Prevalence of Birth Defects

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PUBLIC HEALTH IMPORTANCE

The prevalence of birth defects varies considerably with respect to type of defect, time, place, and other demographic, genetic, and environmental factors. In this chapter, we describe the prevalence of birth defects as determined by two CDC surveillance systems, the Birth Defects Monitoring Program (BDMP) and the Metropolitan Atlanta Congenital Defects Program (MACDP). For additional information about related topics and surveillance activities, see the State Use of Birth Defects Surveillance, Infant Mortality, Neonatal and Postneonatal Mortality, and Fetal Alcohol Syndrome chapters.

HISTORY OF DATA COLLECTION

In the early 1950s, the fact that rubella can cause birth defects became clear. A decade later came the discovery that maternal use of thalidomide had caused an epidemic of limb reduction deformities. Thus, in the 1960s, the realization emerged that infectious and other environmental factors could cause birth defects, and this realization resulted in the establishment of birth defects surveillance programs in a number of countries.

CDC was an early participant in this surveillance activity, starting the MACDP in 1967 and the BDMP in 1974. The New York State Health Department also began an early surveillance program, based on birth certificates. In 1974, CDC and representatives from nine other surveillance programs, primarily from Europe, formed the International Clearinghouse for Birth Defects Monitoring Systems (ICBDMS). Today, the ICBDMS comprises 24 programs. Many of these programs are based in Europe, and some programs are from Australia, China, New Zealand, and Japan. Over the past decade, several state health departments have begun their own birth defects surveillance systems (these state-based activities are described in detail in the State Use of Birth Defects Surveillance chapter).

CDC SURVEILLANCE ACTIVITIES

CDC's two systems for assessing the prevalence of birth defects—the BDMP and the MACDP—are both overseen by CDC's National Center for Environmental Health (NCEH) (*1*).

The BDMP, a national program to monitor congenital malformations, uses hospital discharge data on newborns gathered by the Commission on Professional and Hospital Activities (CPHA), based in Ann Arbor, Michigan. Data from this system cover both live-born and stillborn infants in participating member hospitals from 1970 to the present. The database includes information on >17 million births occurring in 1200 predominately midsized community hospitals across the United States. The system covers approximately 405,000 births annually—>10% of all births occurring in the nation—although the coverage proportion varies considerably by state. Because participation is voluntary, the sampling is not random; thus, the degree of representativeness is an issue to be considered in interpreting the data. The data are derived from newborn discharge information provided to CPHA by participating member hospitals. CPHA processes these data, conducting range

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and consistency edit checks for input accuracy. Diagnoses made for readmissions are not included, because to do so could introduce duplicate counting of infants. Semiannually, CPHA provides CDC with data tapes that include the following information: state and county of birth occurrence, year and month of birth, live-born/stillborn status, race, sex, birth weight, up to 31 *International Classification of Diseases, Ninth Revision, Clinical Modification (ICD-9-CM)* procedure codes, and up to 31 ICD-9-CM diagnostic codes (*2*).

Because the BDMP is a surveillance system with passive case ascertainment based on hospital discharge summaries of newborns, the proportion of cases it detects depends on the severity of the specific defect; less severe defects can be overlooked in the newborn period, whereas more severe defects are more likely to result in prompt and accurate diagnoses. An additional problem is the declining number of participating hospitals. CDC researchers are investigating new avenues for national birth defects surveillance, including collaboration among state birth defects monitoring programs.

The MACDP is one of the oldest birth defects surveillance systems in the country (*1*). This population-based birth defects surveillance system was founded by the Georgia Mental Health Institute, Emory University School of Medicine, and CDC. Day-to-day program operations are the responsibility of NCEH.

The MACDP monitors all births—approximately 38,000 births a year—occurring in the five-county metropolitan Atlanta area. The program collects information on all stillborn and live-born infants diagnosed with at least one major birth defect within the first year of life, with diagnoses ascertained within the first 5 years of life.

The MACDP has served as a prototype for numerous birth defects surveillance systems. MACDP researchers have encouraged the development of uniform methods of birth defects surveillance, developed a more defect-specific coding system and a uniform set of variables for data collection, and provided a focus for collaborative studies between surveillance systems with active case ascertainment.

MACDP researchers gather data using an in-house coding form (Figure 1). They use the precise diagnosis and written description of defects collected and classified according to the six-digit MACDP code, which permits improved classification of birth defects and improves researchers' ability to study specific types of malformations. Case ascertainment includes a review of maternal and infant medical records in multiple sources, including birth hospitals, pediatric referral hospitals, cytogenetic laboratories, specialty clinics, and vital statistics from the Georgia Department of Human Resources. Multiple sources of ascertainment are used to identify potential cases. Hospital records reviewed include obstetric, nursery, pediatric, surgery, autopsy, and laboratory logs as well as cardiac catheterization records and disease indexes. MACDP staff review charts of all infants who are stillborn, die shortly after birth, weigh <2,500 g, or are born before 37 weeks of gestational age. Similar data from pediatric referral hospitals are reviewed as are laboratory service records. In addition, birth and death certificates are reviewed to search for previously unidentified cases.

MACDP case records include basic demographic information (identification of the case infant, case mother, and case father as well as the infant's race, sex, plurality, live-born/stillborn status, date of birth, birth weight, hospital of birth, and date of first diagnosis), laboratory examination results, specific written diagnoses, six-digit MACDP codes, cytogenetic data, complications of birth, prenatal data, pregnancy history, family history, and other birth-related and risk factor information.

These data are computer processed in monthly batches that undergo a variety of edit checks. From 1968 to the present, the MACDP has ascertained the occurrence of birth defects for approximately 725,000 births. MACDP staff monitor birth defects rates and trends by conducting quarterly reviews and analysis of data, and they make temporal and geographic comparisons to search for significant changes in birth defects rates.

