Congenital Heart Disease in the General Population
Changing Prevalence and Age Distribution

Ariane J. Marelli, MD; Andrew S. Mackie, MD, SM; Raluca Ionescu-Ittu, MSc; Elham Rahme, PhD; Louise Pilote, MD, MPH, PhD

Background—Empirical data on the changing epidemiology of congenital heart disease (CHD) are scant. We determined the prevalence, age distribution, and proportion of adults and children with severe and other forms of CHD in the general population from 1985 to 2000.

Methods and Results—Where healthcare access is universal, we used administrative databases that systematically recorded all diagnoses and claims. Diagnostic codes conformed to the International Classification of Disease, ninth revision. Severe CHD was defined as tetralogy of Fallot, truncus arteriosus, transposition complexes, endocardial cushion defects, and univentricular heart. Prevalence of severe and other CHD lesions was determined in 1985, 1990, 1995, and 2000 using population numbers in Quebec. Children were subjects <18 years of age. The prevalence was 4.09 per 1000 adults in the year 2000 for all CHD and 0.38 per 1000 (9%) for those with severe lesions. Female subjects accounted for 57% of the adult CHD population. The median age of all patients with severe CHD was 11 years (interquartile range, 4 to 22 years) in 1985 and 17 years (interquartile range, 10 to 28 years) in 2000 (P<0.0001). The prevalence of severe CHD increased from 1985 to 2000, but the increase in adults was significantly higher than that observed in children. In the year 2000, 49% of those alive with severe CHD were adults.

Conclusions—The prevalence in adults and median age of patients with severe CHD increased in the general population from 1985 to 2000. In 2000, there were nearly equal numbers of adults and children with severe CHD. (Circulation. 2007;115:163-172.)

Key Words: adults ■ children ■ epidemiology ■ heart defects, congenital ■ prevalence

The reported prevalence of congenital heart disease (CHD) varies between 4 and 10 per 1000 live births.1-5 Advances in pediatric cardiac care have resulted in an increasing number of adults with CHD being followed up in tertiary care centers.1,6,7 These data have been important in generating interest in adult CHD as a new subspecialty of cardiology.6,7

Clinical Perspective p 172

Although estimates of the prevalence of CHD in adults have been generated,2,8 no empirical data are available on the epidemiology of CHD in the general adult population. To date, no studies have documented the prevalence of CHD in adults and children in the same population. Such data are important determinants of how healthcare resources should be allocated to patients with CHD.

Where access to health care is universal, diagnoses are recorded for the population at large.9 For congenital cardiac lesions, we used administrative databases in a geographically focused region to determine the prevalence of CHD. Our goals were to measure the observed prevalence, age, and proportions of adults relative to children with CHD in the general population from 1985 to 2000.

Data Acquisition

In Quebec (Canada), every individual is assigned a unique Medicare number in the first year of life. All diagnoses and health services rendered are systematically recorded until death. Demographic records on all patients include the date of birth and death. Two administrative databases were used: the physician’s services and claims database (Régie de l’Assurance Maladie du Québec) from 1983 to 2000 and the hospital discharge summary database (Med-Echo) from 1987 to 2000. Through the use of encrypted numbers, both databases were linked for each patient. Diagnoses conform to the International Classification of Disease, ninth revision (ICD-9), which has 24 codes to designate CHD anomalies. We created a population-based CHD database containing data files of all patients diagnosed with CHD who came into contact with the healthcare system in Quebec from January 1, 1983, to December 31, 2000 (inclusive). At every point in time, patients were divided on the basis of age; children were <18 years of age, and adults were ≥18 years of age.

Permission for the study was obtained from the McGill University Health Center ethics board and government agencies responsible for privacy of access to information in Quebec.

