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RESEARCH ARTICLE

An investigation of the utility of the Australian Guide to the Diagnosis of Fetal Alcohol Spectrum Disorder in young children

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Abstract

Background: Early diagnosis of children with fetal alcohol spectrum disorder (FASD) assists in implementing critical early support. The challenge lies in having a diagnostic process that enables valid and reliable assessment of domains of functioning in young children, with the added complexity that many children will also have co-occurring exposure to childhood adversity that is likely to impact these domains.

Methods: The aim of this study was to test a diagnostic assessment of FASD in young children using the Australian Guide to the Diagnosis of FASD. Ninety-four children (aged 3 to 7 years) with confirmed or suspected prenatal alcohol exposure were referred to two specialist FASD clinics for assessment in Queensland, Australia.

Results: There was a significant risk profile with 68.1% (n = 64) children having had contact with child protection services, and most children living in kinship (n = 22, 27.7%) or foster (n = 36, 40.4%) care. Forty-one percent of the children were Indigenous Australians. The majority (64.9%, n = 61) of children met criteria for FASD, 30.9% were classified as "At Risk" for FASD (n = 29), and 4.3% received no FASD diagnosis (n = 4). Only 4 (4%) children were rated as severe for the brain domain. Over 60% of children (n = 58) had two or more comorbid diagnoses. Sensitivity analyses indicated that the removal of comorbid diagnoses in the Attention, Affect Regulation, or Adaptive Functioning domains resulted in a change in 7 of 47 cases (15%) to an "At Risk" designation.

Conclusions: These results highlight the complexity of presentation and the extent of impairment in the sample. The use of comorbid diagnoses to substantiate a "severe" designation in specific neurodevelopmental domains raises the question of whether there were false-positive diagnoses. The complexity of determining causal relationships between exposure to PAE and early life adversity on developmental outcomes continues to be a challenge in this young population.

KEYWORDS
fetal alcohol spectrum disorder, neurodevelopment, prenatal alcohol exposure

INTRODUCTION

The consequences for children born with birth defects and developmental disabilities encompassed by fetal alcohol spectrum disorder (FASD) are profound. Neurodevelopmental difficulties can include impairment in executive functioning, attention and information processing, language, and motor problems (Mattson et al., 2019). These difficulties contribute to a range of adverse outcomes that may be considered secondary to the neurodevelopmental challenges, including poor educational attainment, social and behavioral problems (Easey et al., 2019; Jacobson et al., 2021; Sessa et al., 2022; Tsang et al., 2016), and physical health issues (Caputo et al., 2016) across childhood, adolescence, and adulthood (Wilhoit et al., 2017), highlighting the importance of robust and rigorous assessment processes that allow for early identification and tailored intervention.

A range of diagnostic frameworks have been developed, which provide a systematic approach to the assessment and diagnosis of the complex array of symptoms that are present in individuals with PAE (Hemingway et al., 2019). The key element to all diagnostic systems is the careful and thorough assessment of PAE, in addition to the assessment of growth parameters, brain structure and neurology, and sentinel facial features (Coles et al., 2016). Typically, diagnostic systems also include assessment of neurodevelopmental impairment, although the exact configuration of impairment, domains of impairment, and criteria for a classification of severe impairment differ across the systems (Coles et al., 2016). The Australian Guide to the diagnosis of fetal alcohol spectrum disorders (Bower & Elliott, 2016) was developed in response to a growing awareness that FASD was under-recognized and often misdiagnosed in Australia (Bower et al., 2017). It builds on earlier diagnostic systems and incorporates key components of both the Canadian (Cook et al., 2016) and American guidelines (Astley, 2004), although some changes may occur as a consequence of a current review (Hayes et al., 2022). When PAE is confirmed, a diagnosis of FASD can be made if (i) an individual has severe impairment in three or more of the 10 domains of central nervous system structure or function; (ii) other causes or conditions have been excluded; and (iii) the "potential influence of other exposures or events" have been considered (p. 36). The diagnosis of FASD can be further divided into one of two subcategories: FASD with three sentinel features or FASD with less than three sentinel features (Bower & Elliott, 2016).

The 10 domains assessed are as follows: brain structure/neurology, motor skills, cognition, language, academic achievement, memory, attention, executive function, affect regulation, and adaptive behavior/social skills and social communication. Detailed guidance is provided with a distinction made between direct assessment using established and well-validated psychometric assessment tools and indirect assessment involving the completion of domain-specific parent/caregiver and teacher reports of behavior. A domain is classified as "severe" if the score obtained is either (i) two or more standard deviations below the mean or below the third percentile for the particular test/report; or (ii) there is a discrepancy between composite measures of functions with a domain that is both clinically and statistically significant (i.e., base rate of <3% and lower score is at least one standard deviation below the mean; Bower & Elliott, 2016). This follows the guidance provided in the Canadian Guide. While concerns were acknowledged about the potential conservativeness of the clinical cutoff were considered and noted in the Canadian Guide (p. 23), the threshold was justified as having alignment with "severe" deficit in other related guidelines such as the DSM-IV and DSM-5. There are also provisions for a "severe" rating to be given in the presence of a number of diagnoses. A diagnosis of attention-deficit/hyperactivity disorder (ADHD, inattentive or combined) based on DSM-5 satisfies criteria for severe impairment for the Attention domain (Bower & Elliott, 2016, p. 27). A diagnosis of autism spectrum disorder or social (pragmatic) communication disorder, as per the DSM-5, may be used to substantiate a rating of "severe" for the domain related to adaptive behavior and social communication (shortened to Adaptive Functioning for this paper; Bower & Elliott, 2016, p. 31). While a diagnosis of conduct disorder and/or severe oppositional defiant disorder cannot be used solely to substantiate a rating of "severe" in the Adaptive Functioning domain, it can be used as supporting evidence for impairment. However, the child also needs to meet other criteria to demonstrate severe impairment in other areas of adaptive function (e.g., on an indirect measure such as the Vineland Rating Scales). Both the Australian and Canadian Guide introduced an "Affect Regulation" domain, with Australian Guide requiring a formal diagnostic assessment for a range of mood and anxiety disorders. A "severe" rating cannot be based on clinical impression or derived from information obtained from rating scales alone.

The Australian Guide provides specific advice on the assessment of FASD in infants and young children. This reflects a growing recognition that early accurate diagnosis of young children provides the best opportunity to tailor interventions to match critical developmental needs. This is particularly pertinent given that the absence of early diagnosis remains a key predictor of adverse outcomes (Flannigan et al., 2019; Streissguth et al., 2004). A formal diagnosis of FASD defines service eligibility in some countries, while early intervention is available in others based on functional impairment in the absence of specific diagnoses. The Australian Guide specifies that children and infants under the age of 6 years can be given a diagnosis of FASD if there is both microcephaly and three sentinel facial features, regardless of whether PAE is confirmed. Children can also receive a diagnosis of FASD if they meet the diagnostic criteria for severe global developmental delay that is confirmed on a standardized measure of child development (e.g., Griffiths Development Scales, Stroud et al., 2016 or Bayley Scales of Infant and Toddler Development, Bayley, 2006).

