Guidelines for Identifying and Referring Persons with Fetal Alcohol Syndrome
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Guidelines for Identifying and Referring Persons with Fetal Alcohol Syndrome

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Summary

Fetal alcohol syndrome (FAS) results from maternal alcohol use during pregnancy and carries lifelong consequences. Early recognition of FAS can result in better outcomes for persons who receive a diagnosis. Although FAS was first identified in 1973, persons with this condition often do not receive a diagnosis. In 2002, Congress directed CDC to update and refine diagnostic and referral criteria for FAS, incorporating recent scientific and clinical evidence. In 2002, CDC convened a scientific working group (SWG) of persons with expertise in FAS research, diagnosis, and treatment to draft criteria for diagnosing FAS. This report summarizes the diagnostic guidelines drafted by the SWG, provides recommendations for when and how to refer a person suspected of having problems related to prenatal alcohol exposure, and assesses existing practices for creating supportive environments that might prevent long-term adverse consequences associated with FAS. The guidelines were created on the basis of a review of scientific evidence, clinical expertise, and the experiences of families affected by FAS regarding the physical and neuropsychologic features of FAS and the medical, educational, and social services needed by persons with FAS and their families. The guidelines are intended to facilitate early identification of persons affected by prenatal exposure to alcohol so they and their families can receive services that enable them to achieve healthy lives and reach their full potential. This report also includes recommendations to enhance identification of and intervention for women at risk for alcohol-exposed pregnancies. Additional data are needed to develop diagnostic criteria for other related disorders (e.g., alcohol-related neurodevelopmental disorder).

Introduction

Prenatal exposure to alcohol during pregnancy damages the developing fetus and is a leading preventable cause of birth defects and developmental disabilities (1–3). Children exposed to alcohol during fetal development can suffer multiple negative effects, including physical and cognitive deficits. Although the number and severity of negative effects can range from subtle to serious, they are always lifelong.

Referral and diagnosis for fetal alcohol syndrome (FAS) can be made throughout the lifespan. However, the majority of persons with FAS are referred and receive a diagnosis during childhood. Thus, the terms “child” or “children” as used in these guidelines are not intended to preclude referral, assessment, and diagnosis of older persons.

Background

The effects of prenatal exposure to alcohol and basic diagnostic features of FAS were first described in 1973 (4–8). In 1981, the U.S. Surgeon General issued a public health advisory warning that alcohol use during pregnancy could cause birth defects (9); this warning was reissued in 2004 (10). In 1989, Congress mandated that language warning of the consequences of drinking during pregnancy be included on alcohol product labels (11).

Despite the known adverse effects of prenatal exposure to alcohol (4,5), children who experience these effects often do not receive a correct diagnosis or referral for diagnostic evaluation because of the absence of uniformly accepted diagnostic criteria and guidelines for referral. Early identification and diagnosis of FAS in affected persons are essential components to providing health, education, and social services that promote optimal well-being. In 2002, Congress directed CDC to 1) develop guidelines for diagnosing FAS and other negative birth outcomes resulting from prenatal exposure to alcohol, 2) incorporate these guidelines into curricula for medical and allied health students and practitioners, and 3) disseminate curricula concerning these guidelines to facilitate training of medical and allied health students and practitioners.
These guidelines represent a consensus of opinion from persons with expertise in relevant scientific and clinical fields, with input from service professionals and families affected by FAS. Information that served as the basis for the development of these guidelines was obtained from published scientific literature, clinical knowledge of participants, and the experience of families affected by FAS.

CDC staff initially identified reports and other documents that were used as the scientific basis for creating diagnostic guidelines. On the basis of this information, and in coordination with the National Taskforce on Fetal Alcohol Syndrome and Fetal Alcohol Effect (NTFFAS/FAE), other federally funded FAS programs, and nongovernment organizations concerned with FAS, CDC formed a scientific working group (SWG) of persons with expertise in research and clinical practice regarding prenatal exposure to alcohol to develop diagnostic guidelines for FAS.

Guidelines were formulated on the basis of consensus among SWG members and NTFFAS/FAE. To assist in defining the dysmorphic features most useful for identifying persons with FAS, SWG members assembled a matrix of the major and associated dysmorphic features of non-FAS syndromes that had one or more features in common with FAS. This matrix was used to determine a combination of dysmorphic features most discriminative for FAS. To assist deliberations concerning central nervous system (CNS) abnormalities associated with FAS, persons with expertise in the science, assessment, and treatment of psychological aspects of FAS were asked to identify the CNS abnormalities and other neurobehavioral domains most common among persons affected by prenatal alcohol exposure. These responses formed the basis for discussion and the resulting guidelines for CNS abnormalities for persons with FAS.

This report summarizes the guidelines drafted as a result of the SWG’s deliberations, provides recommendations for when and how to refer a person suspected of having problems related to prenatal alcohol exposure, and assesses existing practices for creating supportive environments that might prevent long-term adverse consequences associated with FAS.

Prevalence

Varied FAS prevalences (range: 0.2–1.5 cases per 1,000 live births) have been reported worldwide (12–15). Other studies that used different ascertainment methodologies have produced different estimates (range: 0.5–2.0 cases per 1,000 live births) (16–22). These rates are comparable with or higher than rates for other common developmental disabilities (e.g., Down syndrome or spina bifida) (23). On the basis of these prevalence estimates, approximately 4 million infants are born each year with prenatal alcohol exposure, and an estimated 1,000–6,000 are born with FAS.