Diagnostic Codes and Classification of Disease

Imitating the Report of the New England Regional Infant Cardiac Program, we established a hierarchy of CHD diagnoses and surgical
TABLE 1. Hierarchy of CHD Diagnostic Codes

<table>
<thead>
<tr>
<th>Categorical Hierarchy Block</th>
<th>Categorical Diagnosis</th>
<th>ICD-9 Codes</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>AVCD</td>
<td>7456</td>
</tr>
<tr>
<td></td>
<td>TOF</td>
<td>7452</td>
</tr>
<tr>
<td></td>
<td>Univentricular heart</td>
<td>7453</td>
</tr>
<tr>
<td></td>
<td>Transposition complex*</td>
<td>7451</td>
</tr>
<tr>
<td></td>
<td>Truncus arteriosus</td>
<td>7450</td>
</tr>
<tr>
<td></td>
<td>Hypoplastic left heart syndrome</td>
<td>7467</td>
</tr>
<tr>
<td>2</td>
<td>ASD</td>
<td>7455</td>
</tr>
<tr>
<td></td>
<td>VSD</td>
<td>7454</td>
</tr>
<tr>
<td></td>
<td>PDA</td>
<td>7470</td>
</tr>
<tr>
<td></td>
<td>Aortic coarctation</td>
<td>7471</td>
</tr>
<tr>
<td></td>
<td>Ebstein’s anomaly</td>
<td>7462</td>
</tr>
<tr>
<td>3</td>
<td>Unspecified defect of septal closure</td>
<td>7459</td>
</tr>
<tr>
<td>4</td>
<td>Anomalies of the pulmonary artery</td>
<td>7473</td>
</tr>
<tr>
<td></td>
<td>Anomalies of the pulmonary valve</td>
<td>7460</td>
</tr>
<tr>
<td></td>
<td>Congenital tricuspid valve disease</td>
<td>7461</td>
</tr>
<tr>
<td></td>
<td>Congenital aortic stenosis</td>
<td>7463</td>
</tr>
<tr>
<td></td>
<td>Congenital aortic insufficiency</td>
<td>7464</td>
</tr>
<tr>
<td></td>
<td>Congenital mitral stenosis</td>
<td>7465</td>
</tr>
<tr>
<td></td>
<td>Congenital mitral insufficiency</td>
<td>7466</td>
</tr>
<tr>
<td></td>
<td>Anomalies of great veins</td>
<td>7474</td>
</tr>
<tr>
<td>5</td>
<td>Other unspecified anomalies of the heart</td>
<td>7468/9</td>
</tr>
<tr>
<td></td>
<td>Other unspecified anomalies of circulation</td>
<td>7479</td>
</tr>
<tr>
<td></td>
<td>Other unspecified anomalies of the aorta</td>
<td>7472</td>
</tr>
</tbody>
</table>

*Complete or congenitally corrected transposition.

interventions for patients with several diagnostic and surgical codes.5 We classified the 24 ICD-9 diagnostic codes in hierarchical blocks 1 through 5 (Table 1). We designated as specific CHD procedures those from which a CHD diagnosis could be inferred (Table 2). For each diagnosis made and/or procedure (invasive and noninvasive) performed, physicians were classified by specialty as a cardiovascular (CV) specialist (pediatric, medical or surgical), primary care physician (general or family physician, pediatrician or internist), or echocardiographer.

Two categories of CHD severity were defined. We classified as severe those lesions with an ICD-9 diagnosis that reflected the highest probability of being associated with cyanosis or requiring surgical intervention early in life. These included all patients with a block 1 diagnosis (Table 1). All remaining lesions were classified as other CHD.

TABLE 2. Hierarchy of CHD Surgical Codes

<table>
<thead>
<tr>
<th>Surgical Procedures Billed by a Cardiovascular Surgeon</th>
<th>Inferred CHD Diagnosis</th>
</tr>
</thead>
<tbody>
<tr>
<td>Atrioventricular canal repair</td>
<td>AVCD</td>
</tr>
<tr>
<td>Atrial switch</td>
<td>Transposition complex</td>
</tr>
<tr>
<td>Arterial switch</td>
<td>Transposition complex</td>
</tr>
<tr>
<td>Blalock-Hanlon procedure</td>
<td>Transposition complex</td>
</tr>
<tr>
<td>Coarctation repair</td>
<td>Aortic coarctation</td>
</tr>
<tr>
<td>Fontan procedure</td>
<td>Univentricular heart</td>
</tr>
<tr>
<td>Fallot repair</td>
<td>TOF</td>
</tr>
<tr>
<td>Patent ductus ligation</td>
<td>PDA</td>
</tr>
<tr>
<td>Shunt closure</td>
<td>Unspecified defect of septal closure</td>
</tr>
</tbody>
</table>

