

INCREASING DIAGNOSTIC CAPACITY

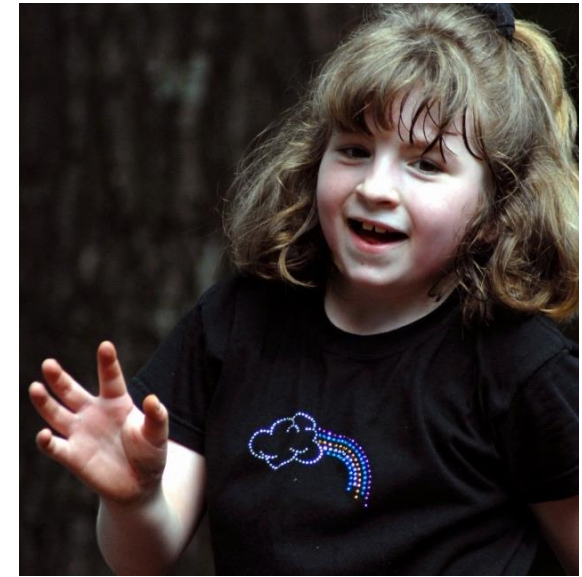
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FASD Impact Week, September 30, 2026



DIAGNOSTIC DILEMMAS

- **Accessing services and care are limited by availability of health practitioners available to make an FASD diagnosis**
- **Historically, an FASD diagnosis was only done in the context of complex multi-disciplinary teams**
 - **Limited expertise (national shortage of geneticists and developmental pediatricians)**
 - **Limited expertise within psychologists and other healthcare providers to contribute to the diagnosis**
 - **These have limited the growth of multidisciplinary teams so that only about a handful of clinics have been actively doing this across the country**
- **Paying for these assessments has also been complicated**
 - **Time and effort associated with running the multidisciplinary teams does mix well with current healthcare models of service delivery (RVU demands)**
 - **Q86.0 is seen by some payors as a “medical diagnosis” and claims have been rejected when used by mental health professionals**

Ethically, greater diagnostic availability can be viewed as an advancement in equitable access to healthcare, reducing disparities in the recognition and treatment of affected individuals

