

CLINICAL GUIDELINES FOR DIAGNOSING FASD IN MINNESOTA

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Introduction

Prenatal alcohol exposure may result in serious health, behavioral, and cognitive challenges for affected individuals. The spectrum of effects is described by the term fetal alcohol spectrum disorder (FASD). While this term is not a diagnosis, it includes fetal alcohol syndrome (FAS) – the most visible presentation of the disability – as well as the more hidden disabilities of partial FAS (PFAS) and alcohol related neurodevelopmental disorder (ARND). FASD presents serious health, educational, social, and justice system challenges, and should be considered a lifelong disability.

FASD is evaluated by assessing five areas:

1. Facial anomalies
2. Growth anomalies
3. Brain growth and physiology
4. Neurobehavioral impairments
5. History of prenatal alcohol exposure

The most important of these indicators is neurobehavioral impairment. Comprehensive assessment is performed to determine the degree of brain dysfunction and the probability of prenatal alcohol exposure (PAE) related neurodevelopmental differences. Additionally, the resulting assessment data provides information to professionals that can be used to guide interventions and foster the strengths of individuals diagnosed with FASD.

We evaluate individuals to provide:

1. Evidence-based diagnosis of the full spectrum of FASD;
2. Risk evaluation for future challenges (medical, neurodevelopmental, and psychosocial) and prevention of adverse outcomes;
3. Intervention recommendations to optimize the life functioning of individuals we assess;
4. Visibility/recognition of the neurodevelopmental impacts of prenatal alcohol in order to promote the growth and development of support services for families and children and to support prevention efforts.

The diagnostic guidelines on the following pages are derived from existing diagnostic systems (primarily Hoyme et al., 2016) with slight modifications developed by a consensus group to increase clarity around the definition of cognitive/behavioral effects. Differential diagnoses descriptions are outlined in Appendix C.

| Fetal Alcohol Syndrome (FAS) Requires features A-D (confirmation of PAE NOT required) | |
|---|--|
| A. Face | 3 facial features required: <ul style="list-style-type: none"> • Short palpebral fissures (\leq 10th percentile) • Thin vermilion (4 or 5 lip philtrum guide, University of Washington) • Smooth philtrum (4 or 5 lip philtrum guide, University of Washington) |
| B. Growth* | Height and/or weight \leq 10th percentile (not due to another known cause) |
| C. Brain Growth & Neurophysiology | \geq 1 of the following (not due to another known cause): <ul style="list-style-type: none"> • Head circumference \leq 10th percentile • Structural brain anomalies identified on imaging • Neurological abnormalities (e.g. seizure disorder, spasticity, hemiparesis) |
| D. Neurobehavioral Impairment** | For individuals under 3 years of age: <ul style="list-style-type: none"> • Developmental delay \geq 1.5 SD below mean on a norm-referenced developmental assessment (\geq standard score 78 or scaled score 6) For individuals 3 years or older: <ul style="list-style-type: none"> • Cognitive/behavioral impairment as indicated by either: <ul style="list-style-type: none"> ○ Overall IQ \geq 1.5 SD below the mean (\geq standard score 78 or scaled score 6) –or– ○ Cognitive or behavioral function \geq 1.5 SD below mean on 2 or more specific norm-referenced measures (\geq standard score 78 or scaled score 6): <ul style="list-style-type: none"> ▪ Specific IQ index ▪ Attention/Executive functioning ▪ Language ▪ Learning disabilities/Achievement ▪ Memory ▪ Social cognition / Pragmatic language ▪ Motor/Visual Motor ▪ Informant-reported impairment*** \geq 1.5 SD from the mean: <ul style="list-style-type: none"> • Mood or behavioral regulation • Attention • Impulse control • Social/Adaptive Functioning • Sensory processing |
| E. Prenatal Alcohol Exposure (PAE) | Confirmed or unconfirmed |

*Anytime during the child's life; age and gender adjusted; parent adjusted when data is available.