Inclusion and Exclusion Criteria

We included patients who had at least 1 ICD-9 CHD code and/or a surgical procedure specific to these codes between 1983 and 2000. Diagnostic codes were cross-referenced between the hospital discharge summary database, those found in the physician’s services and claims database, and those inferred from the surgical procedure. When >1 ICD-9 code was recorded, we chose the most frequent diagnosis in the diagnostic hierarchical block with the lowest numerical value (Table 1), giving priority to the diagnosis made by a CV specialist. When diagnoses were discrepant between databases, the records were manually reviewed by 1 pediatric cardiologist (A.S.M.) and 1 adult CHD specialist (A.J.M.).

We excluded patients who had severe (block 1) or nonspecific (block 5) CHD codes with a diagnosis that was not made by a CV specialist and who had not been hospitalized for CHD (Table 1). We excluded patients with diagnoses in blocks 2 through 4 that had not been made by a primary care physician, CV specialist, or echocardiographer (Table 1). We also excluded patients in whom a CHD procedure had been claimed by a physician other than a CV surgeon without an ICD-9 CHD diagnosis (Table 2).

Statistical Analysis

Prevalence is defined as the proportion of individuals in a population who have a disease at a specific point in time.10 We measured the observed prevalence of CHD corresponding to the number of patients observable in our database between 1983 and 2000 by way of an ICD-9 CHD diagnosis in a given year divided by the Quebec population in that year. Because CHD is present since birth, a patient contributed to the prevalence of all years he or she was alive regardless of when the diagnosis was made during that person’s life. When a patient died, he or she no longer contributed to the prevalence.

Absolute numbers, proportions, and prevalence of adults and children are reported on the basis of the subject’s age for the years 1985, 1990, 1995, and 2000. For age- and sex-related calculations, both the CHD and Quebec populations had the same age and sex distribution. In 2000, the total population in Quebec was 7 357 029, of whom 5 760 295 were adults.11 Prevalence reported among children and adults for both severe and other CHD represents weighted averages of prevalence in more specific age groups and CHD lesions. To investigate trends in subgroups, we measured CHD prevalence in age groups of 1 to 12, 13 to 17, 18 to 25, 26 to 40, and >40 years and in atrial septal defect (ASD), ventricular septal defect (VSD), endocardial cushion defect (AVCD), and tetralogy of Fallot (TOF).

Poisson bivariate models were used to quantify the change in CHD prevalence from 1985 to 2000 and differences between males and females. Changes in prevalence were measured as prevalence ratios and 95% CIs. Age and sex distributions of different cohorts were summarized using medians (interquartile range [IQR]) and proportions. Medians and proportions in different CHD cohorts were compared through the use of Wilcoxon and χ² tests.

To investigate the likelihood of misclassification of patients with unspecified defects, we compared subjects with diagnostic codes in block 5 with those having severe CHD (block 1) and those with diagnoses in blocks 2 through 4 (Table 1). We performed 2 logistic regression models using cardiac hospitalization and CHD specific surgery from 1983 to 2000 as dependent variables and age, sex, and duration of follow-up as covariates. Results are reported using odds ratios (ORs) and 95% CIs. Analyses were performed with SAS statistical software (version 8.02, Cary, NC).

The authors had full access to the data and take full responsibility for their integrity. All authors have read and agree to the manuscript.

