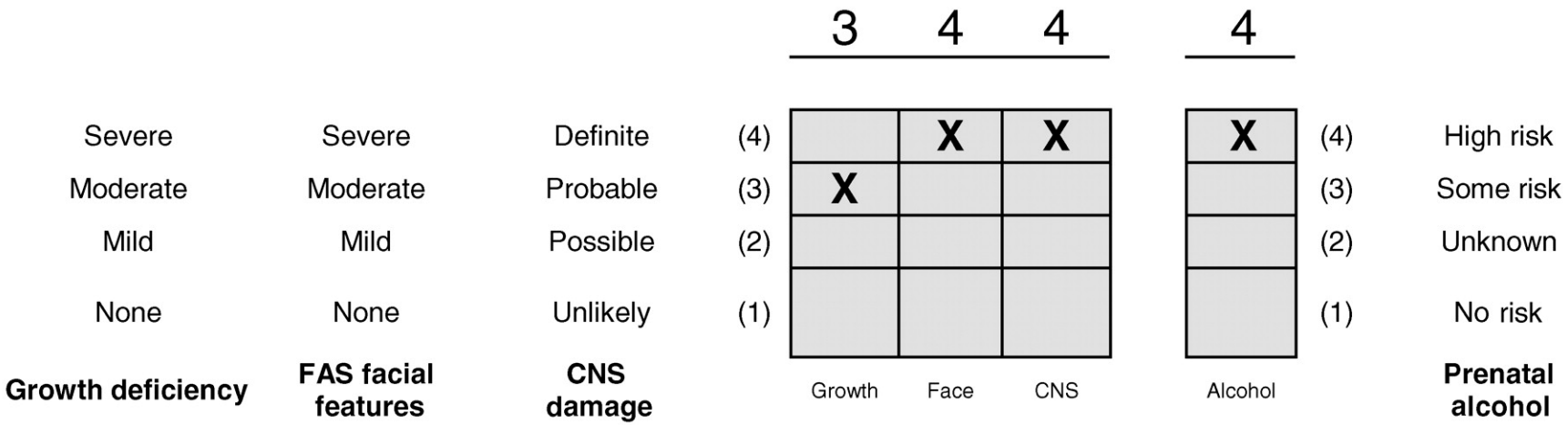


Central Nervous System Damage

- "Hard" Neurological Signs
  - Decreased brain size
    - Corpus callosum
    - Basal ganglia
    - Cerebellum
  - Microcephaly
- "Soft" Neurological Signs
  - Cognition
  - Memory
  - Executive Functioning
  - Motor
  - Communication
- Other CNS Deficit Clues
  - Sleep disturbance
  - Attention deficits
  - Learning disabilities



4-Digit Diagnostic Code (Astley, 2004)



"3444" = Fetal alcohol syndrome (alcohol exposed)

- Growth "3" = Birth length/weight, growth deficiency
- Face "4" = Palpebral Fissure, Philtrum, Upper Lip
- CNS "4"= Micocephaly and/or "Hard" neurological findings and "Soft" neuropsychological findings
- Alcohol "4" = Alcohol use during pregnancy is confirmed and exposure pattern is severe

FASD Diagnostic Algorithm (Hoyme et al., 2016)