**Consider historical data indicating neurobehavioral impairment as well as current test data.

***Informant-ratings should only be used to account for one domain of impairment. At least one domain should be accounted for by an objective performance test.

| Partial Fetal Alcohol Syndrome (Partial FAS) For children with confirmed PAE, a diagnosis of PFAS requires features A & D: For children with unconfirmed PAE, a diagnosis of PFAS requires A, D, & B <u>or</u> C | |
|---|---|
| A. Face | 2 facial features required: <ul style="list-style-type: none"> • Short palpebral fissures (\leq 10th percentile) • Thin vermilion (4 or 5 lip philtrum guide, University of Washington) • Smooth philtrum (4 or 5 lip philtrum guide, University of Washington) |
| B. Growth* | <ul style="list-style-type: none"> • Height and/or weight \leq 10th percentile (not due to another known cause) |
| C. Brain Growth & Neurophysiology | \geq 1 of the following (not due to another known cause): <ul style="list-style-type: none"> • Head circumference \leq 10th percentile • Structural brain anomalies identified on imaging • Neurological abnormalities (e.g. seizure disorder, spasticity, hemiparesis) |
| D. Neurobehavioral Impairment** | <p>For individuals under 3 years of age:</p> <ul style="list-style-type: none"> • Developmental delay \geq 1.5 SD below mean on a norm-referenced developmental assessment (\geq standard score 78 or scaled score 6) <p>For individuals 3 years or older:</p> <ul style="list-style-type: none"> • Cognitive/behavioral impairment as indicated by either: <ul style="list-style-type: none"> ○ Overall IQ \geq 1.5 SD below the mean (\geq standard score 78 or scaled score 6) –or– ○ Cognitive or behavioral function \geq 1.5 SD below mean on 3 or more specific norm-referenced measures (\geq standard score 78 or scaled score 6): <ul style="list-style-type: none"> ▪ Specific IQ index ▪ Attention/Executive functioning ▪ Language ▪ Learning disabilities/achievement ▪ Memory ▪ Social cognition / Pragmatic language ▪ Motor/Visual Motor ▪ Informant-reported impairment*** <p>\geq 1.5 SD from the mean:</p> <ul style="list-style-type: none"> • Mood or behavioral regulation • Attention • Impulse control • Social/Adaptive Functioning • Sensory processing |
| E. Prenatal Alcohol Exposure (PAE) | Confirmed or unconfirmed |

*Anytime during the child's life; age and gender adjusted; parent adjusted when data is available.

**Consider historical data indicating neurobehavioral impairment as well as current test data.

***Informant-ratings should only be used to account for one domain of impairment. At least two domains should be accounted for by an objective performance test.

| Alcohol Related Neurodevelopmental Disorder (ARND) Requires features D & E (confirmation of PAE required) | |
|---|--|
| A. Face | Not required |
| B. Growth | Not required |
| C. Brain Growth & Neurophysiology | Not required |
| D. Neurobehavioral Impairment* | <p>For individuals under 3 years of age:</p> <ul style="list-style-type: none"> • Developmental delay ≥ 1.5 SD below mean on a norm-referenced developmental assessment (\geq standard score 78 or scaled score 6) <p>For individuals 3 years or older:</p> <ul style="list-style-type: none"> • Cognitive/behavioral impairment as indicated by either: <ul style="list-style-type: none"> ○ Overall IQ ≥ 1.5 SD below the mean (\geq standard score 78 or scaled score 6) <li style="text-align: center;">–or– ○ Cognitive or behavioral function ≥ 1.5 SD below mean on 3 or more specific norm-referenced measures (\geq standard score 78 or scaled score 6): <ul style="list-style-type: none"> ▪ Specific IQ index ▪ Attention/Executive functioning ▪ Language ▪ Learning disabilities/achievement ▪ Memory ▪ Social cognition / Pragmatic language ▪ Motor/Visual Motor ▪ Informant-reported impairment** <p>≥ 1.5 SD from the mean:</p> <ul style="list-style-type: none"> • Mood or behavioral regulation • Attention • Impulse control • Social/Adaptive Functioning • Sensory processing |
| E. Prenatal Alcohol Exposure (PAE) | Confirmed |

*Consider historical data indicating neurobehavioral impairment as well as current test data.

**Informant-ratings should only be used to account for one domain of impairment. At least two domains should be accounted for by an objective performance test.