Results

Derivation of Patient Cohort

Healthcare records were obtained from the administrative databases on 61 386 patients. Of the 45 960 patients included in our CHD database, 32 481 (71%) had a unique ICD-9
In 2000, 42,542 patients were alive: 18,979 children (45%) and 23,563 adults (55%). Of these, we included 11,207 patients with unspecified CHD codes (block 5) as patients with “other” CHD. The patients with unspecified CHD codes had diagnoses made by a CV specialist or had been hospitalized with a CHD diagnosis. Comparing these patients with the 4,521 patients classified as severe CHD (block 1) shows ORs and 95% CIs as follows: cardiac hospitalization, 0.20 (95% CI, 0.18 to 0.23); and CHD-specific surgery, 0.02 (95% CI, 0.02 to 0.03). Comparing these patients with the 26,814 patients with diagnoses in blocks 2 through 4 shows the following ORs and 95% CIs: cardiac hospitalization, 0.57 (95% CI, 0.53 to 0.60); and CHD-specific surgery, 0.04 (95% CI, 0.03 to 0.05). Therefore, patients with unspecified defects had cardiac hospitalizations and CHD rates that were 80% and 98% lower, respectively, than those with severe CHD, indicating that misclassification was unlikely.

We excluded 15,426 (25%) patients whose median year of birth was 1945 (IQR, 1925 to 1967). Of these, 14,285 (93%) had never been hospitalized for CHD and had a diagnosis in block 1 or 5 never made by a CV specialist or a diagnosis in blocks 2 through 4 never made by a CV specialist, primary care physician, or echocardiographer (Table 1). We also excluded 1,141 patients (7%) who had a CHD surgical procedure billed by a non-CV surgeon (Table 2). The numbers of excluded patients in each diagnostic block are summarized in Figure 1.

Prevalence of CHD
In 2000, the prevalence of CHD was 11.89 per 1000 children, 4.09 per 1000 adults, and 5.78 per 1000 in the general population. The prevalence of severe CHD was 1.45 per 1000 children and 0.38 per 1000 adults, accounting for 12% and 9% of all CHD lesions in children and adults, respectively (Table 3). In children and adults, conotruncal anomalies and AVCD were the most prevalent lesions among those with severe lesions, whereas ASD, VSD, and patent ductus arteriosus (PDA) were the most common lesions among those with other CHD (Table 3).

Sex Distribution of CHD
In 2000, females accounted for 52% and 57% of the CHD population in children and adults, respectively. The proportion of females in the CHD population was significantly higher than that of the Quebec population: 57% versus 51% in adults (P<0.0001) and 52% versus 49% in children (P<0.0001). The prevalence of all CHD in adults was significantly higher in females than in males (4.55 per 1000 females versus 3.61 per 1000 males; P<0.0001). In those with severe disease, the prevalence was higher in female adults (0.41 per 1000 versus 0.35 per 1000; P=0.0001) but
not in female children. Shunt lesions (ASD, VSD, PDA, and AVCD) were more common in female adults (2.13 per 1000 versus 1.46 per 1000, \(P<0.0001\)) and children (9.95 per 1000 versus 7.92 per 1000; \(P<0.0001\)). Transposition complexes and coarctation were more common in males in adults (0.05 per 1000 versus 0.03 per 1000, \(P=0.01\); 0.08 per 1000 versus 0.05 per 1000, \(P<0.0001\), respectively) and children (0.31 per 1000 versus 0.22 per 1000, \(P=0.0002\); 0.30 per 1000 versus 0.19 per 1000, \(P<0.0001\), respectively).

In 1985, CHD of all ages was also more prevalent in females than in males (4.83 per 1000 versus 3.94 per 1000; \(P<0.0001\)). For severe lesions, the prevalence was higher in female adults (0.25 per 1000 versus 0.16 per 1000; \(P<0.0001\)) but not in female children. Shunts, including ASD, VSD, PDA, and AVCD, were more common in female adults (2.13 per 1000 versus 1.46 per 1000, \(P<0.0001\)) and children (9.95 per 1000 versus 7.92 per 1000; \(P<0.0001\)). Transposition complexes and coarctation were more common in males in adults (0.05 per 1000 versus 0.03 per 1000, \(P=0.01\); 0.08 per 1000 versus 0.05 per 1000, \(P<0.0001\), respectively) and children (0.31 per 1000 versus 0.22 per 1000, \(P=0.0002\); 0.30 per 1000 versus 0.19 per 1000, \(P<0.0001\), respectively).

