

Estimating Prevalence of Fetal Alcohol Syndrome (FAS): Effectiveness of a Passive Birth Defects Registry System

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BACKGROUND: Fetal alcohol syndrome (FAS) is a preventable birth defect, easiest to recognize in children two through eleven years and more difficult to recognize in newborns. In New York State, two systems ascertain FAS cases, the statewide birth defects registry and the Fetal Alcohol Syndrome Surveillance Network (FASSNet) system. The accuracy of FAS reports to the birth defects registry was assessed through a comparison with the FASSNet system. **METHODS:** The birth defects registry mandates reporting up to age two, including FAS with an ICD-9 code of 760.71. FASSNet is a population-based, multi-source surveillance and uses a standard definition to determine FAS case status. **RESULTS:** Among 33 children reported to the registry with FAS, 19 (58%) met FASSNet criteria for FAS. FASSNet identified 24 additional children with FAS facial features documented before the child's second birthday that should have been reported to the birth defects registry. FAS prevalence rate for the birth defects registry was 0.28 per 1,000 live births but would have been 0.37 if all children diagnosed before age two were included. **CONCLUSIONS:** Almost 60% of children reported to a birth defects registry with FAS from 1995 to 1998 were confirmed as FAS based on a more intensive surveillance. Additional children with FAS were not reported to the CMR. FAS prevalence calculated from birth defects registries, relying on the ICD-9 code 760.71, include false positives and underestimate the true prevalence. Age limits for reporting FAS to registries further contribute to under ascertainment. *Birth Defects Research (Part A) 67:604–608, 2003.* © 2003 Wiley-Liss, Inc.

INTRODUCTION

Fetal alcohol syndrome (FAS), caused by heavy maternal drinking during pregnancy, has been described as the most preventable birth defect (Abel, '90). At the community, state, and national level, various programs and strategies are being implemented to prevent FAS, ranging from targeted interventions for high-risk women or subgroups to universal public awareness campaigns to promote health and well being of all women (Hankin, '02; Stratton et al., '96). While tracking the prevalence of FAS poses unique problems (Cordero et al., '94; Hymbaugh et al., '02), it is important that successful surveillance systems be in place to evaluate the effectiveness of a growing number of prevention efforts.

Deriving the prevalence of FAS is difficult because there are no pathognomonic features; expression of the phenotype is heterogeneous among cases and varies at different ages within a case (Clarren, '81; Streissguth et al., '91). FAS is diagnosed using a combination of findings including growth deficiency (pre- or postnatal), central nervous system (CNS) abnormalities, facial dysmorphism and maternal alcohol use during pregnancy (Aase, '94; Stratton et al., '96). For accurate diagnosis, recognition of the pattern of anomalies is important and recognition of FAS facial features is key (Abel et al., '93; Jones, '03; Stoler and Holmes, '99). Experienced diagnosticians are more accurate in detecting the syndrome (Jones, '99; Stoler and Holmes, '99). FAS is more easily recognized in children ages two through eleven and is more difficult to diagnose in newborns (Aase, '94; Abel et al., '93; Jones, '99; Jones, '03;

Larkby and Day, '97; Little et al., '90; Stratton et al., '96; Streissguth et al., '91; Stoler and Holmes, '99).

In addition to diagnostic issues, surveillance for FAS presents challenges because of variation with case finding methods and study population characteristics (CDC, '02b; Cordero et al., '94; Hymbaugh et al., '02; May and Gossage, '01). Researchers have generally used three different approaches to estimate FAS prevalence: clinic-based studies, population-based active case ascertainment, and passive surveillance. Each method has differing strengths and weaknesses (May and Gossage, '01; Stratton et al., '96). These studies have produced baseline estimates of FAS prevalence in the range of 0.2 to 2.0 per 1,000 live births (Abel, '95; Abel and Sokol, '91; CDC, '93a; CDC, '93b; CDC, '95; CDC, '97; CDC, '02b).

Many states report prevalences of FAS using data from their birth defects surveillance system (NBDPN, '02). The purpose of this analysis was to estimate completeness and validity of a birth defects passive surveillance system, using as the criterion or "gold standard", a system specifically developed to ascertain FAS. The passive system was a statewide birth defects registry with mandated reporting of children with birth defects including FAS. The FAS surveillance system was a population-based, multi-source system where records of children with FAS or with known

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or suspected prenatal exposure to alcohol were actively ascertained and abstracted (Hymbaugh et al., '02).