While not included in the diagnostic algorithm (p. 6), the Guide also specifies four circumstances in which an "At Risk of FASD" designation may be given: (i) if neurodevelopmental assessment is incomplete or inconclusive; (ii) if, despite confirmed PAE, neurodevelopmental impairment is present in fewer than three domains; (iii) if neurodevelopmental impairment is present in three or more domains but impairment is not sufficiently severe to meet criteria; and (iv) comprehensive, age-appropriate neurodevelopmental assessment is

impossible or unavailable (e.g., in infants and very young children, p. 19). These circumstances would lead to an "At Risk" designation for children below 6 years of age. The final additional circumstance in which an "At Risk" designation is appropriate is where there is neither microcephaly nor impairment on three developmental domains, but the child has three sentinel facial features, whether PAE is confirmed or not (p. 20). This aligns with the revised Canadian guidelines (also adopted by Scotland), which state that infants and young children should be classified as "At Risk for neurodevelopmental disorder and FASD, associated with prenatal alcohol exposure" if they have confirmed PAE but the assessment of the neurodevelopmental domains is inconclusive (child too young or assessment incomplete). Further, the revised Canadian guidelines also state that children with three sentinel facial features without microcephaly require referral to clinical genetics (Cook et al., 2016, p. 194). The inclusion of the "At Risk" designation is of particular importance for younger children and should trigger the provision of services that address current and emerging needs (Cook et al., 2016).

There are significant challenges in assessing young children. The problems of measuring neurodevelopmental domains, in particular executive functions (EF), are widely acknowledged (Flannigan et al., 2019). This is due in part to the staggered development of EFs in typically developing children and the complexity of measuring specific domains independent of others (e.g., sustained attention, working memory, and planning; Best & Miller, 2010; Nelson et al., 2016). Even with the ongoing development of age-normed EF tasks (e.g., NIH Toolbox, Zelazo et al., 2013), significant challenges remain in assessing some neurodevelopmental domains in young children.

Another significant challenge lies in determining the relative contribution of PAE and early life exposure to adverse childhood experiences to delays in achieving neurotypical developmental trajectories. While some children are adopted at birth, a significant proportion of children with PAE have early life experiences that include exposure to trauma, caregiving instability, and engagement in the child protection system (Flannigan et al., 2021). The consequences of exposure to early life adversities on neurodevelopment are well documented (Cabrera et al., 2020; Cross et al., 2017; Johnson et al., 2021; Kavanaugh et al., 2017; Lang et al., 2020; Naughton et al., 2013). Thus, even for children with PAE, significant environmental adversity may contribute in part or wholly to impairments in domains that are assessed as part of an FASD diagnosis, such as cognition, attention, executive functions, affect regulation, and social communication.

The Australian Guide specifies that FASD is "not necessarily a diagnosis of exclusion" (p. 17) and prompts clinicians to consider pre-existing diagnoses as "indicators of impairment in their relative domains" (p. 17). For example, the Guide allows for a rating of severe in the Attention domain if ADHD combined or inattentive is present; a rating of severe in the Affect Regulation domain if a specific anxiety disorder is present; and a rating of severe in the adaptive behaviour/social skills and communication domain if autism spectrum disorder is diagnosed. Yet, difficulty lies in disentangling whether these diagnoses are due to PAE or are due to other causes such as early childhood adversity (McLennan & Braunberger, 2018). For example, both anxiety disorders (including separation anxiety) and ADHD are more prevalent in children whose experience of adversity has resulted in engagement in the child protection system compared with the general population (Bronsard et al., 2016). Children exposed to neglect can have significant deficits associated with language development and communication skills (Krier et al., 2018; Snow et al., 2020). In this context, the presence of these disorders may result in a misdiagnosis of FASD if PAE is not an unequivocal etiologic factor. This does leave open the possibility that, for some children, a diagnosis of FASD based on comorbid diagnoses may be a false-positive diagnosis (McLennan, 2015).

The purpose of the current study is to investigate the diagnostic outcomes and profiles of children aged 3 to 7 years with confirmed or suspected PAE using the Australian Guide to the Diagnosis of FASD (Bower & Elliott, 2016). We examined the diagnostic status and neurodevelopmental profiles for the total sample and then divided the sample into very young children (aged 3 to 5 years 11 months) and young children (aged 6 to 7 years 8 months) to determine whether there were differences in diagnostic status. Our prediction is that there will be less certainty in young children and thus a greater number who will have an "At Risk" designation. This is warranted given the emphasis in the revised Canadian Guidelines (Cook et al., 2016), which was followed by the Australian Guidelines, regarding the special considerations for assessing children who are aged under 6 years of age. Finally, sensitivity analyses were undertaken to examine whether using specific comorbid diagnoses to arrive at a rating of "severe" would impact FASD diagnostic outcomes.

MATERIALS

The Australian Guide to the diagnosis of FASD was used to structure the assessment process (Bower & Elliott, 2016) and establish whether each domain should be rated as "severe" impairment, "some" impairment, or "none." Domains were assessed using a combination of recommended measures from the Australian Guide (Table S1) and additional validated standardized measures that assess specific constructs for young children (e.g., NIH Toolkit; Zelazo et al., 2013). Academic achievement was assessed for children who had engagement in the education system for at least 6 months of a year, including preparatory year (aged 5 years plus), provided they had been in a stable care environment. The Kiddie Schedule for Affective Disorders and Schizophrenia for School Aged Children (K-SADS-PL DSM-5; Kaufman et al., 2016) was used as the structured diagnostic interview to determine whether any of the diagnoses specified under the Affect Regulation or Adaptive Function domains were present. When using the K-SADS-PL directly with children, an age limit of 6 years is recommended. However, as all K-SADS-PL interviews were conducted with caregivers for this study, the K-SADS-PL was used to guide the diagnostic assessment of the specified disorders in the

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Australian Guide for all cases, regardless of age. Children with a pre-existing diagnosis that was also counted as "severe" on the relevant domain were not reassessed for that diagnosis. FASD diagnoses were derived using the diagnostic algorithm provided in the Australian Guide (p. 6) with reference to special considerations for assessing infants and young children (p. 19; see Table 1).

Sentinel facial features were assessed using computerized analysis of digital photos (Astley, 2016; Astley & Clarren, 2001). Palpebral fissure length was compared with the Stromland norms for children under 6 years of age (Bower & Elliott, 2016). As locally available norms are currently not available, African American lip-philtrum norms were used to assess Aboriginal and Torres Strait Islander children (Tsang et al., 2017).

METHODS

Participants

Ninety-four children aged between 3 and 7 years were recruited from two specialist FASD Clinics that were part of a publicly funded child development service in Queensland, Australia. Children were included in the study if there was suspected or evidenced PAE via (i) direct report of biological mother; (ii) report from relative or carer who knew the biological mother and was able to report accurately on prenatal alcohol exposure; and/or (iii) clinical, health, or child protection records that clearly documented maternal alcohol use during pregnancy.

Procedure

Children were referred for assessment by health clinicians (e.g., general practitioners and pediatricians) and child protective services. Children with suspected or confirmed PAE (Table 1) underwent a diagnostic assessment by a multidisciplinary team comprised of a clinical interview with carers/parents, a pediatric medical review (including growth measurements), and assessment of the 10 neurodevelopmental domains of functioning (Table S1). Assessments were typically undertaken across consecutive days with long breaks at lunch and time for relaxation and comfort breaks to reduce the burden on young children. For some children, three or more assessment sessions were required and these were scheduled a week apart. Ethics approval was provided by relevant hospital and university Human Research Ethics Committees (HREC/17/QRCH/318).

