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Profile of children diagnosed with a fetal alcohol spectrum disorder: A retrospective chart review

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Abstract

Introduction and Aims: Fetal alcohol spectrum disorder (FASD) is a significant public health concern and growing recognition in Australia led to the establishment of a specialist service for young children. The aim of the current study was to report on the diagnostic profile of a group of children who attended the service, to document the extent to which there were comorbid diagnoses and to provide information on the neurocognitive functioning of the children.

Design and Methods: This study was a retrospective chart review conducted by the diagnostic service. A pre-formulated tool to extract the diagnostic data from the client files was designed, a chart review was performed and the required data was extracted. FASD diagnoses were made using the 4-Digit Diagnostic Code.

Results: 31 families participated and the majority of children were diagnosed with static encephalopathy (alcohol exposed) or neurobehavioral disorder (alcohol exposed) (11 children each; 36%). Only one child was diagnosed with fetal alcohol syndrome (alcohol exposed) and five children were diagnosed with partial fetal alcohol syndrome (alcohol exposed). Twenty-six children (84%) had a comorbid diagnosis, with 19 (61%) having a comorbid diagnosis of ADHD. While the majority of children were not found to display growth deficiency or significant facial features, 18 children (58%) had significant central nervous system dysfunction.

Discussion and Conclusions: The current study demonstrates that with the relevant training and expertise, assessment and diagnosis of FASD can be embedded within the existing health services available in Australia.

Keywords: Fetal alcohol spectrum disorder; assessment; diagnosis, Australia

Introduction

The term fetal alcohol spectrum disorder (FASD) is used to describe the broad spectrum of disabilities that can result from prenatal alcohol exposure [1]. Prevalence estimates of FASD vary considerably [2] with recent estimates in the USA suggesting a prevalence of 24 to 48 per 1000 children [3], although for particular sub-populations these rates are considerably higher [4, 5]. Over the past 20 years, multidisciplinary FASD diagnostic clinics have been established in many countries including, Canada, USA, New Zealand and most recently Australia. Diagnostic processes have evolved over time and there are now a number of published diagnostic guidelines available. These include, the 4-Digit Diagnostic Code [6], the updated Institute of Medicine guidelines [7], the updated Canadian Guidelines [1] and most recently the Australian Guide to the Diagnosis of FASD [8]. The 4-Digit Code [6] is a widely use diagnostic system in which FASD is viewed as an umbrella term to encompass the diagnostic categories (e.g., FAS, pFAS, static encephalopathy). Whereas, the Australian Guide [8], following the updated Canadian guidelines [1], now use FASD as a diagnostic term and divide diagnosis into two categories: (i) FASD with three sentinel facial features and (ii) FASD with less than three sentinel facial features.

Growing recognition of FASD across policy and service delivery platforms in Australia led to the establishment of a specialist service for young children within a state government-funded health care system. This service was the first of four currently available in Australia, and the only publically provided service in Queensland. The aim of the current study was threefold: (i) to report on the diagnostic profile of a group of children diagnosed with a FASD, (ii) to document the extent to which there were comorbid diagnoses and (iii) to provide information on the neurocognitive functioning of children who were diagnosed with a FASD.

Method

Assessment process

Children were referred to the service by general practitioners if there were concerns regarding behaviour or development and reports from caregivers or documented evidence of prenatal alcohol exposure. The multidisciplinary team provides assessment, diagnosis and follow-up as follows: (1) a comprehensive clinical intake with the family, liaison with health care providers, education and, if necessary, statutory child protection services, and review of available medical and educational records; (2) an assessment of the key FASD features to derive a diagnosis using the 4-Digit Code (one and a half to two days of assessment per child); (3) a comprehensive report written by the multi-disciplinary team and discussed with

caregivers and health care providers; (4) a school meeting to develop an educational support plan; and (5) long-term follow-up by the Paediatrician as required. The clinic had the capacity to assess two children per month.

