

“A Window of Opportunity”: The Proposed Inclusion of Fetal Alcohol Spectrum Disorder in the DSM-5

Abstract

FASD is a lifelong disability resulting in clinically significant dysfunction that frequently leads to adverse life outcomes for those affected. Yet, FASD diagnoses are often insufficient to obtain the services and supports that clients need to prevent these secondary disabilities. A Diagnostic and Statistical Manual of Mental Disorders (DSM) diagnosis is widely considered a gateway to treatment and support. Efforts to include FASD in the DSM appear to have begun in the last 10 years. A review of these efforts suggests that reimbursement issues and the enabling of services have been much of the impetus for promoting in the inclusion of FASD in the DSM. Criteria for a FASD-related condition, Neurobehavioural Disorder Associated with Prenatal Alcohol Exposure (ND-PAE), have been proposed in section III of the DSM-5 as a condition that requires further research. Criteria include the presence of neurocognitive impairment, impairment in self-regulation, deficits in adaptive functioning, and confirmation of more than minimal alcohol exposure any time during gestation. This is a departure from current FASD diagnostic guidelines, which assess growth restriction and facial features in addition to central nervous system dysfunction and prenatal alcohol confirmation.

Researchers have identified a host of cognitive deficits associated with exposure to alcohol in utero, including deficits in intellectual abilities, learning and memory, language, attention, executive functioning, visuospatial abilities, motor skills, and adaptive and social functioning (Riley & McGee, 2005). These primary cognitive deficits have been shown to be associated with a host of secondary disabilities, including mental illness (Famy, Streissguth, & Unis, 1998), psychiatric disorders (O'Connor & Paley, 2009), and numerous adverse life outcomes (Streissguth, Bookstein, Barr, Sampson, O'Malley, & Young, 2004).

Fetal Alcohol Syndrome (FAS), as a diagnostic term, emerged with the discovery of the negative effects of alcohol on the developing fetus (Jones & Smith, 1973). FAS conveys the most salient consequences of prenatal alcohol exposure (PAE) including facial dysmorphology, growth restriction, and central nervous system (CNS) impairment. It was soon recognized that alcohol exposure in utero could impact the developing brain even in the absence of characteristic facial features and growth restriction (Aase, Jones, & Clarren, 1995). Fetal Alcohol Spectrum Disorder (FASD) is an umbrella term used to describe an array of neurocognitive and neurodevelopmental disabilities associated with PAE (Chudley,

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Conry, Cook, Loock, Rosales, & LeBlanc, 2005). Diagnostic terms such as Alcohol-Related Neurodevelopmental Disorder (ARND), Partial FAS, and Alcohol Related Birth Defects fall under the umbrella of FASD.

Although PAE is associated with these significant and costly deficits, diagnostic categories for FASD are not available in the Diagnostic and Statistical Manual of Mental Disorders, Fourth Edition – Text Revision (DSM-IV-TR) (American Psychiatric Association; APA, 2000). FAS is a diagnosis included in the International Classification of Diseases – 10th edition (ICD-10), under the heading Congenital Malformation Syndromes due to Known Exogenous Causes, Not Elsewhere Classified (World Health Organization; WHO, 2004). Other disorders on the Fetal Alcohol Spectrum such as ARND are not included in the ICD-10.

A disorder describing the negative consequences of PAE has been proposed for inclusion in DSM-5. In May, 2012, criteria for a newly-described condition, Neurobehavioural Disorder Associated with Prenatal Alcohol Exposure (ND-PAE), were proposed in Section III of DSM-5 as a condition that requires further research, under the category of Substance Use and Addictive Disorders (APA, n.d.). Criteria include, the presence of neurocognitive impairment, impairment in self-regulation, deficits in adaptive functioning, and confirmation of more than minimal alcohol exposure any time during gestation.

