# Racial and Temporal Variations in the Prevalence of Heart Defects

Lorenzo D. Botto, MD; Adolfo Correa, MD, PhD; and J. David Erickson, DDS, PhD

**ABSTRACT.** *Background.* **Documenting the prevalence and trends of congenital heart defects provides useful data for pediatric practice, health-care planning, and causal research. Yet, most population-based studies use data from the 1970s and 1980s. We sought to extend into more recent years the study of temporal and racial variations of heart defects occurrence in a well-defined population.**

*Methods.* **We used data from the Metropolitan Atlanta Congenital Defects Program, a population-based registry with active case ascertainment from multiple sources. Heart defects were identified among liveborn infants up to 1 year old, among stillborn infants, and among pregnancy terminations to mothers residing in metropolitan Atlanta.**

*Results.* **From 1968 through 1997, the registry ascertained 5813 major congenital heart defects among 937 195 infants, for a prevalence of 6.2 per 1000. The prevalence increased to 9.0 per 1000 births in 1995 through 1997. The prevalence of ventricular septal defects, tetralogy of Fallot, atrioventricular septal defects, and pulmonary stenosis increased, whereas that of transposition of the great arteries decreased. For some defects, prevalence and trends varied by race.**

*Conclusions.* **The prevalence of congenital heart defects is increasing. Whereas most findings likely result from improved case ascertainment and reporting, others might be because of changes in the distribution of risk factors in the population. The basis of the racial variations is incompletely understood.** *Pediatrics* 2001;107(3). URL: http://www.pediatrics.org/cgi/content/full/107/3/e32; *heart defects, whites, blacks, epidemiology, prevalence.*

---

ABBREVIATIONS. MACDP, Metropolitan Atlanta Congenital Defects Program; ICD-9-CM, *International Classification of Diseases, Ninth Revision, Clinical Modification.*

---

Congenital heart defects are frequent and serious anomalies that have a major impact on pediatric morbidity, mortality, and health care costs.[1–4] Knowing the expected prevalence and distribution of these serious anomalies can help pediatricians and public health professionals better assess health care needs, evaluate potential clusters, and conduct causal research.

Current prevalence estimates are often based on population-based studies conducted between the 1960s and 1980s.[5–7] In such studies, the prevalence at birth of congenital heart defects varied from 2.0 to 5.5 per 1000 births, with higher rates usually associated with later studies or with the use of broader diagnostic criteria.[5–7] Such figures, however, may underestimate the current impact at birth of heart defects because some of these studies,[5,8] as well as later studies,[9–11] documented an increase over time in the occurrence of some heart defects. For example, an increased occurrence rate of ventricular septal defects was reported in some areas of the United States during the 1970s[9] and 1980s.[8] A similar increase was reported for pulmonic stenosis during the 1980s.[8] In some cases, temporal trends seemed to vary with race.[12]

We assessed the temporal and racial variations in heart defect occurrence during a 30-year period in a well-defined population. Our objectives were to: 1) provide a more recent estimate of the impact of major heart defects than is currently available; 2) evaluate past temporal trends and assess whether such trends are still occurring; and 3) assess whether such trends differed by race.

To decrease bias and enhance generalizability, we based our study on a population-based monitoring system with active case ascertainment from multiple sources.

## METHODS

### Case Ascertainment

We used data from the Metropolitan Atlanta Congenital Defects Program (MACDP). MACDP is a population-based registry that actively monitors birth defects among infants born to mothers residing in the 5-county metropolitan Atlanta area. Trained abstractors from the registry actively ascertain structural birth defects among liveborn infants up to 1 year old, among stillborn infants, and among pregnancies terminated at or after 20 weeks of gestation. The abstractors regularly visit hospitals, pediatric and specialty wards, cytogenetic laboratories, and the vital records office in the 5-county area. Data sources include medical records, hospital logs, and birth and death certificates. The abstracted information is reviewed by the medical staff of MACDP, which includes geneticists and pediatricians. Congenital defects are coded in accordance with a modified *International Classification of Diseases, Ninth Revision, Clinical Modification* (ICD-9-CM) coding scheme.

### Case Classification

We selected all cases for which a major cardiac defect was recorded in MACDP (ICD9-CM codes 745.00 to 747.49). Each case participant received a principal cardiac diagnosis, according to a hierarchical classification system[13] (Table 1). In such system, defects placed toward the top of the list are postulated to occur earlier in embryogenesis than those placed further down the list (the exception being the group of "other major defects," which,

**TABLE 1.** Prevalence and Distribution of Major Congenital Heart Defects by Birth Status and During Specified Time Periods, Metropolitan Atlanta, 1968–1997