METHODS

New York State Congenital Malformations Registry

The New York State (NYS) Congenital Malformations Registry (CMR) in the Department of Health (DOH) is one of the largest statewide, population-based birth defects registries in the nation. Hospitals and physicians are mandated to report children born in NYS and diagnosed with birth defects, including FAS, up to the age of two. While the CMR requires reporting of the narrative diagnosis, the *International Classification of Diseases, Ninth Revision, Clinical Modification* (ICD-9-CM) (World Health Organization, '89) of 760.71 ("noxious influences affecting fetus or newborn via placenta or breast milk, specifically alcohol; including fetal alcohol syndrome") is generally what hospitals use to identify children with FAS to report to the CMR. A description of the CMR has been published previously along with an evaluation of the surveillance system (Druschel, '01; Sekhobo and Druschel, '01).

Fetal Alcohol Syndrome Surveillance Network

Since 1997, New York has been part of the Fetal Alcohol Syndrome Surveillance Network (FASSNet) established with funding from the Centers for Disease Control and Prevention (CDC) (Hymbaugh et al., '02). Four states (Alaska, Arizona, Colorado, and New York) have worked cooperatively with the CDC to either establish or enhance a population-based FAS surveillance system using a multiple-source methodology. Children born in 1995 or later, with known or suspected fetal alcohol exposure, are identified from diagnostic and service programs in nine western counties of New York. These sources include the CMR, genetics clinics, developmental disability clinics, hospitals (through hospital discharge codes), early intervention programs, private provider and special high-risk clinics, and vital records. Information on a single child can be obtained from several sources. With current FASSNet sources, younger rather than older children are more likely to be identified.

FAS case status is determined using a standard definition developed by FASSNet, which is based on the Institute of Medicine's criteria developed in 1996 (Stratton et al., '96). For FASSNet, a child is considered to have the FAS facies if two of three specific facial features (short palpebral fissures, long/smooth philtrum and thin upper lip) are noted (CDC, '02b; Hymbaugh et al., '02). A case is considered "confirmed" if facial criteria are met with documentation of both CNS and growth deficiency. A "probable" case status is assigned with facial criteria and either CNS or growth deficiency. Cases can be confirmed or probable with or without documentation of maternal alcohol use during pregnancy. For our analysis here, a child with a FASSNet case status of confirmed or probable was considered to have FAS by the FASSNet method. Hymbaugh et al. ('02) describes the methods used by FASSNet, including the multiple-source methodology, surveillance case definition, data collection variables, and the record abstraction process. Prevalence rates obtained by this surveillance system have been published for birth years 1995 through 1997 (CDC, '02b).

Estimating Validity of a Passive Birth Defects Surveillance System

Children born during 1995 through 1998 residing in the FASSNet surveillance area and reported to the CMR as having FAS were identified. These children were matched with children in the FASSNet database to determine the FASSNet case status. In addition, children with FAS were identified in the FASSNet database that had not been reported to the CMR. The FASSNet system contained diagnostic information on each child, including the actual diagnoses, name and specialty of diagnostician, child's age at diagnosis, and information on FAS facial features, including age of child when FAS facies was documented. For this analysis, we defined an expert as a dysmorphologist, clinical geneticist, or developmental pediatrician. As the CMR includes only children diagnosed to age two years, only FASSNet cases with FAS facial features documented before the child's second birthday were included in the analysis.

RESULTS

CMR Cases

In the nine counties of the New York FASSNet, 33 children born during 1995–1998 were reported to the CMR with FAS (Fig. 1). Nineteen (58%) of these children also had FAS by the FASSNet method; all had documentation in the medical records of characteristic FAS facies during the first 30 days of life and 12 (63%) were identified within the first five days of life. Of the 19, 16 (84%) were examined by an expert who felt the child had FAS.

The remaining 14 CMR cases (42%) that did not meet the FASSNet criteria for FAS had no documentation of the characteristic FAS facies; eight (57%) were examined by an expert, with five not thought to be FAS and three for whom there was insufficient documentation.

FAS Cases Not Reported to the Birth Defects

Through FASSNet, 24 additional children with documentation of FAS facial features before age two were found who were not reported to the CMR (Fig. 2). For 11 of these (46%), FAS facies was documented within the first 30 days of life. Of the 24 children, 23 (96%) were examined by an expert and 20 of these were considered to be FAS.