GENERAL FINDINGS

In this chapter, we focus on the prevalence of a selected set of 26 birth defects reported through

FIGURE 1.

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES
Public Health Service
Centers for Disease Control and Prevention (CDC)
Atlanta, Georgia 30093

REPRODUCTIVE OUTCOMES CASE RECORD

FORM APPROVED
OMB No. 0920-0010
EXP. DATE 12/92

(1-) ROCR

STATE (5-) 11	**I.D. No.** (7-) ___ ___ ___ ___ ___ ___	**INFORMATION RECORDED:** Mo Da Yr **INITIALS** (13-) ___ ___ ___ **DATE** (16-) ___ ___ - ___ ___ - ___ ___ **HOSP.** (22-) ___ ___ ___ ___

PATIENT NAME: (26-) LAST FIRST MIDDLE	**MOTHER'S NAME** (50-) LAST FIRST (MAIDEN)	**AGE AT BIRTH** (74-) __ ___
RESIDENCE AT BIRTH (76-)	**FATHER'S NAME:** (108-) LAST FIRST MIDDLE	**AGE AT BIRTH** (132-) ___ ___

CITY (134-)	**COUNTY** (150-) ___ ___ ___	**ZIP** (153-) ___ ___ ___ ___ ___	**CENSUS TRACT** (158-) ___ ___ ___ ___ ___ ___	**HOME PHONE** (164-) ___ ___ ___ / ___ ___ ___ - ___ ___ ___ ___

MOTHER'S BIRTH DATE (174-) ___ ___ - ___ ___ - ___ ___ (MDY)	**MOTHER'S SSN** (180-) ___ ___ ___ - ___ ___ - ___ ___ ___ ___	**FATHER'S BIRTH DATE** (189-) ___ ___ - ___ ___ - ___ ___ (MDY)	**FATHER'S SSN** (195-) ___ ___ ___ - ___ ___ - ___ ___ ___ ___

MOTHER'S RACIAL OR ETHNIC GROUP (204)
☐ 1 WHITE, NOT HISP ☐ 3 HISPANIC ☐ 5 ASIAN OR PACIFIC ISLANDER
☐ 2 BLACK, NOT HISP ☐ 4 AMERICAN INDIAN OR ALASKAN NATIVE ☐ 9 NOT STATED

PENDING (206) ☐ 1 YES ☐ 2 NO

SEX (214)
☐ 1 MALE ☐ 3 AMBIGUOUS
☐ 2 FEMALE ☐ 9 NOT STATED

PLURALITY (215)
☐ 1 SINGLE ☐ 3 OTHER MULTIPLE BIRTH
☐ 2 TWIN ☐ 9 NOT STATED

OUTCOME OF DELIVERY (216)
☐ 1 LIVE BORN ☐ 3 INDUCED AB
☐ 2 STILLBORN ☐ 9 NOT STATED

CO-TWIN SEX (217)
☐ 1 MALE ☐ 3 AMBIGUOUS
☐ 2 FEMALE ☐ 9 NOT STATED

CO-TWIN CONCORDANCE (218)
☐ 1 CO-TWIN NORMAL ☐ 3 CO-TWIN WITH OTHER DEFECT
☐ 2 CO-TWIN WITH SAME DEFECT ☐ 9 NOT STATED

CO-TWIN LB/SB (219)
☐ 1 CO-TWIN LB ☐ 9 NOT STATED
☐ 2 CO-TWIN STILL BORN

APGAR SCORE 1 MIN (220-)___ ___ 5 MIN (222-)___ ___

DATE OF BIRTH Mo Da Yr (224-)___ ___ - ___ ___ - ___ ___

BIRTH WEIGHT
(2300)___ ___ ___ ___ GRAMS
OR
(234-) ___ ___ LBS. ___ ___ OZS.

HOSPITAL OR PLACE OF FIRST DIAGNOSIS (238-)___ ___ ___ ___

DATE OF FIRST DIAGNOSIS Mo Da Yr (242-) ___ ___ - ___ ___ - ___ ___

HEAD CIRCUMFERENCE (251) ☐ 1 CM
(248-) ___ ___ . ___ ☐ 2 IN

LENGTH (255) ☐ 1 CM
(252-)___ ___ . ___ ☐ 2 IN

MOTHER'S HEMATOCRIT (256-) ___ ___

DX CODE	DIAGNOSIS
(258-) ___ ___ ___ - ___ ___ ___	
(264-) ___ ___ ___ - ___ ___ ___	
(270-) ___ ___ ___ - ___ ___ ___	
(276-) ___ ___ ___ - ___ ___ ___	
(282-) ___ ___ ___ - ___ ___ ___	
(288-) ___ ___ ___ - ___ ___ ___	

GEST. AGE BY NEONATAL EXAM (294-) ___ ___ WKS.

DUBOWITZ EXAM (296)
☐ 1 YES ☐ 3 NOT APPLICABLE
☐ 2 NO ☐ 9 NOT STATED

ULTRASOUND DATE Mo Da Yr (297-) ___ ___ - ___ ___ - ___ ___

ULTRASOUND DATING (303-) ___ ___ WKS.

DATE OF Mo Da Yr
LMP (305-) ___ ___ - ___ ___ - ___ ___
EDC (311-) ___ ___ - ___ ___ - ___ ___

SYNDROME (317-) ___ - ___ ___ ___ ___ ___

CYTOGENETICS: (323)
☐ 1 NORMAL ☐ 4 NOT DONE
☐ 2 ABNORMAL ☐ 9 NOT STATED
☐ 3 PENDING

LABORATORY (324-) ___ ___ ___ ___

DIAGNOSIS (328-) ___ - ___ ___ ___ ___ ___

TO BE INTERVIEWED (334)
☐ 1 YES ☐ 2 NO

ACTION CODE (335)
☐ 1 ORIG. ☐ 3 CORR.
☐ 2 CONT. ☐ 4 DELE.