| Defect Type | Liveborn | | Stillborn | | Total 1968–1997* | | | Change During Study | | | Trend† (P Value) |
|---|---|---|---|---|---|---|---|---|---|---|---|
| | No. | % | No. | % | No. | % | Rate‡ | Rate in 1995–1997‡ | Rate Increase§ | Contribution to Total Change (%) | |
| Heterotaxias | 78 | 1.4 | 2 | 1.5 | 81 | 1.4 | 0.9 | 0.9 | 0.5 | 0.8 | |
| Corrected (L) transposition | 39 | 0.7 | 0 | 0.0 | 39 | 0.7 | 0.4 | 0.7 | 0.4 | 0.6 | |
| Outflow tract defects | | | | | | | | | | | |
| Tetralogy of Fallot | 345 | 6.1 | 6 | 4.5 | 353 | 6.1 | 3.8 | 4.7 | 2.2 | 3.7 | *** |
| D-transposition of the great arteries | 226 | 4.0 | 4 | 3.0 | 231 | 4.0 | 2.5 | 2.4 | −0.4 | | |
| Double outlet right ventricle | 96 | 1.7 | 3 | 2.2 | 101 | 1.7 | 1.1 | 2.2 | 2.0 | 3.4 | *** |
| Truncus arteriosus | 54 | 1.0 | 4 | 3.0 | 60 | 1.0 | 0.6 | 0.6 | 0.2 | 0.3 | |
| Atrioventricular septal defect (atrioventricular canal) | | | | | | | | | | | |
| With Down syndrome | 138 | 2.4 | 2 | 1.5 | 140 | 2.4 | 1.5 | 2.4 | 1.7 | 3.0 | *** |
| Without Down syndrome | 104 | 1.8 | 7 | 5.2 | 111 | 1.9 | 1.2 | 1.0 | 0.6 | 1.1 | * |
| Total anomalous pulmonary venous return | 66 | 1.2 | 0 | 0.0 | 66 | 1.1 | 0.7 | 0.7 | 0.1 | 0.2 | |
| Ebstein anomaly | 36 | 0.6 | 4 | 3.0 | 40 | 0.7 | 0.4 | 0.6 | 0.5 | 0.8 | ** |
| Right obstructive defects | | | | | | | | | | | |
| Tricuspid atresia | 27 | 0.5 | 3 | 2.2 | 31 | 0.5 | 0.3 | 0.3 | 0.0 | −0.1 | |
| Pulmonary atresia, intact septum | 48 | 0.8 | 2 | 1.5 | 50 | 0.9 | 0.5 | 0.7 | 0.2 | 0.4 | |
| Pulmonic stenosis, atresia | 357 | 6.3 | 0 | 0.0 | 358 | 6.2 | 3.8 | 6.0 | 4.3 | 7.4 | *** |
| Peripheral pulmonary stenosis | 333 | 5.9 | 0 | 0.0 | 333 | 5.7 | 3.6 | 7.0 | 6.9 | 11.7 | *** |
| Left obstructive defects | | | | | | | | | | | |
| Hypoplastic left heart | 189 | 3.3 | 5 | 3.7 | 196 | 3.4 | 2.1 | 2.1 | 0.5 | 0.8 | |
| Coarctation of the aorta | 267 | 4.7 | 1 | 0.7 | 268 | 4.6 | 2.9 | 3.5 | 0.7 | 1.2 | |
| Aortic arch atresia or hypoplasia | 57 | 1.0 | 1 | 0.7 | 58 | 1.0 | 0.6 | 0.6 | 0.3 | 0.4 | |
| Aortic valve stenosis | 90 | 1.6 | 0 | 0.0 | 90 | 1.5 | 1.0 | 0.8 | 0.1 | 0.1 | |
| Septal defects | | | | | | | | | | | |
| Ventricular septal defect | 1514 | 26.8 | 39 | 29.1 | 1559 | 26.8 | 16.6 | 25.0 | 17.1 | 29.2 | *** |
| Atrial septal defect | 395 | 7.0 | 34 | 25.4 | 431 | 7.4 | 4.6 | 10.0 | 8.8 | 15.0 | *** |
| Patent ductus arteriosus | 617 | 10.9 | 0 | 0.0 | 617 | 10.6 | 6.6 | 8.1 | 5.3 | 9.0 | *** |
| Other major heart defects | 580 | 10.3 | 17 | 12.7 | 598 | 10.3 | 6.4 | 9.7 | 6.8 | 11.7 | *** |
| | | | | | | | | | | | *** |
| Total | 5656 | | 134 | | 5813 | | 62.0 | 90.2 | 58.7 | 100.0 | *** |

\* Includes cases of pregnancy terminations and of unknown birth status.
† P values, by Cochrane-Armitage test: * .01 < P < .05; ** .001 < P < .01; *** P < .001.
‡ Rate per 10 000 births.
§ Difference in rate, 1995–1997 versus 1968–1972.

although listed last, precedes in coding the category of patent ductus arteriosus). In practice, this system assigns to each infant only one cardiac diagnosis; an infant with multiple cardiac defects is assigned to the defect group that is listed closer to the top of the list. For example, an infant with tetralogy of Fallot, atrial septal defect, and patent ductus arteriosus is counted as (and only as) a case of tetralogy of Fallot.

In this study, ventricular septal defects included perimembranous, muscular, or unspecified defects. We did not analyze these subgroups separately because the ability to distinguish such subgroups depends on available technology and likely changed over the study. Atrioventricular septal defects also included cases of primum atrial septal defects and primum ventricular septal defects. Atrial septal defects included all atrial septal defects, including the generic atrial septal defects, but excluded primum defects.

### Inclusions and Exclusions

We included cases of confirmed heart defects as documented by the medical records and notes. Not all cases, particularly in the earlier years of the study, were necessarily verified by echocardiography, catheterization, surgery, or autopsy. This approach reduces differences caused by changes in technology over the years and provides data that closely reflect the clinical burden of disease.

