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Update on Overall Prevalence of Major Birth Defects --- Atlanta, Georgia, 1978--2005

Major structural or genetic birth defects affect approximately 3% of births in the United States, are a major contributor to infant mortality (*1,2*), and result in billions of dollars in costs for care (*3*). Although the causes of most major birth defects are unknown, concerns have been raised that certain factors, such as an increase in the prevalence of diabetes among women, might result in increased prevalence of birth defects over time (*4*). This report updates previously published data from the Metropolitan Atlanta Congenital Defects Program (MACDP), the oldest population-based birth defects surveillance system in the United States with active case ascertainment (*5*). For the period 1978--2005, CDC assessed the overall prevalence of major birth defects and their frequency relative to selected maternal and infant characteristics. The MACDP results indicated that the prevalence of major birth defects in metropolitan Atlanta, Georgia, remained stable during 1978--2005 but varied by maternal age and race/ethnicity, birthweight, and gestational age. Tracking the overall prevalence of major birth defects can identify subgroups that are affected disproportionately; additional measures focused on these subgroups might improve preconception care and care during pregnancy to prevent birth defects.

State-based surveillance programs monitor the prevalence of certain birth defects through various methods, including passive hospital-based reporting and active medical-record abstraction (*6*). These data are used for prevention, education, policy, and health-care planning (*7*). However, most state-based surveillance programs were established in recent years and only monitor certain types of defects; therefore, population-based estimates of the overall prevalence of all defects and data on long-term trends are lacking in the United States. MACDP, established in 1967 by CDC, Emory University, and the Georgia Mental Health Institute, monitors the prevalence of all major structural or genetic defects at the time of delivery among live births, stillbirths, and pregnancies electively terminated after prenatal diagnosis of defects at ≥20 weeks' gestation in the five central counties of metropolitan Atlanta (*5*). MACDP defines major structural or genetic birth defects as conditions that 1) result from a malformation, deformation, or disruption in one or more parts of the body, a chromosomal abnormality, or a known clinical syndrome; 2) are present at birth; and 3) have a serious, adverse effect on health, development, or functional ability.

To collect data on birth defects, trained MACDP records abstractors visit birth and pediatric hospitals and genetic laboratories to review in-patient medical records of infants and fetuses of ≥20 weeks' gestation. Systematic case-finding by the abstractors at each hospital includes review of labor and delivery logs, nursery and intensive-care logs (including neonatal intensive-care logs), stillbirth and pathology logs, and disease indices. The medical records for each infant or fetus with a potential birth defect are then examined to identify those with defects that meet the MACDP case definition. Information about identified defects among live births is updated until age 6 years. However, the system might miss certain defects, including those that 1) occur among children whose families move away from the Atlanta area before diagnosis, 2) are managed on an outpatient basis only, 3) are unrecognized among stillbirths, or 4) are diagnosed prenatally among pregnancies subsequently terminated outside a hospital setting. Denominator data on the number of live births to residents of the five counties are obtained from vital records of the Georgia Department of Human Resources. Such data have included maternal Hispanic ethnicity only since 1990. Data on birthweight have been available since 1978 for the offspring of white mothers and black mothers and since 1997 for Hispanic mothers; data on gestational age have been available since 1988 for the offspring of white mothers and black mothers and since 1997 for Hispanic mothers.

For MACDP purposes, prevalence is defined as the number of infants and fetuses with a major birth defect that were delivered during a specified period divided by the number of live births during that period. For this report, the overall prevalence of major defects per 100 live births was estimated for each of three periods (1978--1987, 1988--1996, and 1997--2005) and by the following characteristics: maternal race/ethnicity (white, black, or Hispanic), maternal age (<35 years or ≥35 years), infant birthweight (<2,500 g or ≥2,500 g), gestational age (20--36 weeks or ≥37 weeks), and sex. The three periods were chosen because they corresponded to available denominator data for birthweight and gestational age and enabled comparisons of periods of approximately equal length. Data for 2005 are preliminary because abstractions for defects that were not diagnosed or did not require hospitalization until the child was several months of age might not yet have been processed. Data for racial/ethnic groups other than whites, blacks, and Hispanics were not included in this report because of small numbers. Prevalence ratios (PRs) and 95% confidence intervals (CIs) were calculated. Trend over time in overall prevalence was evaluated using the Mantel-Haenszel test for trend.

The overall prevalence of major defects was stable from 1978 (2.8 per 100 live births) to 2005 (3.0 per 100) (test for trend p = 0.19) (Figure). During this period, the number of births in the metropolitan Atlanta area more than doubled, from 24,396 in 1978 to 51,400 in 2005. Prevalence of defects generally was lower among births to black mothers (PR = 0.94, CI = 0.93--0.95) and Hispanic mothers (PR = 0.89, CI = 0.86--0.93) than to white mothers.