Disorders Caused by Prenatal Alcohol Exposure – A Significant Issue

Due to the worldwide availability of alcohol, understanding the effects of PAE and its costs is a significant international and social issue (Clarren, Salmon, & Jonsson, 2011). Prevalence of FAS has been studied in numerous countries throughout the world, with those countries whose citizens are at higher risk of binge drinking and lower socioeconomic status yielding the highest prevalence rates (Abel, 1995). Although prevalence estimates of FAS and FASD have been more conservative in the past, it is thought that they may be as high as 2-5%

amongst younger school children in the United States and some Western European countries (May et al., 2009). Support services are often needed for those diagnosed with the disorder, but many individuals in need of support do not qualify for such services (Carmichael Olson et al., 2009). For example, based on their sample of 415 patients with FASD, Streissguth and colleagues (2004) found that only 13% of them would qualify for disability services based on the typical requirement of an IQ less than 70, despite marked deficits in adaptive behaviour. Paradoxically, individuals with the disorder that appear higher functioning may actually fare more poorly in the long run due to difficulty accessing services. Having a diagnostic category dedicated to disorders caused by PAE in a nosological giant such as the DSM would considerably enhance diagnostic communication and enable access to much needed services for those diagnosed, their families, and their communities.

“A Window of Opportunity”: The Campaign to Include FASD in the DSM

Although the deleterious effects of PAE have been recognized and well-researched since the 1970s, limited attention was paid to the inclusion of criteria for prenatal alcohol disorders in the DSM for the following two decades. FAS, by name, appeared as an etiologic factor to disability in DSM-III, but was removed in DSM-III-R (APA, 1987). FAS was mentioned again briefly in DSM-IV and DSM-IV-TR as a general medical condition frequently associated with Learning Disorder, and as a medical consequence of heavy drinking during pregnancy (APA, 1994; APA, 2000). Mention of FAS in the DSMs do not include diagnostic criteria or codes but were indicated as contextual information. Discussion of FAS and related disorders with regard to DSM often focussed on how individuals with the disorder might fit within the multiaxial classification system of DSM-III and DSM-IV (Streissguth & O'Malley, 2000). In a newsletter for the FAS Family Resource Institute, several prominent FASD diagnosticians indicated in interviews that they utilized the DSM-IV-TR for their clients who presented with a concurrent

mental disorder but typically considered FASD general medical conditions (DeVries, 2006).

Momentum in promoting the inclusion of FASDs in the DSM-5 emerged through the presence of existing organizations. In 1996 the Interagency Coordinating Committee on Fetal Alcohol Syndrome (ICCFAS) was formed, chaired by the National Institute on Alcohol Abuse and Alcoholism (NIAAA). The intent of the ICCFAS (later renamed ICCFASD) was to improve communication and collaboration through the coordination of federal-level activities in the United States to address FASD-related issues, including health, education, developmental disability, research, justice, and social services. Numerous federal-level agencies participate on the ICCFASD, including the Department of Health and Human Services, the Centers for Disease Control and Prevention (CDC), Department of Education, and Department of Justice.

The ICCFAS hosted an Early Childhood Neurobehavioural Assessment workshop in March, 2000 to further develop understanding of ARND and to address related diagnostic issues. A special session on Revising the Diagnostic and Statistical Manual of Mental Disorders: Relevance to ARND was facilitated by Dr. Michael First, Editor of Text and Criteria for DSM-IV and co-chair and editor for work groups of DSM-IV-TR. Dr. First suggested that there was a “window of opportunity” for new diagnostic criteria for FAS to be included in DSM-5, and suggested several options for how this might be incorporated. First, the APA could include a “behavioural disorders due to prenatal exposure” category in DSM-5. Second, prenatal alcohol exposure could be listed in Axis III as a physical condition, with other diagnoses, such as Learning Disorder, listed as primary diagnoses. Third, FAS criteria could be included within the category of alcohol-induced disorders. Although disorders within this category were typically used to describe behaviour of adults under the influence of alcohol, it was suggested that there may be flexibility to consider alcohol within the context of a toxic exposure (First, 2000, March).