We excluded cases of heart defects that were noted in the medical records as being possible or probable cases and those for which a specific diagnosis was not made (eg, cases of murmur, congenital heart defect not otherwise specified, cyanotic heart defect not otherwise specified). We also excluded the following cases: 1) those based on fetal ultrasound examination alone, without postnatal confirmation; 2) isolated patent ductus arteriosus among stillborn infants, pregnancy terminations, and among in-

fants whose birth weight was below 2500 g or whose gestational age was <37 weeks; 3) anomalies of the peripheral vascular system (ICD-9-CM codes 747.50–747.99), such as an aberrant subclavian artery or a peripheral arteriovenous malformation, unaccompanied by defects of the heart or the great vessels.

### Statistical Analysis

We used the number of liveborn infants for the appropriate time and racial/ethnic group as the denominator of the rates. Because most newborns were white or black, we restricted our assessment of racial variation to these 2 groups.

We used SAS Software (SAS, Cary, NC) for the statistical analyses. We used the Cochran-Armitage test[14,15] to analyze time trends and generate the associated P values. The time trends figures are based on 5-year moving averages. Time trend statistics were computed on actual yearly data.

### RESULTS

#### Overall Findings

From 1968 through 1997, MACDP registered 5813 cases of major congenital heart defects among 937 195 infants (Table 1), for an overall prevalence of 62.0 per 10 000 births (.6%). Of these cases, 97% were among liveborn infants and 2% were among stillborn infants. The total count also included 21 pregnancy terminations and 2 cases of unknown pregnancy outcome.

Most defect types were reported in similar propor-

tions among liveborn and stillborn infants, although atrial septal defects seemed to be more common in the latter group (Table 1). Overall, ventricular septal defects accounted for 27% of all cases, and atrial septal defects and patent ductus arteriosus for an additional 18%. Among the more severe defects, tetralogy of Fallot and transposition of the great arteries accounted for ~10% of cases.

Atrioventricular septal defects associated with Down syndrome accounted for 2.4% of all cases. Conversely, over half of all cases of atrioventricular septal defects (56%) occurred in infants with Down syndrome. Among these infants, atrioventricular septal defects accounted for ~40% of all cases (140/359) of heart defects (data not shown).

Pulmonary hypertension in the newborn was reported in a small fraction of cases of heart defects, including atrial septal defects (1.9%) and patent ductus arteriosus (2.5%). Diaphragmatic hernia accounted for <10% of cases of pulmonary hypertension associated with either atrial septal defects or patent ductus arteriosus.

### Temporal Variations

The overall occurrence rate of heart defects in the last 3 years of the study (1995–1997) was 90.2 per 10 000 (0.9%), an increase of 58.7 per 10 000 above the rate at the beginning of the study (1968–1972; Table 1). Ventricular septal defects, atrial septal defects, and peripheral pulmonic stenosis accounted for approximately half of such increase (Table 1). However, the rate of severe defects, such as tetralogy of Fallot, double-outlet right ventricle, and atrioventricular septal defects also increased significantly and accounted for ~11% of the overall increase. Among infants with Down syndrome, we noted an increase in the rate of both atrioventricular septal defects (from 0.7 to 2.4 per 10 000; Table 1) and of other heart defects (from 1.7 to 4.1 per 10 000; data not shown).

For some defects (eg, atrial and ventricular septal defects, valvular and peripheral pulmonic stenosis) the increases seemed to be continuing through the end of the study (Fig 1). For others (eg, atrioventricular septal defects) the increase seems to have been mostly confined to the 1970s. Other severe defects such as pulmonary atresia with intact ventricular septum and hypoplastic left heart syndrome did not vary appreciably during the study.

Anomalies often thought to share a common origin also showed evidence of heterogeneity (Fig 1). For example, tetralogy of Fallot increased, whereas transposition of the great arteries decreased. This distinction is acknowledged in a proposed pathogenetic classification of heart defects.[16] Moreover, among the defects of the pulmonary outflow, the rate of pulmonic atresia with intact ventricular septum did not increase, whereas the rate of pulmonic stenosis increased 3-fold, and that of peripheral pulmonic stenosis increased >10-fold (Fig 1).

### Racial Variations

The occurrence rate and distribution of heart defects varied between blacks and whites (Table 2). The higher overall occurrence rate for blacks than for whites was primarily driven by a higher occurrence of peripheral pulmonic stenosis and atrial septal defects. Other defects, however, occurred more often among whites; these included some outflow tract defects (transposition of the great arteries, truncus arteriosus) and some left obstructive defects (coarctation of the aorta, aortic stenosis). Racial differences were seen also within some broad defect categories (Table 2). For example, transposition of the great arteries, but not tetralogy of Fallot, was more common among whites. Similarly, among the left-side obstructive defects, coarctation of the aorta, but not hypoplastic left heart, was more common among whites.

Temporal trends also showed racial variations (Fig 2), although the overall rate of heart defects increased among both blacks and whites. However, ventricular septal defects increased more rapidly among whites than among blacks, whereas the reverse was true for peripheral pulmonary stenosis (Fig 2). The rate of coarctation of the aorta was stable among whites, but increased among blacks (P for increase = .036), whereas the rate of Ebstein anomaly was stable among blacks but increased among whites (P = .015).