CDC 84.1A REV. 11-92 (SEE REVERSE)

The Centers for Disease Control is authorized to collect this information, including the Social Security number (if applicable), under provisions of the Public Health Service Act, Section 301 (42 U.S.C. 241). Supplying the information is voluntary, and there is no penalty for not providing it. The data will be used to increase understanding of disease patterns, develop prevention and control programs, and communicate new knowledge to the health community. Data will become part of CDC Privacy Act system 09-20-0136, "Epidemiologic Studies and Surveillance of Disease Problems" and may be disclosed to appropriate State or local public health departments and cooperating medical authorities to deal with conditions of public health significance; to private contractors assisting CDC in analyzing and refining records; to researchers under certain limited circumstances to conduct further investigations; to organizations to carry out audits and reviews on behalf of HHS; to the Department of Justice for litigation purposes, and to a congressional office assisting individuals in obtaining their records. An accounting of such disclosures that have been made by CDC will be made available to the subject individual upon request. Except for these and other permissible disclosures expressly authorized by the Privacy Act, no other disclosure may be made without the subject individual's written consent.

FIGURE 1.—continued

PRENATAL DX TEST (336)
☐ 1 DONE ☐ 2 NOT DONE ☐ 9 NOT STATED

TYPE TEST (337-) ___ ___

DATE (339-) Mo Da Yr ___ ___ - ___ ___ - ___ ___

PLACE (345-) ___ ___ ___ ___

COMPLICATIONS OF BIRTH

	YES 1	No 2	NS 9	
(349)	☐	☐	☐	RLF
(350)	☐	☐	☐	TORCH
(351)	☐	☐	☐	NEONATAL SEPSIS/MENINGITIS
(352)	☐	☐	☐	NEONATAL SEIZURES

EXPIRED (362)
☐ 1 YES ☐ 2 NO ☐ 9 NOT STATED

DATE OF DEATH Mo Da Yr
(363) ___ ___ - ___ ___ - ___ ___

PLACE OF DEATH
(369-) ___ ___ ___ ___

AUTOPSY
(373) ★❶1 YES, REVIEWED ★❶3 YES, PENDING
★❶2 NO ★❶9 NOT STATED

Mo Da Yr
DATE (374-) ___ ___ - ___ ___ - ___ ___
PLACE (380-) ___ ___ ___ ___

TOTAL NUMBER OF PREVIOUS PREGNANCIES (Present not included) (384-) ___ ___

NUMBER OF LIVE BIRTHS (386-) ___ ___

NUMBER OF STILL BIRTHS (388-) ___ ___

NUMBER OF INDUCED AB (390-) ___ ___

NUMBER OF SPONTANEOUS AB (392-) ___ ___

NUMBER OF UNSPECIFIED TYPE AB (394) ___ ___

HOSPITAL OF BIRTH
(395-) ___ ___ ___ ___

CHART NUMBER: MOTHER
(399-) ___ ___ ___ ___ ___ ___ ___ ___ ___ ___

CHART NUMBER: INFANT
(409-) ___ ___ ___ ___ ___ ___ ___ ___ ___ ___

HOSPITAL OF SECOND ADMISSION
(419-) ___ ___ ___ ___

CHART NUMBER: INFANT
(423-) ___ ___ ___ ___ ___ ___ ___ ___ ___ ___

(433) ☐ 1 READMISSION ☐ 2 TRANSFER

SECOND ADMISSION DATE Mo Da Yr
(434-) ___ ___ - ___ ___ - ___ ___

HOSPITAL OF LABOR
(440-) ___ ___ ___ ___

CHART NUMBER: MOTHER
(444-) ___ ___ ___ ___ ___ ___ ___ ___ ___ ___

LABOR ADMISSION DATE Mo Da Yr
(454-) ___ ___ - ___ ___ - ___ ___

NAMES OF PHYSICIANS (LAST FIRST INIT.)

OBSTETRICIANS _____ (460-) _____ (490-)

PEDIATRICIANS _____ (519-) _____ (543-) _____ (567-)

OTHERS _____ (591-) _____ (615-) _____ (639-)

PRESENT PREGNANCY INFORMATION (663-)

PREVIOUS PREGNANCY AND MEDICAL HISTORY (743-)

FAMILY HISTORY (823-)

INFANT HISTORY, OTHER INFORMATION (903-962)

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the BDMP and 25 birth defects reported through the MACDP (Rh hemolytic disease is not reported through the MACDP) (Tables 1 and 2). These defects were chosen to reflect a variety of organ systems and the wide range of occurrence rates for individual birth defects.

Many of the overall birth defects rates mask important temporal trends, as is evident in the BDMP prevalence rates for 1970–1971 and 1990–1991 as well as the mean annual percent change in rates between these two periods (Table 1). MACDP data for 1968–1970 and 1989–1991 reveal important trends (Table 2).

BDMP data indicate that the four birth defects with the largest mean annual percentage declines in 1970–1991 were anencephalus, spina bifida without anencephalus, anophthalmos/microphthalmos, and Rh hemolytic disease. According to the MACDP, the four birth defects with the largest declines in 1968–1991 were anencephalus, spina bifida without anencephalus, hydrocephalus without spina bifida, and clubfoot without central nervous system (CNS) defects. Both reporting systems found that the two central nervous system defects, anencephalus and spina bifida without anencephalus, declined substantially; anencephalus declined the most, averaging approximately 7% per year, whereas spina bifida declined a mean of 3%–5% per year. BDMP data reveal that the prevalence of combined anophthalmos and microphthalmos declined an average of 1.8% per year from 1970 to 1991, but virtually all of the decrease occurred before 1975. According to the BDMP, Rh hemolytic disease of the newborn declined on average approximately 6% per year between 1970 and 1991 (Table 1), and almost all of the decline occurred before 1980. MACDP data indicate that the prevalence of hydrocephalus declined a mean of 2.6% and the prevalence of clubfoot without CNS defects fell a mean 3.5% per year (Table 2).