Studies have reported consistently that >50% of all U.S. women of childbearing age report alcohol consumption during the previous month (1,24–28). The majority of these women drank only occasionally, but ≥13% could have been classified as moderate or heavy drinkers. In addition, 12% of women reported binge drinking (i.e., consuming five or more drinks on one occasion) during the preceding month (1,25,27,28). Approximately half of all U.S. pregnancies are unintended, and millions of women of childbearing age are sexually active while not using adequate contraception (24–28). Recent data from the Behavioral Risk Factors Surveillance System indicate that an estimated 12%–13% of U.S. women aged 18–44 years are sexually active, do not use contraception effectively, and drink alcohol frequently or binge drink, thereby putting them at risk for an alcohol-exposed pregnancy (24). Because data are available for all subpopulations, prevalences might be greater than these data indicate.

Fetal Alcohol Spectrum Disorder

Multiple terms are used to describe the continuum of effects that result from prenatal exposure to alcohol, including fetal alcohol effects, alcohol-related birth defects (ARBD), alcohol-related neurodevelopment disorder (ARND), and, most recently, fetal alcohol spectrum disorders (FASDs) (29). In April 2004, the National Organization on Fetal Alcohol Syndrome (NOFAS) convened a meeting of representatives from three federal agencies (the National Institutes of Health [NIH], CDC, and the Substance Abuse and Mental Health Services Administration [SAMHSA]) and persons with expertise in the field to develop a consensus definition of FASDs. The resulting definition, which is used in this report, defined FASDs as the range of effects that can occur in a person whose mother drank alcohol during pregnancy, including physical, mental, behavior, and learning disabilities, with possible lifelong implications. As this definition indicates, multiple diagnostic categories (e.g., FAS, ARND, and ARBD) are subsumed under the term FASDs. However, FASDs is not a diagnostic category and should be used only when referring to the collection of diagnostic terms resulting from prenatal exposure to alcohol.

Recommendations

Diagnostic Criteria

For the majority of health-care providers, the key indicator of FAS is the set of characteristic facial features first described
in 1973 (4). Alcohol is a teratogen that results in dysmorphia, growth problems, and abnormalities of the central nervous system in multiple ways (30,31).

Confirmation and documentation of prenatal alcohol exposure can be difficult to establish. For birth mothers, admission of alcohol use during pregnancy can be stigmatizing. The situation can be further complicated if the woman continues to use alcohol, especially at high consumption rates. Clinicians might need to obtain information regarding alcohol use from other reliable informants, such as a relative.

Clinicians often have to evaluate a child or adult for FAS without definitive information regarding the mother’s use of alcohol during pregnancy. This situation occurs frequently for children in foster and adoptive homes. In such cases, every effort should be made to obtain the necessary information, but lack of confirmation of alcohol use during pregnancy should not preclude a diagnosis of FAS if all other criteria are present. In rare instances, absence of exposure will be confirmed. Documentation that the birth mother did not drink any amount of alcohol from conception through birth would indicate that a FAS diagnosis is not appropriate. This finding typically implies either that the birth mother knew the date of conception (e.g., a planned pregnancy) and did not consume alcohol from that day forward or that she was prevented from drinking for a certain reason (e.g., incarceration).

Because of the imprecise nature of exposure information, the following two qualifying terms are suggested for a finding of prenatal alcohol exposure:

- FAS with confirmed prenatal alcohol exposure requires documentation of the alcohol consumption patterns of the birth mother during the index pregnancy on the basis of clinical observation; self-reports; reports of heavy alcohol use during pregnancy by a reliable informant; medical records documenting positive blood alcohol levels or alcohol treatment; or other social, legal, or medical problems related to drinking during the index pregnancy.
- FAS with unknown prenatal alcohol exposure indicates neither a confirmed presence nor a confirmed absence of exposure. Examples include situations in which the child is adopted, and any prenatal exposure is unknown; the birth mother is an alcoholic, but confirmed evidence of exposure during pregnancy does not exist; or conflicting reports regarding exposure exist that cannot be reliably resolved.

Prenatal exposure to alcohol alone is not sufficient to warrant a diagnosis of FAS. Despite the heterogeneity of expression for features related to prenatal exposure to alcohol, a diagnosis of FAS requires documentation of three findings:

1) three specific facial abnormalities; 2) growth deficit; and 3) CNS abnormalities (Appendix) (30,31) (Box).

BOX. Characteristics for diagnosing fetal alcohol syndrome

**Facial dysmorphia**
On the basis of racial norms (i.e., those appropriate for a person’s race), the person exhibits all three of the following characteristic facial features:

- smooth philtrum (University of Washington Lip-Philtrum Guide* rank 4 or 5*),
- thin vermilion border (University of Washington Lip-Philtrum Guide rank 4 or 5), and
- small palpebral fissures (<10th percentile).

**Growth problems**
Confirmed, documented prenatal or postnatal height, weight, or both ≤10th percentile, adjusted for age, sex, gestational age, and race or ethnicity.

**Central nervous system abnormalities**

**Structural**

- Head circumference ≤10th percentile, adjusted for age and sex
- Clinically meaningful brain abnormalities observable through imaging (e.g., reduction in size or change in shape of the corpus callosum, cerebellum, or basal ganglia)

**Neurologic**

- Neurologic problems (e.g., motor problems or seizures) not resulting from a postnatal insult or fever, or other soft neurologic signs outside normal limits

**Functional**

Test performance substantially below that expected for a person’s age, schooling, or circumstances, as evidenced by either:

- global cognitive or intellectual deficits representing multiple domains of deficit (or substantial developmental delay in younger children) with performance below the third percentile (i.e., two standard deviations below the mean for standardized testing); or
- functional deficits <16th percentile (i.e., one standard deviation below the mean for standardized testing) in at least three of the following domains:
  - cognitive or developmental deficits or discrepancies,
  - executive functioning deficits,
  - motor functioning delays,
  - problems with attention or hyperactivity,
  - social skills, or
  - other (e.g., sensory problems, pragmatic language problems, or memory deficits).