### Pregnancy Terminations

Pregnancy terminations accounted for a small proportion of heart defects in this study and such proportion changed over time. For example, pregnancy terminations accounted for .03% of cases overall but for 1.3% of cases in 1995 to 1997. This proportion did not vary appreciably by race.

### DISCUSSION

The occurrence rate of major heart defects nearly tripled in metropolitan Atlanta from 1968 to 1997, when major heart defects were reported in 0.9% or 1 in 110 infants. This figure, although higher than previous estimates,[5–7] is probably conservative because it excludes common conditions such as patent ductus arteriosus in preterm infants as well as some cases occurring among pregnancy terminations. The increase in prevalence does not seem to have ceased yet.

Although the increase can be attributed primarily to a few common defects (eg, ventricular and atrial septal defects, peripheral pulmonic stenosis), severe defects such as tetralogy of Fallot and double-outlet right ventricle also seem to be increasing.

These findings must be interpreted in the context of the limitations and strengths of the study. Diagnostic and reporting practices may have changed during the study, improving ascertainment and reducing the average age at diagnosis. Conversely, some cases may have been missed if diagnosed in outpatient clinics or outside the study area. Although the registry collects data on pregnancy terminations, their ascertainment is likely incomplete. For example, we only had data on pregnancy terminations that occurred at 20 gestational weeks or later, and, of these, only diagnoses confirmed after delivery were included. The impact of prenatal diagnosis on heart
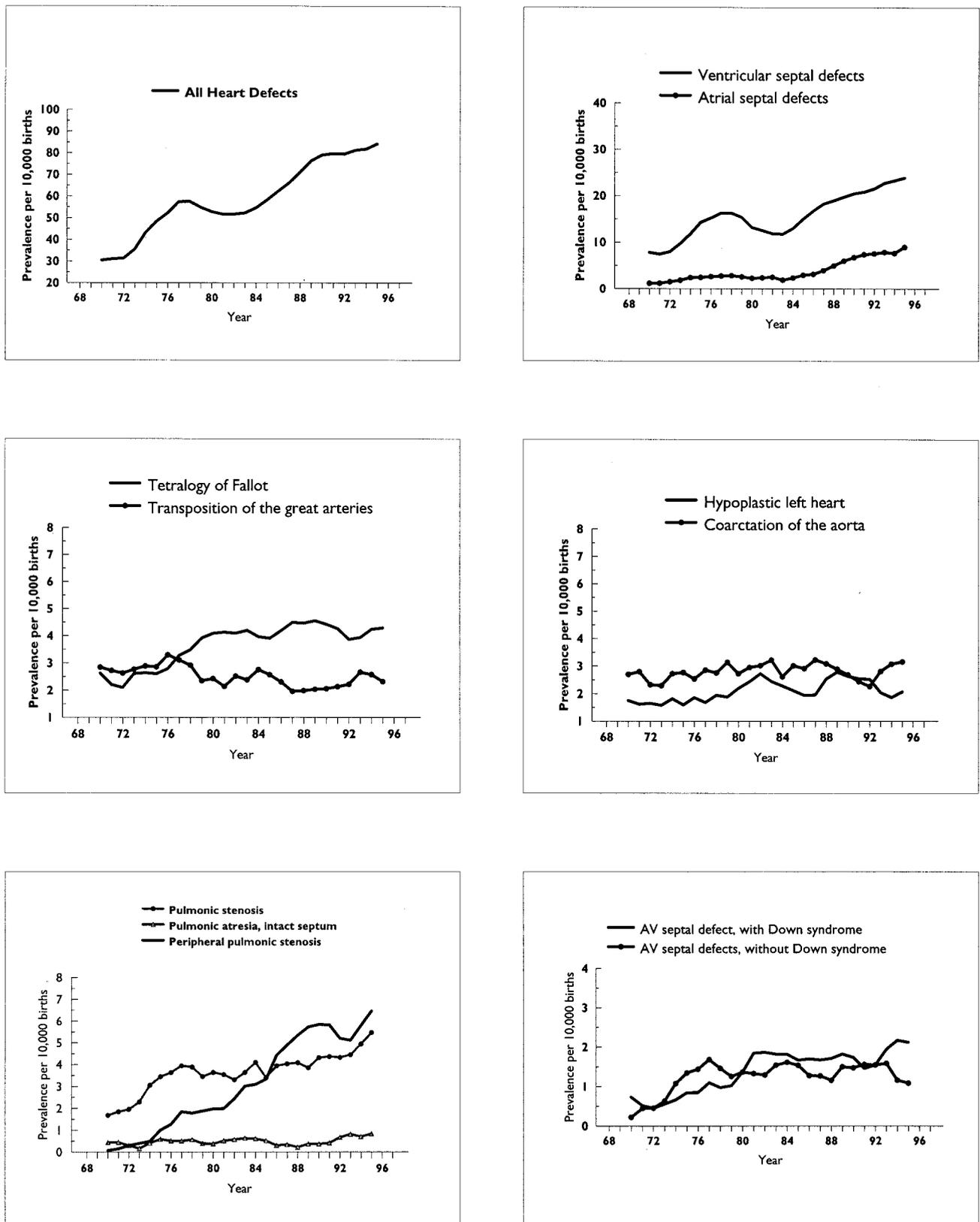
**Fig 1.** Prevalence of heart defects by year, Metropolitan Atlanta, 1968–1997. Prevalence is by 10 000 births and is presented as a 5-year moving average.