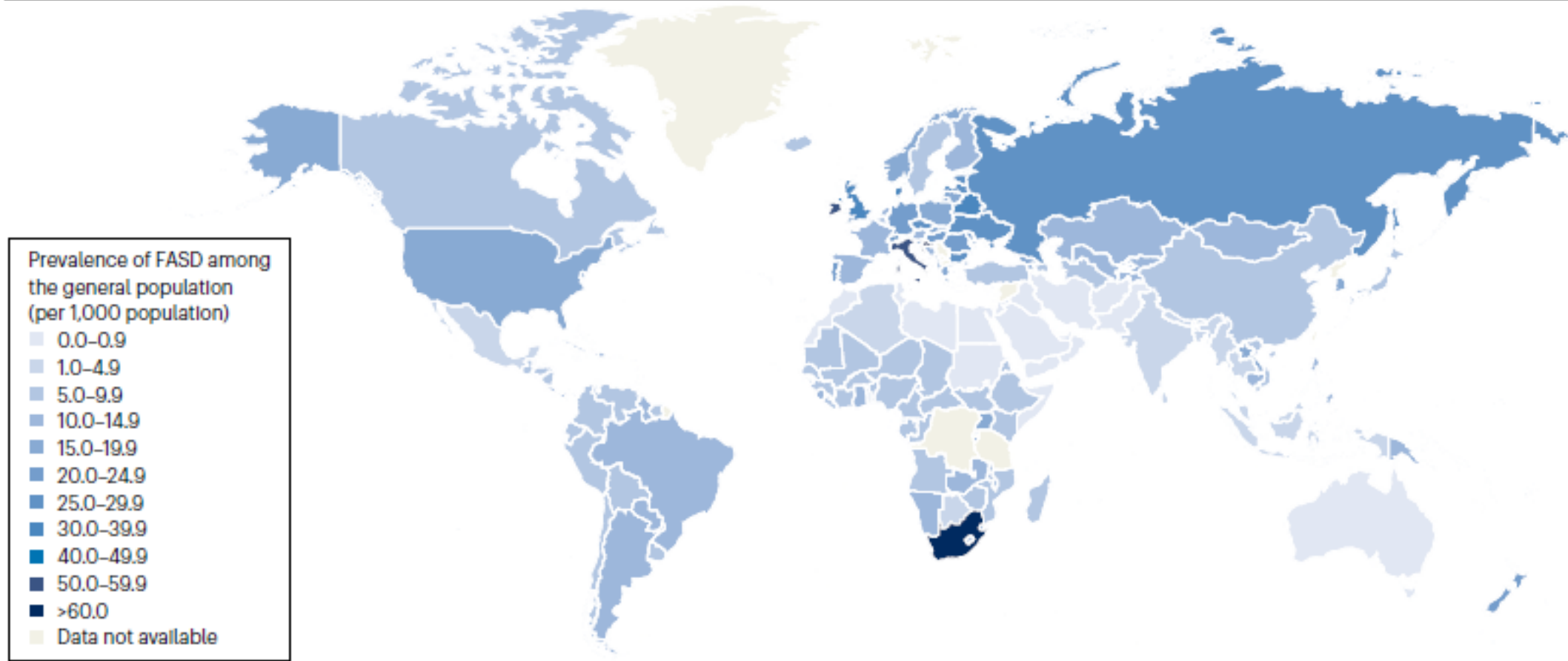


Fig. 2 | Prevalence of FASD among the general population (per 1,000 population). In line with the prevalence of alcohol use during pregnancy, the highest pooled prevalence (per 1,000) of fetal alcohol spectrum disorders (FASD) was in the WHO European Region (19.8 per 1,000 population, 95% CI 14.1–28.0), followed by the Region of the Americas (8.8 per 1,000 population, 95% CI 6.4–13.2), the African Region (7.8 per 1,000 population, 95% CI 5.4–10.7), the Western

Pacific Region (6.7 per 1,000 population, 95% CI 4.5–11.7) and the South-East Asia Region (1.4 per 1,000 population, 95% CI 0.6–5.3), and the lowest prevalence was estimated in the Eastern Mediterranean Region (0.1 per 1,000 population, 95% CI 0.1–0.5). The pooled global prevalence of FASD was estimated to be 7.7 (95% CI 4.9–11.7) per 1,000 in the general population. Data from refs. ^{25,48}.

LACK OF CONSENSUS IN MAKING A FASD DIAGNOSIS

- **Methods and procedures for making a diagnosis are not uniform internationally or even within the United States**
- **Recent research has identified that these different methods result in substantial disagreement about who should make a diagnosis.**
- **The International Consensus Committee for the Research Classification of FASD (ICC) has been working towards developing standards for improving our diagnostic methodology-but the progress has been slow**
 - **Normative comparison charts for defining palpebral fissure length have varied and have been based on limited data sets**

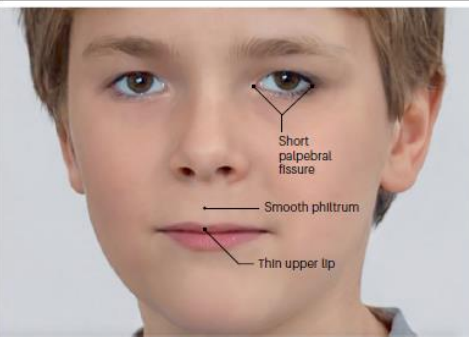


Fig. 6 | Sentinel facial features of fetal alcohol syndrome. Fetal alcohol syndrome has three characteristic (sentinel) facial features: thin upper lip (with absent cupid bow), smooth philtrum (with absence of the normal midline vertical groove and lateral ridges extending from the base of the nose to the vermillion border of the upper lip) and short palpebral fissures (the space between the medial and lateral canthus of the open eye). Image created by Ria Chockalingam using an image from Generated Photos and modified with Adobe Photoshop.

Table 1 | Comparison of key diagnostic criteria for the three most commonly used diagnostic systems

Diagnostic categories	Diagnostic criteria						
	Confirmed PAE ^a	Sentinel facial features	Growth deficiency	Structural brain abnormality (OFC) ^b	Structural brain abnormality (brain imaging) ^b	Functional brain abnormality ^c	Other birth defects
Fetal alcohol syndrome							
CoFASP ¹⁰ — fetal alcohol syndrome	–	2 of 3 ^d	≤Tenth centile	≤Tenth centile ^b	Or ^{b,a}	≤1.5 SD ^f	–
4-Digit Diagnostic Code ¹⁶⁶ — fetal alcohol syndrome	–	3 of 3	≤Tenth centile	≤Third centile ^b	Or ^{b,g}	Or ≤2 SD ^{b,h}	–
Canadian ¹⁶⁷ — FASD with sentinel facial features	–	3 of 3	–	≤Third centile ⁱ	Or ^b	≤2 SD ⁱ	–
Partial fetal alcohol syndrome							
CoFASP ¹⁰	Required	2 of 3	–	–	–	≤1.5 SD	–
CoFASP ¹⁰	–	2 of 3	≤Tenth centile ^b	Or ≤tenth centile ^b	Or ^b	≤1.5 SD ^b	–
4-Digit Diagnostic Code ¹⁶⁶	Required	3 of 3 ^j	–	≤Third centile ^b	Or ^{b,g}	Or ≤2 SD ^b	–
FASD without sentinel facial features							
CoFASP ¹⁰ — alcohol-related neurodevelopmental disorder	Required	–	–	–	–	≤1.5 SD	–
CoFASP ¹⁰ — alcohol-related birth defects	Required	–	–	–	–	–	Required
4-Digit Diagnostic Code ¹⁶⁶ — static encephalopathy, alcohol exposed	Required	–	–	–	Required ^k	Or ≤2 SD ^b	–
4-Digit Diagnostic Code ¹⁶⁶ — neurobehavioural disorder, alcohol exposed	Required	–	–	–	–	Moderate	–
Canadian ¹⁶⁷ — FASD without sentinel facial features	Required	–	–	–	–	≤2 SD	–