Births to women aged ≥35 years had a greater prevalence of defects than births to women aged <35 years (PR = 1.28, CI = 1.24--1.31), with this excess prevalence increasing over time (Table). During 1978--2005, the overall prevalence was greater among infants with birthweight <2,500 g (PR = 2.97, CI = 2.90--3.04) and among infants with gestational age of 20--36 weeks (PR = 2.53, CI = 2.47--2.59). Prevalence was greater among males than among females (PR = 1.17, CI = 1.16--1.18); however, the higher prevalence among males decreased when defects that occur almost exclusively in males (e.g., hypospadias) were excluded (PR = 1.04, CI = 1.02--1.05).

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Editorial Note:

The findings in this report indicate that the overall prevalence of major birth defects in metropolitan Atlanta did not change significantly during 1978--2005. This finding suggests that, over time, no changes occurred in major risk factors that affect birth defects overall. This information can be useful in assessing the success of prevention interventions for defects overall.

However, although the overall prevalence did not change significantly, a greater prevalence of birth defects among infants of low birthweight and preterm gestation might signal a need for increased prenatal health care and planning for the extended-care requirements. The greater prevalence of defects among the offspring of women aged ≥35 years likely reflects an upward trend in maternal age distribution and the progressive association of certain defects as maternal age increases beyond 35 years (*8,9*). The lower prevalence of defects among black and Hispanic infants might reflect an actual lower prevalence among these groups; however, racial and ethnic variation in health-insurance coverage, diagnosis of nonsymptomatic defects through pediatric and subspecialty care, and ascertainment of these defects by MACDP's hospital-based methods also might affect differences in defect prevalence. Further evaluation of these differences is needed.

The stable overall prevalence of major birth defects in Atlanta is consistent with the trend observed for many individual defects (*5*). However, the prevalence of certain defects in Atlanta has changed over time. For example, a decline in the prevalence of anencephaly and spina bifida might reflect fortification of the U.S. grain supply with folic acid and increased consumption of folic acid vitamin supplements. Progressive declines in the prevalence of clubfoot not associated with spina bifida and of cleft lip (with or without cleft palate) also have been observed. In contrast, the prevalence of Down syndrome and other autosomal trisomies among the offspring of mothers aged ≥35 years has increased over time, likely reflecting the increase in age distribution of mothers aged ≥35 years in metropolitan Atlanta (CDC, unpublished data, 2007). The prevalence of ventricular septal defect, atrial septal defect, and valvar pulmonic stenosis also have increased progressively, likely reflecting increased use of bedside echocardiography to diagnose heart defects among newborns (*5*).

The findings in this report are subject to at least four limitations. First, because childbearing women in Atlanta might differ from women in other areas of the United States with respect to characteristics that might be associated with the risk for birth defects, the observed prevalence of major birth defects in metropolitan Atlanta might not be generalizable to other populations. Second, the specific defect inclusion and exclusion criteria used by MACDP might differ from those used by other surveillance programs, resulting in differences in prevalence estimates (*10*). For example, the MACDP case definition does not include developmental, functional, or other types of congenital disorders (e.g., nonstructural or genetic disorders not detected in children aged ≤6 years). Third, data in this report do not include defects diagnosed prenatally among pregnancies electively terminated before 20 weeks' gestation or outside a hospital setting. Failure to ascertain these pregnancies might result in underestimation of the prevalence of major defects (*9*). Finally, data on age of mother and sex of offspring were available by maternal Hispanic ethnicity only since 1990, and data on birthweight and gestational age of offspring were available by maternal Hispanic ethnicity only since 1997.

Population-based data on the overall prevalence of major birth defects can be used to identify subgroups that are affected disproportionately, evaluate prevention measures (e.g., promotion of preconception health and health care use), and recommend additional health-care services and resources where needed. These Atlanta findings should encourage surveillance programs elsewhere to monitor the overall prevalence of major defects in their areas, assess their public health burden, and examine the variability of defects among specific populations.