Prevalence, Sensitivity and Predictive Value Positive

The FAS prevalence rate for births in 1995 through 1998, diagnosed before age two years, derived from the CMR was 0.28 per 1,000 live births; through FASSNet, it was 0.37 per 1,000 births. The FASSNet prevalence was 25% higher than the CMR-derived prevalence.

Using the FASSNet surveillance approach as the "gold standard", sensitivity and predictive value positive (PVP) were derived. Sensitivity was defined here as the proportion of FAS cases identified by the CMR. Of all children with documented FAS facial features before age two, the sensitivity of the CMR was 0.44 (19/(19+24)) (Fig. 3). The PVP represents the proportion of children identified with FAS by the CMR that actually have FAS. The predictive value positive for the CMR was 0.58 (19/(19+14)) (Fig. 3).

DISCUSSION

The purpose of this analysis was to examine the ability of a state birth defects registry to document the prevalence

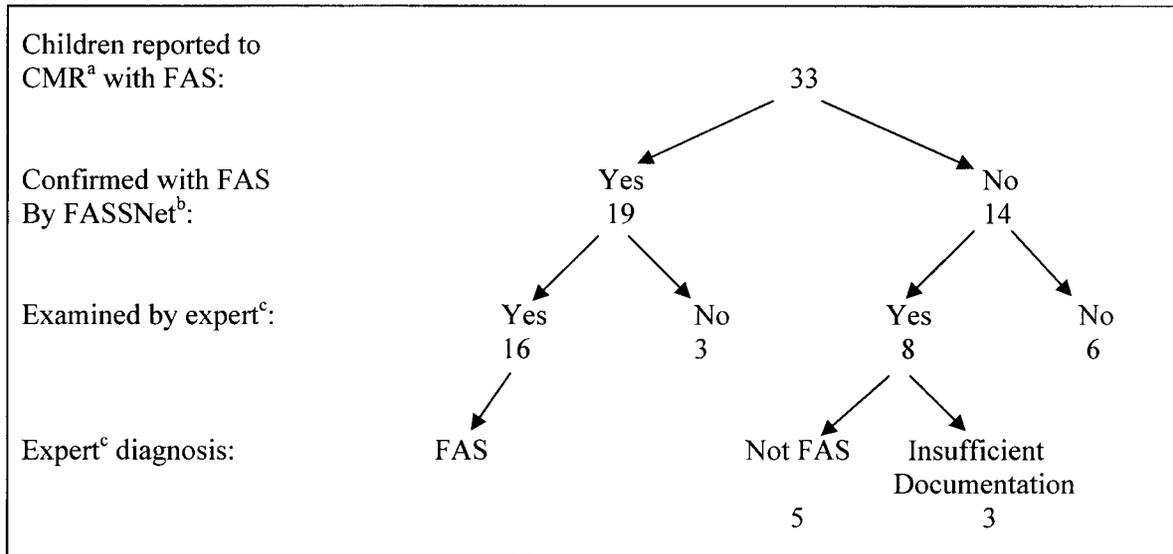


Figure 1. Characteristics of western NY children reported to the CMR^a with fetal alcohol syndrome (FAS)

^aCongenital Malformations Registry (CMR), the New York State birth defects registry

^bFetal Alcohol Syndrome Surveillance Network (FASSNet)

^cAn expert is defined as a dysmorphologist, clinical geneticist or developmental pediatrician.

of FAS by comparing it with a system specifically designed for FAS surveillance (FASSNet). Almost 60% of children (19 of 33) born in Western New York from 1995 to 1998, and reported to the NY Congenital Malformations Registry (CMR) with FAS, were found to have FAS by FASSNet methods. FASSNet identified 24 additional children with FAS that had FAS facial features documented prior to their second birthday; 11 of these had FAS facies documented in the first month after birth.