Changing Age Distribution of Adults and Children With CHD

The median age of all patients with severe CHD was 11 years (IQR, 4 to 22 years) in 1985 and 17 years (IQR, 10 to 28 years) in 2000 (\(P<0.0001\)). Among adults in the year 2000, the median age of the CHD population was 40 years (IQR, 27 to 60 years) with a median age of 29 years (IQR, 22 to 39 years) in those with severe disease versus 42 years (IQR, 28 to 62 years) in those with other CHD.

Changing Proportions of Adults and Children With CHD

When all CHD lesions were considered, there have been more adults than children with CHD since 1985 (Figure 2A). For severe CHD, the ratio of adults to children increased so that in 2000, 49% (95% CI, 47 to 50) of patients with severe lesions were adults (Figure 2B).

Changing Prevalence of CHD in Adults and Children

The prevalence of all CHD increased from 1985 to 2000 in both adults and children (Figure 3A). However, the prevalence of severe CHD in adults increased by 85% (prevalence ratio for year 2000 versus year 1985, 1.85; 95% CI, 1.72 to
2.00) from 0.21 to 0.38 per 1000 adults, whereas the prevalence of severe disease in children increased by only 22% (prevalence ratio for year 2000 versus year 1985, 1.22; 95% CI, 1.15 to 1.30) from 1.19 to 1.45 per 1000 children over the same time period (Figure 3B). The highest increase in prevalence occurred among adolescents 13 to 17 years of age, followed by adults in the age group of 18 to 40 years (Figure 4).

The prevalence of ASD increased at higher rates in children compared with adults (Figure 5A), whereas that of AVCD and TOF increased at higher rates in adults compared with children (Figure 5B). The prevalence of VSD increased at the same rate in both children and adults. The highest increase in prevalence was among children with ASD and adults with AVCD, with a 2-fold increase in prevalence from 1985 to 2000 (prevalence for children with ASD, 3.3; 95% CI, 3.1 to 3.4; prevalence for adults with AVCD, 2.5; 95% CI, 2.2 to 2.8) (Figure 5).

Discussion
We report a population-based prevalence of CHD in children observed for up to 18 years of life: 11.89 per 1000 for all CHD and 1.45 per 1000 for severe disease in the year 2000. Previous studies documented the prevalence of CHD among infants using surveillance registries.\textsuperscript{3-5,12} Reported rates varied from 4 to 10 per 1000 live births.\textsuperscript{3,4} For severe CHD, the New England Infant Cardiac Registry reported a prevalence of 1.5 per 1000.\textsuperscript{5} Referral-based participation may result in overascertainment of severe disease and underascertainment of mild lesions more likely to become manifest after the first year of life. Not surprisingly, the population-based prevalence of CHD in children in this study is higher than previously found. For severe CHD, which is expected to become manifest in the first year of life regardless of the ascertainment method, our results for children are in line with the findings of others.\textsuperscript{5}

The prevalence of CHD in adults has previously been estimated\textsuperscript{1,2,8} but not measured. For the year 2000, we determined a population prevalence of 4.09 per 1000 for all CHD in adults. If we assume that the 15,425 patients excluded from our sample represent a margin of error, including them increases the overall prevalence of CHD in adults to 6.02 per 1000. Extrapolating a prevalence of 4.09 per 1000 to a Canadian population of 24 million adults\textsuperscript{11} and a US population of 209 million adults\textsuperscript{13} corresponds to \( \approx 96,000 \) patients in Canada and \( \approx 856,000 \) patients in the United States. Estimates of the number of adults with CHD have been generated using prevalence at birth and estimated

\[ \text{CHD} = \text{congenital heart disease} \]

Figure 2. Numbers and proportion of adults and children with all CHD (A) and severe CHD (B) in 1985, 1990, 1995, and 2000.
changes in survival.\textsuperscript{1,2,8} For the US population, the range of estimates varies from 787 800\textsuperscript{1} to 1.3 million.\textsuperscript{8} Therefore, our findings in terms of overall numbers of adults with CHD are in line with published estimates.\textsuperscript{1,2,8}