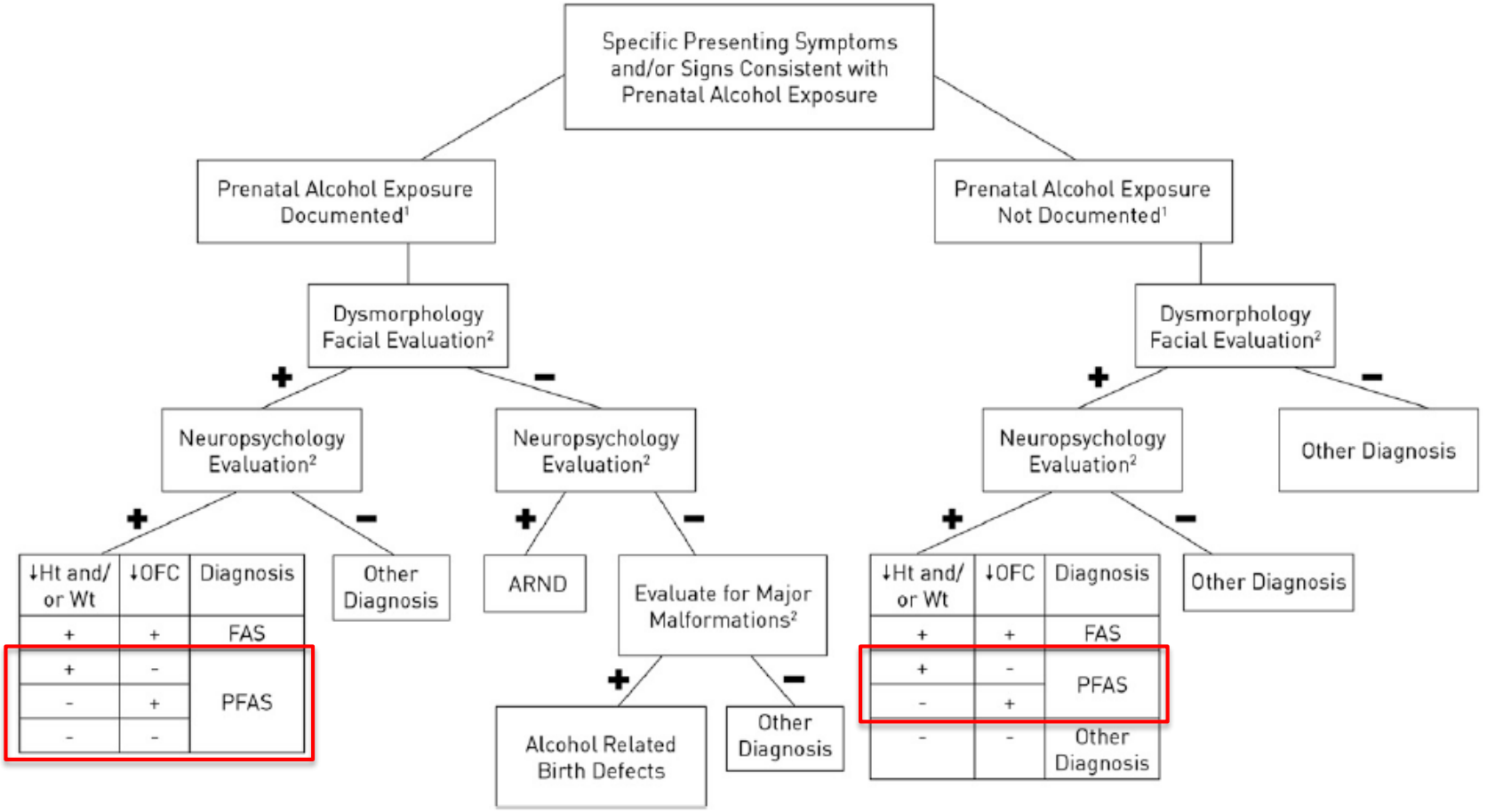


FIGURE 1
FASD diagnostic algorithm. See text for complete discussion. A positive dysmorphology facial evaluation requires 2 of the 3 cardinal facial features of FASD (short palpebral fissures, smooth philtrum, and this vermilion border of the upper lip). Cutoffs for neuropsychological testing are –1.5 SD. Cutoffs for stature, weight, and head circumference are at the 10th percentile.

Partial FAS

1. Facial abnormalities (>2 of the following, all 3 if alcohol exposure is not documented)
   - Short palpebral fissures (<10th percentile)
   - Thin vermilion border of the upper lip
   - Smooth philtrum
2. Prenatal and/or postnatal growth deficiency (required if prenatal alcohol exposure is not documented)
   - Height and/or weight, OR deficient brain growth/anomalies
3. Neurobehavioral impairment (>1.5 SD)
   - Global impairment
   - Behavioral deficit in at least 2 domains (>1.5 SD)

FASD Diagnostic Algorithm (Hoyme et al., 2016)