In that same year (2000), the CDC hosted the first National Task Force on Fetal Alcohol Syndrome and Fetal Alcohol Effect. At this

meeting there was some debate as to whether it would be advantageous to have a DSM code for FAS since the condition was listed in the ICD-9, and it was argued that the worst diagnosis for insurance reimbursement was a mental health code. Yet, it was further argued that addressing the potential inclusion of FAS in the DSM would make the condition diagnosable, thus reimbursable for adults, and that further consideration of the DSM for the Task Force was needed (CDC, 2000, December).

In 2004, a newly formed work group of the National Task Force, the Post-Exposure Work Group, advocated for recognition of FASD within government and organizational bodies. The work group asserted that FAS needed to be a part of DSM-5 and drafted a letter from the Task Force to the APA inquiring about the inclusion of FAS (CDC, 2004, June). In addition to advocating the inclusion of FAS in the DSM, the work group focussed attention upon ways of promoting eligibility for services for those with FASD. These included efforts to promote the inclusion of FAS in the Individuals with Disabilities Education Act (IDEA) (CDC, 2005, June), and to advocate for the provision of life-long supports for adults with FAS, particularly in addressing psychological, behavioural, and housing needs (CDC, 2004, June). The efforts of the work group continued until the eventual disbanding of the National Task Force in 2007.

In May, 2005, Dr. Susan Rich, a psychiatrist, co-authored an APA action paper with Dr. Roger Peele outlining issues related to FASD and recommended that the disorder be considered for future editions of the DSM (Rich, 2005). Action papers are documents that may be submitted by APA members to the APA Assembly for review and possible implementation within the association (Mamah, 2006). The authors proposed a diagnostic classification system similar to the Pervasive Developmental Disorders category of DSM-IV-TR, in that the term FASD would serve as the categorical heading but not as a diagnostic term. Rather, diagnostic terms would include existing terms FAS, ARND, and a newly proposed category called Fetal Alcohol-Induced Disorders. The action paper was endorsed by the National Organization on Fetal Alcohol Syndrome (NOFAS), NIAAA, and CDC (CDC, 2005, June). The APA Assembly 2005 approved the action paper unanimously,

signalling an official starting point for the APA in exploring the possibility of including disorders caused by prenatal alcohol exposure in the DSM. Yet, momentum within the APA created by this action paper and its unanimous approval by the Assembly appeared to wane; by 2008, Rich (2008) indicated that the action paper lay “dormant” within the DSM-5 Task Force.

In 2008, Dr. Howard Moss, a psychiatrist associated with NIAAA, submitted a white paper advocating for the inclusion of Alcohol-Related Neurobehavioural Syndrome in DSM-5. Dr. Moss argued that while FASD had not been a part of psychiatric taxonomy, psychiatrists routinely worked with these patients. The inability of psychiatrists to bill for services and diagnose patients with FASD was deemed to be a barrier to service (Brunk, 2009, August).

Michael First emphasized in the March, 2000 Early Childhood Neurobehavioural Assessment workshop that the APA tended to ignore reimbursement matters when revising the DSM. Yet, it appears that much of the impetus for including FASD in the DSM has been based predominantly on reimbursement issues and enabling of services. Many of the efforts of the Post-Exposure Work Group of the National Task Force were aimed at enabling services for individuals with FAS, and Moss emphasized that a DSM code was needed to add flexibility in the services psychiatrists could provide. In addition, advocacy groups, such as NOFAS and the Minnesota Organization on Fetal Alcohol Syndrome (MOFAS), issued position statements emphasizing that the absence of FASD in the DSM has resulted in treatment delivery problems and resource limitations for those with the disorder (MOFAS, n.d.; NOFAS, n.d.). It is not surprising that service provision and reimbursement issues were central to advocacy efforts to include FASD in the DSM, given that services and reimbursements are elusive for those impacted by prenatal alcohol exposure (Carmichael Olson et al., 2009), and that a DSM code is often considered a gateway to treatment and support (Goldman & Grob, 2006; Rappo, 2002).