The prevalence of severe CHD in a general population of adults was 0.38 per 1000 or 9\% of the total in this study. Without exclusion of the 15 425 patients, the prevalence of adults with severe CHD in our population increases from 0.38 per 1000 to 0.53 per 1000 in 2000, and the proportion of subjects with severe CHD remains constant at 0.53 of 6.02 (9\%). Extrapolating a prevalence of 0.38 per 1000 corresponds to ~9000 patients in Canada and ~80 000 patients in the United States who in the year 2000 can be expected to have had severe CHD. Based on requirements for expert care,\textsuperscript{1} the range of published estimated number of adults with complex CHD in the United States in 2000 varied from 117 000 to 180 000, depending on the denominator used.\textsuperscript{1,8}

The proportion of adults with complex CHD\textsuperscript{1} varied from 14\% to 16\% when survival estimates of referred patients in the first year of life were used.\textsuperscript{1,2,8} Approximating the

Figure 3. Change in CHD prevalence of children and adults from 1985 to 2000 among patients with all CHD (A) and severe CHD (B).

Figure 4. Change in prevalence of CHD from 1985 to 2000 among patients of different age groups.
combination of lesions that we have in common with the categories of “complex” and “moderate” disease used at the 32nd Bethesda Conference (TOF, truncus arteriosus, AVCD, transposition of the great arteries, univentricular heart, aortic coarctation, Ebstein’s anomaly, PDA, pulmonary artery and pulmonary vein anomalies, VSD) amounts to a prevalence of 1.38 out of 4.09, or 34% of adult patients in our study. Published estimates on the combined proportion of adults with complex and moderate CHD vary: 31%, 44%, and 53%, with a wide range of denominators. Our population prevalence rates for severe and moderate CHD are therefore in the lower range of estimates derived from tertiary care centers. The fact that our numbers can be observed in the general population is an indication that adult CHD will increasingly come to general medical attention.

We report the frequency of specific CHD in children and adults in the same population. In previous studies, the 3 most common lesions found in infants were VSD, D-transposition of the great arteries, and TOF. In this study, at all ages, the most common defects were conotruncal anomalies and AVCD for severe CHD and ASD and VSD for other CHD. We found that ASD was the most common congenital lesion in adults. Consistent with what has been published in children, conotruncal anomalies, including TOF, were the most common severe congenital lesions in adults.

New data on age and sex differences in the CHD population are presented. The median age of patients with severe CHD increased from late childhood to late adolescence from 1985 to 2000. In 2000, there were significantly more females in the adult and pediatric CHD populations. For subjects with severe disease, a female predominance was observed in 1985 and 2000 in adults but not in children. Mortality among males has been reported to be 5% greater than in females in a population of high-risk CHD infants. Our results in terms of

![Figure 5](http://circ.ahajournals.org/figure) Change in prevalence of specific lesions in children vs adults from 1985 to 2000 among subjects with septal defects (A) and severe congenital heart lesions (B).
sex differences for specific lesions in children are consistent with prevalence-at-birth studies showing that transposition of the great arteries complexes, aortic coarctation, and lesions of the left ventricular outflow tract are more prevalent in males, whereas septal defects are more prevalent in females. Further studies are required to determine whether sex differences can be demonstrated in mortality rates in CHD patients. The rising prevalence of severe CHD in a predominantly female population, in conjunction with higher transmission rates, may change the incidence of CHD in years ahead.

To the best of our knowledge, this study is the first to document the changing prevalence of adults with CHD in a North American, predominantly white population during a period of major progress in management of CHD. The rise in absolute numbers and prevalence of severe CHD in adults exceeded that observed in children. By the year 2000, adults and children constituted nearly equal proportions of those alive with severe CHD, with an 85% increase in prevalence observed in adults between 1985 and 2000. The largest increase in prevalence was in adolescents and young adults, suggesting that the prevalence of severe CHD has yet to peak in adults.