Appendix A: Confirmed/Documented Prenatal Alcohol Exposure (PAE) Guidelines

Precise information about prenatal alcohol exposure amounts, timing, etc. is often not available. Clinical judgment is frequently necessary to decide on this particular diagnostic criterion. General guidance follows:

- Consider the source(s) of information carefully. Medical records, social service records, and police records are examples with higher credibility but these can be incomplete and/or imperfect.
- Records indicating maternal treatment for alcohol use disorder in close proximity and/or during the pregnancy is useful documentation.
- Maternal acknowledgment of alcohol use in close proximity to the pregnancy and/or during the pregnancy is often useful for documentation. Maternal denial of alcohol use should generally be respected unless there is sufficient evidence otherwise.
- Pregnancy begins at conception (prior to recognition of the pregnancy), and prenatal alcohol exposure between conception and recognition is considered high risk.
- First-hand (eyewitness) reports of maternal alcohol consumption (ex. from a family member or someone in the home) can be useful documentation. Second-hand/third-hand reports should be considered less reliable and corroborating information should be sought.
- Patterns of alcohol use differ for individuals. Repeated exposure to alcohol or even a single episode of binge drinking (high blood alcohol level) are both considered to be high risk to the neurodevelopment of the fetus. As an example of repeated exposure, some diagnostic criteria set a threshold of 6-8 drinks per week early in pregnancy or for more than 2 weeks. Binge drinking (to intoxication) is generally defined as consumption of 4 or more drinks within a 2 hour period for a woman of average height/weight.
- Generally, very low levels of alcohol consumption below these thresholds are not considered to meet the criterion for diagnosis of FASD. For example, a single drink on one or two occasions is not thought to be associated with high risk for FASD. Nonetheless, there is no safe level of alcohol consumption during pregnancy.

The following are examples of specific criteria used by three different diagnostic systems. These examples are included for illustrative purposes and context only.

Hoyme/Revised IOM Guidelines (2016):

One or more of the following conditions must be met to constitute documented PAE during pregnancy (including drinking levels reported by the mother 3 months before her report of pregnancy recognition or a positive pregnancy test documented in the medical record). The information must be obtained from the biological mother or a reliable collateral source (e.g., family member, social service agency, or medical record):

- ≥ 6 drinks/week for ≥ 2 week during pregnancy*
- ≥ 3 drinks per occasion on ≥ 2 occasions during pregnancy*
- Any incidence of binge drinking (4 or more drinks consumed on one occasion within a 2 hour period)
- Documentation of alcohol-related social or legal problems in proximity to (before or during) the index pregnancy (e.g. history of citation(s) for driving while intoxicated or history of treatment of an alcohol-related condition)

- Documentation of intoxication during pregnancy by blood, breath, or urine alcohol content testing
- Positive testing with established alcohol-exposure biomarker(s) during pregnancy or at birth (e.g., analysis of fatty acid ethyl esters, phosphatidylethanol, and/or ethyl glucuronide in maternal hair, fingernails, urine, or blood, or placenta, or meconium)
- Increased prenatal risk associated with drinking during pregnancy as assessed by a validated screening tool of, for example, T-ACE (tolerance, annoyance, cut down, eye-opener) or AUDIT (alcohol use disorders identification test)

*These criteria for maternal drinking are based on large epidemiologic studies that demonstrate adverse fetal effects from ≥ 3 drinks per occasion and others that indicate 1 drink/day as a threshold measure for FASD.

Hoyme, H. E., Kalberg, W. O., Elliott, A. J., Blankenship, J., Buckley, D., Marais, A. S., ... May, P. A. (2016). Updated clinical guidelines for diagnosing fetal alcohol spectrum disorders. *Pediatrics*, 138(2), e20154256. doi: 10.1542/peds.2015-4256

Kable et al. Guidelines (2016):

Although both animal and human studies have documented adverse effects with low levels of drinking, identifying a threshold of PAE remains challenging. Current data suggest that a history of more than minimal gestational alcohol exposure (e.g., light drinking) prior to pregnancy recognition and/or following pregnancy recognition should be required. Light drinking has been defined as 1 to 13 drinks per month, with no more than 2 drinks per drinking occasion. Confirmation of more than minimal gestational alcohol exposure may be obtained from maternal self-report of alcohol use in pregnancy; a spouse/partner, relative, or friend who observed the biological mother drinking alcohol during pregnancy; and/or documentation in medical or other records.