In 2008, the ICCFASD developed a Diagnostic Issues Work Group with the intent to revise and enhance current FASD diagnostic guidelines for research and medical practice. Much of the focus of the work group centred on

ARND diagnosis, and whether it can be differentiated from other disorders given the absence of the characteristic facial features of FAS (ICCFASD, 2011, Oct-Nov). Early in 2010, the DSM revision group issued a call for comments on an early draft of DSM-5 that included FAS as a “Condition Proposed by Outside Sources.” Although the Diagnostic Issues Work Group was not developed for the purpose of incorporating FASD into the DSM, the work group did develop a DSM Revision Subcommittee to address the issue. The subcommittee responded to the call for comments with a white paper that included preliminary diagnostic criteria in the style of DSM, based in part on data presented at a scientific workshop hosted by ICCFASD in May, 2009. The subcommittee subsequently submitted revised criteria to the DSM-5 Neurodevelopmental Disorders Work Group, and to the Substance-Related Disorders Work Group. In May, 2012 DSM-5 draft criteria were proposed for ND-PAE as a condition requiring further study under the category Substance Use and Addictive Disorders (APA, n.d.).

From FASD Guidelines to ND-PAE Criteria

In existing FASD diagnostic guidelines (Astley & Clarren, 2000; Chudley et al., 2005) four key domains are assessed; physical growth restriction, facial dysmorphism, CNS dysfunction, and confirmation of alcohol exposure. In the proposed ND-PAE criteria made publicly available from APA (n.d.), only two of those domains, CNS dysfunction and confirmation of alcohol exposure, were retained. This likely relates back to the underlying ideology of the DSM, which tends to avoid incorporating physical signs in criteria. Rather, DSM criteria focus on behaviourally observable symptoms.

The criteria identified in the proposed revisions addressing maternal alcohol confirmation states:

- A. More than minimal exposure to alcohol at any time during gestation, including prior to pregnancy recognition. Confirmation of gestational exposure to alcohol may be obtained from any of the following sources: maternal self-report of alcohol use in pregnancy, collateral reports, or medical or other records. (APA, n.d.)

Confirmation of gestational alcohol exposure is the keystone of the FASD diagnostic process. Without confirmation of PAE, the presence of FASD cannot be confirmed. According to some FASD guidelines, however, a FAS diagnosis can be given when growth deficiency and facial features are fully expressed in the presence of CNS dysfunction (Chudley et al., 2005).

With regard to CNS dysfunction specifically, behaviourally observable symptoms and core features are listed below:

B. Neurocognitive impairment, as evidenced by one (or more) of the following:

1. global intellectual impairment (i.e., IQ of 70 or below, or a standard score of 70 or below on a comprehensive developmental assessment)
2. impairment in executive functioning (e.g., poor planning and organization; difficulty changing strategies or inflexibility; difficulty with behavioural inhibition)
3. impairment in learning (e.g., lower academic achievement than expected for intellectual level; requires special education services; specific learning disability)
4. impairment in memory (e.g., problems remembering information learned recently; repeatedly making the same mistakes; difficulty remembering lengthy verbal instructions)
5. impairment in visual spatial reasoning (e.g., disorganized or poorly planned drawings or constructions; problems differentiating left from right; problems aligning numbers in columns)

C. Impairment in self-regulation in one (or more) of the following:

1. impairment in mood or behavioural regulation (e.g., mood lability; negative affect or irritability; frequent behavioural outbursts)
2. attention deficit (e.g., difficulty encoding new information; difficulty shifting attention; difficulty sustaining mental effort)

3. impairment in impulse control (e.g., difficulty waiting turn; difficulty complying with rules; confabulating; taking possessions of others)

D. Deficits in adaptive functioning as manifested in two (or more) of the following, including at least one of (1) or (2):

1. communication deficit (e.g., delayed acquisition of language; difficulty understanding spoken language; difficulty using language to express self so that the listener understands)
2. social impairment (e.g., overly friendly with strangers; difficulty reading social cues; difficulty understanding social consequences; acting “too young”)
3. impairment in daily living (e.g., delayed toileting, feeding, or bathing; problems following rules of personal safety; difficulty managing daily schedule)
4. motor impairment (e.g., poor fine motor development; delayed attainment of gross motor milestones or ongoing deficits in gross motor function; problems in coordination and balance) (APA, n.d.)