The four birth defects with the largest increases in prevalence in 1970–1991 were endocardial cushion defect, patent ductus arteriosus, pulmonary artery anomaly, and lung agenesis and hypoplasia, according to BDMP data. In comparison, MACDP findings indicate that the four birth defects with the largest increases in prevalence in 1968–1991 were atrial septal defect, endocardial cushion defect, patent ductus arteriosus, and pulmonary artery anomaly. Three of these birth defects are common to both reporting systems: endocardial cushion defect, patent ductus arteriosus, and pulmonary artery anomaly. Atrial septal defect, another cardiovascular defect, was among the four birth defects with the largest increases, according to MACDP data, and it also increased by a substantial 8.9% according to the BDMP findings. These data clearly show that birth defects with the largest increases in prevalence over these two periods are concentrated in the cardiovascular organ system (Tables 1 and 2).

TABLE 1. Reported prevalence of selected birth defects and mean annual percentage change in prevalence — Birth Defects Monitoring Program, 1970–1991*

Birth defect	Rate			Mean annual percentage change
	1970–1991	1970–1971	1990–1991	
CNS				
Anencephalus	3.6	5.48	1.19	-7.4
Spina bifida without anencephalus	5.4	7.55	4.31	-2.8
Hydrocephalus without spina bifida	4.9	4.81	5.01	0.2
Encephalocele	1.2	1.20	0.88	-1.5
Eye				
Anophthalmos/microphthalmos	0.7	0.97	0.67	-1.8
Congenital cataract	0.8	0.64	1.09	2.7

TABLE 1. Reported prevalence of selected birth defects and mean annual percentage change in prevalence — Birth Defects Monitoring Program, 1970–1991* — continued

Birth defect	Rate			Mean annual percentage change
	1970–1991	1970–1971	1990–1991	
Cardiovascular				
Common truncus	0.3	0.28	0.40	1.8
Transposition of great arteries	1.1	0.76	2.23	5.5
Tetralogy of Fallot	1.0	0.57	2.49	7.7
Ventricular septal defect	12.1	4.45	23.78	8.7
Atrial septal defect	2.5	1.91	10.48	8.9
Endocardial cushion defect	0.5	0.08	1.40	15.4
Patent ductus arteriosus	20.2	3.96	52.10	13.8
Coarctation of aorta	0.7	0.42	1.46	6.4
Pulmonary artery anomaly	1.3	0.38	3.52	11.8
Respiratory				
Lung agenesis and hypoplasia	1.9	0.17	3.71	16.7
Orofacial				
Cleft palate without cleft lip	5.2	5.05	5.32	0.3
Cleft lip with or without cleft palate	9.1	9.91	8.54	-0.7
Gastrointestinal				
Tracheoesophageal anomalies	1.9	1.67	2.60	2.2
Rectal and intestinal atresia	3.5	3.75	3.72	-0.0
Genitourinary				
Renal agenesis and dysgenesis	1.4	0.71	2.54	6.6
Bladder exstrophy	0.3	0.35	0.29	-0.9
Musculoskeletal				
Clubfoot without CNS defects	25.5	27.49	23.85	-0.7
Limb reduction deformity	3.5	3.16	3.69	0.8
Chromosomal				
Down's syndrome	8.3	8.17	9.93	1.0
Other				
Rh hemolytic disease	20.6	42.28	12.01	-6.1
Number of births	17,736,971	1,730,257	816,496	

*Rates per 10,000 total births.

TABLE 2. Reported prevalence of selected birth defects and mean annual percentage change in prevalence — Metropolitan Atlanta Congenital Defects Program, 1968–1991*

Birth defect	Rate			Mean annual percentage change
	1968–1991	1968–1970	1989–1991	
CNS				
Anencephalus	5.0	9.69	2.26	-6.7
Spina bifida without anencephalus	7.2	11.96	4.26	-4.8
Hydrocephalus without spina bifida	8.2	10.05	5.73	-2.6
Encephalocele	1.9	1.56	1.22	-1.2
Eye				
Anophthalmos/microphthalmos	3.4	2.39	3.04	1.2
Congenital cataract	2.1	0.72	1.74	4.3
Cardiovascular				
Common truncus	0.8	0.48	0.78	2.3
Transposition of great arteries	4.3	3.47	3.91	0.6
Tetralogy of Fallot	3.4	2.51	4.34	2.6
Ventricular septal defect	21.1	12.08	26.15	3.7
Atrial septal defect	19.4	5.26	41.53	10.3
Endocardial cushion defect	3.0	1.56	4.00	4.6
Patent ductus arteriosus	44.6	10.89	39.79	6.4
Coarctation of aorta	3.9	3.47	4.69	1.4
Pulmonary artery anomaly	5.1	1.44	9.21	9.2
Respiratory				
Lung agenesis and hypoplasia	5.1	2.63	4.69	2.8
Orofacial				
Cleft palate without cleft lip	5.4	3.95	4.78	0.9
Cleft lip with or without cleft palate	10.4	10.53	9.12	-0.7
Gastrointestinal				
Tracheoesophageal anomalies	2.2	2.03	1.74	-0.7
Rectal and intestinal atresia	4.0	4.78	3.91	-1.0
Genitourinary				
Renal agenesis and dysgenesis	3.3	2.27	3.65	2.3
Bladder exstrophy	0.3	0.24	0.17	-1.6
Musculoskeletal				
Clubfoot without CNS defects	27.7	32.78	14.33	-3.5
Limb reduction deformity	5.5	7.54	4.60	-2.3

TABLE 2. Reported prevalence of selected birth defects and mean annual percentage change in prevalence — Metropolitan Atlanta Congenital Defects Program, 1968–1991* — continued

Birth defect	Rate			Mean annual percentage change
	1968–1991	1968–1970	1989–1991	
Chromosomal				
Down's syndrome	10.0	8.85	10.95	1.0
Number of births	696,057	83,599	115,105	

*Rates per 10,000 live births.

