Epidemiology of Congenital Diaphragmatic Hernia, Hawaii, 1987-1996

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Congenital diaphragmatic hernias (CDHs) in Hawaii between 1987 and 1996 were examined with data from a birth defects surveillance system. There were 51 cases of CDH (prevalence 2.45 per 10,000 births). Forty-nine percent of livebirths survived an increase over the rate reported in Hawaii in 1975-1982. These results are similar to those reported by other population-based studies.

Introduction

Congenital diaphragmatic hernia (CDH), a birth defect in which an incompletely developed diaphragm allows the abdominal organs to herniate into the chest, occurs in approximately 3-4 per 10,000 births.1-3 Although the defect can be repaired surgically, the mortality rate from CDH is still high.1-3 One study has estimated that the treatment and care of each individual with CDH would cost approximately $250,000 in direct and indirect costs over his or her lifetime.4

Little is known about the etiology of CDH. Epidemiologic studies have noted an association between CDH and chromosomal abnormalities, and that a high proportion of CDH cases also have other structural defects.1-3,5,7 One study reported a non-significant increase in CDH prevalence over time, and greater prevalence of the defect among males, but no differences by ethnic group, maternal age, or parity.2

CDH can be detected in-utero by ultrasonography,8 and the detection of serious birth defects in pregnancy has led to the elective termination of affected pregnancies.3,5 A number of the birth defect surveillance systems currently in operation were created prior to the common use of prenatal screens and diagnostic tests. Many of these systems do not include in their ascertainment elective pregnancy terminations following the prenatal diagnosis of birth defects. As a consequence, most of these systems cannot evaluate the impact of prenatal diagnosis and elective termination on CDH rates. Several studies have reported from 1 to 21 percent of prenatally diagnosed and 1 to 5 percent of all CDH pregnancies are electively terminated.2,3,5

In this study, data collected by the Hawaii Birth Defects Program (HBDP) was used to examine the CDH mortality rate over a 10-year period (1987-1996) in Hawaii. It investigates the effect of demographic factors such as maternal ethnicity and age on the prevalence of the defect and the proportion of cases with chromosomal and other structural defects. The study also evaluates the impact of prenatal diagnosis and elective termination on the prevalence of CDH. Because the HBDP has nearly universal access to prenatal diagnostic information and follow-up data on elective terminations, Hawaii is an ideal setting in which to study their effects on the prevalence rates of birth defects.

Methods

Data were from the Hawaii Birth Defects Program (HBDP), a population-based, active surveillance system for birth defects and other adverse reproductive/pregnancy outcomes for the entire state of Hawaii. All pregnancies are included in the HBDP regardless of outcome (livebirth, fetal demise, or elective termination) or the gestational age of the fetus at the end of the pregnancy. In order to be included in the registry: 1) the pregnancy must be affected by one or more moderate to severe birth defects or other adverse conditions such as antenatal maternal substance abuse, neoplasms, and congenital infections; 2) the end of the pregnancy must occur in Hawaii; and 3) the diagnosis must be made prenatally or within one year after delivery.

Affected pregnancies were identified by examining lists of medical record diagnostic codes and other reports provided by hospitals where births and terminations due to fetal anomalies occur, tertiary care facilities, and clinics and laboratories that perform prenatal diagnostic screening, testing, or follow-up counseling. In addition to clinical information about each reported infant or fetus, the HBDP collected data on demographic characteristics, health behaviors, and

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The analysis included ten years of data collected from 1987 through 1996 on all reported cases with congenital absence or hernia of the diaphragm. Cases with a probable or possible diagnosis which could not be confirmed were excluded from the analysis, as were cases of eversion of the diaphragm. The pregnancy outcome, year of birth, place of residence, sex, plurality, side of defect, and maternal age and race/ethnicity for each case were determined. Ethnicity was grouped as white, Far East Asian (Japanese, Chinese, and Korean), Pacific Islander, and Filipino. Cases belonging to other races/ethnicities (black, Hispanic, etc.) were excluded from the race/ethnicity analysis because of their relatively small numbers in Hawaii’s birth population. The presence of chromosomal abnormality, or other major structural anomalies were identified. Diagnoses of hypoplastic lung, malrotation, and defects associated with prematurity (patent ductus arteriosus, patent foramen ovale) were excluded in the analysis of additional anomalies.