ICC-CONSENSUS PFL CHARTS-IN PROGRESS- BASED ON >9,000 CASE RECORDS



ICCFASD Diagnostic Issues Work Group DSM-5 Revision Subcommittee

- Julie Kable (Emory University)
- Mary O'Connor (U. California at Los Angeles)
- Heather Carmichael Olson (U. of Washington)
- Sarah Mattson (San Diego State University)
- Blair Paley (U. California at Los Angeles)
- Edward Riley (San Diego State University)
- Sally Anderson (NIAAA, NIH)
- Kenneth R. Warren (NIAAA, NIH)

Special thanks to Bridget Grant

WHY WAS A DSM-5 DIAGNOSIS NEEDED FOR INDIVIDUALS WITH AN FASD?

- ⦿ There was no specific mental health code that adequately documents the cognitive and mental health impact of PAE
 - ❖ Intellectual Deficiency
 - ❖ Cognitive impairment, NOS
 - ❖ Unspecified emotional and behavioral disturbance
 - ❖ ADD-ADHD
- ⦿ The existing diagnostic codes (760.710 do not adequately capture their mental health needs)
- ⦿ Children with FASD may not respond to treatment regimens developed using the existing codes similarly, which may lead to inappropriate treatments
- ⦿ When seeking mental health care (assessments or interventions), providers and families often struggle with obtaining appropriate reimbursement for habilitative care

AMERICAN PSYCHIATRIC ASSOCIATION



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For Immediate Release:

December 1, 2012

Release No. 12-43

Approves *DSM-5*

May 2013 publication

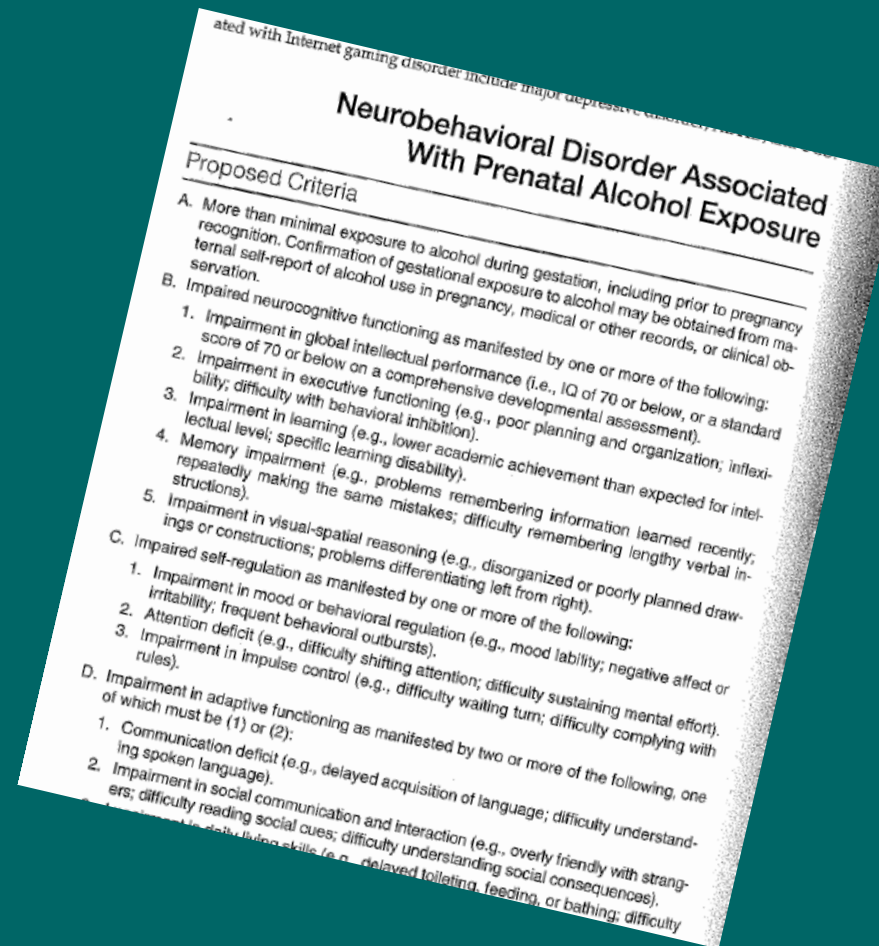
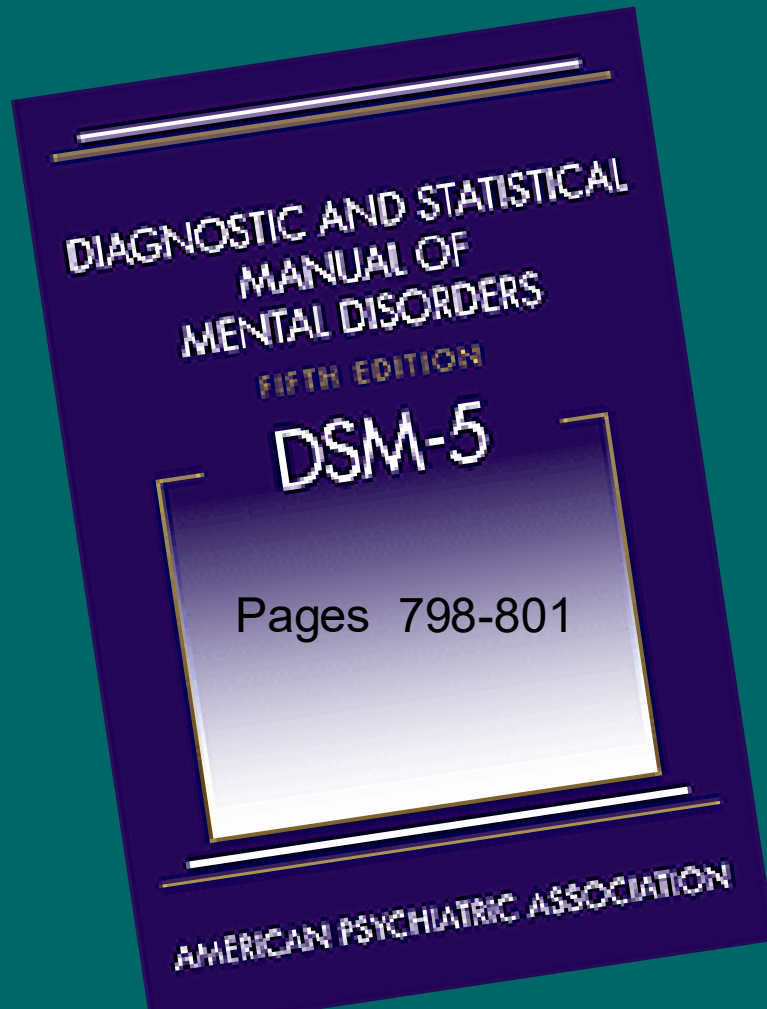
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 edition of the *Diagnostic and*
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ence for *DSM-5*,” said Dilip
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Source: Elizabeth Ghaffari and Barbara McNurlin, "The Technology C
unpublished paper.

DSM-5 Released May 2013



Other Neurodevelopmental Disorders

Other Specified Neurodevelopmental Disorder

315.8 (F88)