Considerations When Diagnosing FAS

Because FAS is a syndrome rather than a specific disease, additional features can be present. For example, in addition to the key facial dymorphic features, maxillary hypoplasia is often noted for persons with FAS (3). Features often change with age or development. After puberty, the characteristic facial features associated with FAS can become more difficult to detect (32). However, the key features remain constant for the majority of persons with FAS (33,34).

Changes in growth pattern across development also lead to variability in presentation. For certain affected persons, growth problems might occur at a younger age but not be present at the time of the diagnostic evaluation. This is particularly important when considering prenatal growth retardation or early growth problems caused by failure to thrive. Because multiple treatments exist for growth problems (e.g., use of feeding tubes or hormone therapy), any history of growth retardation, including prenatal growth deficiencies, is consistent with the criteria for diagnosing FAS (35). The clinician should be certain that the child was not nutritionally deprived at the single point in time when the growth deficit was present. The adopted threshold for growth (<10th percentile) represents an attempt to maximize sensitivity, even though it reduces specificity.

CNS Abnormalities

The diagnostic criteria for CNS abnormality require documentation of one of three types of deficits or abnormalities (i.e., structural, neurologic, and functional). A person might have more than one CNS abnormality (36). Identifying CNS abnormalities resulting from prenatal alcohol exposure can be the most difficult aspect of a FAS diagnosis because of the heterogeneity of expression for these deficits across persons (Appendix).

Approximately one fourth of persons who receive a diagnosis of FAS perform at two standard deviations below the mean (which approaches substantial impairment [i.e., mental retardation]) on standardized measures of cognition (37). To capture the full spectrum of effects adequately, two levels of functional deficits are consistent with the criteria for a CNS abnormality: 1) performance below the third percentile (i.e., two standard deviations below the mean) on a measure of global cognitive functioning or 2) performance <16th percentile (i.e., one standard deviation below the mean) on standardized measures of three functional domains. Thus, persons scoring below the normal range on a global measure of intelligence or development and persons scoring in the below-average range on standardized measures of three specific functional domains would be consistent with the criteria for functional CNS abnormality for diagnostic purposes.

Because of the importance of documenting CNS abnormalities and the variability in functional deficits, the diagnostic process should include a thorough neuropsychologic evaluation that assesses multiple domains. Extensive standardized testing might not be readily available in all diagnostic settings. Clinicians are encouraged to supplement their observations by obtaining standardized testing through early intervention programs, public schools, and psychologists in private practice. Such testing will facilitate the development of appropriate personalized treatment plans for persons who receive a diagnosis of FAS. These guidelines recommend that functional domains be assessed by using norm-referenced standardized measures. Assessments should be conducted by professionals using reliable and validated instruments.

Differential Diagnosis

Individual dysmorphic features are not unique to any particular syndrome. Even rare defects or certain clusters of dysmorphic features can appear in multiple syndromes. Therefore, a process of differential diagnosis is essential in making an accurate FAS diagnosis. Features that discriminate these disorders from FAS have been described (38). Certain syndromes have single overlapping features with FAS. With the exception of toluene embryopathy, no other known syndrome has the full constellation of small palpebral fissures, thin vermilion border, and smooth philtrum. However, for certain syndromes (e.g., Williams syndrome, Dubowitz syndrome, or fetal dilantin syndrome), the overall constellation of features (primary, occasional features, or both) is similar to FAS, and these syndromes should be considered in particular when completing the differential diagnosis.

Growth retardation and deficiencies occur among children, adolescents, and adults for multiple reasons. Insufficient nutrition could be a particular problem for infants with poor sucking responses who fail to thrive. In addition, certain genetic disorders result in specific growth deficiencies (e.g., dwarfism). Prenatal growth retardation 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. Both environmental and genetic bases for growth retardation should be considered for differential diagnosis when considering a FAS diagnosis. Finally, because a threshold of <10th percentile (rather than the lower threshold of the third percentile commonly used to denote growth retardation) was adopted, certain children will be classified as being consistent with this
criterion for reasons other than prenatal exposure to alcohol (e.g., parents having short stature). However, because the diagnosis of FAS is made only when facial dysmorphia and CNS abnormalities also are present, the increased sensitivity achieved with the 10th percentile was selected.

Differential diagnosis of CNS abnormalities involves not only ruling out other disorders but also specifying simultaneously occurring disorders. CNS deficits associated with FAS (especially functional deficits) can be produced by multiple factors in addition to prenatal alcohol exposure. Observed functional deficits should be determined not to be better explained by other causes. In addition to other organic syndromes that produce deficits in one or more of the previously cited domains (e.g., Williams syndrome and Down syndrome), disrupted home environments or other external factors can produce functional deficits in multiple domains that overlap those affected by FAS. In making a differential diagnosis of FAS, the clinician should evaluate CNS abnormalities in conjunction with dysmorphia and laboratory findings. CNS abnormalities resulting from environmental influences (e.g., abuse or neglect, disruptive homes, and lack of opportunities) are harder to differentiate. To assist with differential diagnosis between FAS and environmental causes for CNS abnormalities, clinicians should obtain a complete, detailed history for the person and family members.