The various demographic factors were examined and compared with livebirth and stillbirth data provided by the Hawaii State Department of Health/Office of Health Status Monitoring, as derived from birth and fetal death certificates. Prenatal diagnosis, elective termination, and survival rates were calculated. Ninety-five percent confidence intervals were calculated by Poisson probability. Secular trends were analyzed by the Chi-square tests for trend.

### Results

The study identified 51 confirmed cases of congenital diaphragmatic hernia (CDH) in Hawaii between 1987 and 1996 for a prevalence of 2.45 per 10,000 births (95% confidence interval CI 1.82-3.22). All were the posterolateral type of diaphragmatic hernia. Fifteen (36.1%) of the cases occurred in the City and County of Honolulu, with similar numbers in the rest of the state (35.3%). The prevalence was highest in Hawaiian ethnicity (49.2%), followed by Filipinos, Far East Asians, and Pacific Islanders. Ethnicities such as black, Hispanic, etc. were excluded from the analysis because of their relatively small numbers in Hawaii’s birth population. The prevalence of CDH in Hawaii between 1987 and 1996 was calculated to be 2.45 per 10,000 births. This is lower than the prevalence reported in other states: Iowa (2.7), California (3.3), and Utah (3.9).

Of the 45 liveborn CDH cases, 22 were known to be alive at one year of age, giving a survival rate of 48.9 percent. None of the 7 cases which had been prenatally diagnosed and not electively terminated survived to one year, while 57.9 percent of the cases not prenatally diagnosed survived. Fifteen (53.6 percent) of the liveborn cases with isolated CDH survived, while only 7 (41.2 percent) of those with other congenital abnormalities survived.

### Table 1.—Prenatal diagnosis, pregnancy outcome, and survival rate (for at least one year) of congenital diaphragmatic hernia by year, Hawaii, 1987-1996.

<table>
<thead>
<tr>
<th>Year</th>
<th>Total rate*</th>
<th>Prenatally diagnosed No.</th>
<th>Elective terminations No.</th>
<th>Survival rate %</th>
</tr>
</thead>
<tbody>
<tr>
<td>1987</td>
<td>1.99</td>
<td>1</td>
<td>0</td>
<td>25</td>
</tr>
<tr>
<td>1988</td>
<td>2.92</td>
<td>0</td>
<td>0</td>
<td>67</td>
</tr>
<tr>
<td>1989</td>
<td>4.77</td>
<td>2</td>
<td>1</td>
<td>44</td>
</tr>
<tr>
<td>1990</td>
<td>2.69</td>
<td>1</td>
<td>1</td>
<td>80</td>
</tr>
<tr>
<td>1991</td>
<td>2.30</td>
<td>1</td>
<td>0</td>
<td>50</td>
</tr>
<tr>
<td>1992</td>
<td>2.33</td>
<td>1</td>
<td>0</td>
<td>50</td>
</tr>
<tr>
<td>1993</td>
<td>2.37</td>
<td>1</td>
<td>0</td>
<td>50</td>
</tr>
<tr>
<td>1994</td>
<td>0.96</td>
<td>1</td>
<td>1</td>
<td>100</td>
</tr>
<tr>
<td>1995</td>
<td>2.53</td>
<td>2</td>
<td>1</td>
<td>25</td>
</tr>
<tr>
<td>1996</td>
<td>1.55</td>
<td>0</td>
<td>0</td>
<td>67</td>
</tr>
<tr>
<td>Total</td>
<td>2.45</td>
<td>12</td>
<td>5</td>
<td>49</td>
</tr>
</tbody>
</table>

*per 10,000 births

### Discussion

The prevalence of CDH in Hawaii between 1987 and 1996 was calculated to be 2.45 per 10,000 births. This is lower than the prevalence reported in other states: Iowa (2.7), California (3.3), and Utah (3.9). Only the Iowa study did not include elective terminations in its analysis, so differences in pregnancy outcomes cannot account for the prevalence differences between the studies.

Since this study identified a slight decline, albeit not statistically significant, in the prevalence over the ten-year period of study, and several of the other studies covered time periods preceding that of the current study, it could be suggested that the CDH prevalence in Hawaii was higher in the past and thus similar to that observed in the past.
the gestation when identifying the defect would have been even more difficult. The prenatal diagnosis of CDHs demonstrated a slight increase during the ten-year period of study, so the relatively poor record of prenatal diagnosis of CDH in Hawaii may be improving.