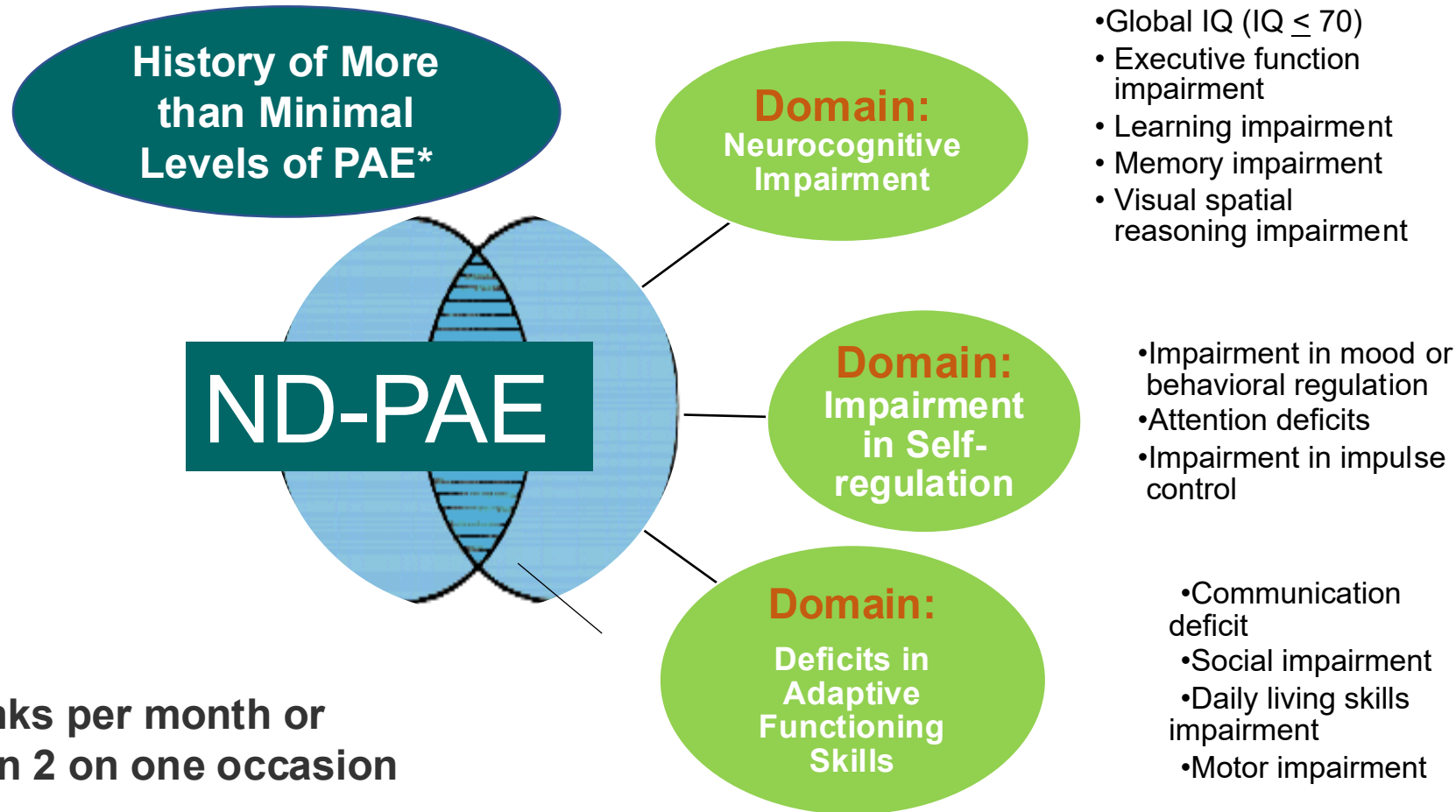
This category applies to presentations in which symptoms characteristic of a neurodevelopmental disorder that cause impairment in social, occupational, or other important areas of functioning predominate but do not meet the full criteria for any of the disorders in the neurodevelopmental disorders diagnostic class. The other specified neurodevelopmental disorder category is used in situations in which the clinician chooses to communicate the specific reason that the presentation does not meet the criteria for any specific neurodevelopmental disorder. This is done by recording "other specified neurodevelopmental disorder" followed by the specific reason (e.g., "neurodevelopmental disorder associated with prenatal alcohol exposure").

An example of a presentation that can be specified using the "other specified" designation is the following:

Neurodevelopmental disorder associated with prenatal alcohol exposure: Neurodevelopmental disorder associated with prenatal alcohol exposure is characterized by a range of developmental disabilities following exposure to alcohol in utero.

NEUROBEHAVIORAL DISORDER ASSOCIATED WITH PRENATAL ALCOHOL EXPOSURE

Symptoms



* >13 drinks per month or more than 2 on one occasion

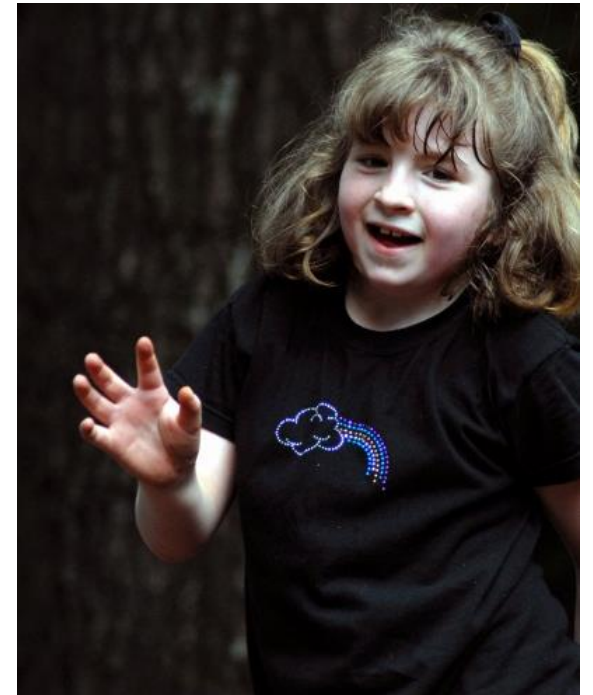
History of More than Minimal Levels of PAE

- Amount not specified in actual criteria, but guidelines are in the supporting text
>13 drinks per month or more than 2 on one occasion
- If child meets criteria for full FAS then ND-PAE can be diagnosed without documented exposure
- Documentation can be from maternal self-report, medical and other records, or clinical observation