In addition to ruling out other causes for CNS abnormalities, a complete diagnosis should identify and specify other disorders that can coexist with FAS (e.g., autism, conduct disorder, or oppositional defiant disorder). A particular person might have a conduct disorder in addition to FAS; however, not all persons with FAS have conduct disorders, and not all persons with conduct disorders have FAS. Certain functional deficits might lead to additional behavior problems. For example, a child with an attention problem also could have conduct disorder. Clinicians should consider organic causes, environmental contributions, and comorbidity for both inclusive and exclusive purposes when evaluating a person for a FAS diagnosis (32,39). Because differential diagnosis for CNS abnormalities within a FAS diagnosis is difficult, the evaluation should be conducted by professionals trained in both the features of FAS and those of a broad array of birth defects and developmental disabilities.

**Conditions Consistent with a Subset of Diagnostic Criteria for FAS**

The majority of persons with deficits resulting from prenatal exposure to alcohol do not express all the features necessary for a FAS diagnosis (36). Sufficient scientific evidence is not available to define diagnostic criteria for any prenatal alcohol-related condition other than FAS. Persons who have the neurodevelopmental deficits required for a FAS diagnosis but who do not have all three facial features or growth deficits might not receive a diagnosis and so not be provided with services. Ongoing funding has been provided by the National Institute on Alcohol Abuse and Alcoholism to conduct research that might lead to evidence-based diagnostic criteria for persons with other conditions caused by prenatal alcohol use. CDC is using a collaborative database of neurodevelopment data from five intervention studies to explore the nature of persons who could be considered in the diagnostic category of alcohol related neurodevelopment disorder, as well as data from a prospective cohort study in Denmark of children aged 5 years. FAS is the only diagnostic category with scientific evidence to support clinical criteria at this time. As future data become available, these guidelines can be refined and expanded to delineate other conditions resulting from prenatal alcohol exposure.

**Mental Health Problems and Other Lifelong Consequences**

FAS has lifelong consequences. Common FAS-related mental health conditions (excluding attention problems) reported include conduct disorders, oppositional defiant disorders, anxiety disorders, adjustment disorders, sleep disorders, and depression (37,40–44). Although attention problems can be classified as a mental health issue or psychiatric condition, in these guidelines, they are treated as a primary deficit resulting from alcohol-related CNS damage rather than a secondary mental health concern (45). Decreased adaptive skills and increased problems with daily living abilities have been documented (e.g., dependent living conditions, disrupted school experiences, poor employment records, and encounters with law enforcement, including incarceration) among persons with FAS (37). These mental health–related consequences should not be used for diagnosis. However, they are prevalent among persons with FAS and are likely to result in referral and comprehensive diagnostic evaluation.

**Referral Considerations**

Providers of medical, educational, and social services often must decide whether to refer a child, person, or family to a specialist for a full FAS diagnostic evaluation. This decision can be difficult. For biologically related family members, social stigma might be associated with any evaluation concerning prenatal alcohol exposure. In adoptive or foster families, alcohol use during pregnancy might be suspected, but direct information might not be available.
The following guidelines were developed to assist service providers in making referral decisions. Each case should be evaluated individually. When in doubt, providers should refer persons for a full evaluation by a multidisciplinary team with experience in evaluating prenatal alcohol exposure.

The following circumstances should prompt a diagnostic referral:

- When prenatal alcohol exposure is known, a child should be referred for full FAS evaluation when substantial prenatal alcohol use (i.e., seven or more drinks per week, three or more drinks on multiple occasions, or both) has been confirmed. If substantial prenatal alcohol exposure is known, in the absence of any other positive criteria (i.e., dysmorphia, growth deficits, or CNS abnormalities), the primary health-care provider should document this exposure and monitor the child's ongoing growth and development closely.

- When information regarding prenatal alcohol exposure is unknown, a child should be referred for full FAS evaluation for any one of the following:
  
  - any report of concern by a parent or caregiver (e.g., foster or adoptive parent) that a child has or might have FAS;
  
  - presence of all three facial features (i.e., smooth philtrum, thin vermillion border, and small palpebral fissures);
  
  - presence of one or more of these facial features, with growth deficits in height, weight, or both;
  
  - presence of one or more facial features, with one or more CNS abnormalities; or
  
  - presence of one or more facial features, with growth deficits and one or more CNS abnormalities.

In addition to specific features associated with a FAS diagnosis, certain social and family history factors have been associated with prenatal alcohol exposure (46). The possibility of prenatal alcohol exposure should be considered fully for persons who are experiencing or have experienced one or more of the following:

- premature maternal death related to alcohol use (either disease or trauma),
- living with an alcoholic parent,
- current or previous abuse or neglect,
- current or previous involvement with child protective services agencies (PSAs),
- a history of transient caregiving situations, or
- foster or adoptive placements (including kinship care).

Although such situations might have a negative impact on the development of any child, evidence exists that children with FAS or a related disorder are particularly likely to experience negative situations that involve a dysfunctional family unit (46), especially if the biologic mother abuses alcohol.

### Services for Persons with FAS

For persons with developmental disabilities and their families, diagnosis is never an endpoint. This is particularly true for persons with FAS, their families, and their communities. The diagnostic process (especially the neuropsychologic assessment) should be part of a continuum of care that identifies and facilitates appropriate health-care, education, and community services. Early diagnosis and a stable, nurturing home environment have been identified as strong protective factors for persons with FAS (46). Limited information is available regarding strategies for interventions specific to persons with FAS. Information available has been gathered primarily from the experience of persons with other disabilities and from that of parents gained through trial and error and shared through informal networks. Treatments currently employed to reduce the risk for adverse effects of FAS have not been evaluated systematically or scientifically. In 2001, CDC provided the first federal funding to develop and test systematic, scientifically developed interventions specific to FAS (e.g., a modified mathematics curriculum or a program to develop peer friendship skills). These projects are in their final stages, and findings will be published.