Surveillance of FAS based on newborn diagnosis or focused on diagnosis of FAS at younger ages would result in

under ascertainment of cases and lower sensitivity (Abel et al., '93; Jones, '99; Jones, '03; Larkby and Day, '97; Stoler and Holmes, '99). Although physicians are included in the mandated reporting to the NY CMR, in actuality, over 95% of reports come from hospitals. All of the CMR cases that were confirmed with FAS by FASSNet had FAS facial features evident within the first 30 days, many within five days. Unless a child with FAS was diagnosed in the newborn period, an FAS diagnosis, even before age two years, would most likely not be reported, because a diagnosis of FAS in itself would not lead to hospitalization. Many of the

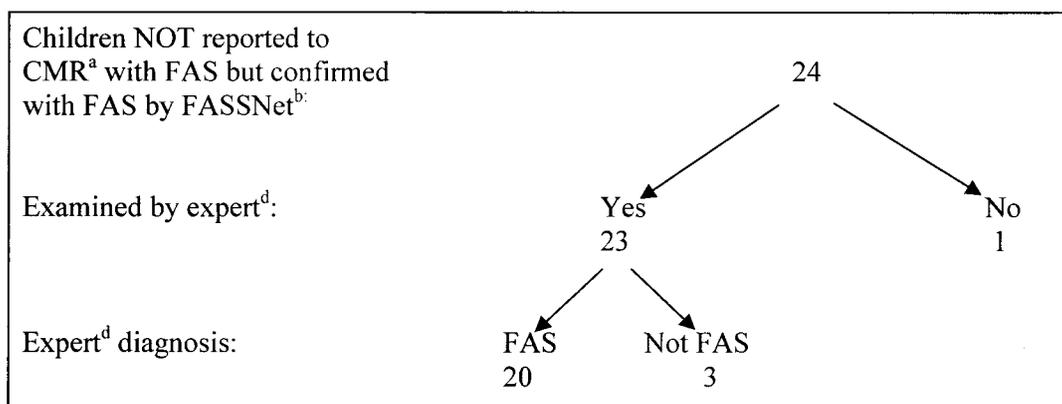


Figure 2. Characteristics of western NY children NOT reported to the CMR^a with fetal alcohol syndrome (FAS) identified through FASSNet^b with FAS facies^c documented before age two years

^aCongenital Malformations Registry (CMR), the New York State birth defects registry

^bFetal Alcohol Syndrome Surveillance Network (FASSNet)

^cA child is considered to have the FAS facies if two of three specific facial features (short palpebral fissures, long/smooth philtrum and thin upper lip) are noted.

^dAn expert is defined as a dysmorphologist, clinical geneticist or developmental pediatrician.

		Children with FAS by FASSNet ^c Method		Total
		Yes	No	
Children reported to CMR ^b with FAS	Yes	19	14	33
	No	24		
Total		43		

Figure 3. 2 × 2 table of children with fetal alcohol syndrome (FAS) facial characteristics^a documented by age two years
^aA child is considered to have FAS facial characteristics if two of three specific facial features (short palpebral fissures, long/smooth philtrum and thin upper lip) are noted.
^bCongenital Malformations Registry (CMR), the New York State birth defects registry
^cFetal Alcohol Syndrome Surveillance Network (FASSNet)

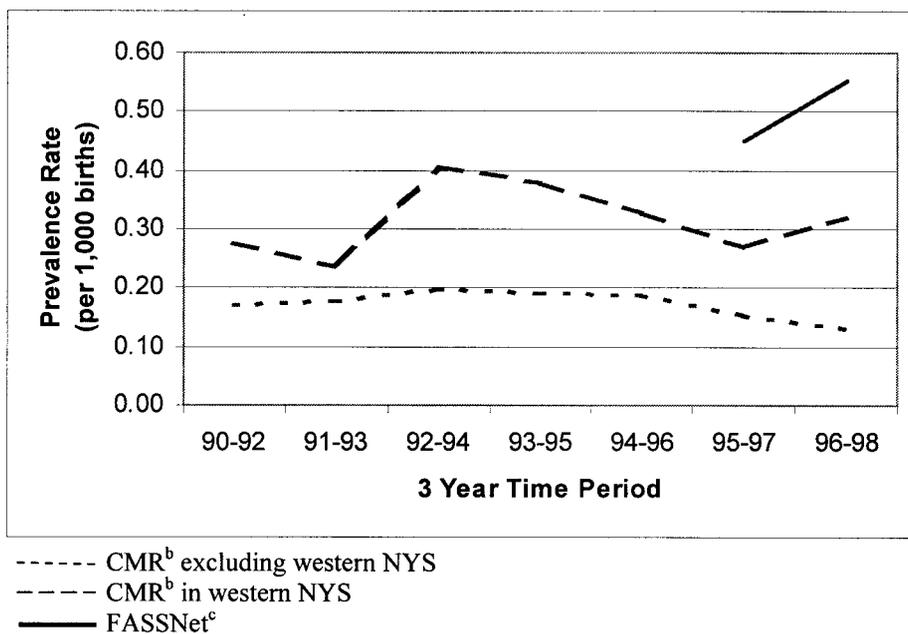
24 FASSNet children not in the CMR were most likely missed because facial features were identified after newborn discharge. The sensitivity of the CMR for FAS was 44% (19/19+24) based on diagnosis by age two years. This value, representing the proportion of FAS cases identified, was comparable to the 38% reported by Miller et al. ('95) for hospital discharge data where cases were identified by the ICD-9 code of 760.71. However, FASSNet also identified 18 additional children with FAS facies documented after their second birthday. If all children with FAS regardless of age at diagnosis (i.e., FASSNet cases) are included, the sensitivity of the CMR dropped to 31% (19/19+42) as 30% were diagnosed after their second birthday. For children diagnosed after their second birthday, it is possible