Diagnosis using the 4-Digit Diagnostic Code

This study predated the release of the Australian Guide to the diagnosis of FASD. The 4-Digit Code was one of the most extensively researched diagnostic guidelines available. One Clinical Psychologist attended training with the Washington State FAS Diagnostic Prevention Network and all other members of the multi-disciplinary team completed the 4-Digit Code online training course and training in North America in harmonisation of the 4-Digit Code and the Revised IOM criteria. The four digits of the code reflect the four key diagnostic features of FASD: (1) growth deficiency, (2) facial phenotype, (3) central nervous system (CNS) structural/functional abnormalities, and (4) prenatal alcohol exposure. The degree of expression of each of these features is ranked independently on a 4-point Likert scale, with 1 reflecting the absence of the feature and 4 reflecting the strong presence of the feature. Further, facial phenotype was assessed using FAS Facial Photographic software [9]. See Supplementary Tables S1, S2, S3 and S4 for further details regarding diagnosis using the 4-Digit Code.

Procedure

The current study was approved by the Queensland Health Human Research Ethics Committee and conducted by the diagnostic service. This was a convenience sample; families who had previously been assessed by the service or current clients provided written consent to be included in the study. A pre-formulated spreadsheet detailing the relevant demographic and diagnostic data was designed, a chart review was performed by the first author and the required data was extracted.

Results

There were 37 children assessed between March 2014 and December 2015, and 31 families consented to be included. Six families could not be contacted due to a range of difficult life circumstances (e.g., placement breakdowns, death of family members). The mean age of the children was 8.5 years (range 6 – 13 years; *SD* = 1.71). Seventeen children were Caucasian Australian, 10 children from Aboriginal or Torres Strait Islander backgrounds and four children from other varied backgrounds. Eleven caregivers were foster parents, eight were legal guardians (i.e., foster or kinship caregivers who had long-term full parental responsibilities for a child), five were adoptive or kinship caregivers and the remaining seven were biological parent(s). The majority of children in the current sample had

experienced one care placement (16), the remaining eight had experienced two or more placements. Fourteen caregivers reported that their child had experienced trauma or neglect during their lifetime.

FASD diagnostic profile

All children received a FASD diagnosis; eleven children were each diagnosed with Static Encephalopathy Alcohol Exposed (SE/AE; 35%) or Neurobehavioral Disorder Alcohol Exposed (ND/AE; 35%). One child was diagnosed with FAS; five children with pFAS; and one child was diagnosed with Sentinel Physical Findings/ND/AE. Two other diagnoses were "alcohol exposure unknown" (ND/alcohol exposure unknown & SE/alcohol exposure unknown), thus a Rank 2 (Unknown Risk) was given (Table 1).

Comorbid diagnoses

Twenty-six children (84%) had a comorbid diagnosis, of which the most common was attention-deficit hyperactivity disorder (19 children). The next most prevalent comorbid conditions were Intellectual Disability (7 children) and Autism Spectrum Disorder (4 children).

Neurocognitive functioning

Although the majority of children were not found to display growth deficiency or significant facial features, 18 children (58%) were found to have significant CNS dysfunction, thus receiving a Rank 3 on the 4-Digit Code (see Tables 1 & 2).

Mean Full Scale Intelligence Quotient (FSIQ) on the Wechsler Intelligence Scale for children (WISC-IV) was found to be 85.7 [Low Average] (*SD* = 17.9, range 49 [Extremely Low] – 123[Superior]). The Mean General Adaptive Composite (GAC) on the Adaptive Behaviour Assessment System-Second Edition (ABAS-2) was 67.5 [Extremely Low] (*SD* = 15.3, range 41[Extremely Low] – 104[Average]). This indicates a significant difference between children's general cognitive and adaptive abilities, t (27) = 5.2, p <0.01. Over 90% of children scored in the Clinical Range for behaviour problems (T score > 63) on the Child Behaviour Checklist (see Table 3).