Data analysis

Parametric and descriptive analyses using information about child and family characteristics, neurobehavioral outcomes, and clinical characteristics were undertaken using SPSS. Chi-square analyses were used to examine whether FASD diagnostic status, number of

TABLE 1 Demographic characteristics for children undergoing FASD diagnostic assessment ($N = 94$)

Gender ($n = 94$)	
Male	52 (55.3%)
Female	42 (44.7%)
Mean age, months (SD; $n = 94$)	71.94 (14.99)
Age category	
3 years 0 month to 5 years 11 months	40 (42.55%)
6 years 0 month to 7 years 8 months	54 (57.45%)
Ethnicity ($n = 94$)[a]	
Caucasian/White	48 (51.6%)
Aboriginal/Torres Strait Islander	38 (40.9%)
Pacific Islander/Asian	2 (2.2%)
Other	5 (5.4%)
Weight at time of assessment	
≤3rd percentile	1 (1.1%)
≤10th >3rd percentile	2 (2.1%)
Height at time of assessment	
≤3rd percentile	0 (0.0%)
≤10th >3rd percentile	7 (7.5%)
Primary carer ($n = 94$)	
Biological parent	28 (29.8%)
Kinship carer	26 (27.7%)
Foster care	38 (40.4%)
Adopted Parents/Legal Guardian	2 (2.1%)
Any contact with child protection system (yes)	64 (68.1%)
Current contact with child protection system (yes)	56 (59.6%)
Out-of-home placements at any time (yes)[b]	69 (73.4%)
Kinship only	22 (23.4%)
Foster only	36 (38.3%)
Both	9 (9.6%)
Other	2 (2.1%)
Mean (SD) Age (months) at first entry into out-of-home placement ($n = 68$)[c]	15.64 (19.92)
# of children entering out-of-home placement before age 1 year	42 (60.9%)
# of children entering out-of-home placement after age 1 year	27 (39.1%)
Prenatal alcohol exposure	
Mean (SD) AUDIT-C score during pregnancy ($n = 53$)	7.53 (2.80)
Source of reported information on alcohol use ($n = 94$)	
Birth mother only	22 (23.4%)
Other (e.g., official records, relative, biological father)	59 (62.8%)
Birth mother and other sources	13 (13.8%)
Reliability of information on alcohol exposure ($n = 94$)	
Unknown	9 (9.6%)
Low	5 (5.3%)
High	80 (85.1%)

(Continues)

TABLE 1 (Continued)

Classification of prenatal alcohol exposure risk ($n = 94$)	
Unknown exposure	3 (3.2%)
Confirmed exposure	36 (38.3%)
Confirmed high-risk exposure	55 (58.5%)

Note: Using growth calculators described in Chou et al. (2020).

[a]$n = 1$ missing and/or unknown.

[b]Includes placements facilitated by child protection agencies, informal kinship placements, and/or placements arranged through Family Court. Missing data for two participants.

[c]$n = 1$ cases where age of first entering out-of-home placement is missing and/or unknown.

TABLE 2 Diagnostic outcome for children assessed for FASD ($N = 94$)

	Total sample	Age <6.0 years ($n = 40$)	Age ≥6 years ($n = 44$)
FASD with 3 sentinel facial features	10 (10.6%)	5 (12.5%)	5 (9.3%)
FASD with <3 sentinel facial features	51 (54.3%)	23 (57.5%)	28 (51.9%)
At Risk of FASD	29 (30.9%)	11 (27.5%)	18 (33.3%)
No FASD diagnosis	4 (4.3%)	1 (2.5%)	3 (5.5%)

TABLE 3 Sentinel facial features for children assessed for FASD ($N = 93$)

	Total sample	Age <6.0 years ($n = 39$)	Age ≥6 years ($n = 54$)
Number of sentinel facial features			
0	25 (26.6%)	10 (25.0%)	15 (27.9%)
1	28 (29.8%)	10 (25.0%)	18 (33.3%)
2	27 (28.7%)	12 (30.0%)	15 (27.8%)
3	13 (13.8%)	7 (17.5%)	6 (11.1%)

Note: Number of Sentinel Facial Features refers to the number of Palpebral Fissure Lengths 2 SD or more below the mean, philtrum rank 4 or 5, and upper lip rank 4 or 5. One case did not have SFF measurements. Three cases with 3 SFF received an "At Risk" designation because they were either ≥6 years, had no microcephaly, and did not have "severe" impairment in ≥ neurodevelopmental domains.

sentinel facial features, or distribution of "severe" impairments differed by age group (<6.0 years vs. ≥6.0 years).

RESULTS

Demographic characteristics

A total of 94 children were assessed between March 2018 and August 2020. All children spoke English as their first language and had received some schooling or early childhood education at an appropriate year level for their chronological age. Table 1 provides the full sociodemographics for the study sample.

TABLE 4 Impairment in neurodevelopmental domains for children assessed for FASD (N = 93)

	Total sample	Age <6.0 years ($n = 40$)	Age ≥6 years ($n = 53$)
N domains with severe impairment			
0	6 (6.5%)	3 (7.7%)	3 (5.7%)
1	15 (16.1%)	6 (15.4%)	9 (17.0%)
2	10 (10.8%)	3 (7.7%)	7 (13.2%)
3	14 (15.1%)	8 (20.5%)	6 (11.3%)
4	20 (21.5%)	12 (30.8%)	8 (15.1%)
5	10 (10.8%)	2 (5.1%)	8 (15.1%)
6	12 (12.9%)	4 (10.3%)	8 (15.1%)
7	6 (6.5%)	2 (5.1%)	4 (7.5%)

Note: Domains assessed: brain structure/neurology, motor skills, cognition, language, academic achievement, memory, attention, executive function, affect regulation, and adaptive behavior. One case had an attempted but incomplete assessment, so is not included in the table.

Diagnostic outcome and sentinel facial features

Of the 94 children, 10 (10.6%) children met criteria for FASD with 3 sentinel facial features, 51 (54.3%) met criteria for FASD with <3 sentinel facial features, 29 (30.9%) were classified as "At Risk of FASD," and 4 (4.3%) did not meet diagnostic thresholds for FASD or "At Risk" (Table 2). There was no statistical difference in the distribution of diagnostic categories across the two age groups ($\chi^2(1, 94) = 1.12$, $p = 0.290$). Thirteen (14%) children had three sentinel facial features (Table 3). There was no statistical difference in the distribution of sentinel facial features across the two age groups ($\chi^2(3, 93) = 1.31$, $p = 0.727$).

Distribution of severe impairment across neurodevelopmental domains

Sixty-five percent of the total sample ($n = 62$) had "severe" impairment in three or more neurodevelopmental domains and nearly one third of the sample had "severe" impairment in five or more domains ($n = 28$, see Table 4). The two age groups (<6.0 vs. ≥6.0 years) significantly differed in the distribution of "severe" neurodevelopmental domains, as classified into <3, 3 to 4, or ≥5 "severe" domains ($\chi^2(2, 93) = 6.08$, $p = 0.048$). Nearly 50% of children <6.0 years received a "severe" rating for 3 to 4 domains compared to 26% of children aged ≥6.0 years. However, this pattern shifted to 20% of children <6.0 years receiving a "severe" rating for ≥5 domains compared to 38% of children aged ≥6.0 years (Table 5).

Six children did not score in the "severe" range for any of the domains despite having confirmed PAE and between 0 and 2 sentinel

facial features. Three of these children were not given an FASD diagnosis as their level of impairment was not classified as either "severe" or "some" on neurodevelopmental domains. The other three were given an "At Risk" designation as their level of functioning in ≥3 domains was above the cutoff for a "severe" rating yet was rated as not typically developing (i.e., fell within the 16th to 3rd percentile, see Table 6).