Although the specific symptoms (i.e., impairment in executive functioning or memory) are addressed in the current FASD diagnostic guidelines, the ND-PAE criteria imply that neurocognitive impairment, impairment in self-regulation, and deficits in adaptive functioning are core features shared by all individuals with ND-PAE. This differs from current guidelines where significant deficits in CNS domains imply CNS dysfunction, but not a core set of features. For example, using current guidelines, one client may receive a diagnosis based on significant deficits in learning, memory, and executive functioning, whereas another client can receive an identical diagnosis based on significant deficits in attention, communication, and daily living skills. In this way, ND-PAE is a step closer to defining a neurobehavioural profile shared by all individuals with FASD.

Like most disorders in the DSM, ND-PAE includes contextual criteria that relate to onset,

functional impact, and distinctions from other conditions:

- E. The onset of the disturbance (symptoms in Criteria B, C, and D) is before 18 years of age.
- F. The disturbance causes clinically significant distress or impairment in social, occupational, or other important areas of functioning.
- G. The disturbance is not better explained by the direct physiological effects associated with postnatal use of a substance (e.g., medication, alcohol or other drugs), another medical condition (e.g., traumatic brain injury, delirium, dementia), other known teratogens (e.g., fetal hydantoin syndrome), a genetic condition (e.g., Williams syndrome, Down syndrome, Cornelia de Lange syndrome), or environmental neglect. (APA, n.d.)

Another challenge in the diagnosis of FASDs involves differentiating effects of PAE from deficits caused by other factors, which can result in false positive diagnoses. Children exposed to alcohol in utero are often exposed to other risk factors that can lead to cognitive and behavioural problems (McLennan, 2011). For example, the majority of children with a FASD are raised in unstable home environments or in adoptive/foster care, which environments which are often associated with childhood learning disabilities (Evans, 2001; Sullivan & Knutson, 2000).

Conclusion

The formal process of developing the criteria for ND-PAE began relatively recently. Formed in 2008, the Diagnostic Issues Work Group of the ICCFASD developed a DSM Revision Subcommittee to work on the criteria. Other efforts to advocate the inclusion of FASD in the DSM were documented in meetings of the National Task Force on FAS and FAE, facilitated by the CDC, and with advocacy groups such as NOFAS and MOFAS. Psychiatrists Susan Rich and Howard Moss also initiated efforts to include FASD in the DSM. Central to the efforts to incorporate FASD in the DSM was the recognition that FASD diagnoses are currently insufficient to obtain the services and supports clients and their significant others require (Carmichael Olson et al., 2009). A DSM

diagnosis is understood to be more effective in opening doors to services (Goldman & Grob, 2006; Rappo, 2002).

FASD is a lifelong disorder that causes clinically significant impairment and that results in numerous adverse life outcomes for those affected. Efforts to include FASD in the DSM have recognized the significant impact of PAE, the need for services and supports, and the potential for a DSM diagnosis to enable services. In addition, these efforts have promoted the need for standardized diagnosis to assist in building diagnostic capacity, establishing prevalence, informing prevention efforts, and facilitating research. Continued efforts to make ND-PAE a diagnosable condition in DSM-5 will further enhance understanding of this disorder and support those that are affected.

Key Messages From This Article

People with disabilities: You deserve to receive the supports you need to stay safe. We need to work with researchers and the community to find ways to help you access those services.

Professionals: Individuals with FASD often need support to achieve and maintain positive life outcomes. We need to find ways to advocate for these individuals to help them access the support they need.

Policymakers: Individuals with FASD often do not receive the support they need, even with a diagnosis. We need to find ways to help these individuals access these supports in order to promote positive life outcomes.

Author's Note

DSM-5 was published in May, 2013, which included criteria for ND-PAE in section III as a condition requiring further study. There were editorial changes in wording but no substantive changes in the criteria from those that were proposed.

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