Neurocognitive Impairment

B. Neurocognitive impairment, as evidenced by 1 (or more) of the following:

- **Global intellectual impairment**
- **Impairment in executive functioning**
- **Impairment in learning**
- **Impairment in memory**
- **Impairment in visual spatial reasoning**



Impairment in Self-regulation



C. Impairment in self-regulation in 1 (or more) of the following:

- Impairment in mood or behavioral regulation (e.g., mood lability; negative affect or irritability; frequent behavioral outbursts).
- Attention deficit (e.g., difficulty encoding new information; difficulty shifting attention; difficulty sustaining mental effort).
- Impairment in impulse control (e.g., difficulty waiting turn; difficulty complying with rules; confabulating; taking possessions of others).

Deficits in Adaptive Functioning Skills

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

- **Communication deficit (e.g., delayed acquisition of language; difficulty understanding spoken language; difficulty using language to express self so that the listener understands).**
- **Social impairment (e.g., overly friendly with strangers; difficulty reading social cues; difficulty understanding social consequences; acting too young).**
- **Impairment in daily living (delayed toileting, feeding, or bathing; problems following rules of personal safety; difficulty managing daily schedule).**
- **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).**



-
- 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), a general medical condition (e.g., traumatic brain injury, delirium, dementia), other known teratogens (e.g., Fetal Hydantoin syndrome), genetic condition (e.g., Williams syndrome, Down syndrome, Cornelia de Lange syndrome), or environmental neglect and/or abuse.

EARLY VALIDATION ATTEMPTS ON ND-PAE CRITERIA

58 children referred for multidisciplinary FASD assessment were evaluated.

Used factor analysis to validate symptoms

Concluded that ND-PAE has **some empirical support**, but the proposed DSM-5 criteria require further refinement and validation and noted that adaptive behavior may be a particularly important component of the diagnosis.

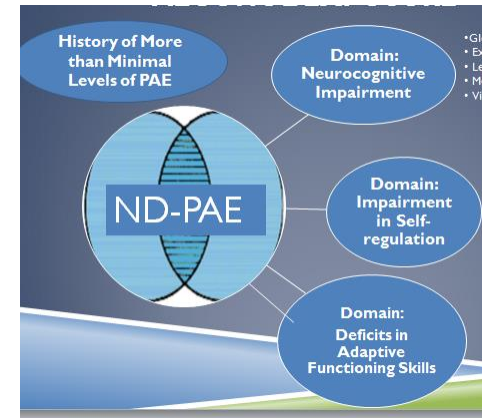
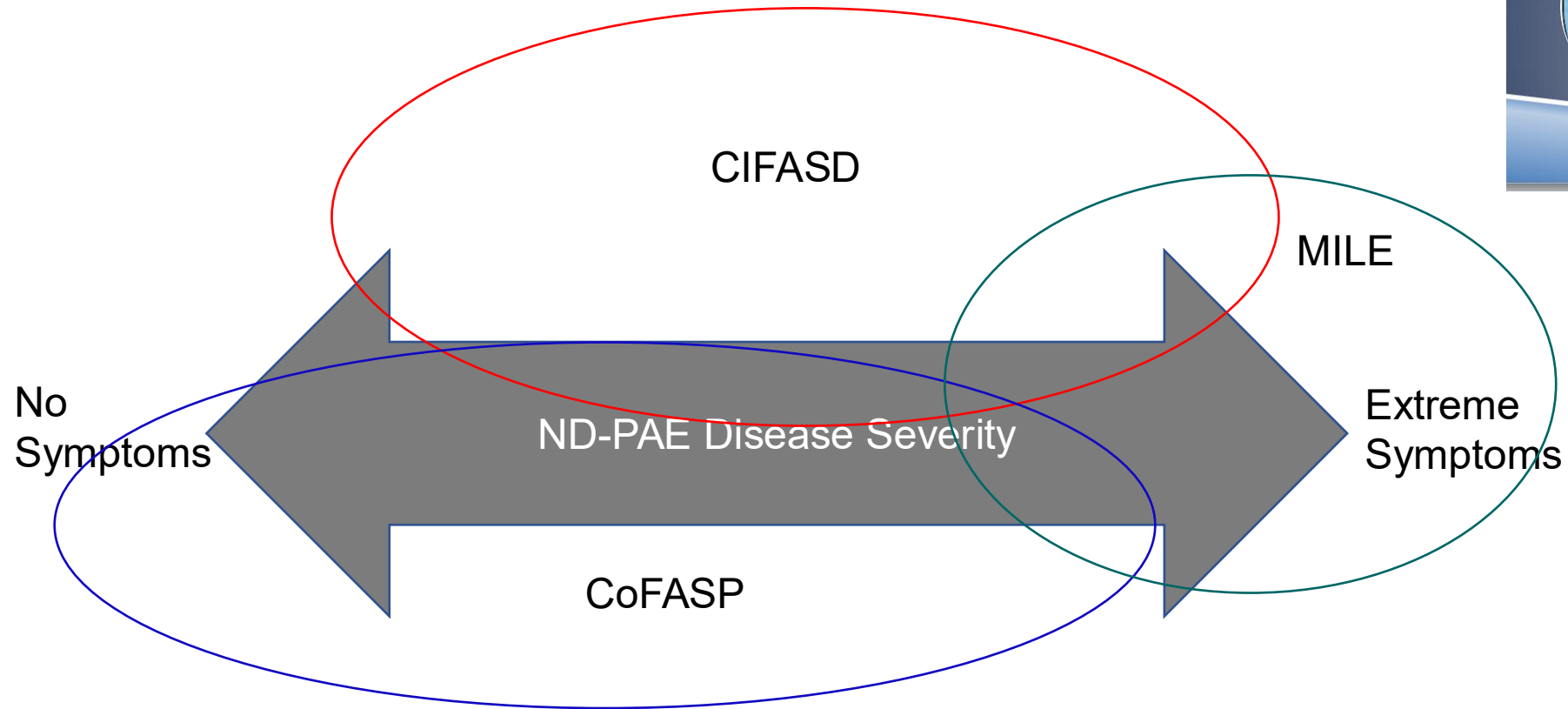
Sanders, J.L., Netelenbos, N., & Dei, S.O. (2020). Construct and factorial validity of Neurobehavioral Disorder associated with Prenatal Alcohol Exposure (ND-PAE). *BMC Psychology*, 8, 53.