Tables 7 and 8 display the patterns of impairment across the two age groups, number of sentinel facial features, and final diagnosis. These tables also indicate where a diagnosis was used to support a "severe" rating, as per the Australian Guide. Specifically, 48 children (51.6%) received a diagnosis of ADHD (Inattentive or Combined), resulting in a rating of "severe" impairment for the Attention domain. Twenty-four children (25.8%) received one or more mood or anxiety disorder diagnoses listed in the Australian Guide under the Affect Regulation domain, thereby resulting in a "severe" rating. Five children (5.4%) either had a pre-existing diagnosis of autism spectrum disorder or received this diagnosis as a result of the FASD assessment, which led to these children receiving a rating of "severe" impairment for the Adaptive Behavior, Social Skills, or Social Communication domain. Of the 62 children receiving ≥1 of the Guide's specified diagnoses to establish a "severe" rating in either the Attention, Affect Regulation or Adaptive Behavior domains, 8 (12.9%) received a diagnosis of FASD with 3 sentinel facial features, 39 (62.9%) received a diagnosis of FASD with <3 sentinel facial features, 14 (22.6%) received a designation of "At Risk" of FASD, and 1 received no diagnosis. [Correction added on March 03, 2023, after first online publication: In the preceding sentence, 'diagnosis' has been changed to 'designation'.]

Comorbid neurodevelopmental and mental health diagnoses

Comorbidity with FASD was high with only 10 (10.8%) children having a sole diagnosis of FASD. Almost one quarter (n = 21, 22.6%) of children had at least one other diagnosis, while a notable proportion had either 2 (n = 18, 19.4%) or >2 diagnoses (n = 40, 43.0%) in addition to FASD (Table 9). Just over half of the children (n = 48, 51.6%) met diagnostic criteria for ADHD and close to a quarter met criteria for a behavioral disorder (i.e., oppositional defiant disorder and conduct disorder; n = 21 (22.6%)) or an anxiety disorder (i.e., separation anxiety disorder, generalized anxiety disorder, selective mutism, panic disorder, and/or agoraphobia; n = 20 (21.5%)). Thirteen children (13.98%) also received an intellectual disability diagnosis. Rates of mood disorders, trauma- or stressor-related disorders, and autism spectrum disorder were relatively low and were all below 10% of the sample (Table 10).

Thirty of the 94 children (31.91%) presented with one or more of the diagnoses that the Australian Guide specifies can be used to substantiate a severe rating on the Attention, Affect Regulation, and/or Adaptive Functioning domains. Of these 30 children, 28 had pre-existing diagnoses of ADHD, 4 had pre-existing diagnoses of mood or anxiety disorders (as per the Guide), and 4 had a diagnosis of autism spectrum disorder.

For the 28 children with a pre-existing diagnosis of ADHD, 26 were medicated at the time of assessment and many did not arrive with sufficient detail to verify the ADHD classification (i.e., inattention, hyperactive, or combined). Therefore, all 28 children had their diagnosis verified either by direct or indirect assessments, or via collateral information gathered from their treating pediatrician. In two cases, the ADHD diagnosis was not confirmed and so an ADHD diagnosis was not used to substantiate a severe rating for the Attention domain.

For the four children with a pre-existing mood or anxiety disorder that would warrant a rating of severe for the Affect Regulation domain, we used the KSADs semistructured interviews and other indirect measures (e.g., CBCL) to verify the diagnosis. For one child, their existing anxiety disorder diagnosis was not supported by the assessment process and was not used to substantiate a rating of severe on the Affect Regulation domain.

For the four children with a pre-existing diagnosis of Autism, we either verified the diagnosis via the KSADs semistructured interview and other indirect measures (e.g., ABAS) or gathered collateral from previous practitioners to ascertain whether the diagnosis was valid. For one child, the Autism diagnosis was not verified during the assessment process and so the diagnosis was not used to verify a severe rating on the Adaptive Functioning domain.

TABLE 5 Impairment in neurodevelopmental domains for children assessed for FASD (N = 93)

	Total sample	Age <6.0 years (n = 40)	Age ≥6 years (n = 53)
N domains with severe impairment			
<3	31 (33.3%)	12 (30.0%)	19 (35.8%)
3 to 4	34 (36.6%)	20 (50.0%)	14 (26.4%)
≥5	28 (30.1%)	8 (20.0%)	20 (37.7%)

Note: One case had an attempted but incomplete assessment, so is not included in the table.

Sensitivity analysis

Sensitivity analyses were undertaken to examine whether using specific comorbid diagnoses to arrive at a rating of "severe" would impact FASD diagnostic outcomes. Of the 61 children receiving an FASD diagnosis, 47 received a diagnosis in at least one of the Attention, Affect Regulation, or Adaptive Functioning domain to substantiate a "severe" rating (Table 11). The most common was a diagnosis of ADHD (inattentive or combined; n = 37), followed by an eligible mood and/or anxiety disorder for the Affect Regulation domain (n = 20), and then autism spectrum disorder for the Adaptive Functioning domain (n = 7).

For 7 of the 47 children (14.9%), their diagnosis of FASD would change to an "At Risk" designation. Although another two children (Cases 53 and 18) had a diagnosis in the Attention and Adaptive Functioning domains, respectively, both would retain their "severe" rating in each domain because the direct and/or indirect assessment results met the threshold for a rating of "severe." For the Attention

TABLE 6 Australian guideline diagnostic algorithm

	Diagnostic categories		
Diagnostic criteria	**FASD with 3 SFF**	**FASD with <3 SFF**	**At risk**
Prenatal alcohol exposure	Confirmed or unknown	Confirmed	Confirmed
Neurodevelopmental domains 1. Brain structure/ neurology 2. Motor skills 3. Cognition 4. Language 5. Academic achievement 6. Memory 7. Attention 8. Executive function (including impulse control and hyperactivity) 9. Affect regulation 10. Adaptive behavior, social skills, or social communication	Severe impairment in ≥3 domains (≤2 SD or <3rd percentile) If <6 years: Diagnosis can be made if there is a "severe" global developmental delay diagnosis based on standardized assessment OR where there is microcephaly and 3 SFF (regardless of PAE or impairment based on assessment) OR severe impairment in ≥3 domains	Severe impairment in ≥3 domains (≤2 SD or < 3rd percentile) If <6 years: Diagnosis can be made if there is a "severe" global developmental delay diagnosis based on standardized assessment OR severe impairment in ≥3 domains	Under age 6 without microcephaly or impairment in ≥3 domains, but 3 SFF present Age-appropriate neurodevelopmental assessment is complete, inconclusive, or impossible Despite confirmed PAE, impairment is present in <3 domains Impairment in ≥3 domains, but not sufficient to meet clinical cutoff for "severe"[a]
Sentinel facial features 1. Short palpebral fissure 2. Smooth philtrum 3. Thin upper lip	Presence of 3 SFF	Presence of 0, 1, or 2 SFF	Presence of 0 to 3 SFF