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Table

TABLE. Overall number and prevalence* of major structural or genetic birth defects,† by selected maternal and infant characteristics and maternal race/ethnicity — Metropolitan Atlanta Congenital Defects Program (MACDP), 1978–2005§

Characteristic	Period	Maternal race/ethnicity: White No.	White Prevalence	Black No.	Black Prevalence	Hispanic¶ No.	Hispanic¶ Prevalence	Total** No.	Total** Prevalence	Prevalence ratio	(95% CI††)
Total		**15,448**	**2.92**	**10,971**	**2.62**	**2,224**	**2.57**	**29,769**	**2.76**	—	—
Age of mother (yrs)											
<35	1978–1987	4,141	2.69	3,007	2.97	—	—	**7,554**	**2.80**	Referent	—
≥35		371	2.94	117	3.66	—	—	**572**	**3.19**	1.14	(1.05–1.23)
<35	1988–1996	4,366	2.81	3,021	2.31	293	2.25	**7,953**	**2.47**	Referent	—
≥35		878	3.35	349	3.15	28	2.64	**1,305**	**3.27**	1.24	(1.18–1.31)
<35	1997–2005	3,949	3.00	3,622	2.43	1,703	2.55	**9,793**	**2.64**	Referent	—
≥35		1,432	3.70	783	3.70	193	3.37	**2,540**	**3.62**	1.31	(1.26–1.35)¶¶
Birthweight (g)¶¶											
≥2,500	1978–1987	3,825	2.30	2,224	2.40	—	—	**6,168**	**2.34**	Referent	—
<2,500		944	9.04	958	7.21	—	—	**1,935**	**8.05**	3.02	(2.90–3.15)
≥2,500	1988–1996	4,297	2.33	2,213	1.78	—	—	**7,003**	**2.19**	Referent	—
<2,500		927	8.42	1,150	6.42	—	—	**2,225**	**7.48**	2.96	(2.87–3.10)
≥2,500	1997–2004	3,868	2.71	2,608	1.97	1,237	2.12	**8,131**	**2.29**	Referent	—
<2,500		918	9.21	1,243	6.68	342	9.50	**2,631**	**7.69**	2.93	(2.83–3.04)
Gestational age (wks)***											
≥37	1988–1996	4,141	2.40	2,304	2.00	—	—	**6,937**	**2.33**	Referent	—
20–36		990	7.26	998	4.94	—	—	**2,137**	**6.14**	2.33	(2.25–2.42)
≥37	1997–2005	4,043	2.62	2,973	2.02	1,433	2.14	**8,963**	**2.29**	Referent	—
20–36		1,217	7.60	1,357	5.95	424	7.57	**3,141**	**7.12**	2.68	(2.60-2.77)
Sex											
Female	1978–1987	1,933	2.25	1,422	2.72	—	—	**3,416**	**2.43**	Referent	—
Male		2,857	3.13	1,753	3.26	—	—	**4,700**	**3.18**	1.13	(1.11–1.16)
Female	1988–1996	2,003	2.26	1,386	1.98	145	2.10	**3,663**	**2.14**	Referent	—
Male		3,241	3.48	1,971	2.74	176	2.44	**5,580**	**3.12**	1.19	(1.17–1.21)
Female	1997–2005	2,148	2.58	1,795	2.14	855	2.40	**5,050**	**2.33**	Referent	—
Male		3,244	3.73	2,609	3.01	1,043	2.83	**7,294**	**3.25**	1.17	(1.15–1.18)

* Per 100 live births.
† MACDP defines major structural or genetic birth defects as conditions that 1) result from a malformation, deformation, or disruption in one or more parts of the body, a chromosomal abnormality, or a known clinical syndrome; 2) are present at birth; and 3) have a serious, adverse effect on health, development, or functional ability.
§ 2005 data are preliminary.
¶ Data on age of mother and sex of offspring have been available by maternal Hispanic ethnicity only since 1990. Data on birthweight and gestational age of offspring have been available by maternal Hispanic ethnicity only since 1997. Mothers categorized as Hispanic might be of any race.
** Includes racial/ethnic populations other than white, black, and Hispanic.
†† Confidence interval.
¶¶ Trend in major defect prevalence over time is statistically significant (p<0.05) using Mantel-Haenszel test for trend.
§§ 2005 birthweight data were not available.
*** Data on gestational age were available for the offspring of white mothers and black mothers only since 1988.

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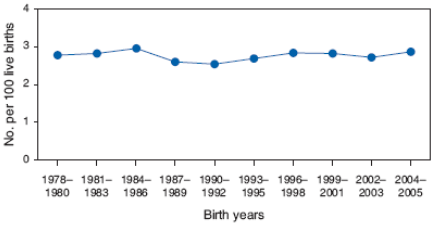
Figure

FIGURE. Overall prevalence of major structural or genetic birth defects,* by selected maternal and infant characteristics and maternal race/ethnicity — Metropolitan Atlanta Congenital Defects Program (MACDP), 1978–2005†



* MACDP defines major structural or genetic birth defects as conditions that 1) result from a malformation, deformation, or disruption in one or more parts of the body, a chromosomal abnormality, or a known clinical syndrome; 2) are present at birth; and 3) have a serious, adverse effect on health, development, or functional ability.
† 2005 data are preliminary. Mantel-Haenszel test for trend, p = 0.19.

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