Misdiagnosis and Missed Diagnoses in Foster and Adopted Children With Prenatal Alcohol Exposure

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abstract

OBJECTIVE: The purpose of this article is to assess the rate of misdiagnosis and missed diagnoses of fetal alcohol spectrum disorders (FASD) among a population of foster and adopted youth referred to a children's mental health center.

METHODS: Data were collected from a sample of 547 children who underwent a comprehensive multidisciplinary diagnostic evaluation. Utilizing current diagnostic criteria, children were diagnosed, as appropriate, with fetal alcohol syndrome, partial fetal alcohol syndrome, alcohol-related neurodevelopmental disorder, or alcohol-related birth defects. Changes in rates of alcohol exposure-related diagnoses and cooccurring mental health disorders pre- and postassessment were analyzed by using McNemar's test for dependent proportions.

RESULTS: Among 156 children and adolescents who met criteria for a diagnosis within the fetal alcohol spectrum, 125 had never been diagnosed as affected by prenatal alcohol exposure, a missed diagnosis rate of 80.1%. Of the 31 who had been recognized before referral as affected by prenatal alcohol exposure, 10 children's FASD diagnoses were changed within the spectrum, representing a misdiagnosis rate of 6.4%. The remaining 21 (13.5%) children's diagnoses stayed the same. There also were significant changes in the rate of mental health diagnosis, and learning disorders, communication disorders, and intellectual disability, objective signs of neurocognitive damage, were not recognized in a significant number of children with FASD.

CONCLUSIONS: Within this clinical sample, 86.5% of youth with FASD had never been previously diagnosed or had been misdiagnosed. These high rates of missed diagnoses and misdiagnosis have significant implications for intervention and therapeutic services.

WHAT'S KNOWN ON THIS SUBJECT: Researchers speculate that children with fetal alcohol spectrum disorders often are not recognized or diagnosed correctly.

WHAT THIS STUDY ADDS: This is the first study to assess the rate of missed diagnoses and misdiagnosis in foster and adopted children with fetal alcohol spectrum disorders.

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In children with a history of prenatal alcohol exposure, the diagnosis of fetal alcohol syndrome (FAS) is based on 3 criteria: prenatal and/or postnatal growth retardation, central nervous system impairment, and characteristic facial dysmorphology.[1–3] However, neurodevelopmental deficits among children who have confirmed prenatal exposure to alcohol but who do not meet diagnostic criteria for FAS are much more common. Partial FAS[4] (pFAS) and alcohol-related neurodevelopmental disorder (ARND)[4] are the most common diagnoses, and alcohol-related birth defects (ARBDs)[4] is relatively rare.[5] In 2004, a group of federal agencies developed a consensus definition of a more comprehensive term, fetal alcohol spectrum disorders (FASD). The term FASD is "an umbrella term describing the range of effects that can occur in an individual whose mother drank during pregnancy."[3]

Unfortunately, many children and adolescents with FASD go unrecognized and untreated[6–8]; this is due to multiple factors, including unknown maternal history of alcohol use during pregnancy,[9,10] lack of consistent facial dysmorphology and growth impairment across all diagnoses within the fetal alcohol spectrum,[3,4,10,11] and the high rate of cooccurring mental health disorders.[12] Within our clinic's population at the Children's Research Triangle (CRT), it was noted that large numbers of children with FASD had been incorrectly diagnosed before referral. The purpose of this article is to assess the rate of misdiagnosis and missed diagnoses of FASD among a population of children and adolescents referred to a children's mental health center for assessment and treatment. We hypothesized that the majority of children with FASD referred to the center would not have been diagnosed with FASD at the time of referral and that, of those that were identified as having an alcohol diagnosis within FASD, a significant number would have been inaccurately diagnosed.

METHODS

The clinic at CRT is a mental health center specializing in the assessment and treatment of high-risk populations of children and adolescents, especially those in the child welfare system. CRT is not an "FAS clinic"; there are no screening criteria for referral to the clinic, but the most common reason for referral is behavioral problems. Almost all of the children are referred through the Illinois Department of Children and Family Services (DCFS), and all children are in the care of a pediatrician or other children's primary health care provider, as required by DCFS. An average of 200 children per year undergo a comprehensive medical, mental health, and neurodevelopmental assessment at the center. Approximately 30% of the children evaluated each year receive a diagnosis within the fetal alcohol spectrum.