Although these criteria provide general guidelines to define confirmed more than minimal gestational alcohol exposure, questions regarding how much alcohol is needed to impact neurodevelopmental outcome remain unanswered. The impact of low levels of PAE remains controversial. Consumption levels may vary on a daily and weekly basis, even among heavy alcohol users. Dosage level interacts with the period of fetal development, maternal and fetal genetics, and maternal physical status and metabolic systems. Maternal characteristics such as older age at delivery, smoking, certain obstetric problems and medical conditions (especially liver disease) all increase the probability of bearing affected offspring. Despite extensive research, no specified “safe” level of drinking during pregnancy has been identified. Public health recommendations in multiple countries state that women should not drink when planning pregnancy or throughout pregnancy to prevent teratogenic effects, including those that may be more subtle.

Clinicians may be uncomfortable with asking patients and their families about gestational alcohol exposure histories but this is a necessary criterion for making the diagnosis. Guidelines as well as professional training programs to facilitate asking about gestational alcohol exposure have been developed and have been found to increase the clinicians’ comfort and skill at asking these questions. Clinicians should not make a diagnosis of FASD if they are not confident in the validity of information they obtain documenting a more-than-minimal level of gestational alcohol exposure.

Kable, J. A., O'Connor, M. J., Olson, H. C., Paley, B., Mattson, S. N., Anderson, S. M., & Riley, E. P. (2016). Neurobehavioral Disorder Associated with Prenatal Alcohol Exposure (ND-PAE): Proposed DSM-5 Diagnosis. *Pediatrics*, 138(2), e20160914. doi: 10.1542/peds.2016-0914

The 4-Digit Diagnostic Code Guidelines (2004)

Alcohol exposure is ranked according to the quantity, timing, frequency, and certainty of exposure during pregnancy. The case definitions for the four Ranks address two important issues: 1) that exposure information in a clinical setting can be of limited availability or of unknown accuracy; and 2) a clear consensus is not available concerning the amount of alcohol that can actually be toxic to each individual fetus (Stratton et al., 1996).

The case definitions for prenatal alcohol exposure differentiate four clinically meaningful groups:

- Rank 4: confirmed exposure to high levels of alcohol
- Rank 3: confirmed exposure, but the level is less than Rank 4 or the level is unknown;
- Rank 2: unknown exposure (neither confirmed absent nor confirmed present); and
- Rank 1: confirmed absence of exposure from conception to birth).

High exposure is defined generally to be a blood alcohol concentration of greater than 100 mg/dL (a level that typically can be reached by a 55-kg (121 lb.) woman consuming six to eight beers) weekly, early in pregnancy. In the absence of a clear consensus on the amount of alcohol that can actually be toxic to the fetus, this general definition should only serve as a guide, not a threshold.

One example of a 'Rank 4' exposure is the birth mother reported drinking to the point of intoxication weekly throughout pregnancy. Two examples of 'Rank 3' exposures include: 1) birth mother was observed to be drinking during pregnancy, but the amount is unknown, 2) birth mother reported drinking a glass of wine weekly, but stopped drinking as soon as she learned she was pregnant at 3 months. Two examples of when alcohol exposure is ultimately unknown and thus coded as Rank 2 include: 1) the child is adopted and the records are closed, and 2) the birth mother is known to have a problem with drinking, but there are no records or direct observation of her drinking during the index pregnancy. A Rank 1 classification (confirmed absence of drinking from conception to birth) is relatively rare in the general population since it is unlikely to occur unless a pregnancy is either planned or the woman never drinks.

Astley, S. J. (2004). Diagnostic guide for fetal alcohol spectrum disorders: The 4-digit diagnostic code (3rd ed.). University of Washington.