[a]For children who did not receive a rating of "severe" in ≥3 domains, decision rules were used to ensure consistency between "At Risk" and no diagnosis. While the Australian Guide allows for an "At Risk" designation when there is impairment in ≥3 domains that is not at a level to permit a "severe" classification, there is no specification of what level of impairment would be sufficient for cases where the child has no or <3 "severe" ratings. [Correction added on March 03, 2023, after first online publication: In the preceding sentence, 'diagnosis' has been changed to 'designation'.] Informed by the Washington Guide and the "Clinical judgment" considerations in the Australian Guide, the following decision rules were developed to decide if "some" impairment was present in these cases with <3 "severe" domains: (a) whether an index or composite fell ≥1 < 2 SD below the mean (Rank 2 impairment (i.e., mild/moderate) in Washington Guide, 16th to 3rd percentile); and/or (b) whether the 95% confidence intervals for an index or composite contained a score that met the "severe" threshold. We were also conscious of the Guide stating that "a domain should not be considered impaired on the basis of a single subtest score" (p. 19).

domain—and excluding diagnoses in other domains—3 of the 37 children would change from an FASD diagnosis to "At Risk" if an ADHD diagnosis was not used to substantiate a "severe" rating (Cases 32, 54, 73). For the Affect domain, and excluding diagnoses in other domains, 2 of the 20 children would receive an "At-Risk" designation if a mood/anxiety diagnosis was not used to substantiate a "severe" rating (Cases 32, 27). While seven children also had a diagnosis of Autism, this alone did not contribute to their diagnostic status as at least three other domains met criteria for severe impairment.

For 13 children, an eligible diagnosis was used to substantiate a "severe" rating in two or more of the Attention, Affect Regulation, or Adaptive Functioning domains (Table 11). If all eligible diagnoses were not used for substantiating a "severe" rating in these cases, four cases would change from an FASD diagnosis to an "At-Risk" designation (Cases 16, 29, 32, 47).

Of the 29 children receiving an "At Risk" designation, 13 received a diagnosis in either the Attention, Affect Regulation, or Adaptive Functioning Domain to substantiate a "severe" rating (Table 11). Regardless of whether another diagnosis was used to establish a "severe" rating, all of children would retain their diagnostic status due to impairment in multiple other domains (either "severe" or "some"). The single case with no diagnosis where an ADHD diagnosis was used to substantiate a "severe" rating for the Attention domain would retain their diagnostic status as PAE was unknown.

DISCUSSION

The early identification of atypical development in infants and children is vital in ensuring early intervention is provided at a critical stage in the development of neurocognitive functioning (Flannigan et al., 2019). The inclusion of processes to identify severe functional impairment in younger children (aged <6 years), in both the Australian and Canadian Guides, provides a valuable opportunity for early diagnosis and intervention with this age group. The current study reports on the diagnostic outcomes and neurodevelopmental profiles of children aged 3 to 7 years with suspected or confirmed PAE, both as a total sample and then divided into very young (age 3 to 5 years 11 months) and young children (aged 6 to 7 years 8 months). We comment on the complexities inherent in determining

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TABLE 7 Diagnostic summary across domains for children assessed for FASD aged <6 years

Case ID	Age (months)	PAE	SFF	BRAIN	MO	CO	LA	AC	ME	AT	EF	AR	AD	N severe domains	Diagnosis
1	37	Confirmed high-risk	2							DX				4	FASD < 3 SFF
2	38	Confirmed high-risk	2										DX	5	FASD < 3 SFF
3	40	Confirmed high-risk	3											3	FASD 3 SFF
4	41	Confirmed high-risk	3										DX	6	FASD 3 SFF
5	41	Confirmed high-risk	UK											1	At risk of FASD
6	42	Confirmed high-risk	0											4	FASD < 3 SFF
7	43	Confirmed high-risk	3											3	FASD 3 SFF
9	46	Confirmed high-risk	2											3	FASD < 3 SFF
8	46	Confirmed exposure	0											0	At risk of FASD
10	49	Confirmed exposure	1							DX				4	FASD < 3 SFF
11	51	Confirmed exposure	2											1	At risk of FASD
12	53	Confirmed high-risk	3											1	At risk of FASD
13	54	Confirmed high-risk	1											3	FASD < 3 SFF
14	56	Confirmed exposure	1							DX				4	FASD < 3 SFF
15	56	Confirmed high-risk	1											1	At risk of FASD
17	57	Confirmed high-risk	2							DX	*	DX		5	FASD < 3 SFF
16	57	Confirmed exposure	0							DX		DX		4	FASD < 3 SFF
20	58	Confirmed exposure	0							DX				4	FASD < 3 SFF
21	58	Confirmed high-risk	2											1	At risk of FASD
19	58	Confirmed high-risk	1									DX		4	FASD < 3 SFF
18	58	Confirmed exposure	0										DX	3	FASD < 3 SFF
22	62	Confirmed exposure	0							DX				2	At risk of FASD
23	62	Confirmed high-risk	1							DX				6	FASD < 3 SFF
24	62	Confirmed exposure	1											4	FASD < 3 SFF
25	62	Confirmed exposure	2											1	At risk of FASD
26	63	Confirmed high-risk	0							DX	*			7	FASD < 3 SFF
29	64	Confirmed high-risk	2							DX		DX		4	FASD < 3 SFF
28	64	Confirmed exposure	2								*			6	FASD < 3 SFF
27	64	Confirmed high-risk	3									DX		3	FASD 3 SFF
30	65	Confirmed high-risk	3									DX		4	FASD 3 SFF
31	66	Confirmed high-risk	2											3	FASD < 3 SFF
32	68	Confirmed exposure	0							DX		DX		3	FASD < 3 SFF
33	68	Confirmed high-risk	1											0	At risk of FASD
34	69	Confirmed high-risk	1							DX				2	At risk of FASD
35	69	Confirmed exposure	0									DX		4	FASD < 3 SFF
36	70	Confirmed exposure	2											0	No diagnosis
37	71	Confirmed exposure	0							DX		DX		6	FASD < 3 SFF
38	71	Confirmed exposure	1							DX	*	DX		7	FASD < 3 SFF
40	71	Confirmed high-risk	2							DX				4	FASD < 3 SFF
39	71	Confirmed exposure	3							DX				2	At risk of FASD

Note.

= No impairment … = Some impairment = Severe impairment No impairment

DX = specific diagnosis from Australian Guide present. * = Direct measures of EF used to substantiate 'severe' rating.

Note: : No impairment; : some impairment; : severe impairment; : no impairment. DX: specific diagnosis from Australian Guide present.

*Direct measures of EF used to substantiate "severe" rating.