The learning and life skills affected by prenatal alcohol exposure vary among persons, depending on the amount, timing, and pattern of exposure and on each person's current and past environment (47,48). As a result, services needed for persons with FAS and their families vary according to the parts of the brain affected, the person's age or level of maturation, the health or functioning of the family, and the person's overall living environment. Thus, the service needs of affected persons and their families should be individualized (49). Certain general areas of service and specific services have been identified as helpful to persons with FAS and their families (32).

Interventions should include strategies that stabilize home placement and improve parent-child interaction (47). One means of accomplishing this goal is to increase the understanding of the disorder among parents, teachers, law enforcement personnel, and other professionals who might become involved with the affected person. Children with FAS often need specialized parenting techniques because of their difficulty with cause-and-effect reasoning and other executive functioning skills (47). Caregiver education should highlight and explain differences in the thought processes of children with FAS compared with typically developing children and children with other developmental disabilities. This knowl-
edge would enable parents to avoid potentially difficult situations (e.g., overly stimulating environments) and better manage problems when they do arise. Overall, a better functioning family that results from caregiver education promotes the stable, nurturing home that has been demonstrated to be a protective factor for children with FAS (50).

Professionals who work with persons affected by FAS could benefit from better understanding of the disorder and services available for affected persons and their families (39). These professionals can help link families with needed community resources and ensure that affected children receive maximum benefit from services provided. Interacting with social and educational service agencies can be overwhelming and confusing, and each agency typically uses a specialized vocabulary (i.e., jargon) that is difficult for nonspecialists to understand. In addition to being able to diagnose FAS, clinicians should help parents and caregivers identify available services, determine which ones are effective for their children, and understand how to work productively with service providers (32).

Prenatally exposed infants and children often enter the foster or adoptive care system at an early age. The prevalence of children with FAS or a related disorder in the foster care system is estimated to be 10 times that of the general population (51). Although PSAs might have information regarding a child’s prenatal history, PSA staff generally do not know about FAS, understand how FAS affects the child, or communicate with other service systems regarding the child’s FAS status (51). As a result, foster and adoptive families typically are not educated regarding the long-term effects of FAS and are unprepared to meet their children’s needs.

The majority of PSAs require foster parents to take a specified number of educational courses annually. These courses should include education regarding the effects and developmental needs of children with FAS because the majority of foster parents will encounter at least one child with FAS or a related disorder during their time as a foster parent (51). Projects funded by CDC have developed FAS curricula for parents, educators, and juvenile justice systems; information regarding these curricula is available at http://www.cdc.gov/ncbddd/fas/awareness.htm.

The assessment process is integral to both the FAS diagnosis and the development of an effective treatment plan. Such a treatment plan minimizes risk factors for lifelong negative consequences and promotes protective factors that maximize developmental potential. Clinicians and service providers must ensure that assessments include communication and social skills, emotional maturity, verbal and comprehension abilities, language usage, and, if appropriate, referral for medication assessments. Finally, the health and development of children with disabilities, including children with FAS, can be promoted by public support for programs that provide access to school, recreational, and social activities.

Alcohol Use During Pregnancy

Because no safe threshold of alcohol use during pregnancy has been established, CDC and NTFFAS/FAE recommend that women who are pregnant, planning a pregnancy, or at risk for pregnancy should not drink alcohol. Women of childbearing age who are not pregnant should drink no more than seven drinks per week and no more than three drinks on any one occasion.

Federal, state, and local agencies; clinicians and researchers; educational and social service professionals; and families should work together to educate women of childbearing age and communities countrywide regarding the risks of drinking alcohol during pregnancy. Women who have had at least one child with FAS are at especially high risk for giving birth to a second affected child (2,52). Universal screening for alcohol use among all women of childbearing age might help identify women who drink above recommended levels as well as those who drink and might become pregnant. Screening can be performed in clinicians’ offices or in community health settings. Screening techniques that include measures of quantity, frequency, and heavy episodic drinking, as well as behavioral manifestations of risk drinking, have proven to be most beneficial; simple questionnaires have been developed to screen for problematic alcohol use among adults in multiple populations and settings (53).

Effective prevention programs frequently employ a multi-component approach that combines cognitive-behavioral techniques with norms clarification, education, and motivational enhancement interventions. For women who screen positive for hazardous alcohol use or abuse, brief interventions that use time-limited, self-help, and preventative strategies to promote reductions in alcohol use in nondependent persons and that facilitate referral of dependent persons to specialized treatment programs are low-cost, effective treatment alternatives (54–57). The acronym FRAMES is used to encompass six key elements of the majority of successful brief interventions as follows: 1) feedback of personal risk, 2) responsibility for personal control, 3) advice to change, 4) menu of ways to reduce or stop drinking, 5) empathetic counseling style, and 6) self-efficacy or optimism regarding reducing or stopping drinking (58). Preconception counseling of women of childbearing age who are at risk for an alcohol-exposed pregnancy and who are not using effective contraception has been demonstrated as a promising method of prevention (59). Project

Summary of Recommendations

On the basis of a review of current scientific and clinical evidence, the following recommendations are made concerning referral of children and diagnosis of FAS:

Diagnosis of FAS

- A diagnosis of FAS should be made if documentation exists of 1) all three dysmorphic facial features (i.e., smooth philtrum, thin vermilion border, and small palpebral fissures), 2) prenatal or postnatal growth deficit in height or weight, and 3) CNS abnormality.
- The diagnosis should be classified on the basis of available history as confirmed prenatal alcohol exposure or unknown prenatal alcohol exposure.
- CNS abnormality may be documented as structural, neurologic, or functional (Box).