defect rates in Atlanta is difficult to estimate. A special study on prenatal diagnosis of heart defects in this same Atlanta population between 1990 and 1994 reported a rapid increase in the fraction of cases of heart defects diagnosed prenatally, so that by 1994 an estimated 12% of heart defects were diagnosed in utero.[17] However, that study also reported that only few such affected pregnancies (8%) were

| Defect Type | Whites | | | Blacks | | | Rate Ratio† | 95% Confidence Interval | P‡ | Rate Difference§ |
|---|---|---|---|---|---|---|---|---|---|---|
| | No. | % | Rate* | No. | % | Rate | | | | |
| Heterotaxias | 39 | 1.2 | 0.71 | 38 | 1.7 | 1.11 | 1.55 | 0.99–2.42 | | 0.4 |
| Corrected (L) transposition | 24 | 0.7 | 0.44 | 11 | 0.5 | 0.32 | 0.73 | 0.36–1.49 | | −0.1 |
| Outflow tract defects | | | | | | | | | | |
|   Tetralogy of Fallot | 209 | 6.4 | 3.82 | 124 | 5.4 | 3.61 | 0.94 | 0.76–1.18 | | −0.2 |
|   D-transposition of the great arteries | 161 | 5.0 | 2.95 | 61 | 2.7 | 1.77 | 0.60 | 0.45–0.81 | *W | −1.2 |
|   Double outlet right ventricle | 58 | 1.8 | 1.06 | 36 | 1.6 | 1.05 | 0.99 | 0.65–1.50 | | 0.0 |
|   Truncus arteriosus | 41 | 1.3 | 0.75 | 14 | 0.6 | 0.41 | 0.54 | 0.30–1.00 | *W | −0.3 |
| Atrioventricular septal defect (atrioventricular canal) | | | | | | | | | | |
|   With Down syndrome | 73 | 2.2 | 1.34 | 62 | 2.7 | 1.80 | 1.35 | 0.96–1.89 | | 0.5 |
|   Without Down syndrome | 61 | 1.9 | 1.12 | 47 | 2.1 | 1.37 | 1.23 | 0.84–1.79 | | 0.3 |
| Total anomalous pulmonary venous return | 39 | 1.2 | 0.71 | 21 | 0.9 | 0.64 | 0.90 | 0.53–1.51 | | −0.1 |
| Ebstein anomaly | 31 | 1.0 | 0.57 | 7 | 0.3 | 0.20 | 0.36 | 0.16–0.82 | *W | −0.4 |
| Right obstructive defects | | | | | | | | | | |
|   Tricuspid atresia | 23 | 0.7 | 0.42 | 9 | 0.4 | 0.23 | 0.55 | 0.25–1.24 | | −0.2 |
|   Pulmonary atresia, intact septum | 29 | 0.9 | 0.53 | 20 | 0.9 | 0.58 | 1.10 | 0.62–1.94 | | 0.1 |
|   Pulmonic stenosis, atresia | 189 | 5.8 | 3.46 | 154 | 6.7 | 4.48 | 1.30 | 1.05–1.60 | | 1.0 |
|   Peripheral pulmonary stenosis | 134 | 4.1 | 2.45 | 184 | 8.0 | 5.35 | 2.18 | 1.75–2.73 | *B | 2.9 |
| Left obstructive defects | | | | | | | | | | |
|   Hypoplastic left heart | 107 | 3.3 | 1.96 | 77 | 3.4 | 2.24 | 1.14 | 0.85–1.53 | | −1.1 |
|   Coarctation of the aorta | 182 | 5.6 | 3.33 | 81 | 3.5 | 2.36 | 0.71 | 0.55–0.92 | *W | 1.8 |
|   Aortic arch atresia or hypoplasia | 33 | 1.0 | 0.60 | 22 | 1.0 | 0.64 | 1.06 | 0.62–1.82 | | −0.7 |
|   Aortic valve stenosis | 72 | 2.2 | 1.32 | 17 | 0.7 | 0.49 | 0.38 | 0.22–0.64 | *W | −16.0 |
| Septal defects | | | | | | | | | | |
|   Ventricular septal defect | 904 | 27.9 | 16.54 | 574 | 25.1 | 16.69 | 1.01 | 0.91–1.12 | | 10.8 |
|   Atrial septal defect | 210 | 6.5 | 3.84 | 200 | 8.7 | 5.82 | 1.51 | 1.25–1.84 | *B | 0.3 |
| Patent ductus arteriosus | 324 | 10.0 | 5.93 | 262 | 11.5 | 7.62 | 1.29 | 1.09–1.51 | *B | −51.7 |
| Other major heart defects | 302 | 9.3 | 5.52 | 266 | 11.6 | 7.74 | 1.40 | 1.19–1.65 | *B | 7.7 |
| Total | 3245 | 100 | 59.35 | 2287 | 100 | 66.51 | 1.12 | 1.06–1.18 | *B | 66.5 |

W indicates excess rate among whites; B, excess rate among blacks.
* Rate per 10 000 births.
† Rate among blacks/rate among whites.
‡ P < .05.
§ Rate among blacks minus rate among whites.

terminated.[17] This is in contrast to other regions in the United States, where the proportion of cases among pregnancy terminations may be higher. For example, a hospital-based study in Boston reported that the proportion of heart defects that were accounted for by pregnancy terminations increased during the late 1970s and the 1980s to ~22% in 1990. The impact of selective pregnancy terminations on prevalence at birth may also vary internationally, particularly for certain heart defects. For example, the proportion of cases of transposition of the great arteries or hypoplastic left heart that are accounted for by pregnancy terminations was as high as 21% and 53%, respectively, in certain regions in France.[18]

Some findings of our study may be driven by underlying diagnostic challenges. For example, the higher proportion of atrial septal defects reported among stillborn infants (Table 1) may be because of the difficulty in distinguishing a true atrial septal defect from a patent foramen ovale among these infants.