TABLE 8 Diagnostic summary across domains for children assessed for FASD aged ≥6 years

Case ID	Age (months)	PAE	SFF	BRAIN	MO	CO	LA	AC	ME	AT	EF	AR	AD	N severe domains	Diagnosis
42	72	Confirmed exposure	2							DX				5	FASD < 3 SFF
41	72	Confirmed high-risk	0							DX				2	At risk of FASD
43	72	Confirmed exposure	1							DX				2	At risk of FASD
44	73	Unknown exposure	0							DX	*			4	No diagnosis
45	73	Confirmed high-risk	1									DX		1	At risk of FASD
46	73	Confirmed exposure	2											1	At risk of FASD
47	74	Confirmed exposure	2							DX		DX		4	FASD < 3 SFF
49	74	Confirmed high-risk	2											3	FASD < 3 SFF
48	74	Confirmed exposure	0								*			5	FASD < 3 SFF
50	74	Confirmed exposure	2											0	No diagnosis
52	75	Confirmed high-risk	3							DX				5	FASD 3 SFF
53	75	Confirmed exposure	2							DX				3	FASD < 3 SFF
51	75	Confirmed exposure	0											1	At risk of FASD
54	76	Confirmed exposure	1							DX				3	FASD < 3 SFF
56	77	Confirmed high-risk	3											UK	At risk of FASD
57	77	Unknown exposure	3							DX	*			6	FASD 3 SFF
55	77	Confirmed high-risk	1							DX				4	FASD < 3 SFF
58	78	Confirmed high-risk	0							DX				1	At risk of FASD
59	79	Confirmed high-risk	0							DX				6	FASD < 3 SFF
60	81	Confirmed high-risk	2							DX				1	At risk of FASD
61	81	Unknown exposure	3									DX		5	FASD 3 SFF
62	82	Confirmed high-risk	1							DX	*	DX		5	FASD < 3 SFF
63	82	Confirmed exposure	0							DX				4	FASD < 3 SFF
64	82	Confirmed high-risk	3										DX	4	FASD 3 SFF
65	83	Confirmed high-risk	0							DX	*			6	FASD < 3 SFF
66	84	Confirmed high-risk	1							DX	*			2	At risk of FASD
67	85	Confirmed exposure	1								*			5	FASD < 3 SFF
68	85	Confirmed exposure	1											3	FASD < 3 SFF
70	86	Confirmed high-risk	0							DX				4	FASD < 3 SFF
71	86	Confirmed exposure	0							DX	*		DX	6	FASD < 3 SFF
69	86	Confirmed high-risk	2											0	At risk of FASD
73	87	Confirmed high-risk	1							DX				3	FASD < 3 SFF
72	87	Confirmed high-risk	0							DX		DX	DX	7	FASD < 3 SFF
74	87	Confirmed high-risk	2							DX	*	DX		7	FASD < 3 SFF
77	87	Confirmed high-risk	1							DX		DX		7	FASD < 3 SFF
75	87	Confirmed exposure	1							DX	*			6	FASD < 3 SFF
76	87	Confirmed exposure	2							DX				5	FASD < 3 SFF
79	88	Confirmed high-risk	2							DX				4	FASD < 3 SFF
80	88	Confirmed high-risk	0							DX				1	At risk of FASD
78	88	Confirmed exposure	1								*			6	FASD < 3 SFF
84	89	Confirmed high-risk	0							DX				6	FASD < 3 SFF
88	89	Confirmed high-risk	3							DX	*			5	FASD 3 SFF
82	89	Confirmed high-risk	0									DX		2	At risk of FASD
89	89	Confirmed high-risk	2											1	At risk of FASD
90	89	Confirmed high-risk	2											2	At risk of FASD
86	89	Confirmed high-risk	1											3	FASD < 3 SFF
81	89	Confirmed high-risk	2									DX		2	At risk of FASD
83	89	Confirmed exposure	1											2	At risk of FASD
85	89	Confirmed exposure	0											1	At risk of FASD
87	89	Confirmed high-risk	1									DX	DX	6	FASD < 3 SFF
91	90	Confirmed high-risk	1							DX				1	At risk of FASD
93	90	Confirmed high-risk	1							DX				7	FASD < 3 SFF
92	90	Confirmed exposure	2											0	No diagnosis
94	92	Confirmed high-risk	1							DX				4	FASD < 3 SFF

Note.

= No impairment … = Some impairment = Severe impairment No impairment

DX = specific diagnosis from Australian Guide present. * = Direct measures of EF used to substantiate 'severe' rating.

Note: : No impairment; … : some impairment; : severe impairment; : no impairment. DX: specific diagnosis from Australian Guide present.

*Direct measures of EF used to substantiate "severe" rating.

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TABLE 9 Comorbid mental health diagnoses in children assessed for FASD ($N = 93$)

	Total sample	Age <6.0 years ($n = 40$)	Age ≥6 years ($n = 53$)
No diagnosis	3 (3.2%)	1 (7.5%)	2 (3.8%)
Diagnosis/diagnoses other than FASD	1 (1.1%)	0 (0.0%)	1 (1.9%)
FASD diagnosis only	10 (10.8%)	6 (15.0%)	4 (7.5%)
FASD plus 1 comorbid diagnosis	21 (22.6%)	10 (25.0%)	11 (20.8%)
FASD plus 2 comorbid diagnoses	18 (19.4%)	7 (17.5%)	11 (20.8%)
FASD plus >2 comorbid diagnoses	40 (43.0%)	16 (40.0%)	24 (45.3%)

Note: One case had an incomplete assessment and no verification of comorbid diagnoses.

TABLE 10 Comorbid mental health diagnoses in children assessed for FASD ($N = 93$)

	Total sample	Age <6.0 years ($n = 40$)	Age ≥6 years ($n = 53$)
Attention-deficit/hyperactivity disorder	48 (51.6%)	16 (40.0%)	32 (60.4%)
Mood disorder[a]	4 (4.3%)	0 (0.0%)	4 (7.5%)
Anxiety disorder[b]	20 (21.5%)	10 (25.0%)	10 (18.9%)
Trauma- or stressor-related disorder	9 (9.7%)	2 (5.0%)	7 (3.8%)
Intellectual disability	13 (14.0%)	5 (12.5%)	8 (15.1%)
Autism spectrum disorder	7 (7.5%)	3 (7.5%)	4 (7.5%)
Behavior disorder[c]	21 (22.6%)	9 (22.5%)	12 (22.6%)
Other un/specified neurodevelopmental disorder	5 (5.4%)	1 (2.5%)	4 (7.5%)

Note: Some children have multiple diagnoses, so comorbidities do not sum to the total sample size; $n = 1$ case had an incomplete assessment and no verification of comorbid diagnoses.

[a]Includes disorders listed in the Australian Guide under Affect Regulation domain: Major depressive disorder (with recurrent episode), persistent depressive disorder, disruptive mood regulation disorder.

[b]Includes disorders listed in the Australian Guide under Affect Regulation domain: Separation anxiety disorder, selective mutism, social anxiety disorder, panic disorder, agoraphobia, generalized anxiety disorder.

[c]Includes disorders listed in the Australian Guide under the adaptive behavior, social skills, or social communication domain: oppositional defiance disorder, conduct disorder.

a diagnosis of FASD in the context of potential comorbidity. Finally, sensitivity analyses were undertaken to examine whether using specific comorbid diagnoses to establish a rating of "severe" would impact FASD diagnostic outcomes.

Ninety-four children underwent a comprehensive, multidisciplinary assessment for FASD. Most children referred to the study received a diagnosis of FASD with either three sentinel facial features or less than three sentinel facial features (64.9%). A smaller proportion (30.9%) received an "At Risk" for FASD designation with reassessment recommended in the future, while only a small number (4.3%) did not receive a diagnosis of FASD. Notably, there was no difference in the two age groups in overall diagnostic status, a finding that was somewhat surprising as the potential for an "At Risk" designation would appear to be more likely in younger children given the complexities of assessing neurodevelopment in young children. It is notable that nearly 40% of children over age 6 years had severe impairment in five or more domains compared to 20% of children <6 years. This cannot be taken as evidence of increasingly compromised neurodevelopment given the cross-sectional nature of the analysis, but may suggest the influence of age-related cognitive skill differentiation on the assessment and diagnostic process. Nevertheless, it does underscore the importance of obtaining longitudinal data from early childhood to determine whether patterns of impairment are influenced by environmental and contextual factors such as stability in care and appropriate support.