Appendix B: Physical Assessment Guidelines & Resources

Physical assessment and measurements should be completed by an individual who has received training in these methods. Standardized references for measurements to be used in Minnesota are:

1. Palpebral fissure length as determined by Stromland/Scandinavian charts:

[Stromland K, Chen Y, Norberg T, Wennerstrom K, Michael G. Reference values of facial features in scandinavian children measured with a range-camera technique. Scand J Plast Reconstr Hand Surg 1999;33:59-65.](#)

A tool for computing z-scores for palpebral fissures is available at the University of Washington FASD Diagnostic Clinic website:

<https://depts.washington.edu/fasdnp/htmls/diagnostic-tools.htm#pfl>

2. Height, weight, & head circumference:

For children under 24 months, use WHO norms for height, weight, and head circumference:

https://www.cdc.gov/growthcharts/who_charts.htm#The%20WHO%20Growth%20Charts

For children 2 to 18 years of age, use Nellhaus for head circumference:

<https://www.ped.med.utah.edu/pedsintranet/clinical/references/growthCharts/nellhaus.pdf>

For children 2 years to 18 years, use CDC norms for height and weight:

https://www.cdc.gov/growthcharts/clinical_charts.htm

3. Lip-Philtrum Guide (Astley, 2014), Caucasian/African American:

<https://depts.washington.edu/fasdnp/htmls/lip-philtrum-guides.htm>

Useful links for calculating percentiles:

<https://www.infantchart.com/child/>

<https://simulconsult.com/resources/measurement.html>

<https://depts.washington.edu/fasdnp/htmls/diagnostic-tools.htm#pfl>

Appendix C: Examples of Neuropsychological Tests by Domain

Note: This is not an exhaustive list. We recommend using the most recent edition of tests when possible.

Global Cognitive Functioning

- Bayley Scales of Infant and Toddler Development
- Wechsler Intelligence Scale for Children
- Wechsler Adult Intelligence Scale
- Wechsler Abbreviated Scale of Intelligence
- Wechsler Preschool and Primary Scale of Intelligence
- Kaufman Assessment Battery for Children
- Differential Ability Scales-Second Edition-School Age
- Comprehensive Test of Nonverbal Intelligence
- Stanford Binet Intelligence Scales
- Leiter International Performance Scale

Learning Disabilities/Achievement

- Woodcock-Johnson Tests of Achievement
- Wechsler Individual Achievement Test
- Comprehensive Test of Phonological Processing
- Gray Oral Reading Tests
- Test of Written Language
- Bracken Basic Concept Scale

Attention/Executive Functioning

- NEPSY Developmental Neuropsychological Assessment
- Conners Continuous Performance Tests
- Delis-Kaplan Executive Function System
- Test of Variables of Attention – Visual/Auditory
- Test of Everyday Attention for Children
- Behavior Rating Inventory of Executive Functioning, Parent and Teacher Report
- Wisconsin Card Sorting Test

Language

- Clinical Evaluation of Language Fundamentals
- Comprehensive Assessment of Spoken Language
- Expressive Vocabulary Test
- Peabody Picture Vocabulary Test
- Preschool Language Scale

Pragmatics

- Social Language Development Test, Elementary
- Social Language Development Test, Adolescent
- Test of Pragmatic Language

Memory

- California Verbal Learning Test, Children's Version
- California Verbal Learning Test
- Children's Memory Scale

- Wechsler Memory Scales
- Child and Adolescent Memory Profile
- Rey Complex Figure Test and Recognition Trial
- Wide Range Assessment of Memory and Learning

Motor/Visual Motor

- Purdue Pegboard
- Grooved Pegboard
- Beery VMI Developmental Tests
- NEPSY Developmental Neuropsychological Assessment

Social/Adaptive Functioning

- Vineland Adaptive Behavior Scales
- Adaptive Behavior Assessment System
- Social Communication Questionnaire
- Social Responsiveness Scale

Dysregulation of Mood/Behavior

- Behavior Assessment System for Children, Parent and Teacher Report
- Behavior Rating Inventory of Executive Function
- Child Behavior Checklist
- Children's Depression Inventory,
- Multidimensional Anxiety Scale for Children
- Revised Children's Manifest Anxiety Scale
- Beck Depression Inventory
- Beck Anxiety Inventory
- Minnesota Multiphasic Personality Inventory
- Trauma Symptom Checklist for Children