Discussion

The current study describes the profile of children who had been referred to the first FASD diagnostic service permanently operating within an Australian public health service. The diagnostic profile and associated features were consistent with previous reports that have also implemented the 4-Digit Code [e.g.,10, 11]. Notably, the majority of children did not have the physical features of FASD, a finding that is markedly similar to a major US study of 1,400 individuals with prenatal alcohol exposure, where 4% were diagnosed with FAS; 7%

pFAS; 28% SE/AE; and 52% ND/AE [10]. It is important to note that the range of diagnoses found in this and other studies of clinical outcomes are not always assessed in prevalence studies. For example, the only published Australian prevalence study to date, using active case ascertainment, reported only FAS and pFAS [5]. It is likely that higher rates of FASD will be found in similar, highly exposed Australian populations when including the full range of diagnoses in the FASD spectrum.

Comorbid diagnoses are frequently found for children with FASD [e.g., 10] with approximately half having a diagnosis of ADHD [12]. Similar results were obtained in this Australian sample, highlighting the importance of considering both diagnoses when children present to health professionals. Again consistent with overseas findings [e.g., 10], the majority of children experienced significant CNS dysfunction and significant delays in their adaptive behaviour compared to their IQ performance. Notably, having fewer physical features of FASD and a higher IQ has been associated with more behavioural problems [13] and poorer long-term outcomes [14].

Additionally, children performed poorly on a range of tests assessing executive functions (EF) with none receiving a ranking in the normal range. As these difficulties may underpin adaptive functioning impairments [15], identification and remediation is extremely important to avoid life-long difficulties across multiple domains. The growing evidence base indicates that amelioration of these difficulties is possible [16], highlighting the importance of accurate diagnosis and access to appropriate supports.

Finally, it is important to consider the potential compounding effects of other negative psychosocial risk factors [17]. In keeping with previous studies [e.g.,14], a large proportion of children in the current study had also experienced exposures to other substances prenatally, childhood trauma or neglect, and sometimes multiple care placements. Thus, service development and treatment planning needs to consider that the range of adversities children and their families may have experienced. Importantly, receiving an early FASD diagnosis has been found to be an important protective factor for children against potential adverse life outcomes, such as incarceration, drug/alcohol, and mental health problems [14].

The strengths of the current study are that it involved a well-validated diagnostic system, which included a comprehensive multi-disciplinary assessment and provided the first clinic-based outcomes on a sample of Australian children diagnosed with a FASD. However, the total number of children reported in this study is small and patterns may change with a larger sample. Detailed information regarding specific patterns of alcohol consumption during pregnancy was not available as many children were in foster care.

While prevention of FASD remains a vital public health concern, it is imperative that the assessment and diagnosis of FASD is expanded in Australia. The recent release of the Australian Guide to the Diagnosis of FASD [8] may provide the impetus for future development of services. Importantly, the current study demonstrates that the establishment of a multi-disciplinary FASD assessment and diagnostic service can be embedded within an existing Child Development Service, once appropriate training in diagnosis has been obtained.

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Conflict of interest: None to declare

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Table 1.
Growth, facial, CNS outcomes and alcohol/other substance exposure

Characteristic		Frequency
Growth deficiency Rank	Rank 1	24
	Rank 2	4
	Rank 3	3
FAS facial phenotype Rank	Rank 1	9
	Rank 2	15
	Rank 3 or 4	7
Philtrum Smoothness Rank	Rank 1-very deep	<3
	Rank 2- somewhat deep	3
	Rank 3- normal	15
	Rank 4- moderately smooth	13
	Rank 5- completely smooth	<3
Upper Lip Thinness Rank	1-very thick or 2 moderately thick	5
	3- normal	18
	4- moderately thin or 5- very thin	8
Z-scores for Mean Palpebral Fissure Lengths	≤ -2 SD	11
	>-2SD and ≤ -1SD	7
	> -1SD	13
Probability of CNS dysfunction Rank	1 No evidence of damage	<3
	2 - Mild to Moderate	13
	3 Significant or 4 Definite Dysfunction	18
Prenatal alcohol exposure rank	2 –unknown level of exposure	<3
	3- confirmed moderate	14
	4 – confirmed high	15
Source of alcohol information	Biological mother	10
	Person who directly observed mother	8
	Other source (e.g. medical, legal records)	11
Exposure to other substances in-utero	Tobacco	<3
	Cannabis	<3
	Solvents	<3
	Polydrug use (e.g. methamphetamine, Cannabis & Tobacco)	17
	No known other exposure	<3
	Unknown	8