The main strength of the study is its use of the MACDP, a registry with active ascertainment of cases from a geographically well-defined population. Moreover, ascertainment relied on multiple sources of information and extended through the first year of life. Such an intensive population-based approach is likely to have minimized underascertainment of cases and improved the representativeness of the findings.

Our findings complement and extend those from previous reports. Investigators in North America and elsewhere noted an increase in specific heart defects, including pulmonic stenosis and ventricular, atrial, and atrioventricular septal defects,[7–9,19,20] during the 1970s and 1980s.[7–9,11,19,21–23] Our findings document a continuation of such trends well into the 1990s.

The underlying causes of these increases are unclear. Increased availability of 2-dimensional and color-Doppler echocardiography services in the community has often been cited as the main reason for such time trends. In fact, investigators of the Baltimore-Washington Infant Study directly documented an increased reporting of heart defects diagnosed by echocardiography but not of those diagnosed by invasive methods.[8] Although we could not conduct a similar analysis, we found that increases were greater for the less severe defects, a finding consistent with improved diagnosis and reporting. For example (Fig 1), the rate for pulmonary atresia, a severe obstruction of the right outflow tract, was relatively stable; that of pulmonic stenosis, a less severe obstruction of the right outflow tract, increased 3-fold; and that of peripheral pulmonary stenosis, the mildest anomaly of the spectrum, increased 10-fold. The major contribution of ventricular septal defects to the
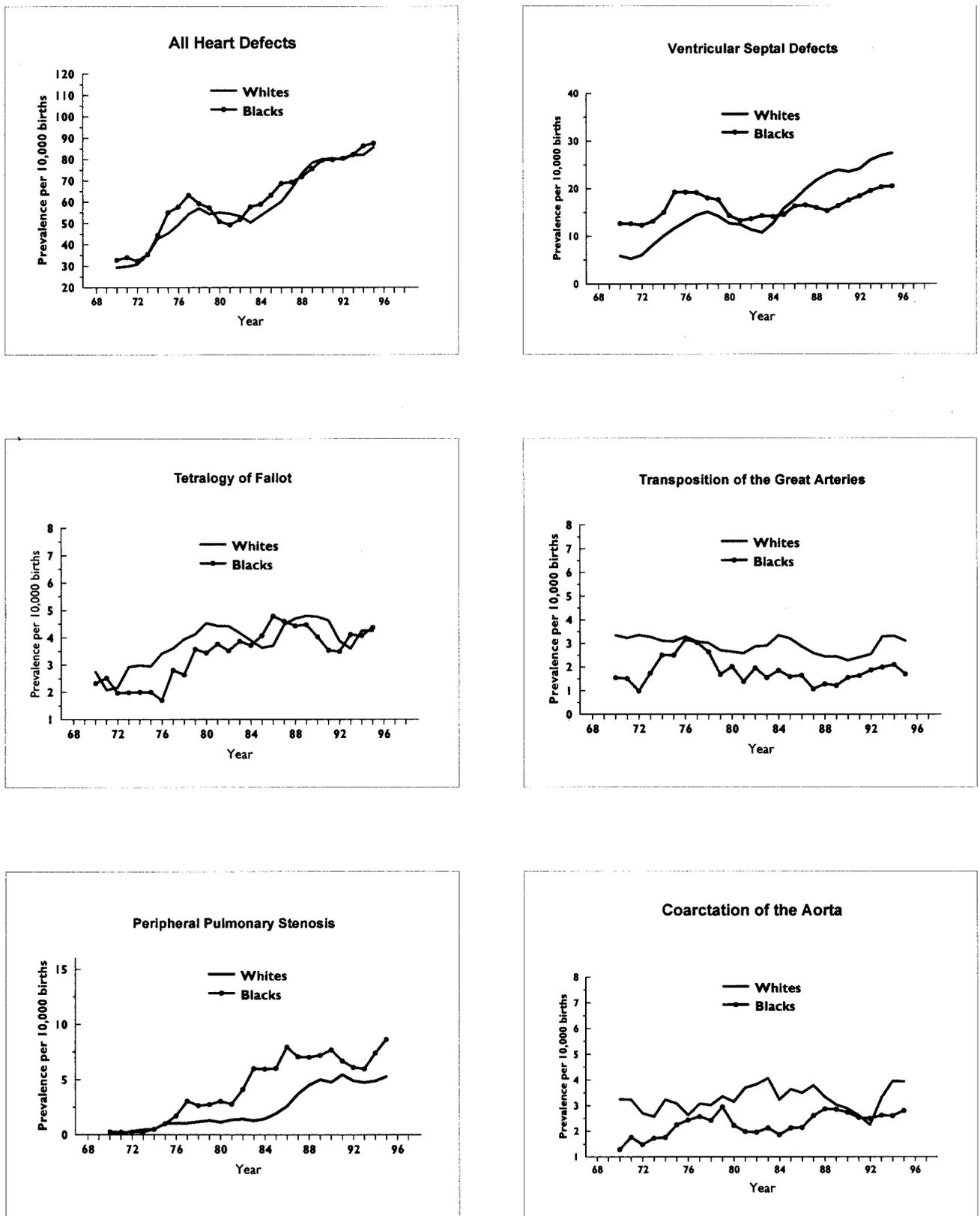
**Fig 2.** Racial variation in the time trends of heart defect occurrence, Metropolitan Atlanta, 1968–1997. Prevalence is by 10 000 births and is presented as a 5-year moving average.