Referral

- If prenatal alcohol exposure is known, a child or person should be referred for full FAS evaluation when alcohol abuse (defined as seven or more alcohol drinks per week or three or more alcohol drinks on multiple occasions, or both) is confirmed.
- If prenatal alcohol exposure is unknown, a child or person should be referred for full FAS evaluation when:
  — a parent or caregiver (foster or adoptive parent) reports that a child has or might have FAS;
  — all three facial features (i.e., smooth philtrum, thin vermilion border, and small palpebral fissures) are present;
  — one or more facial features are present in addition to growth deficits in height, weight, or both; one or more facial features are present and one or more CNS abnormalities; or
  — one or more facial features are present, with growth deficits and one or more CNS abnormalities.
- In addition to specific features associated with the FAS diagnosis, the following social and family history factors associated with prenatal exposures to alcohol might indicate a need for referral:
  — premature maternal death related to alcohol use (either disease or trauma),
  — living with an alcoholic parent,
  — current or previous abuse or neglect,
  — current or previous involvement with child PSAs,
  — a history of transient caregiving situations, or
  — having been in foster or adoptive care (including kinship care).

Services

- The FAS diagnosis and the diagnostic process (especially the neuropsychologic assessment) should be considered as part of a continuum of care that identifies and facilitates appropriate health-care, education, and community services.
- General areas of service needs for persons with FAS and their families should include strategies that stabilize home placement, improve parent-child interaction through caregiver education, advocate for access to services, and educate service professionals involved with affected persons and their families regarding FAS and its consequences.
- Specific intervention services should be tailored to a person's individual needs and deficits. These might include communication and social skills; emotional development; verbal and comprehension abilities; language usage; and, if appropriate, referral for medication assessments.
- The needs of children in adoptive or foster placements should receive particular attention in the diagnostic and referral process.

Prevention

- Federal, state, and local agencies; clinicians and researchers; educational and social service professionals; and families should work together to educate women of childbearing age and communities countrywide regarding the risks of drinking alcohol during pregnancy.
- Universal screening by health-care providers for alcohol use is recommended for all women of childbearing age.
- For women drinking at risk levels and not effectively using contraception, brief interventions have proven effective in reducing the risk for an alcohol-exposed pregnancy.
- Because no safe threshold of alcohol use during pregnancy has been established, women who are pregnant, planning a pregnancy, or at risk for pregnancy should be advised not to drink alcohol. Women who are not pregnant, not planning a pregnancy, or not at risk for unintended preg-
nancy should be advised to drink no more than seven drinks per week and no more than three drinks on any one occasion.

- Additional information regarding these guidelines has been published (60).

References

25. CDC. Alcohol consumption among women who are pregnant or who might become pregnant—United States. MMWR 2004;53:1178–81.
Appendix

Central Nervous System Abnormalities Associated with Fetal Alcohol Syndrome (FAS)

Central nervous system (CNS) abnormalities are classified as structural, neurologic, or functional.

**Structural**
- Documented evidence exists of small or diminished overall head circumference (also known as occipito-frontal circumference [OFC]) (i.e., ≤10th percentile), adjusted for age and sex (including head circumference at birth; (1,2). For children who have overall growth deficiency (i.e., height and weight <10th percentile) to meet this criteria for CNS abnormality, the child's head circumference should be disproportionately small to the child's overall size (i.e., OFC at or below the third percentile).
- Clinically significant brain abnormalities are observable through imaging techniques (e.g., reduction in size or change in shape of the corpus callosum, cerebellum, or basal ganglia), as assessed by an appropriately trained professional (1,3–10). A child could have a structural abnormality that would be consistent with a diagnosis of fetal alcohol syndrome (FAS) but not demonstrate detectable functional deficits.

**Neurologic**
- Documented evidence exists of CNS neurologic damage. Neurologic problems can include seizures not attributable to a postnatal insult or fever or other soft neurologic signs outside normal limits (e.g., coordination problems, visual motor difficulties, nystagmus, or difficulty with motor control) (11–14). As with head circumference, abnormal neurologic findings among younger children can be most predictive of underlying CNS abnormality resulting from prenatal alcohol exposure rather than later environmental factors. Use of norm-referenced measures of neurologic functioning is recommended.

**Functional**
- Assessment findings indicate deficits, problems, or abnormalities in functional CNS skills. Early brain damage is usually generalized rather than specific, with increased specificity of abnormalities revealed as development progresses. Functional abilities affected by prenatal exposure to alcohol vary from person to person, depending on the amount, timing, and pattern of alcohol exposure (e.g., chronic exposure versus binge episodes). Despite this inherent variation in effects, areas of functional vulnerability have been observed consistently by clinicians and researchers with particular damage to corresponding structures reported (e.g., corpus callosum, cerebellum, or basal ganglia).

For functional deficits, multiple locations in the brain (and corresponding functional capability) are generally accepted to be affected by prenatal exposure to alcohol. Functional deficits consistent with CNS abnormality criteria can be identified in two ways: 1) global cognitive deficit (e.g., decreased IQ) or substantial developmental delay in children too young for an IQ assessment or 2) deficits in three or more specific functional domains. These two ways of assessing functional CNS abnormality were adopted because of the composite nature of cognitive, intellectual, and developmental measures (15,16). Decreased performance on a standardized measure of cognition, intelligence, and development assumes deficits in multiple domains. In the absence of such a measure, multiple domains should be assessed individually to determine that multiple functional domains have been affected. For each domain, other agents and environmental factors can produce deficits or outcomes similar to prenatal alcohol exposure, making differential diagnosis essential.