Ten percent of all CDH cases in Hawaii (42 percent of prenatally diagnosed cases) were electively terminated, a much higher proportion than that reported by California (0.8 percent) and Utah (5 percent). Access to prenatal screens and tests has been found to vary by geographical area. However, this could only serve as an explanation for the difference in termination proportions between Hawaii and California. As noted previously, a greater proportion of CDHs were prenatally diagnosed in Utah than in Hawaii. More likely the differences represent variations in attitudes towards and access to elective terminations between the states. This hypothesis is possibly supported by the observation that the percentages of prenatally-diagnosed CDHs which were electively terminated varied widely in North America: Utah (10 percent), Alabama (21 percent), United States and Canada (1 percent).

Maternal race was not found to impact a woman's risk for having a pregnancy affected by CDH, a finding which supports that observed in California. The prevalence of CDH in Metropolitan Honolulu was similar to that for the rest of the state. While the CDH rate was roughly the same in three of the four counties in the state (City and County of Honolulu, Maui County, Kauai County), the rate in Hawaii County was less than one-fifth that of the others. Also, women less than age 20 were approximately twice as likely to have a pregnancy affected by CDH than any other age group. The lack of statistical significance for this observation precludes offering any potential explanation for these differences.

Forty-nine percent of the livebirths with CDH in Hawaii between 1987 and 1996 survived to age one year, a rate compatible with that observed in other states: California (44 percent) and Utah (60 percent). This is also greater than the 33 percent survival rate observed in Hawaii during 1975-1982, suggesting that either the treatment and care of neonates with CDH has improved between the two time periods or cases less likely to survive are now being prenatally diagnosed and electively terminated. A slight decline in survival rate was identified over the ten-year period studied. This would appear to contradict the first hypothesis except that this trend was not statistically significant.

The survival rate has been reported to be lower for CDHs prenatally diagnosed than those detected after delivery, something observed in the current study where none of the livebirths prenatally diagnosed with CDH survived, while 58 percent of those detected after birth survived. Those cases which were prenatally diagnosed and electively terminated would likely suffer a similar higher risk for mortality if they had been allowed to continue. This would tend to support the second hypothesis.

Other studies had found that the survival rate for cases of CDH with other congenital defects was lower than that for isolated defects. In the current study, 54 percent of isolated CDHs survived while 41 percent of those with other defects survived. Thus, if CDHs with additional congenital defects were more likely to be electively terminated, then the second hypothesis would be supported. However, the proportions of electively terminated CDHs and total CDHs with other defects were similar.
Most likely both hypotheses contribute to some extent to the improvement in survival of livebirths with CDH in Hawaii between 1975-1982 and 1987-1996.

The primary limitation with the current study was the small number of CDH cases included. This limited the statistical power of the analyses performed and left any differences and trends observed to be merely suggestive. One way to increase the number of cases, and thus the statistical power of the analyses, would be to extend the time period covered. For logistic reasons, the HBDP cannot consistently collect data prior to 1986, so future years would be needed for additional data. Due to the low prevalence of CDH, another analysis would have to wait for a number of years.

Also, the survival rate for CDH may be overestimated. The HBDP receives information on all deaths of infants less than age one year which occur in the state from the DOH. The DOH also receives information on deaths to state residents which occur in other states, but it is not known if all out-of-state deaths, particularly among infants, are reported to the DOH. Thus if an infant with CDH is transferred to another state for treatment and dies in the other state, this information may not reach the DOH, and through them the HBDP. However, this underestimation is believed to be low. Due to the serious nature of the defect, many infants with the defect expire shortly after delivery. And the critical nature of the condition may make the time necessary to transport the infant across the Pacific Ocean unrealistic and treatment of the CDH in Hawaii a preferable option.

In conclusion, the prevalence of CDH in Hawaii between 1987 and 1996 was found to be slightly lower than that of other population-based studies performed in other states. With the possible exception of county of residence and maternal age, demographic factors did not appear to affect the risk for CDH in the state. A higher proportion of cases were electively terminated than that observed by other studies. Prenatal diagnosis of CDH and the presence of additional congenital defects reduced the chances of survival for an infant with CDH. For several possible reasons, the survival rate for CDH has improved over the last two decades.

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