According to the BDMP, the prevalence of lung agenesis and hypoplasia rose 6.6% per year on average.

Geographic differences in the prevalence of birth defects were evaluated by using data for 1970–1987. Because of the rarity of these conditions, the data had to be smoothed by aggregating groups of counties. The groups were aggregated by superimposing a grid of squares—each representing approximately 40 miles per side—over a U.S. map (Figures 2 and 3). Data from counties whose population centers fell within the same square were combined, resulting in greater stability of prevalence estimates. After indirect adjustment for the year of birth and race, the observed and expected numbers of cases within each square were compared for statistically significant differences under the Poisson assumption. The two birth defects with the most striking geographic clustering were anencephalus and spina bifida without anencephalus, both of which tended to occur more frequently in the eastern part of the country in a band roughly corresponding with the Appalachian mountain region. The clustering of high-prevalence squares in this area was particularly striking for spina bifida without anencephalus. Concomitantly, most of the significantly low-prevalence squares for these birth defects were located in the western states.

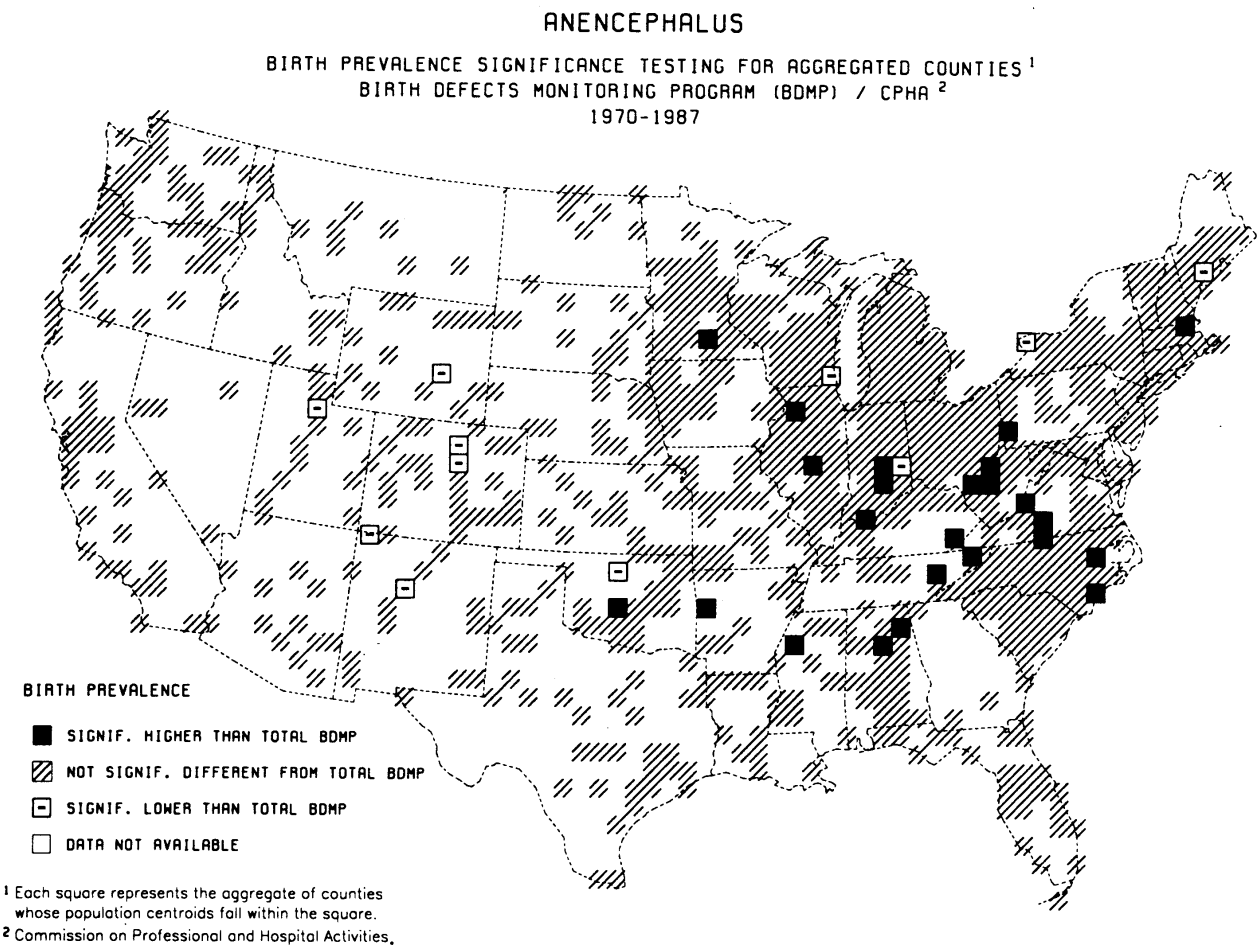
The prevalence of many birth defects vary markedly according to race (Table 3). Rates of almost all CNS defects were lowest for Asians, with the exception of anencephalus rates, which were lowest for blacks. Hispanics had the highest rates of anencephalus and spina bifida without anencephalus, whereas Native Americans had the highest prevalence of hydrocephalus and encephalocele. Compared with other races, Asians were at a decreased risk of the two eye birth defects—anophthalmos/microphthalmos and congenital cataract. Hispanics had the lowest rates of all but three of the nine cardiovascular defects followed; and, among these three conditions, only coarctation of the aorta showed a substantial elevation. For the two orofacial defects, rates were lowest for blacks and highest for Native Americans. Native Americans had the highest rates of the two genitourinary defects—renal agenesis/dysgenesis and bladder exstrophy—whereas Asians had the lowest rates.