In over 80% of cases, the confirmation of PAE was based on information rated as highly reliable and over half of the cases were classified as confirmed high-risk exposure (Table 1). While diagnosis can be reliably made when the characteristic sentinel facial features are present (Wozniak et al., 2019), typically less than 10% of children have all three sentinel facial features (Roozen et al., 2016). The findings in this current sample are consistent with previous findings: 13.8% of children had three sentinel facial features with the majority of children having only one or two (29.8% and 28.7%, respectively). Consequently, the diagnosis of FASD was reliant on accurate ascertainment of impairment across neurodevelopmental domains.

The patterns of neurodevelopmental impairment in the current sample show a diverse profile: while impairments in executive function are particularly notable in both very young and young children (28/40 and 34/54, respectively), a significant proportion had "severe" impairment in the attention (24/40 and 36/54, respectively) and motor domains (15/40 and 24/54, respectively). Notably, and consistent with other findings (e.g., Mattson et al., 2010, 2011), relatively fewer children were rated as "severe" on the cognitive domain (15/40 and 16/54, respectively).

The pattern of findings in the current study raises a number of diagnostic challenges, some of which reflect the complexity of the presentation of FASD. However, others are specific to (i) the diagnostic algorithms and conceptualization of the disorder in the Australian Guide, and (ii) the sensitivity of neuropsychological assessments of children in the early years (Anderson & Reidy, 2012). This issue is of particular relevance when determining whether there was "severe" impairment in executive function (EF). There is considerable variability in the developmental trajectory of executive and related functions in young children (Best & Miller, 2010). Further, the additional challenge of task impurity where a task measures multiple components of EF and other cognitive functions makes assessing specific aspects of executive functioning more challenging in this

TABLE 11 Diagnostic summary across domains for children with comorbid diagnoses to establish severe rating

Case ID	Age (mths)	PAE	SFF	BRAIN	MO	CO	LA	AC	ME	AT	EF	AR	AD	N Severe Domains	Diagnosis
1	37	Confirmed high-risk	2							DX				4	FASD < 3 SFF
2	38	Confirmed high-risk	2										DX	5	FASD < 3 SFF
4	41	Confirmed high-risk	3										DX	6	FASD 3 SFF
10	49	Confirmed exposure	1							DX				4	FASD < 3 SFF
14	56	Confirmed exposure	1							DX				4	FASD < 3 SFF
16	57	Confirmed exposure	0							DX		DX		4	FASD < 3 SFF
17	57	Confirmed high-risk	2							DX		DX		5	FASD < 3 SFF
18	58	Confirmed exposure	0										DX	3	FASD < 3 SFF
19	58	Confirmed high-risk	1									DX		4	FASD < 3 SFF
20	58	Confirmed exposure	0							DX				4	FASD < 3 SFF
22	62	Confirmed exposure	0							DX				2	At risk of FASD
23	62	Confirmed high-risk	1							DX				6	FASD < 3 SFF
26	63	Confirmed high-risk	0							DX				7	FASD < 3 SFF
27	64	Confirmed high-risk	3									DX		3	FASD 3 SFF
29	64	Confirmed high-risk	2							DX		DX		4	FASD < 3 SFF
30	65	Confirmed high-risk	3									DX		4	FASD 3 SFF
32	68	Confirmed exposure	0							DX		DX		3	FASD < 3 SFF
34	69	Confirmed high-risk	1							DX				2	At risk of FASD
35	69	Confirmed exposure	0									DX		4	FASD < 3 SFF
37	71	Confirmed exposure	0							DX		DX		6	FASD < 3 SFF
38	71	Confirmed exposure	1							DX		DX		7	FASD < 3 SFF
39	71	Confirmed exposure	3							DX				2	At risk of FASD
40	71	Confirmed high-risk	2							DX				4	FASD < 3 SFF
41	72	Confirmed high-risk	0							DX				2	At risk of FASD
42	72	Confirmed exposure	2							DX				5	FASD < 3 SFF
43	72	Confirmed exposure	1							DX				2	At risk of FASD
44	73	Unknown exposure	0							DX				4	No diagnosis
45	73	Confirmed high-risk	1									DX		1	At risk of FASD
47	74	Confirmed exposure	2							DX		DX		4	FASD < 3 SFF
52	75	Confirmed high-risk	3							DX				5	FASD 3 SFF
53	75	Confirmed exposure	2							DX				3	FASD < 3 SFF
54	76	Confirmed exposure	1							DX				3	FASD < 3 SFF
55	77	Confirmed high-risk	1							DX				4	FASD < 3 SFF
57	77	Unknown exposure	3							DX				6	FASD 3 SFF
58	78	Confirmed high-risk	0							DX				1	At risk of FASD
59	79	Confirmed high-risk	0							DX				6	FASD < 3 SFF
60	81	Confirmed high-risk	2							DX				1	At risk of FASD
61	81	Unknown exposure	3									DX		5	FASD 3 SFF
62	82	Confirmed high-risk	1							DX		DX		5	FASD < 3 SFF
63	82	Confirmed exposure	0							DX				4	FASD < 3 SFF
64	82	Confirmed high-risk	3										DX	4	FASD 3 SFF
65	83	Confirmed high-risk	0							DX				6	FASD < 3 SFF
66	84	Confirmed high-risk	1							DX				2	At risk of FASD
70	86	Confirmed high-risk	0							DX				4	FASD < 3 SFF
71	86	Confirmed exposure	0							DX			DX	6	FASD < 3 SFF
72	87	Confirmed high-risk	0							DX		DX	DX	7	FASD < 3 SFF
73	87	Confirmed high-risk	1							DX				3	FASD < 3 SFF
74	87	Confirmed high-risk	2							DX		DX		7	FASD < 3 SFF
75	87	Confirmed exposure	1							DX				6	FASD < 3 SFF
76	87	Confirmed exposure	2							DX				5	FASD < 3 SFF
77	87	Confirmed high-risk	1							DX		DX		7	FASD < 3 SFF
79	88	Confirmed high-risk	2							DX				4	FASD < 3 SFF
80	88	Confirmed high-risk	0							DX				1	At risk of FASD
81	89	Confirmed high-risk	2									DX		2	At risk of FASD
82	89	Confirmed high-risk	0									DX		2	At risk of FASD
84	89	Confirmed high-risk	0							DX				6	FASD < 3 SFF
87	89	Confirmed high-risk	1									DX	DX	6	FASD < 3 SFF
88	89	Confirmed high-risk	3							DX				5	FASD 3 SFF
91	90	Confirmed high-risk	1							DX				1	At risk of FASD
93	90	Confirmed high-risk	1							DX				7	FASD < 3 SFF
94	92	Confirmed high-risk	1							DX				4	FASD < 3 SFF

Note.
= No impairment … = Some impairment = Severe impairment No impairment
DX = specific diagnosis from Australian Guide present. * = Direct measures of EF used to substantiate 'severe' rating.

Note: : No impairment; … : some impairment; : severe impairment; : no impairment. DX: specific diagnosis from Australian Guide present.

*Direct measures of EF used to substantiate "severe" rating.

young age group (see Reilly et al., 2022 for a review). Nonetheless, this is also the age group where there is significant development in inhibitory control and cognitive flexibility (Anderson & Reidy, 2012; Best & Miller, 2010).