Table 2.
Functional CNS outcomes

Domain	**Level of dysfunction – Frequency**		
	Rank 1 – Normal Range	**Rank 2 –** Mild to Moderate impairment	**Rank 3-** Significant impairment
Soft neurological signs	12	16	3
Cognitive	11	6	14
Communication	11	8	12
Academic	8	9	14
Memory	11	12	8
Executive Functioning	<3	16	15
Attention	<3	12	17
Adaptive, Social behaviour	0	16	15

Note: Ranking based on scores on standardised assessments; Rank 1 = scores falling within 1 Standard deviation (SD) either side of the mean; Rank 2 = Scores > 1 but < 2 SDs below the means; Rank 3 = Scores equal to or > 2 SDs below the mean.

Table 3.
Frequency of Child Behaviour Checklist Outcomes

Scales and Subscales	**Clinical Range**	**Borderline Range**	**Normal Range**
Total	28	0	<3
Internalising Problems	16	6	8
Externalising Problems	27	<3	<3
Anxious/Depressed	8	7	14
Withdrawn/Depressed	7	7	15
Somatic Complaints	6	4	19
Social Problems	16	6	7
Thought Problems	19	<3	9
Attention Problems	19	5	5
Rule-breaking Behaviour	14	6	10
Aggressive Behaviour	17	5	8

Note: Total, Internalising & Externalising: Clinical Range = T-score > 63, Borderline Clinical Range = T-score 60 – 63; Subscales: - Clinical Range = T-score > 69, Borderline Clinical Range = T-score 65 – 69; All scales/subscales N = 30 except Anxious/Depressed, Withdrawn/Depressed, Somatic Complaints, Social Problems, Thought Problems & Attention Problems N = 29; Note there was 1 missing CBCL report and 1 report missing some data that that could not be included.

Supplementary Table 1.
Overview of the 4-Digit Diagnostic Code Criteria

Rank	Growth deficiency	FAS facial phenotype	Probability of CNS damage	Prenatal exposure to alcohol
4	**Significant** Height and weight <3rd %tile	**Severe** All 3 features: PFL 2 or more SDs below mean Thin lip: rank 4 or 5 Smooth Philtrum: rank 4 or 5	**Definite** Structural and/ or neurologic evidence	**High Risk** Confirmed exposure to high levels i.e. high peak blood alcohol concentrations delivered at least weekly in early pregnancy
3	**Moderate** Height or weight < 3rd %tile	**Moderate** 2.5 features	**Probable** Significant dysfunction across 3 or more domains	**Some Risk** Confirmed exposure. Level of alcohol use is less than Rank 4 or level is unknown
2	**Mild** Height and/or weight >3rd and ≤ 10th or Height or Weight > 10th	**Mild** 1-2 features	**Possible** Evidence of dysfunction, but less than rank 3	**Unknown** Exposure not confirmed present or absent
1	**None** Height and weight > 10th percentile	**None** No features	**Unlikely** No structural neurologic or functional abnormalities	**No Risk** Confirmed absence of exposure from conception to birth

Note: PFL = palpebral fissure length; SD = standard deviation; %tile = percentile
Source: Astley, 2004