The strong relationship between Down's syndrome and maternal age is reflected by MACDP data for 1968–1991 (Table 4). The age-specific rates began to increase substantially after the age of 29 years and attained levels in the range of 1%–2% for women >40 years of age.

INTERPRETATION ISSUES

The diagnosis and reporting of birth defects is rarely perfect, and problems of sensitivity and specificity of ascertainment abound. Thus, completeness and accuracy of birth defects reporting must be considered in the interpretation of nominal rates. For example, the birth prevalences of externally apparent malformations such as

FIGURE 2.



anencephalus, spina bifida, and cleft lip are more secure than those for birth defects of the cardiovascular system, which may not be manifest during the newborn period or which require sophisticated techniques for diagnosis. In addition, birth defects reporting through the MACDP, which uses multiple ascertainment methods, is more complete than reporting through the BDMP, which relies on passive reporting of newborn hospital discharge diagnoses. Often the more relevant occurrence statistic is the change in prevalence over time or geographic-based differences in birth defects rates. Even though the absolute levels in reported prevalence may be highly questionable in certain instances, we may judge that changes or differences in rates are fairly reliable.

The finding that maternal intake of folic acid decreases the risk of anencephalus and spina bifida (*3–6*) suggests that increasingly better nutrition during the past two decades has contributed to the decline in prevalence of these neural tube defects. Although the increasing use of prenatal diagnosis and pregnancy termination may have introduced a downward bias in the birth prevalences of anencephalus and spina bifida, the decline in reported prevalence began, in the 1980s, before these procedures were used significantly. The halving of the prevalence of combined anophthalmos and microphthalmos between 1970 and 1976, followed by subsequent stability of rates, is striking, but we have no explanation for this pattern of rates. An explanation for the marked decline in the prevalence of Rh hemolytic disease is easy to find—the introduction of Rh immunoglobulin in the late 1960s was the undoubted preventive agent. We have no good explanations for declines in the occur-

FIGURE 3.