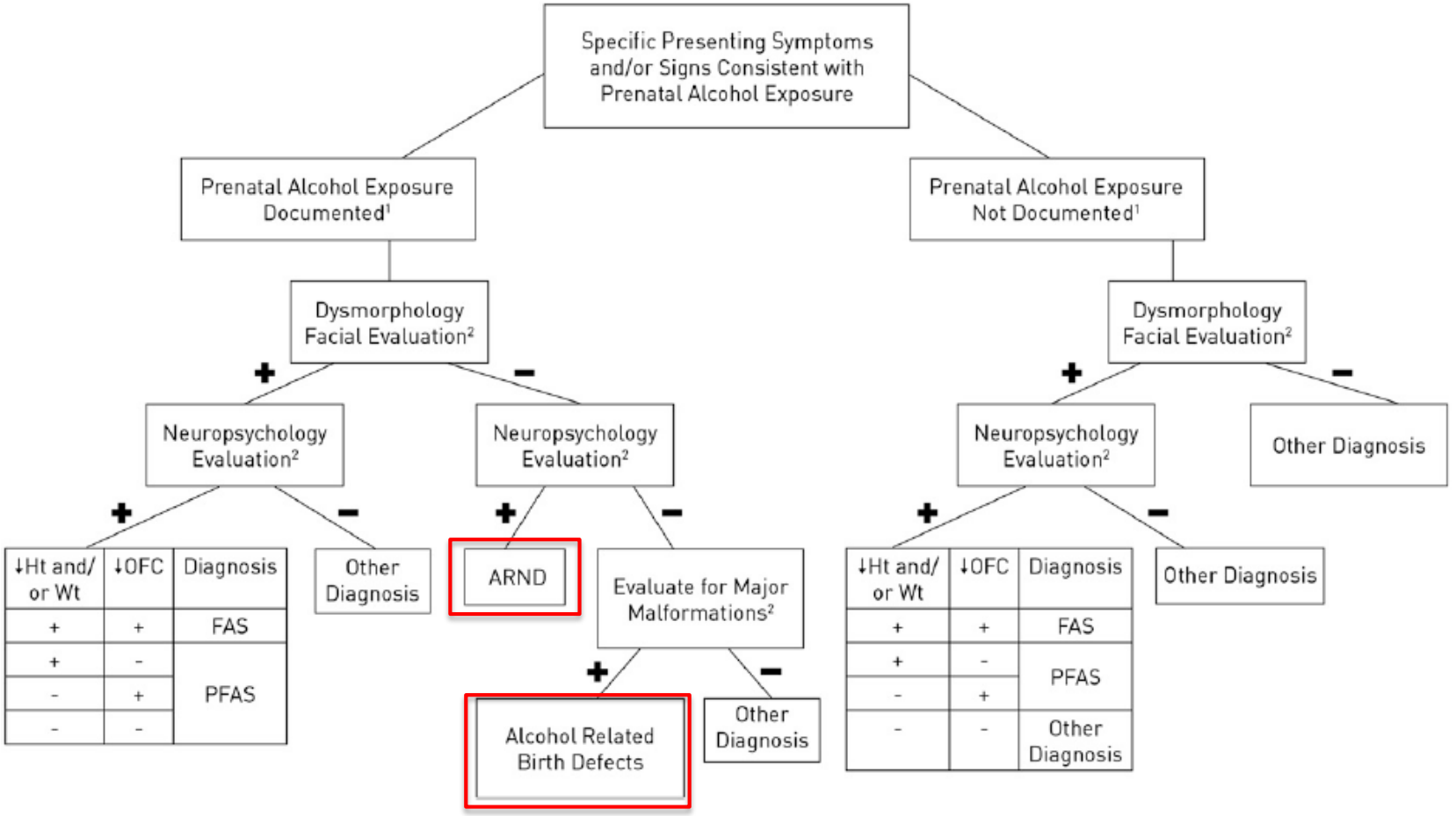


FIGURE 1
FASD diagnostic algorithm. See text for complete discussion. A positive dysmorphology facial evaluation requires 2 of the 3 cardinal facial features of FASD (short palpebral fissures, smooth philtrum, and this vermilion border of the upper lip). Cutoffs for neuropsychological testing are –1.5 SD. Cutoffs for stature, weight, and head circumference are at the 10th percentile.