The specific domains most often cited as areas of deficit or concern for persons with FAS are described below. These descriptions are intended to be suggestive and are examples of likely and possible problems a clinician might encounter and need to assess by using psychometric instruments. The examples are not intended to be exhaustive or to present a list of behaviors to be used as a checklist without reliable and valid assessment.

- **Cognitive deficits or significant developmental discrepancies.** Global deficits or delays can leave the child scoring in the normal range of development but below what would be expected for the child's environment and background (17–22). In addition to formal testing (either through records or current testing), behaviors that might be observed or reported in the clinical setting that suggest cognitive deficits or developmental delays that should be assessed by standardized testing include but are not limited to specific learning disabilities (especially mathematic or visual-spatial deficits), uneven profile of
cognitive skills, low academic achievement, discrepancy between verbal and nonverbal skills, and slowed movements or reaction to persons and stimuli (e.g., difficulty in processing information) (23–27).

• **Executive functioning deficits.** Executive functioning (EF) is defined as the ability to maintain an appropriate problem solving set for attainment of a future goal. This ability includes the more specific skills of inhibition, planning, and mental representation (28). Behaviors that can be observed or reported in the clinical setting that might indicate an EF deficit that should be assessed by standardized testing include but are not limited to inadequate organization, planning, or strategy use; concrete thinking; lack of inhibition; difficulty grasping cause and effect; inability to delay gratification; difficulty following multistep directions; difficulty changing strategies or thinking of things in a different way (i.e., perseveration); impaired judgment; and inability to apply knowledge to new situations (29–33).

• **Motor functioning delays or deficits.** Both gross and fine motor skills can be impaired for persons with FAS (34,35). Visual-motor/visual-spatial coordination is a particularly vulnerable area of functioning (22,36–38). Behaviors that can be observed or reported in the clinical setting that indicate motor problems that should be assessed by standardized testing include but are not limited to delayed motor milestones, difficulty with writing or drawing, clumsiness, balance problems, tremors, and poor dexterity. For infants, a poor suck is often observed (17,38–40).

• **Attention and hyperactivity problems.** Attention problems are often noted for children with FAS, with children frequently receiving a diagnosis of attention-deficit hyperactivity disorder (ADHD) (41). Although such a diagnosis can be applied, attention problems for children with FAS do not appear to be consistent with the classic pattern of ADHD. Persons with FAS tend to have difficulty with the encoding of information and flexibility (shifting) aspects of attention, whereas children with ADHD typically display problems with focus and sustaining attention (42,43). Persons with FAS also can appear to display hyperactivity because their impulsivity might lead to increased activity levels. Behaviors that might be observed or reported in the clinical setting that suggest attention problems related to FAS that should be assessed by standardized testing include but are not limited to being described by adults as “busy,” inattentive, easily distracted, difficulty calming down, being overly active, difficulty completing tasks, and/or trouble with transitions. Parents might report inconsistency in attention from day to day (e.g., “on” days and “off” days) (44–50).

• **Social skills problems.** The executive, attention, and developmental problems described previously often lead to clinically significant difficulty for persons with FAS when interacting with peers and others. Because of the mental representation problems, persons with FAS often have social perception or social communication problems that make it difficult for them to grasp the more subtle aspects of human interactions (51,52). Consistent difficulty understanding the consequence of behavior or inappropriate behavior frequently is described for persons with FAS (53,54). Behaviors that can be observed or reported in the clinical setting that indicate these types of social difficulties that should be assessed by standardized testing include but are not limited to lack of fear of strangers, naiveté and gullibility, being taken advantage of easily, inappropriate choice of friends, preferring younger friends, immaturity, superficial interactions, adaptive skills significantly below cognitive potential, inappropriate sexual behaviors, difficulty understanding the perspective of others, poor social cognition, and clinically significant inappropriate initiations or interactions (55–57). Standardized assessment of social problems can be difficult; social functioning is a multifaceted domain that can require multiple areas of assessment.

• **Other potential domains that can be affected.** In addition to these five most-often-cited problem areas, deficits and problems to be assessed by standardized testing can present in several other areas, including sensory problems (e.g., tactile defensiveness and oral sensitivity), pragmatic language problems (e.g., difficulty reading facial expression, and poor ability to understand the perspectives of others), memory deficits (e.g., forgetting well-learned material, and needing many trials to remember), and difficulty responding appropriately to common parenting practices (e.g., not understanding cause-and-effect discipline). Although abnormalities in these areas have been reported for persons with FAS, deficits in these areas are reported at a lower frequency than are those in the other five specific domains (53).

References


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Goal and Objectives

This report provides updated criteria for diagnosis of fetal alcohol syndrome (FAS) among persons affected by prenatal alcohol exposure. The goal of this report is to provide guidance for health-care providers in determining which persons might need referral for a complete multidisciplinary diagnostic evaluation and information regarding medical, educational, social, and family services appropriate for affected persons. Upon completion of this educational activity, the reader should be able to 1) describe the negative outcomes associated with prenatal alcohol exposure; 2) list the specific criteria that constitute a diagnosis of FAS; 3) identify persons who should receive a referral for a multidisciplinary evaluation for FAS; 4) list services appropriate for a person receiving a FAS diagnosis; and 5) list instruments appropriate for screening women of childbearing age for alcohol use or abuse.

To receive continuing education credit, please answer all of the following questions.