increasing trends is also consistent with improved diagnosis and reporting, a finding that others have noted as well.[8,22] Muscular ventricular defects, in particular, tend to be clinically mild or even asymptomatic and have been detected in as many as 5% of newborns screened by color-Doppler echocardiography.[24] The increased rate of atrioventricular septal defect among infants with Down syndrome in our study is consistent with previous reports[25] and also may reflect a more intensive or earlier cardiologic

evaluation of affected pregnancies, newborns, or young infants.

The racial variations in this study offer additional, if indirect, clues to understanding such time trends. Racial and ethnic differences in the prevalence of specific heart defects are well-documented.[12] Their interpretation in terms of genetic and environmental causation, however, is not straightforward. For example, racial and ethnic variations are often associated with socioeconomic differences, lifestyle variation, cultural factors, and other factors that indicate environmental exposures. We noted an excess of white infants among certain heart defects (eg, transposition of the great arteries, aortic stenosis, coarctation of the aorta). These findings are similar to those reported in a different geographic area[12] and may indicate differences in background risk or genetic susceptibility. In contrast, the similar temporal trends in the overall rate of heart defects among black and white infants (Fig 2, first panel) are consistent with improved reporting.

It is, however, difficult to explain all instances of increased occurrence with improvement in diagnosis and reporting. For example, the timing of the trends is not identical across heart defect types (Fig 1). For ventricular septal defects an increasing trend is evident from the beginning of the study, whereas for atrial septal defects the increase seems to begin later. Likewise, it is also difficult to ascribe solely to diagnostic improvements the increase in severe defects, such as tetralogy of Fallot, or the racial variations in temporal trends (eg, for coarctation of the aorta and ventricular septal defects).

The reason for the possible decrease of transposition of the great arteries is also unclear. Selective pregnancy termination is unlikely to have played a major role.[17] In the study previously discussed from metropolitan Atlanta, only 6% of cases of transposition of the great arteries were diagnosed prenatally, and only 8% of women carrying a fetus with a prenatally diagnosed heart defect terminated the pregnancy.[17]

The findings from this study have some practical implications. First, even if these trends exclusively reflect diagnostic improvements, a high and increasing rate of heart defects among young children and their parents increases the burden on families and health care providers, both in terms of parental anxiety and of the direct costs of diagnostic examinations, follow-up, and possibly treatment. Second, the trends in heart defects magnify the challenge of detecting teratogen-induced epidemics of birth defects. The small increases in rates that are often associated with teratogenic exposures may go undetected if confounded by the larger background trend. Close collaboration between clinical and public health professionals is needed to face this challenge. Finally, much of the temporal and racial variations in heart defect occurrence remain unexplained and underscore the need to study further the genetic and environmental determinants of heart defects. Population-based studies of trends that account for prenatal diagnosis, method of diagnosis, and severity of the lesion may help elucidate the extent to which artifactual and causal factors contribute to the temporal and racial variations in the occurrence of cardiac defects.