The rise in observed prevalence of CHD is likely multifactorial. A true rise in the overall number of patients with all CHD may be due to changing birth prevalence rates related to increasing maternal age or exposure to medication during the first trimester of pregnancy. More likely, this finding can be attributed to the development of cardiac ultrasound since the 1980s, maximizing the possibility of detecting less severe CHD. Our observations in prevalence rates of specific lesions are consistent with this supposition. The prevalence of ASD increased at higher rates in children, whereas that of AVCD and TOF increased at higher rates in adults. Ascertainment bias is more likely to affect the detection of ASDs; survival is more likely to affect TOF and AVCD in adulthood. It is unlikely that ascertainment bias would account for the increase in prevalence of severe CHD observed in adults in excess of that observed in children because there is no reason to believe that echocardiography has been more accessible to adults than children. The observed rise in prevalence of VSDs in children and adults is consistent with increasing detection rates at all ages with Doppler interrogation. Studies examining changing birth rates of CHD and the use of cardiac ultrasound are needed to completely understand the rise in observed prevalence of CHD.

The rise in prevalence and numbers of adults with severe CHD in excess of that observed in children supports, although does not prove, that surgical progress has resulted in decreased mortality because patients with severe lesions correspond to the subset of patients most likely to benefit from improved surgical outcomes. The greater survival to adulthood may result in a shift in mortality beyond 18 years of age. Indeed, our data show that the majority of adults in 2000 were children in 1985 (Figure 2b). This suggests that higher mortality rates can be expected in adults with severe CHD than in children. Further studies are needed to clarify how improved survival in childhood will affect adult mortality.

This study has limitations related to the detection of CHD, depending on contact with the healthcare system. We may have excluded subjects who did not require health services from 1983 to 2000. However, for the population at large, the proportion of people using health services in Quebec is high: 80% to 81% per year from 1998 to 2004. Even so, we may have excluded adults with lesions that can be clinically silent such as ASD, congenitally corrected transposition of the great arteries, or Ebstein's anomaly. This may result in the underestimation of the prevalence of overall CHD. The potential for migration into and out of the province of Quebec may have influenced the number of patients receiving health care within this jurisdiction. During the study period, there was a net immigration into the province of an average of 24 000 people per year. If we use the overall prevalence of 5.78 per 1000 for CHD of all ages determined in this study, we may thereby have overestimated the number of patients with CHD by only 0.05%. Our data came from 1 of 13 Canadian provinces and territories. However, the population of Quebec accounts for 23% to 25% of Canada's children and adults, making our sample large enough to strengthen the interpretation of our results.

Although each person born in Quebec is assigned a unique medical identifier within days or weeks of birth, the timing of this designation may take up to 1 year. Therefore, infants who died before obtaining a Medicare number may not be in our database. In Quebec, in the year 2000, 91 infants died with any congenital malformation in the first year of life, the registration of which is a legal requirement (Vital Statistics Act or equivalent legislation). If we assume that all had CHD and that all were missed in our database, including them would increase the prevalence of CHD only from 11.89 to 11.94 per 1000 children in the year 2000. Even so, the prevalence rates of specific lesions that we report in children cannot be directly compared with those previously reported in infants in the first year of life.

We relied on administrative databases, which are prone to misclassification and lack of specificity. We excluded 25% of our initial sample because our data suggested that they had miscoded CHD and we did not want to inaccurately inflate the overall prevalence of CHD. We decided to include 11 207 subjects who had an unspecified CHD with a diagnosis made by a CV specialist or during a hospitalization. Cardiologists may not be familiar with specific ICD-9 CHD codes, and there are no designated codes for certain lesions. In these subjects, no other information was available to specify a CHD diagnosis. Although some patients with unspecified anomalies may have been misclassified in other rather than severe CHD, we demonstrated significantly lower rates of CHD hospitalization and interventions when these subjects were compared with those with severe CHD, justifying our classification. Of the original 61 386 patients, samples of 17 474 data files were subject to manual review by 1 or 2 authors (A.J.M., A.S.M.). Subsets of patients were audited in all categories of subjects, including those excluded, with severe and other CHD, with unspecified defects, hospitalized (operated and unoperated), and outpatients. After the application of inclusion and exclusion criteria, 71% of the subjects in our CHD database had a unique ICD-9 diagnostic code used...
throughout the study period. Despite limitations, the ICD-9 has been used to investigate CHD-related births and mortality in the United States.3,22