Supplementary Table 2.
Definitions of CNS Ranks 1 through 4

CNS Rank 4: Structural/Neurological Abnormalities Definite Evidence of CNS damage	This rank is selected when the evidence for CNS damage is defined through a traditional medical approach. At least one significant structural or neurological finding is required. Structural evidence may include, but is not limited to: Microcephaly, defined as occipital frontal circumference 2 or more standard deviations below the mean or Significant brain abnormalities observable through imaging. Neurological evidence may include, but is not limited to: Seizures or other hard neurological signs.
CNS Rank 3: Significant Dysfunction Probable evidence of CNS damage	This rank is based on evidence from standardised psychometric assessments that are administered directly to the individual or obtained from a reliable informant. Significant impairment is defined as performance 2 or more standard deviations below the mean on a standardised test (i.e. below the 3rd percentile). Important to also consider whether there are significant differences (i.e. at least 1SD) between test subdomains within the individual's profile that are unusual or occur rarely in the normative population.
CNS Rank 2: Mild to Moderate Delay/Dysfunction Possible evidence of CNS damage	This rank is also based on evidence from standardised psychometric assessments. Scores > 1SD and < 2SDs below the mean. This rank could also be assigned to individuals who have not yet been able to complete all the testing e.g. they are too young to be tested, typically less than 6 years.
CNS Rank 1: No Current evidence of Delay/dysfunction No current evidence of CNS damage	No functional or developmental problems are identified.

Source: Astley, 2004

Supplementary Table 3.
Overview of brain domains and assessment methods

Brain domain	Assessment tools used
Neurological/Motor	Standard neurological examination conducted by a Paediatrician
Cognitive	Wechsler Intelligence Scale for Children – 4th Edition (WISC-IV)
Communication	Clinical Evaluation of Language Fundamentals – 4th Edition (CELF-4)
Academic	Wechsler Individual Achievement Test – 2nd Edition (WIAT-II)
Memory	NEPSY-2 Developmental Neuropsychological Assessment – 2nd Edition (NEPSY-II)
Executive Function	Behaviour Rating Inventory of Executive Function (BRIEF)
	NEPSY-2 Developmental Neuropsychological Assessment – 2nd Edition (NEPSY-II)
	Test of Problem Solving – Elementary (TOPS 3)
Attention	NEPSY-2 Developmental Neuropsychological Assessment – 2nd Edition (NEPSY-II)
	Child Behaviour Checklist for ages 6 – 18 (CBCL)
	Teacher Report Form for ages 6 – 8 years (TRF)
Adaptive, Social behaviour	Adaptive Behaviour Assessment System – 2nd Edition (ABAS-II)
	Social Language Development Test, Elementary (SLDT-E)
	CELF-4 Pragmatics Profile

Supplementary Table 4.
Terminology used to describe the 22 diagnostic categories of the 4-Digit Diagnostic Code

Sentinel Physical findings	This term is used when an individual presents with growth deficiency at the Rank 3 or 4 level and/or FAS facial phenotype at the Rank 3 or 4 level.
Static encephalopathy	This term is used when an individual presents with significant structural, neurological. And/or functional abnormalities that strongly support the presence of underlying CNS damage at the Rank 3 or 4 level. The term does not define or suggest any specific pattern.
Neurobehavioural disorder	This term is used when an individual presents with cognitive/behavioural dysfunction at the Rank 2 level and no evidence of structural, neurological or functional abnormalities at the Rank 3 or 4 levels.
Alcohol Exposed, Not exposed or Exposure Unknown	These terms are used to reflect prenatal alcohol exposure and its potential risk to the unborn child. Alcohol exposure is reported independently of outcomes and does not imply that a causal association exists between the exposure and the outcomes.
Fetal alcohol syndrome (alcohol exposed)	The term FAS is used to refer to an individual who presents with one of the 12 4-digit diagnostic code combinations reflecting growth deficiency; the full facial phenotype; significant structural, neurological, and/or functional CNS abnormalities; and confirmed prenatal alcohol exposure.
Fetal alcohol syndrome (alcohol exposure unknown)	A diagnosis of FAS can be made when prenatal alcohol exposure is unknown. Six possible codes fall under this category.
Partial fetal alcohol syndrome (alcohol exposed)	This term is used for individuals who present with static encephalopathy, most (but not all) of the growth and/or facial features of FAS, and have a confirmed history of prenatal alcohol exposure. 20 possible codes fall under this category
Fetal alcohol syndrome phenocopy (no alcohol exposure)	This term is used when an individual who meets the growth, face and CNS criteria, but has a confirmed absence of alcohol exposure. This has not yet been observed.

Note: The above terms are used in various combinations to name the 22 diagnostic categories that the 256 possible 4-digit Diagnostic Codes fall under.

Source: Astley, 2004