PROGRESS IN VALIDATING THE DISORDER

Samples Studied

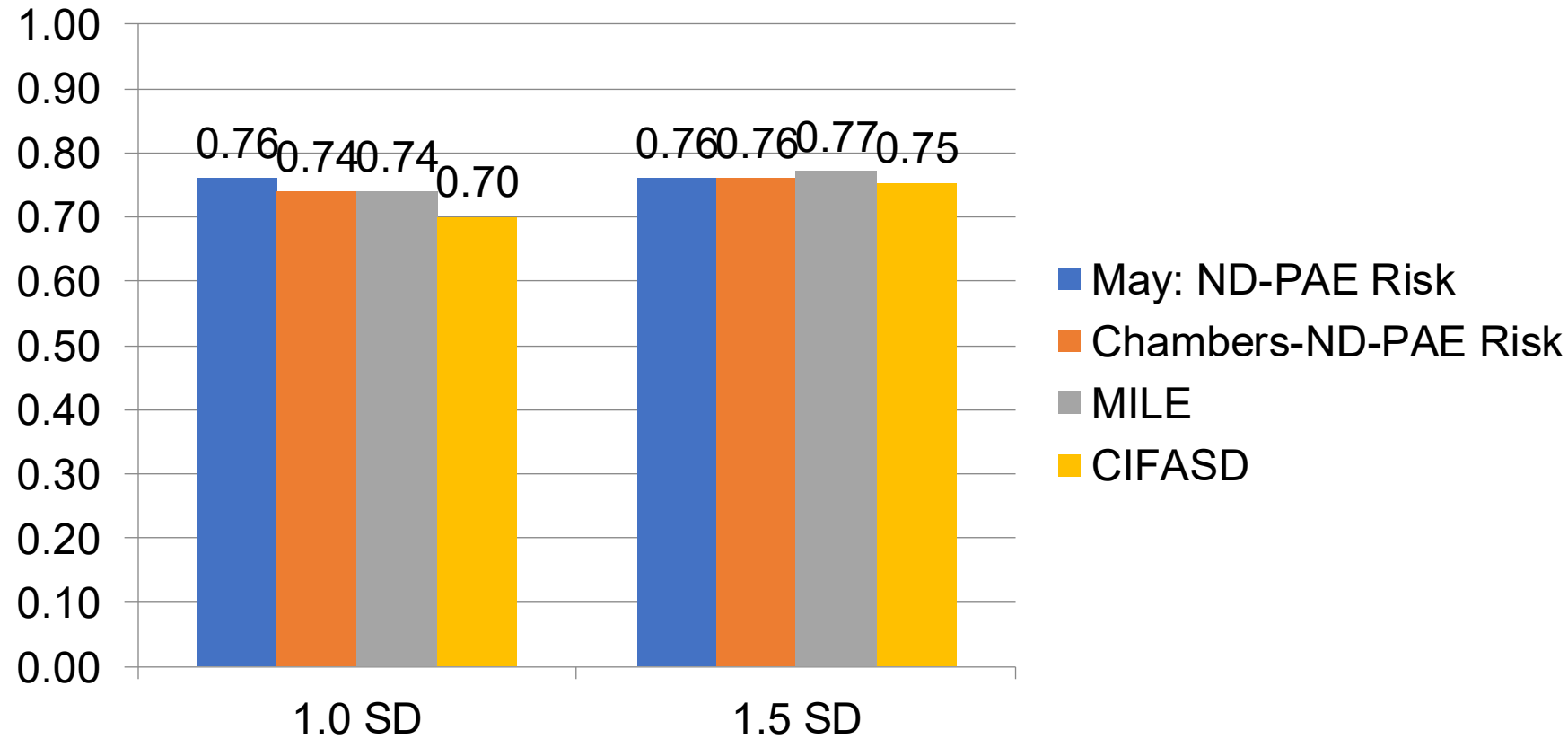
- **MILE Cohort**-A sample of children between 3-10 years of age who had been identified as having FAS or pFAS and had enrolled in math intervention study Funded by a CDC Cooperative agreement # U84-CCU320162-02 and this analysis was carried out with the support of the National Institute of Alcohol Abuse and Alcoholism subcontract #10313752 to grant # U01 AA019879-01)
- **CIFASD**-A multi-site sample of children who had a history of PAE recruited from FAS diagnostic clinics or the community to evaluate alcohol exposed children and youth ages 5 to 17 years in comparison to appropriate contrast groups to identify alcohol-specific teratogenic effects on cognition Funded by the support of support of the National Institute of Alcohol Abuse and Alcoholism subcontract #10313752 to grant # U01 AA019879-01 and U01 AA014834 (Mattson) /Collaborative Initiative on Fetal Alcohol Spectrum Disorders (CIFASD)
- **CoFASP** –A multi-site sample of children who enrolled into a study designed to **to establish the prevalence of FASD in school-aged children** Funded by the support of support of the National Institute of Alcohol Abuse and Alcoholism subcontract #10313752 and U01 AA019879 (Chambers) and U01X (May)/ The Collaboration on FASD Prevalence (CoFASP)