Appendix D: Differential Diagnoses & Additional Considerations

Making a differential diagnosis of FAS requires clinicians to consider several genetic and teratogenic conditions that share some common features with FASD. Individual dysmorphic features are not unique to any particular syndrome. For certain syndromes (e.g. Williams syndrome, Dubowitz syndrome, Cornelia de Lange syndrome, and fetal dilantin syndrome), the overall constellation of features is similar to FAS. These syndromes should be considered when completing the differential diagnosis. In cases with facial dysmorphism, referral to a genetics clinic may be indicated.

Growth deficiencies can occur in children, adolescents, and adults for multiple reasons. Prenatal growth impairment can result from multiple factors, including maternal smoking or other behaviors leading to hypoxia, poor maternal nutrition, or genetic disorders unrelated to maternal alcohol consumption. When making an FASD diagnosis, clinicians should also consider environmental and genetic bases for growth anomalies in their differential diagnosis.

Areas of neurobehavioral impairment associated with FASD can be produced by multiple factors in addition to prenatal alcohol exposure. Organic syndromes (e.g. Williams syndrome, Dubowitz syndrome, Cornelia de Lange syndrome, and fetal dilantin syndrome) and/or environmental influences (e.g., abuse or neglect, changes in caregivers, and other sources of trauma) can impact neurophysiology and neurobehavioral functioning. To assist with diagnosis, clinicians should obtain a complete, detailed history. Clinicians should consider organic causes and environmental factors for both inclusive and exclusive purposes when evaluating a person for an FASD diagnosis.

Bertrand, J., Floyd, R. L., & Weber, M. K. (2005). Guidelines for Identifying and Referring Persons with Fetal Alcohol Syndrome. *Fetal Alcohol Syndrome Prevention Team, Division of Birth Defects and Developmental Disabilities, National Center on Birth Defect and Developmental Disabilities.*

Additional Resources:

Denny, L., Coles, S., & Blitz, R. (2017). Fetal Alcohol Syndrome and Fetal Alcohol Spectrum Disorders. *American Family Physician*, 96(8), 515-522. Retrieved from <https://www.aafp.org/afp/2017/1015/p515.html>

Differential Diagnosis: Growth Defects and Dysmorphic Features:
<https://www.aap.org/en/patient-care/fetal-alcohol-spectrum-disorders/diagnosis/differential-diagnosis-growth-defects-and-dysmorphic-features/>

Appendix E: References

- Astley, S. J. (2004). Diagnostic guide for fetal alcohol spectrum disorders: The 4-digit diagnostic code (3rd ed.). FAS Diagnostic & Prevention Network, University of Washington.
- Bertrand, J., Floyd, R. L., & Weber, M. K. (2005). Guidelines for Identifying and Referring Persons with Fetal Alcohol Syndrome. *Fetal Alcohol Syndrome Prevention Team, Division of Birth Defects and Developmental Disabilities, National Center on Birth Defect and Developmental Disabilities.*
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- Hoyme, H. E., Kalberg, W. O., Elliott, A. J., Blankenship, J., Buckley, D., Marais, A. S., ... & May, P. A. (2016). Updated clinical guidelines for diagnosing fetal alcohol spectrum disorders. *Pediatrics*, 138(2).
- Kable, J. A., O'Connor, M. J., Olson, H. C., Paley, B., Mattson, S. N., Anderson, S. M., & Riley, E. P. (2016). Neurobehavioral Disorder Associated with Prenatal Alcohol Exposure (ND-PAE): Proposed DSM-5 Diagnosis. *Pediatrics*, 138(2), e20160914. doi: 10.1542/peds.2016-0914
- National Task Force on Fetal Alcohol Syndrome and Fetal Alcohol Effects, National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, & Department of Health and Human Services. (2004, July). *Fetal alcohol syndrome: Guidelines for referral and diagnosis.*

Nellhaus, G. (1968). Head circumference from birth to eighteen years: practical composite international and interracial graphs. *Pediatrics*, *41*(1), 106-114.