1. No amount of alcohol, type of alcohol, or time to consume alcohol is safe during pregnancy.
   A. True.
   B. False.

2. The cognitive deficits and behavioral problems resulting from prenatal exposure to alcohol...
   A. are present only at birth.
   B. resolve by age 3 years.
   C. resolve by puberty.
   D. are lifelong.

3. What percentage of sexually active women of childbearing age do not use contraception effectively and drink alcohol frequently or binge drink, putting them at risk for an alcohol-exposed pregnancy?
   A. 1%–2%.
   B. 12%–13%.
   C. 20%–40%.
   D. 50%–75%.

4. The diagnosis of FAS includes which of the following criteria?
   A. Documentation of all three facial abnormalities (i.e., smooth philtrum, thin vermillion, and small palpebral fissures).
   B. Documentation of growth deficits.
   C. Documentation of central nervous system abnormalities.
   D. Documentation of mental retardation.
   E. A, B, and C.
   F. A and D.

5. Which of the following statements is true?
   A. One of the diagnostic criteria for FAS is mental retardation.
   B. Persons who have been exposed to alcohol prenatally but whose physical condition is not consistent with the criteria for FAS might have substantial cognitive deficits.
   C. All persons with FAS have attention deficit hyperactivity disorder.
   D. Persons with FAS are likely to have speech and language impairments but not fine motor deficits.

6. A person should be referred for a complete multidisciplinary diagnostic evaluation when...
   A. all three facial features are present.
   B. any concern is reported by a parent or caregiver that a child has or might possibly have been exposed to alcohol prenatally.
   C. a child is living with an alcoholic parent, or the biological mother died as a result of alcohol-related disease or trauma.
   D. all of the above.

7. The diagnostic process, especially the neuropsychologic assessment, should be part of a continuum of care that identifies and facilitates appropriate health-care, education, and community services.
   A. True.
   B. False.

8. Simple alcohol screening techniques that are most beneficial include...
   A. measure of quantity.
   B. measure of frequency.
   C. measure of heavy episodic drinking.
   D. measure of behavioral manifestations of risk drinking.
   E. all of the above.

9. Which best describes your professional activities:
   A. Physician.
   B. Nurse.
   C. Health educator.
   D. Office staff.
   E. Other

10. I plan to use these recommendations as the basis for...(Indicate all that apply.)
    A. health education materials.
    B. insurance reimbursement policies.
    C. local practice guidelines.
    D. public policy.
    E. other.

11. Overall, the length of the journal article was...
    A. much too long.
    B. a little too long.
    C. just right.
    D. a little too short.
    E. much too short.

12. After reading this report, I am confident I can describe the negative outcomes associated with prenatal exposure to alcohol.
    A. Strongly agree.
    B. Agree.
    C. Undecided.
    D. Disagree.
    E. Strongly disagree.

13. After reading this report, I am confident I can list the specific criteria that constitute a diagnosis of FAS.
    A. Strongly agree.
    B. Agree.
    C. Undecided.
    D. Disagree.
    E. Strongly disagree.

14. After reading this report, I am confident I can identify persons who should receive a referral for a multidisciplinary evaluation for FAS.
    A. Strongly agree.
    B. Agree.
    C. Undecided.
    D. Disagree.
    E. Strongly disagree.
15. After reading this report, I am confident I can list services appropriate for a person receiving a FAS diagnosis.
   A. Strongly agree.  
   B. Agree.  
   C. Undecided.  
   D. Disagree.  
   E. Strongly disagree.

16. After reading this report, I am confident I can list instruments appropriate for screening women of childbearing age for alcohol use or abuse.
   A. Strongly agree.  
   B. Agree.  
   C. Undecided.  
   D. Disagree.  
   E. Strongly disagree.

17. The learning outcomes (objectives) were relevant to the goal of this report.
   A. Strongly agree.  
   B. Agree.  
   C. Undecided.  
   D. Disagree.  
   E. Strongly disagree.

18. The instructional strategies used in this report (text, box, and appendix) helped me learn the material.
   A. Strongly agree.  
   B. Agree.  
   C. Undecided.  
   D. Disagree.  
   E. Strongly disagree.

19. The content was appropriate given the stated objectives of the report.
   A. Strongly agree.  
   B. Agree.  
   C. Undecided.  
   D. Disagree.  
   E. Strongly disagree.

20. The content expert(s) demonstrated expertise in the subject matter.
   A. Strongly agree.  
   B. Agree.  
   C. Undecided.  
   D. Disagree.  
   E. Strongly disagree.

21. Overall, the quality of the journal article was excellent.
   A. Strongly agree.  
   B. Agree.  
   C. Undecided.  
   D. Disagree.  
   E. Strongly disagree.

22. These recommendations will improve the quality of my practice.
   A. Strongly agree.  
   B. Agree.  
   C. Undecided.  
   D. Disagree.  
   E. Strongly disagree.

(Continued on pg CE-4)

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23. The availability of continuing education credit influenced my decision to read this report.
   A. Strongly agree.
   B. Agree.
   C. Undecided.
   D. Disagree.
   E. Strongly disagree.

24. The *MMWR* format was conducive to leaning this content.
   A. Strongly agree.
   B. Agree.
   C. Undecided.
   D. Disagree.
   E. Strongly disagree.

25. Do you feel this course was commercially biased? *(Indicate yes or no; if yes, please explain in the space provided.)*
   A. Yes.
   B. No.

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   B. Advertisement (e.g., fact sheet, *MMWR* cover, newsletter, or journal).
   C. Coworker/supervisor.
   D. Conference presentation.
   E. *MMWR* subscription.
   F. Other.

Correct answers for questions 1–8.
1. A
2. D
3. B
4. E
5. B
6. D
7. A
8. E