that the FAS facies would not have been recognizable before age two, even by an expert.

The FASSNet prevalence of children diagnosed at any age was 0.52 per 1,000 births, compared to 0.37 for FASSNet and 0.28 for the CMR for diagnosis by age two years. However, FASSNet prevalences may still be low as that system is biased towards detection of younger children. Loudenburg et al. ('02) reported that the average age at time of diagnosis for children diagnosed with FAS was 6.5 years and Clarren et al. ('01), conducting a population-based study of first graders, reported a prevalence rate of 3.1 per 1,000 students. All FASSNet children, however, were diagnosed before age six. This may change over time with continued follow up. As children become older and have difficulty in schools, for example, they may be referred to one of the FASSNet sources.

Reliance by birth defects registries on the non-specific ICD-9 code 760.71 to estimate FAS prevalence is problematic and may be the most significant reason that 42% of the CMR reports (14 of 33) were false positives. The code 760.71 can be assigned to a child with prenatal alcohol exposure without requiring the child be examined (Miller et al., '95). Assignment is subjective and influenced by other factors. While passive registries generally report higher prevalences of FAS than active ones (NBDNP, '02), possibly because active registries have more information and exclude cases without FAS documentation, they are still lower than those reported through clinic-based or multiple source surveillance methods. A recent study (CDC, '02a) reported that 13% of pregnant women in 1999 consumed alcohol during pregnancy. If all alcohol-exposed pregnancies were assigned 760.71, we would expect a much higher prevalence. Cases reported to a birth defects registry with 760.71 may instead represent a subset of children with problems at birth (e.g., growth deficiency, small head circumference, preterm birth and/or complications,

Figure 4. Fetal alcohol syndrome (FAS) prevalence rates^a with different methods in New York State (NYS), reported as three-year moving averages^a per 1,000 live births
^bCongenital Malformations Registry (CMR), the New York State birth defects registry
^cFetal Alcohol Syndrome Surveillance Network (FASSNet)



other birth defects, etc.) whose mothers were known to use or abuse alcohol (Abel and Sokol, '91).

Interest and awareness may also affect reporting (Little et al., '90). Within New York State, for example, we have regional differences of FAS reporting to the CMR. For the NYS CMR excluding the western region, the average annual prevalence for FAS from 1990 to 1998 was 0.16 per 1,000 live births, whereas for western NY, the prevalence was 0.32 per 1,000 live births (Fig. 4). The higher prevalence reported in western NY might be attributed to more interest in FAS by several diagnosticians combined with greater accuracy in diagnosis and assignment of 760.71. FAS reports to the CMR from western NY may more likely be FAS which would explain the favorable positive predictive value (PVP) of 58% for the CMR in western New York compared to the 27% reported by Miller et al. ('95) for hospital discharge data.

Many states have active or passive birth defects registries that, if effective, would be useful to estimate FAS trends and monitor prevention efforts nationally. The majority of birth defects registries require reporting before the child's first or second birthday, an appropriate window for most structural, functional, or biochemical abnormalities that registries were originally designed to detect, but an inappropriate window for conditions such as autism, mental retardation, and FAS. Our analysis of the NY CMR suggests that birth defect registries are not effective tools to monitor FAS rates. Diagnosis of FAS, unlike more easily recognizable birth defects, is complex, and is more difficult to identify at particular ages and especially at birth. Birth defects registries have been popular as tools to monitor FAS because of their cost-savings benefits as they rely on existing record collection systems. To effectively monitor FAS and prevention efforts, additional funds would be necessary to develop or maintain surveillance activities specifically focusing on FAS.

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