Study Inclusion

All foster and adopted children 4 to 18 years of age who have undergone comprehensive evaluation for any reason at CRT were eligible for inclusion in the study. An office clerk and interns who had no knowledge of history or diagnosis of any of the children pulled a sample of 547 charts from ~3000 charts.

Child Assessment

The initial evaluation for each child consisted of a full pediatric, neurologic, and dysmorphology examination conducted by 1 of 2 board-certified pediatricians with extensive experience in diagnosing, assessing, and treating children with FASD. Each child's prenatal alcohol exposure (yes/no) was verified through documentation in the child's birth, medical, child welfare, and/or adoption records. In addition, maternal use of tobacco and illicit drugs as documented through maternal admission of use or positive toxicology for the mother or newborn was recorded. Information regarding dosage and frequency of maternal alcohol, tobacco, and/or illicit drug use was not available for most children.

Before 2003, the pediatricians assessed the child's facial features based on published dysmorphic abnormalities consistent with FAS.[1,3] After 2003, a digital facial photograph of each child was taken following the guidelines established by Astley and Clarren,[13] and measurements of palpebral fissure length and intercanthal distance were calculated via the photograph by using the recommended formulae. The philtrum and lip Ranks (Ranks 1 through 5) were assigned by the pediatrician during the examination based on the established grading system[13] and were confirmed through computer-generated upper lip circularity calculations. After the medical examination, the child and family underwent a clinical interview with a licensed psychologist, and the child was evaluated under the direction of a doctoral level psychologist utilizing instruments that assess child psychological and neurodevelopmental functioning across several domains.

Measures

The neurodevelopmental battery with which each child was evaluated included age appropriate instruments and approaches that assessed neurocognitive functioning, including general intelligence, memory, executive functioning, and speech and language; academic achievement; self regulation, including sensory processing, social skills, and behavior; and adaptive behaviors.[14–16]

Diagnostic Assignment

Based upon the completed comprehensive evaluation, children were assigned an alcohol exposure-

related diagnosis that is consistent with the University of Washington's 4-digit code system[10] for diagnosis:

- Growth retardation: current or past weight and/or height less than third percentile adjusted for age and gender. We tightened the growth criteria from 10th percentile, as recommended in the Centers for Disease Control and Prevention diagnostic and referral guidelines,[3] to third percentile, as recommended by Astley,[10] because of the high rates of diagnostic misclassification.[2,17,18] Recent studies have documented that use of third percentile for growth assessment neurodevelopmentally differentiates children within the fetal alcohol spectrum.[10,15] Also, growth criteria below third percentile align more closely with the original definition of FAS.[1]
- Facial dysmorphology: abnormal measurements of the upper lip (rank 4 or 5) and the philtrum (rank 4 or 5) and shortened palpebral fissures based on direct measurement or, after 2003, according to analysis of facial features utilizing the Lip-Philtrum Guide and digital facial photograph based on the criteria of Astley and Clarren.[13,19]
- Central nervous system abnormalities: demonstration of structural, neurologic, or functional central nervous system deficits[20] as documented by the presence of microcephaly (current head circumference below third percentile for age and gender) and/or functional deficits demonstrated as global cognitive delays with performance below the third percentile on standardized testing or 3 or more domains of neurodevelopmental functioning more than 2 SDs below the normed mean on standardized measures of neurocognitive, self-regulatory, or adaptive functioning.

Based on these standards, children who met all physical criteria for growth impairment and facial dysmorphology as well as neurodevelopmental deficits were assigned a diagnosis of FAS. Children with confirmed prenatal alcohol exposure, facial dysmorphology, and neurodevelopmental deficits but with normal growth (height and weight) patterns were diagnosed as pFAS. Children with confirmed exposure, normal growth and neurodevelopmental functioning but major structural abnormalities were diagnosed as ARBDs. Children who had confirmed exposure and met criteria for neurodevelopmental deficits but did not meet criteria for facial dysmorphology and/or growth were classified as ARND. This approach is consistent with the newly published *Diagnostic and Statistical Manual of Mental Disorders, Fifth Edition* mental health criteria for neurobehavioral disorder with prenatal alcohol exposure (ND-PAE),[21] which is the terminology that will replace the term ARND.