## REFERENCES

1. Yoon PW, Olney RS, Khoury MJ, Sappenfield WM, Chavez GF, Taylor D. Contribution of birth defects and genetic diseases to pediatric hospitalizations: a population-based study. *Arch Pediatr Adolesc Med.* 1997;151:1096–1103
2. Yang Q, Khoury MJ, Mannino D. Trends and patterns of mortality associated with birth defects and genetic diseases in the United States, 1979–1992: an analysis of multiple-cause mortality data. *Genet Epidemiol.* 1997;14:493–505
3. Waitzman N, Scheffler R, Romano P. An assessment of total costs and policy implications. In: Waitzman N, Scheffler R, Romano P, eds. *The Cost of Birth Defects: Estimates of the Value of Prevention.* Lanham, MD: University Press of America, Inc; 1996:145–177
4. Ferencz C. On the birth prevalence of congenital heart disease. *J Am Coll Cardiol.* 1990;16:1701–1702
5. Ferencz C, Loffredo CA, Rubin JD, Magee CA. Epidemiology of congenital heart disease: the Baltimore-Washington Infant Study 1981–1989. In: *Perspectives in Pediatric Cardiology.* Mount Kisco, NY: Futura Publishing Company, Inc; 1993
6. Fyler DC. Report of the New England Regional Infant Cardiac Program. *Pediatrics.* 1980;65:375–461
7. Grabitz RG, Joffres MR, Collins-Nakai RL. Congenital heart disease: incidence in the first year of life. The Alberta Heritage Pediatric Cardiology Program. *Am J Epidemiol.* 1988;128:381–388
8. Wilson PD, Correa-Villasenor A, Loffredo CA, Ferencz C. Temporal trends in prevalence of cardiovascular malformations in Maryland and the District of Columbia, 1981–1988. The Baltimore-Washington Infant Study Group. *Epidemiology.* 1993;4:259–265
9. Layde PM, Dooley K, Erickson JD, Edmonds LD. Is there an epidemic of ventricular septal defects in the USA? *Lancet.* 1980;1:407–408
10. Lin AE, Herring AH, Amstutz KS, et al. Cardiovascular malformations: changes in prevalence and birth status, 1972–1990. *Am J Med Genet.* 1999;84:102–110
11. Johnson KC, Rouleau J. Temporal trends in Canadian birth defects birth prevalences, 1979–1993. *Can J Public Health.* 1997;88:169–176
12. Correa-Villasenor A, McCarter R, Downing J, Ferencz C. White-black differences in cardiovascular malformations in infancy and socioeconomic factors. The Baltimore-Washington Infant Study Group. *Am J Epidemiol.* 1991;134:393–402
13. Ferencz C, Loffredo CA, Correa-Villasenor A, Magee CA, eds. Categorization of cardiovascular malformations for risk factors analysis. In: *Genetic and Environmental Risk Factors for Major Cardiovascular Malformations.* 1st ed. Mount Kisco, NY: Futura Publishing Company, Inc; 1997:13–28
14. Armitage P. Test for linear trend in proportions and frequencies. *Biometrics.* 1955;11:375–386
15. Cochran WG. Some methods for strengthening the common chi-square tests. *Biometrics.* 1954;10:417–454
16. Clark EB. Pathogenetic mechanisms of congenital cardiovascular malformations revisited. *Semin Perinatol.* 1996;20:465–472
17. Montana E, Khoury MJ, Cragan JD, Sharma S, Dhar P, Fyfe D. Trends and outcomes after prenatal diagnosis of congenital cardiac malformations by fetal echocardiography in a well defined birth population, Atlanta, Georgia, 1990–1994. *J Am Coll Cardiol.* 1996;28:1805–1809
18. International Center for Birth Defects, World Health Organization, EUROCAT. *World Atlas of Birth Defects.* Geneva, Switzerland: World Health Organization; 1998:52–59
19. Fixler DE, Pastor P, Chamberlin M, Sigman E, Eifler CW. Trends in congenital heart disease in Dallas County births, 1971–1984. *Circulation.* 1990;81:137–142
20. Perry LW, Neill CA, Ferencz C, Rubin JD, Loffredo CA. Infants with congenital heart disease: the cases. In: Ferencz C, Rubin JD, Loffredo

CA, Magee CA, eds. *Epidemiology of Congenital Heart Disease*. 1st ed. Mount Kisco, NY: Futura Publishing Company, Inc; 1993:33–62

21. Martin GR, Perry LW, Ferencz C. Increased prevalence of ventricular septal defect: epidemic or improved diagnosis. *Pediatrics*. 1989;83: 200–203

22. Grech V. Epidemiology and diagnosis of ventricular septal defect in Malta. *Cardiol Young*. 1998;8:329–336

23. Allan LD, Cook A, Sullivan I, Sharland GK. Hypoplastic left heart syndrome: effects of fetal echocardiography on birth prevalence. *Lancet*. 1991;337:959–961

24. Roguin N, Du ZD, Barak M, Nasser N, Hershkowitz S, Milgram E. High prevalence of muscular ventricular septal defect in neonates. *J Am Coll Cardiol*. 1995;26:1545–1548

25. Khoury MJ, Erickson JD. Improved ascertainment of cardiovascular malformations in infants with Down's syndrome, Atlanta, 1968 through 1989: implications for the interpretation of increasing rates of cardio-vascular malformations in surveillance systems. *Am J Epidemiol*. 1992; 136:1457–1464

## Racial and Temporal Variations in the Prevalence of Heart Defects

Lorenzo D. Botto, Adolfo Correa and J. David Erickson

| | |
|---|---|
| **Updated Information & Services** | including high resolution figures, can be found at: http://pediatrics.aappublications.org/content/107/3/e32 |
| **References** | This article cites 20 articles, 2 of which you can access for free at: http://pediatrics.aappublications.org/content/107/3/e32#BIBL |
| **Subspecialty Collections** | This article, along with others on similar topics, appears in the following collection(s): **Cardiology** http://www.aappublications.org/cgi/collection/cardiology_sub |
| **Permissions & Licensing** | Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: http://www.aappublications.org/site/misc/Permissions.xhtml |
| **Reprints** | Information about ordering reprints can be found online: http://www.aappublications.org/site/misc/reprints.xhtml |

**American Academy of Pediatrics**

DEDICATED TO THE HEALTH OF ALL CHILDREN™

# PEDIATRICS®

OFFICIAL JOURNAL OF THE AMERICAN ACADEMY OF PEDIATRICS

**Racial and Temporal Variations in the Prevalence of Heart Defects**

Lorenzo D. Botto, Adolfo Correa and J. David Erickson

The online version of this article, along with updated information and services, is
located on the World Wide Web at:

American Academy of Pediatrics

DEDICATED TO THE HEALTH OF ALL CHILDREN™