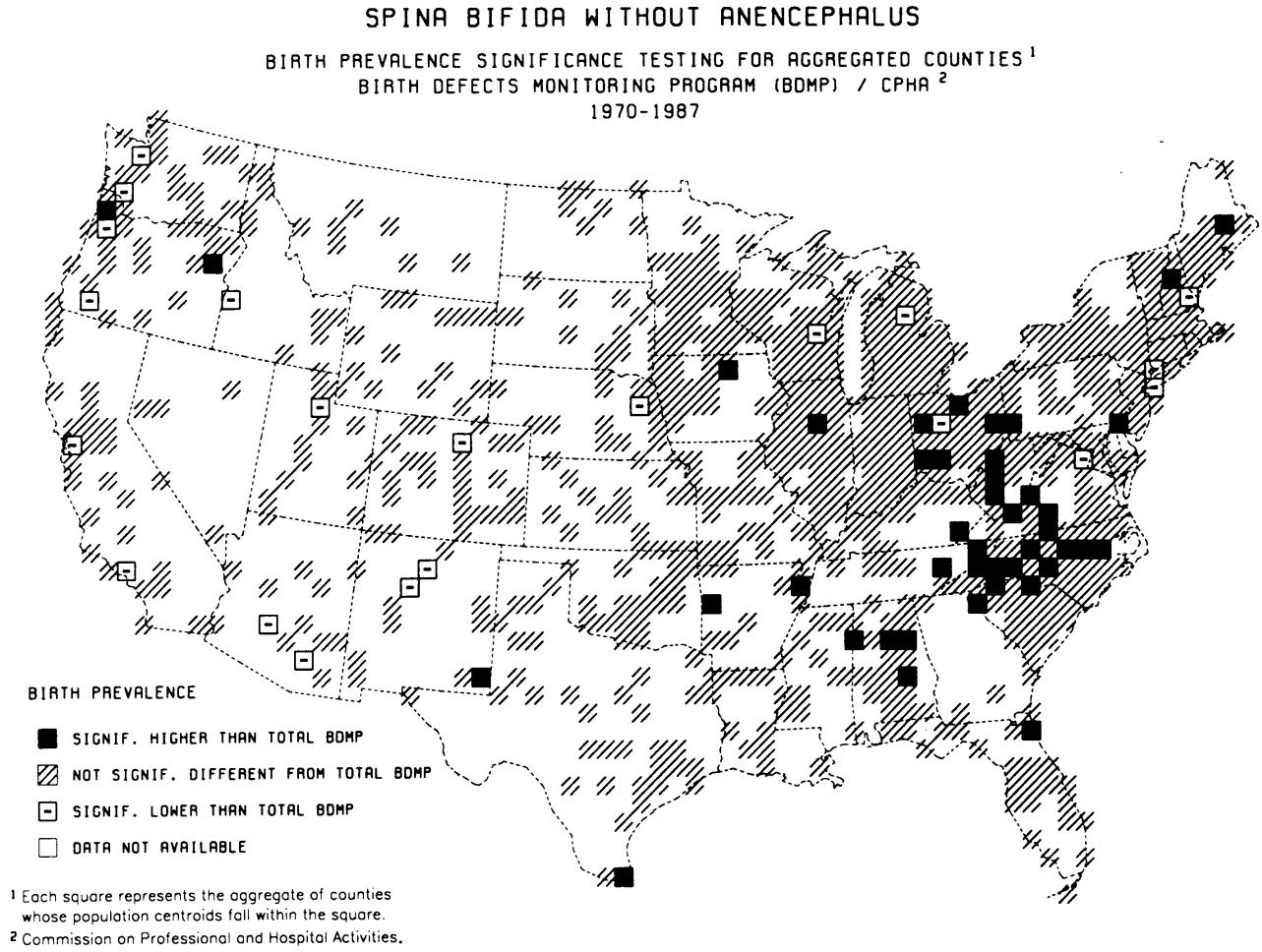


TABLE 3. Prevalence of selected birth defects, by race, and race-specific rate ratios — Birth Defects Monitoring Program, 1981–1991*

	White		Black		Hispanic		Asian		Native American	
Birth Defect	**Rate**	**Ratio**	**Rate**	**Ratio**	**Rate**	**Ratio**	**Rate**	**Ratio**	**Rate**	**Ratio**
CNS										
Anencephalus	2.6	1.36	*1.9*	*1.00*	**3.7**	**1.95**	3.5	1.84	2.8	1.49
Spina bifida without anencephalus	4.8	3.49	3.4	2.49	**5.2**	**3.78**	*1.4*	*1.00*	4.0	2.90
Hydrocephalus without spina bifida	5.4	1.37	8.4	2.13	4.5	1.14	*4.0*	*1.00*	**11.7**	**2.95**
Encephalocele	1.0	1.04	1.1	1.14	1.3	1.27	*1.0*	*1.00*	**4.0**	**4.06**
Eye										
Anophthalmos/microphthalmos	0.8	1.54	0.9	1.80	0.6	1.16	*0.5*	*1.00*	**2.0**	**4.06**
Congenital cataract	1.0	2.06	**1.5**	**2.96**	0.8	1.59	*0.5*	*1.00*	0.8	1.63

TABLE 3. Prevalence of selected birth defects, by race, and race-specific rate ratios — Birth Defects Monitoring Program, 1981–1991* — continued

Birth Defect	White		Black		Hispanic		Asian		Native American	
	Rate	Ratio	Rate	Ratio	Rate	Ratio	Rate	Ratio	Rate	Ratio
Cardiovascular										
Common truncus	0.3	1.67	0.3	1.37	*0.2*	*1.00*	**0.4**	**2.16**	0.0	0.00
Transposition of great arteries	1.4	1.79	*0.8*	*1.00*	0.9	1.11	1.0	1.27	**1.6**	**2.07**
Tetralogy of Fallot	1.4	1.35	1.3	1.28	*1.0*	*1.00*	**1.8**	**1.70**	1.6	1.54
Ventricular septal defect	19.1	1.28	15.7	1.05	*15.0*	*1.00*	**19.9**	**1.33**	18.9	1.26
Atrial septal defect	3.7	1.73	4.4	2.06	*2.1*	*1.00*	6.3	2.01	**5.2**	**2.47**
Endocardial cushion defect	0.9	1.09	0.9	1.10	*0.8*	*1.00*	1.0	1.22	**1.6**	**1.98**
Patent ductus arteriosus	31.2	1.19	**58.9**	**2.24**	*26.2*	*1.00*	31.8	1.21	41.9	1.60
Coarctation of aorta	0.9	2.39	0.8	2.05	0.9	2.25	*0.4*	*1.00*	**1.2**	**3.05**
Pulmonary artery anomaly	1.7	1.04	**5.6**	**3.50**	1.8	1.14	2.2	1.35	*1.6*	*1.00*
Respiratory										
Lung agenesis and hypoplasia	3.3	1.36	3.4	1.38	*2.5*	*1.00*	3.1	1.25	**4.8**	**1.96**
Orofacial										
Cleft palate without cleft lip	5.8	1.56	*3.7*	*1.00*	4.4	1.18	5.2	1.38	**8.5**	**2.27**
Cleft lip with or without cleft palate	9.6	2.17	*4.4*	*1.00*	8.8	1.97	12.0	2.71	**16.9**	**3.82**
Gastrointestinal										
Tracheoesophageal anomalies	**2.5**	**1.91**	*1.3*	*1.00*	1.9	1.50	1.5	1.15	2.0	1.56
Rectal and intestinal atresia	3.7	1.28	3.0	1.01	*2.9*	*1.00*	3.6	1.22	**5.2**	**1.78**
Genitourinary										
Renal agenesis and dysgenesis	2.1	2.39	1.5	1.72	1.7	1.94	*0.9*	*1.00*	**2.4**	**2.71**
Bladder exstrophy	0.3	3.32	0.2	1.62	0.2	1.59	*0.1*	*1.00*	**0.8**	**8.13**
Musculoskeletal										
Clubfoot without CNS defects	**26.9**	**1.91**	19.4	1.38	19.7	1.40	*14.1*	*1.00*	14.5	1.03
Limb reduction deformity	**3.8**	**1.91**	3.7	1.83	3.2	1.60	2.8	1.38	*2.0*	*1.00*
Chromosomal										
Down's syndrome	8.9	1.29	*6.9*	*1.00*	11.7	1.70	**11.8**	**1.72**	8.9	1.29
Other										
Rh hemolytic disease	15.3	3.36	13.8	3.02	**19.1**	**4.18**	*4.6*	*1.00*	10.9	2.39
Number of births	4,887,008		872,816		381,603		100,882		24,821	

* Rates per 10,000 total births. Rates are computed with respect to the smallest race-specific rate greater than zero (italics). Maximum rate ratios for each defect are shown in boldface type.