Data Analytic Approach

Frequency tables and case summaries were generated for each of the hypothesized questions by using SPSS 21.0 (IBM SPSS Statistics, IBM Corporation). Pre- and postassessment proportions of diagnoses were analyzed by using McNemar's test for dependent proportions, again using SPSS 21.0. This statistical test is used on paired nominal data, which are contained in a 2×2 table in studies that have a before and after component.[22]

The Institutional Review Board of DCFS and the Western Institutional Review Board approved all procedures for this archival study.

RESULTS

Descriptive Data

All children in the sample were in a foster or adoptive home at the time of referral and evaluation. Among the 547 children included in the study, the mean age was 9.36 years (SD = 3.93), and 63.8% were boys. Racial (50.6% African American, 1.3% Asian, 32.2% white, 0.7% Native American, 12.2% biracial, 0.2% other, 2.8% unknown) and ethnic distribution (11.7% Hispanic, 82.8% non-Hispanic, 5.9% unknown) reflected the general distribution of children under DCFS supervision.

Referral Diagnoses

By far, the most common reason for referral of the 547 children to the CRT clinic was "behavioral problems." At referral, diagnoses related to prenatal alcohol exposure were relatively rare, with 36 children (6.6%) being referred with a diagnosis of FAS and 15 children (2.7%) being referred with a diagnosis of ARND. The most common mental health diagnosis at the time of referral (Table 1) was attention-deficit/hyperactivity disorder (ADHD), followed by posttraumatic stress disorder, conduct disorder, oppositional defiant disorder, and reactive attachment disorder.

Diagnoses After Evaluation

After the comprehensive multidisciplinary evaluation at CRT and using the diagnostic criteria presented in the Methods section, 156 children (28.5%) met criteria for a diagnosis within FASD: 93 with FAS, 1 with pFAS, 61 with ARND, and 1 with ARBD. In this way, the sample was representative of our overall clinic population in that ~30% of the children evaluated at CRT meet criteria for a diagnosis within FASD. The subset of children with FASD was similar as to mean age and distribution by gender and race/ethnicity to the referral sample of children. Mental health diagnoses assigned after comprehensive assessment changed significantly for the 156 children with FASD (Table 2) and demonstrated a wide range of disorders.

TABLE 1 Changes in Mental Health Diagnoses of Children With FASD After Assessment ($N = 156$)

Diagnosis	Referral Diagnosis		Postassessment Diagnosis		P	Effect Size
	N	%	N	%		Cramer's ν
ADHD	42	26.4	88	55.3	<.001	0.05
Adjustment disorder	3	1.9	20	12.6	.001	0.05
Anxiety disorder	3	1.9	15	9.4	.004	0.11
Autism/pervasive developmental disorder	0	0.0	8	5.0	—	
Bipolar Cyc	3	1.9	3	1.9	.999	0.32
Bipolar NOS	6	3.7	3	1.9	.453	0.22
Communication disorder	6	3.7	20	12.5	.007	0.02
Depression NOS	5	3.2	12	7.7	.065	0.36
Developmental disorder	2	1.3	8	5.0	.109	0.03
Developmental delay	8	5.0	5	3.1	.508	0.29
Learning disability	7	4.4	23	14.5	.002	0.08
Mental retardation	7	4.4	24	15	<.001	0.34
Mood disorder NOS	3	1.9	6	3.8	.508	0.03
Oppositional defiant disorder	8	5.0	4	2.5	.344	0.15
Psychotic disorder	2	1.3	5	3.1	.375	0.30
Posttraumatic stress disorder	10	6.3	26	16.3	.001	0.38
Reactive attachment disorder	9	5.6	14	8.8	.383	0.02
Sensory integration disorder	6	3.8	25	16.0	<.001	0.28
Sleep disorder	1	0.6	7	4.4	.031	0.37
Other diagnosis	7	4.4	20	12.6	.011	0.10

Em dash denotes the following: Because there were no children with this diagnosis at referral, McNemar's test could not be calculated. NOS, not otherwise specified.