Latent Trait Of Disease Severity



Individuals with a history of PAE are anticipated to have variability in ND-PAE symptomatology allowing for a comparison of cohesiveness and necessity of each item to identify the latent trait of disorder severity.

Internal Consistency of 12 ND-PAE Symptoms Across Samples



MILE n=56; San Diego n=854; May ND-PAE Risk n=48; CIFASD n =486

OTHER EVALUATIONS

- 1. Altering the threshold of the symptoms (1.5 vs 2.0 standard deviation units)
- 2. Systematic evaluations of criteria for adaptive function

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Table 5 AUC values for each domain for each SD level

Group	Model	1.0 SD Threshold cut-off level			1.5 SD Threshold cut-off level			2.0 SD Threshold cut-off level		
		NI	SR	AF	NI	SR	AF	NI	SR	AF
Overall sample	AF 1	0.622	0.722	0.923	0.729	0.763	0.929	0.794	0.720	0.955
	AF 2/4	0.595	0.673	0.979	0.702	0.733	0.970	0.771	0.703	0.981
CON group	AF 1	0.634	0.737	0.927	0.745	0.806	0.952	0.821	0.777	0.976
	AF 2/4	0.612	0.698	0.981	0.728	0.784	0.985	0.809	0.767	0.992
AE group	AF 1	0.562	0.646	0.904	0.688	0.658	0.870	0.742	0.611	0.913
	AF 2/4	0.537	0.587	0.973	0.648	0.624	0.939	0.705	0.594	0.963

Significant values are given in bold font

Christina R. Veziris¹ · Matthew T. Hyland¹ · Julie A. Kable² · Jeffrey R. Wozniak³ · Claire D. Coles² · Philip A. May⁴ · Wendy O. Kalberg⁵ · Elizabeth R. Sowell⁶ · Kenneth L. Jones⁷ · Edward P. Riley¹ · Sarah N. Mattson¹

General Conclusions

- **Results indicate that the ND-PAE criteria are useful in identifying affected children-adequate breadth of coverage**
- **Results are limited to children with known exposure history at this point**
- **Criteria had good internal validity, especially after modifying AF criteria-homogenous symptoms across 3 samples**
- **Diagnosis occurs more often in those identified as ND-PAE RISK relative to controls: discriminative validity**
- **Using a 1.0 SD cutoff and the revised AF criteria yielded the strongest model (AUC = .831), although specificity was higher when using the 1.5 SD cutoff-(discriminative validity)**
- **Additional modifications to the criteria (e.g., changing general cognitive cutoff to 1.5 SD?)**

FUTURE

- Nothing in clinical science is “perfect” on-going research and further refinement is always welcome
- Generally, ND-PAE seems to have increased awareness and comfort with providing the diagnosis-thereby expanding access to care
- Still difficult to count for public health purposes the number of individuals impacted



We need our own
code!