TABLE 4. Prevalence of Down's syndrome, by maternal age — Metropolitan Atlanta Congenital Defects Program, 1968–1991*

Maternal age (years)	No. cases	No. births	Rate
<20	81	112,112	7.2
20–24	138	206,003	6.7
25–29	159	210,276	7.6
30–34	176	122,902	14.3
35–39	89	38,120	23.3
40–44	49	5,436	90.1
45+	5	223	224.2
Unknown	6	985	60.9
Total, all ages	703	696,057	10.1

* Rates per 10,000 live births.

rence of hydrocephalus and clubfoot, but physicians' tightening of the diagnostic criteria for these conditions during this period may have contributed to these reductions. Substantial increases in the occurrence of most cardiovascular malformations raise the question of whether these increases may have been related to improvements in case ascertainment. Technological advances in diagnostic techniques, such as in the field of echocardiography, are likely responsible for some portion of these increases. In addition, better survival of affected infants over time increases the probability of a diagnosis being made. However, it would be premature to discount the existence of underlying true increases in the occurrences of these defects. Increases in the prevalence of lung agenesis and hypoplasia between 1970 and 1991 can be attributed partly to 1974 and 1979 coding changes that included additional conditions in this diagnostic category. Continued increases after 1979, however, point to other unknown factors that influence the rates.

The decreasing prevalence of spina bifida from eastern to western states (Figure 3) is consistent with the finding by Hewitt of a similar gradient in infant mortality caused by this birth defect (7). Given the embryologic connection between anencephalus and spina bifida, it is not surprising that anencephalus has a similar geographic gradient in prevalence, although not quite as striking. Whether these patterns of rates are related to genetic or environmental factors is not known. Given the previously mentioned finding that dietary folic acid reduces the risk of these neural tube defects, nutritional differences associated with geography quite possibly may play a role.

The variations in birth defect occurrence according to race could result from differences in risk-related exposures or to race-specific susceptibility (Table 3). We now lack the data needed to judge which of these two possibilities are operative for particular birth defects. We may reasonably surmise that, at least for some defects, both factors could have contributed to the observed differences.

The increased risk of Down's syndrome among women over the age of 30 years has been long recognized. These data underscore the need for increased awareness of this risk among the relevant population and the availability of prenatal testing procedures for detecting affected fetuses.

EXAMPLES OF USING DATA

Birth defects surveillance systems provide current and baseline data that allow investigators to monitor changes in the prevalence of specific malformations on a national or local level. Exploring the occurrence patterns of these birth defects can generate etiologic hypotheses, descriptive epidemiologic studies, follow-up studies, family studies, case-control studies, and cluster investigations.

As a national, hospital-based system, the BDMP has provided researchers, policymakers, and the lay public with time- and place-specific prevalence data. These data have helped to dispel unwarranted concerns about the possibility of increased birth defects risks in a particular area. They have also generated investigations of seemingly unexplained increases in birth defects occurrence. The ability to evaluate geographic differences in rates is especially important in areas that do not have a local birth defects surveillance system. Public health officials can often use this information to help them make decisions and establish policies.

The MACDP is an intensive, population-based system that has served as a prototype for other state and local birth defects surveillance systems. Consistent and systematic surveillance procedures—which include detailed coding, uniform variables, and standard data collection methods—have been developed and enhanced through MACDP and have facilitated collaborative birth defects studies across the country.

Birth defects registries can also help to identify children who may be eligible for special programs or services. This role can lead to the expansion of surveillance programs to incorporate prevention, intervention, and evaluation components into their systems.

FUTURE ISSUES

During the next decade we can expect to see tremendous increases in the ability to make prenatal diagnoses of birth defects. This change in capability will necessitate changes in the methods and data sources used for birth defects surveillance.

Over the past two decades, chromosomal analysis of amniotic fluid cells has become widely available for pregnant women aged 35 years and older, primarily because these women are at increased risk of having a fetus affected by Down's syndrome. Alpha-fetoprotein screening of maternal serum is also widely used, mainly to detect fetuses affected by neural tube defects. More recently, prenatal diagnoses of neural tube defects and other types of malformations have been made by fetal ultrasonographic examination. As prenatal ultrasonography becomes more commonly used, and as instrumentation and techniques improve, we can expect to see a greater proportion and variety of malformations diagnosed prenatally. Advances in the analysis of DNA (i.e., the new genetics) should also increase the numbers of prenatally diagnosed congenital malformations.

Many women who discover that they are carrying a fetus with a defect elect to have their pregnancy terminated. Most current birth defects surveillance programs, including the MACDP and the BDMP, make use of records created in hospitals at the time of birth. Understanding variations observed in the frequency of birth defects at birth will increasingly require a knowledge of the effects of pregnancy terminations that are done as the result of prenatal diagnoses of birth defects.

Methods of collecting birth defects data will also need to change to adapt to revisions in hospital data processing methods. The BDMP was started at a time when, for convenience and economical reasons, small- and medium-sized hospitals had computer service organizations handle their data processing. The advent of more accessible and affordable data processing equipment has reduced the number of hospitals that use these organizations. Therefore, the CPHA, the source of BDMP hospital discharge abstract data, no longer services the large number of hospitals that it once did, and the number of hospitals available for the BDMP has dropped from 1,264 in 1974 to 464 in 1991.

These changes will force us to seek new sources of data. We hope that the much discussed health-care reform brings changes that will improve our prospects for having more accessible data for national birth defects surveillance and thus, for achieving our year 2000 goals to reduce the prevalence of birth defects (for details about these objectives, see the State Use of Birth Defects Surveillance chapter).

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