Changes in Diagnosis Pre- and Postassessment

Among the 547 children, 51 were referred with a diagnosis within the fetal alcohol spectrum; of these children, 20 did not meet criteria for any diagnosis within FASD, and 31 children retained an alcohol exposure-related diagnosis, a significant reduction ($P < .001$, Cramer's $\nu = 0.73$) in the number of children with a diagnosis related to FASD. Specific diagnoses within FASD also demonstrated a high rate of error. Of the 36 children who had been diagnosed with FAS, only 16 of these children met criteria for FAS after their full assessment, 7 met criteria for ARND, and 13 qualified for no alcohol-related diagnosis. Of 15 children with a referral diagnosis of ARND, 5 retained a diagnosis of ARND after comprehensive assessment, 2 received a diagnosis of FAS, and 8 received no alcohol-related diagnoses.

Of the 156 children who after full assessment received a diagnosis within FASD, only 31 (19.9%) had been referred to the clinic with a diagnosis related to prenatal alcohol exposure; 80.1% of children with FASD had not been recognized.

After comprehensive assessment by the multidisciplinary clinical team, there was a significant change in mental health diagnoses for the 156 children with FASD (Table 1). Of note, learning disorders, communication disorders, and intellectual disability, objective signs of significant neurocognitive damage, had not been recognized in a large majority of the children with these disabilities. Among the 156 children with confirmed FASD, 147 (94.2%) received a cooccurring mental health diagnosis, with 104 (66.7%) having 2 or more mental health diagnoses in addition to the alcohol exposure-related diagnosis.

TABLE 2 Changes in Therapeutic Services for Children With FASD After Assessment ($N = 156$)

Therapeutic Service	Referral		Postassessment		P	Cramer's ν
	N	%	N	%		
Occupational therapy	40	25.7	2	1.3	<.001	0.08
Physical therapy	19	11.9	4	2.6	<.001	0.06
Developmental therapy	19	11.9	0	0	—	
Speech/language	45	28.8	16	10.2	<.001	0.09
Family therapy	15	9.5	25	15.7	.002	0.13
Sensory integration	18	11.3	76	48.7	<.001	0.21
Psychotherapy	54	34.6	42	26.9	.008	0.02

Em dash denotes the following: Because there were no children receiving this therapy post assessment, McNemar's test could not be calculated.

Changes in Treatment Pre- and Postassessment

The multidisciplinary assessment led to a significant change in therapeutic approaches for the 156 children with FASD (Table 2). After the evaluation, significantly fewer children required the developmental therapy, physical therapy, and speech/language therapy that they had been receiving and instead needed services, especially family therapy, sensory integration treatment, and psychotherapy, that they previously had not been receiving (Table 2). In addition to the therapeutic modalities presented in Table 2, after assessment, a number of children with FASD required further medical interventions; 27 children (17.3%) required extensive dental work and 8 (5.1%) children needed an ophthalmology evaluation. Also, attachment therapy was recommended for 33 children (21.2%), and educational services were recommended for 109 (69.9%) of the children with FASD.

Recommendations for medication use also changed from the time of referral to the time after diagnosis of FASD. At the time of referral, 11 of the 156 children and youth were on stimulant medications to treat ADHD. After assessment, stimulant medications were recommended to only 1 of these individuals. Twenty-two other children and youth with FASD who

had not presented on medications were prescribed stimulant medications for ADHD after assessment.

Of the 156 children and youth with FASD, before referral 8 had been prescribed psychotropic medications. Of those 8 youth, 1 child's prescription stayed the same and 7 children were taken off psychotropic medication. Six additional youth with FASD were placed on psychotropic medications. Eighteen of the youth came into the clinic with other medication prescriptions, of which 14 were taken off their medication and 4 maintained their medication. Eleven additional youth were placed on new medication.

DISCUSSION

Within this clinical sample, the higher number of children with FAS as opposed to pFAS or ARND most likely is due to the fact that foster and adopted children with severe behavioral disorders frequently are referred to CRT's clinic. However, even with this severity of behavioral problems, 86.5% of children and adolescents with FASD had never been previously diagnosed or had been misdiagnosed. The majority of these youth (80.1%) had a missed diagnosis, whereas the remaining 6.4% of youth had a misdiagnosis (ie, their diagnosis within the FASD spectrum was changed). These findings suggest that FASD frequently go unrecognized; thus, education is most needed in overall awareness of FASD, with additional emphasis on differential diagnosis within the spectrum. This is especially pertinent as ND-PAE replaces ARND as a diagnostic term. In the current study, all 155 children diagnosed as having FAS, pFAS, and ARND met newly published criteria for ND-PAE.[21]