Alcohol Related Neurodevelopmental Disorder (ARND)

1. Documented prenatal alcohol exposure
2. Neurobehavioral impairment (≥1.5 SD)
   - Global impairment, OR
   - Cognitive deficit in at least 2 domains
   - Behavioral deficit in at least 2 domains (≥ 1.5 SD)

- **Few or none of the physical features of FAS**

Alcohol Related Birth Defects (ARBD)

1. Documented prenatal alcohol exposure
2. One or more specific major malformations in the skeletal and/or major organ systems:
   - Abnormalities of the heart (e.g., holes in the heart), eyes, underdeveloped kidneys, and skeleton (e.g., fused bones)

- **Few or none of the physical features of FAS**
- **No significant neuropsychological, cognitive, or behavioral impairments**

UNM Diagnostic Clinic

- CDD FASD Clinic
  - Caregivers or providers can refer
  - Serve individuals from birth to 22 years
  - Clinical services:
    - FASD comprehensive diagnostic evaluation
    - Follow-up neuropsychological evaluations to support educational programming and behavioral interventions
    - Educational consultation (IEPs, IFSPs, transition planning)
  - Contact: 505-272-6157

Helpful Resources

- Hoyme et al. (2016). Updated clinical guidelines for diagnosing fetal alcohol spectrum disorders. *Pediatrics, 138*(2). Retrieved from: http://pediatrics.aappublications.org/content/pediatrics/early/2016/07/25/peds.2015-4256.full.pdf
- Clinical Report: Fetal Alcohol Spectrum Disorders (http://pediatrics.aappublications.org/content/136/5/e1395)
- National Organization on Fetal Alcohol Syndrome (https://www.nofas.org/)
- Collaborative Initiative of Fetal Alcohol Spectrum Disorders (CIFASD) http://cifasd.org/
- Collaboration on FASD Prevalence (CoFASP) http://www.emory.edu/msacd/Research/CoFASP.html
- Fetal Alcohol Syndrome Diagnostic and Prevention Network http://depts.washington.edu/fasdpn

Questions

References

American Psychiatric Association, (2013). *Diagnostic and statistical manual of mental disorders – 5*. Washington, DC: Author.

Astley, S.J. (2004). Diagnostic guide for fetal alcohol spectrum disorders: The 4-digit diagnostic code. Seattle: University of Washington Publication Services. Retrieved from: https://depts.washington.edu/fasdpn/pdfs/guide2004.pdf

Baio, J. (2012). Prevalence of autism spectrum disorders—autism and developmental disabilities monitoring network, 14 sites, United States, 2008, *MMWR Surveillance Summaries, 61,* 1-19.

Center for Disease Control and Prevention (CDC, 2014). Autism spectrum disorders (ASDs). Retrieved from: http://www.cdc.gov/ncbddd/autism/data.html

Hoyme, H. E., Kalberg, W. O., Elliott, A. J., Blankenship, J., Buckley, D., Marais, A. S., ... & Jewett, T. (2016). Updated clinical guidelines for diagnosing fetal alcohol spectrum disorders. *Pediatrics*, e20154256.

Lord C., Rutter M., DiLavore P. C., Risi S., Gotham K., Bishop S. (2012). Autism diagnostic observation schedule, second edition. Torrance, CA: Western Psychological Services.

May, P. A., Baete, A., Russo, J., Elliott, A. J., Blankenship, J., Kalberg, W. O., ... & Adam, M. P. (2014). Prevalence and characteristics of fetal alcohol spectrum disorders. *Pediatrics*, *134*(5), 855-866.

Stratton K, Howe C, Battaglia F.C. (1996). *Fetal alcohol syndrome: diagnosis, epidemiology, prevention, and treatment*. Washington: Institute of Medicine and National Academy Press.

Williams, J.F. & Smith, V.C. (2015). American Academy of Pediatrics clinical report: Fetal alcohol spectrum disorders. *Pediatrics, 136*(5). 10.1542/peds.2015-3113

Contact Info

Ellen F. Geib, M.S.
Clinical Psychology Pre-Doctoral Intern
Track: Neurodevelopmental/Autism
University of New Mexico
Center for Development and Disability
egeib@salud.unm.edu