This raises challenges around the most appropriate measure of EF in this age group. The Australian Guide specifies that a "severe" rating can only be given when there is converging evidence from multiple sources (p. 28). We read this to be at least two sources (as per the instructions provided for direct measures) and that the impairment needed to be in a composite score (as per instructions for other domains). The preponderance of "severe" ratings on EF were obtained by concordance on indirect measures, specifically between teacher and carer/parent Behavior Rating Inventory of Executive Function (BRIEF; 46 of 62 cases). The determination of "severe" where there was convergence across both direct and indirect measures was considerably smaller (12 of 62 cases). Thus, it appears that the neurodevelopmental tests, although carefully selected, had discordance with self-report measures. Though this does not necessarily indicate that direct assessments were not sensitive to executive functioning impairment. The association between ratings on the BRIEF and neuropsychological tests is typically weak (Toplak et al., 2013), and these findings may be simply another example in which reliance on measures that are aligned with everyday life (i.e., show verisimilitude) is recommended (Wallisch et al., 2018). It also aligns with Toplak et al.'s (2013) suggestion that questionnaires and direct assessments should not be used equivalently or interchangeably as they may represent two dissociated constructs. Nevertheless, the extent to which elevated scores on the BRIEF reflected parent/carer perceptions needs to be considered. The BRIEF and BRIEF-P have validity indices, which requires careful consideration if elevated. We did not use a BRIEF that had elevated negativity indices and would only rate the Executive Function domain as "severe" if there was converging evidence from multiple sources, which excluded a BRIEF that was considered invalid (Bower & Elliott, 2016, p. 28). It is also possible that the behaviors reported on in the BRIEF as measures of executive functioning, reflected, in part, other psychological problems. However, the decision rules embedded within the Guide mean that a rating of "severe" on a parent-report measure alone cannot be given.

The high rate of co-occurring mental health diagnoses is notable in this sample of young children (Tables 8 and 9) and show similar patterns to a broader literature on individuals with FASD. Weyrauch et al. (2017), in their systematic review, reported the following: co-occurrence of ADHD (50%); intellectual disability (23%); oppositional defiant disorder (16%) depression and anxiety disorders (16 and 8%, respectively) and PTSD (6%). A later study by Lange et al. (2018) found similarly high rates of ADHD (53%), followed by oppositional defiant disorder (13%), conduct disorder (7%) and also documented rates of autism spectrum disorder at 2.6%. There are two issues relating to co-occurring diagnoses. First, the question as to whether these disorders are indeed separate disorders. Behaviors such as impulsivity, extreme emotional dysregulation, poor attention and concentration and impairment in other executive functions such as working memory, organization and planning are shared diagnostic criteria of ADHD and FASD, have considerable overlap with ODD, and are common in children who have experienced childhood neglect (Maguire et al., 2015) and/or had exposure to child maltreatment and trauma (Hackman & Farah, 2009; Malarbi et al., 2017). Second, whether the presence of a "diagnosis" of these disorders increases the likelihood of a false-positive diagnosis of FASD as these disorders can result in a "severe" rating across three separate neurodevelopmental domains: Affect Domain, Attention, and Adaptive Functioning.

In the current sample, nearly 70% of the children had been involved with child protective services at some point in their life, and approximately 60% were currently involved with child protective services, indicating significant exposure to childhood adversity in this population. While the Guide clearly states that early life trauma (including social and emotional abuse) needs to be considered when determining a diagnosis of FASD (p. 13), in practice this is difficult to tease apart in a cross-sectional assessment, particularly where there may have been disruption in the child's caregiving experience. For example, many of the diagnoses that are assessed under the "Affect Regulation" domain may be a consequence of exposure to disrupted and inconsistent caregiving, thereby providing disruption in attachment: mood and anxiety disorders are highly prevalent in children who have experienced maltreatment (Gardner et al., 2019). While it is important to determine the presence of such disorders to ensure that adequate services are provided, the assumption of a causal link between PAE and these disorders may obscure the clinical picture and, at worse, increase the possibility of a false-positive diagnosis of FASD.

A similar concern can also be raised for ADHD (inattentive or combined). Once again, prevalence of ADHD in children involved in the child protection system is much higher than the general population (Craig et al., 2020) and is associated with neuropsychological deficits in emotional regulation and executive functioning—core deficits in children with FASD. Thus, if ADHD is independent of PAE and occurs in the context of child maltreatment, the potential for a misdiagnosis of both ADHD and FASD is possible.

A comment on the diagnosis of Autism is also necessary. In this sample, seven children had an Autism diagnosis and therefore met the "severe" criteria for the "Adaptive Behaviour" domain. There is no question that children with Autism have significant social and communication challenges but the attribution of this to PAE is controversial (Carpita et al., 2022). While autistic symptomology can be observed in children with FASD (Stevens et al., 2013), autistic symptomology can also be the result of early childhood neglect (Bishop et al., 2007; Carpita et al., 2022; Dodds, 2021). It may be timely to consider whether it is appropriate to continue using a diagnosis of Autism as evidence for "severe" impairment in this domain, particularly given that other behavioral disorders listed by the Guide for this domain cannot be used to establish a "severe" rating (i.e., oppositional defiance disorder and conduct disorder).

The extent to which these issues may result in a false-positive diagnosis of FASD can be informed by the sensitivity analysis. Of the 61 children receiving an FASD diagnosis, 47 received a diagnosis in at least one of the Attention, Affect Regulation, or Adaptive Functioning domains that could be used to substantiate a "severe" rating. Seven of 47 children would revert from an FASD diagnosis to an "At Risk" designation representing a potential false-positive rate of 15% for this subsample. [Correction added on March 03, 2023, after first online publication: In the preceding sentence, 'diagnosis' has been changed to 'designation'.]

CONCLUSIONS AND FUTURE DIRECTIONS

The Australian Guide to the diagnosis of FASD provided an extensive assessment of 10 domains of neurodevelopmental functioning. The Guide provided clear recommendations around assessment measures, and these were used to structure the assessment process. There were no differences in age groups in the proportion of children who received a diagnosis of FASD or "At Risk" designation. The potential challenges associated with using co-occurring diagnosis of neurodevelopmental and mental disorders to fulfill criteria for a "severe" rating in domains of Affect, Attention, and Adaptive Functioning was considered. Notably, sensitivity analyses found that when removed, approximately 15% (7 of 47 diagnoses) changed to an "At Risk" designation. The pattern of impairments found across both very young (≤5 years, 11 months) and young (≥6 years, 0 months) children was diverse, although nearly two thirds of children were rated as "severe" in executive functioning and attention (mostly due to a co-occurring diagnosis of ADHD). Future research and diagnostic algorithms could consider the utility of specifiers if homogenous subgroupings that have shared features (Harris, 2014) could be identified and reliably classified in individuals with FASD. Notably, this idea underpinned the adoption of specifiers for neurodevelopmental disorders in the DSM-5 (Harris, 2014) and has continued to be adopted with the DSM-5-TR. It may be helpful to consider one such subgroup as "FASD with attention and or executive function difficulties." There were also a number of children with "severe" impairment across multiple domains. Even with the caveat around the potential of false positives due to non-PAE-related disorders such as mood and anxiety disorders, there may be a case for consideration of severity ratings, which may then inform the level of supports required for children and families. These suggestions require further research and careful field trials. However, the potential for better tailoring of support and treatment for children less than 7 years old warrants further consideration of how best to advance the diagnosis and assessment of young children with PAE.

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CONFLICT OF INTEREST

The authors declare no conflict of interests.

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SUPPORTING INFORMATION

Additional supporting information can be found online in the Supporting Information section at the end of this article.

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