There are several barriers to early recognition and accurate diagnosis of children and adolescents with FASD. The frequent lack of clear physical findings in children affected by alcohol exposure,[3,5,10,11] the historically confusing language and diagnostic terminology applied to alcohol-affected children,[23] and the perceived stigma against addressing alcohol use by pregnant women[24] most likely contributed to the majority of affected children and adolescents in the current study having been misdiagnosed or missed completely. A survey of American Academy of Pediatrics members indicated that only 50% of respondents felt prepared to make a diagnosis within the fetal alcohol spectrum.[24] Further, children's health providers do not routinely consider prenatal alcohol exposure in the differential diagnosis of behavioral and learning problems.[25]

In the current study, ADHD was the most common referral diagnosis for children who ultimately were diagnosed with FASD. Previous studies have demonstrated that anywhere from 40% to 75% of children with FASD are diagnosed with ADHD.[15,26,27] However, there are qualitative differences in the types of attention problems seen in children with FASD as compared with children with ADHD.[28–30] Children with ADHD and FASD have been shown to have greater deficits in verbal comprehension and perceptual reasoning than children having ADHD without prenatal alcohol exposure.[28] Another study of children with FASD compared with children with ADHD revealed that children with FASD were more likely than the ADHD group to engage in sociopathic behaviors, such as lying and stealing.[29] Greenbaum et al[30] found that children with FASD demonstrate a behavioral profile distinct from children with ADHD, especially related to difficulties in social cognition and emotion processing. These differences and the lack of recognition of FASD have significant implications for the pharmacologic and therapeutic approach to treating the child, since studies have demonstrated differential response to medication for children with FASD.[31,32]

The cooccurring mental health disorders in individuals affected by prenatal alcohol exposure have implications for therapy.[33,34] In the current study, the majority of children and youth ultimately diagnosed with FASD required significant alteration of therapeutic services. The therapies most commonly delivered by early intervention and school systems (developmental, speech/language, occupational, and physical therapies) were the therapies the children with FASD were most frequently receiving. However, upon receiving a comprehensive evaluation, significant numbers of the children and youth did not need these therapies but required more intense forms of mental health therapy addressing attachment difficulties, behavioral difficulties, and sensory processing deficits, as well as the need for the child and family to participate in some form of psychotherapy. In addition, the need for specialized educational services in the school and dental care often had been overlooked for the children who ultimately were diagnosed with FASD.

This study has limitations in that the target population consists of a foster or adopted population referred for a behavioral or mental health assessment. Thus, conclusions may not be generalized to the general population of children with FASD. In addition, it is important to note that some of the mental health diagnoses and the recommended changes to therapeutic approaches may have been due to the child's aging and corresponding evolution of symptoms. Further, children's varying presentations at the assessment session, incomplete access to records, changes in reporting by parents and other auxiliary informants, and foundational differences in training

among professionals (psychologists, psychiatrists, and pediatricians) may also influence the interpretation of diagnostic criteria. However, issues such as intellectual disabilities, communication disorders, and learning disorders (problems that are relatively common among individuals with FASD) do not change over time, are less subjective than many of the mental health diagnoses, and provide concrete evidence of neurologic damage, but were not recognized in a number of the children.

Once children and adolescents with FASD are recognized, there must be an immediate effort to obtain diagnostic and therapeutic services. Early diagnosis, especially before the age of 6 years, coupled with earliest intervention is 1 of the strongest correlates with an improved outcome for the child long-term.[35] Delayed or incorrect diagnosis, especially among children who do not have the sentinel facial dysmorphology associated with FAS, may lead to a higher incidence of secondary disabilities[36] and greater need for special education services.[37] The role of the pediatrician and other children's health care providers is clear: early recognition of the child or adolescent with FASD, referral to a provider who can conduct a full evaluation, and participation in the development of a targeted treatment plan that incorporates mental health treatment, behavioral management strategies, and special education services.

CONCLUSIONS

Although FASD have long been recognized as a leading cause of intellectual disabilities, behavior problems, learning disabilities, and cooccurring mental health disorders,[1,2,5,9,12–15,20,31,32] children and adolescents who have been affected by prenatal alcohol exposure often go undiagnosed or are misdiagnosed. Pediatricians and other children's health care providers have the opportunity to screen children and youth in their practices for FASD and ensure that affected individuals receive the targeted range of services they may need.

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