We were limited to the ICD-9 CHD codes that were available in the administrative databases. We defined as severe CHD those lesions with a description in the ICD-9 that reflected the highest probability of being associated with cyanosis or requiring surgical intervention early in life. We thought this approach was reasonable because cyanosis has been a longstanding component of the classification of CHD lesions,23–25 cyanotic lesions have been linked to determinants of survival in the first year of life,5 and the surgical procedures targeting these lesions from 1955 to 197126–30 are likely to affect observed CHD prevalence from the 1980s on. Our definition differs from other valuable CHD classifications based on the recommended requirements for expert care.1 Our purpose was to provide empirical data on the observed prevalence of CHD in the general population. Our definition of severe CHD could result in misclassification of lesions classified as complex or moderate in the Bethesda classification1 such as Eisenmenger’s syndrome, mitral or tricuspid atresia, and supravalvar and subvalvar aortic stenoses.1 We also may have misclassified patients with defects having a broad spectrum of severity depending on the age of presentation such as Ebstein’s anomaly, congenital aortic and mitral stenoses, and coarctation of the aorta. We minimized misclassification by examining all available data for a given subject, including inpatient and outpatient codes and surgical codes. We established a hierarchy of diagnoses and surgical interventions imitating what was done for the Report of the New England Regional Infant Cardiac Program,5 dividing the ICD-9 diagnoses into blocks of decreasing specificity. When specific CHD surgical interventions were performed, we inferred a more specific CHD diagnosis. Even so the proportion of patients with severe CHD may have been underestimated.

Conclusions
This population-based study yielded new information on the epidemiology of CHD between 1985 and 2000. The median age of those with severe disease increased. There was a female predominance among adults with severe CHD. Overall, in the year 2000, 1 in 84 children had CHD. In the same year, a population prevalence of 4.09 per 1000 or 1 per 245 adults with CHD was determined, of whom 9% had severe lesions. Prevalence rates of severe CHD increased between 1985 and 2000, but the increase was significantly higher in adults, so that in the year 2000, the numbers of adults and children alive with severe CHD were nearly equal. The results of this study provide empirical data on which recommendations for resource allocation in CHD can be anchored. These data underscore the published estimates and recommendations stating that more specialized care facilities for adults with CHD are necessary to meet the needs of this growing patient population.31 Although studies using administrative databases have limitations, we have used a unique and contemporary application of prevalence determination. Further studies are needed to determine whether the increase in prevalence of adults with severe CHD will level off or continue to rise.

Sources of Funding
Dr Marelli was supported by the Heart and Stroke Foundation of Canada (No. G-02-MA-1137). Dr Mackie was supported by the Fonds de la Recherche en Santé du Québec (6498 and 8232). Dr Rahme was supported by The Arthritis Society (TAS02/0007). Dr Pilote was supported by the Canadian Institutes of Health Research and is a William Dawson Chair of Medicine at McGill University.

Disclosures
None.

References
CLINICAL PERSPECTIVE

This is the first study to provide empirical data on the changing epidemiology of congenital heart disease (CHD) in the general population. Advances in pediatric cardiovascular care have resulted in more adults with CHD being followed up in tertiary care centers. Previous estimates of CHD prevalence in adults have been generated using birth prevalence and estimated survival rates. In this study, administrative databases were used to obtain data on children and adults in the province of Quebec, a large geographic region of Canada with universal access to healthcare services. A total of 45,960 patients with International Classification of Disease, ninth revision, diagnoses of severe and other forms of CHD from 1985 to 2000 were included. The prevalence of CHD was 4.09 per 1000 adults in 2000; 9% had severe lesions with a significant female predominance. This corresponds to 96,000 patients in Canada and 856,000 patients in the United States with CHD in 2000. The median age of all patients with severe CHD increased from 11 years in 1985 to 17 years in 2000, and the prevalence of severe CHD increased more in adults than in children during the study period. In 2000, there were nearly equal numbers of adults and children with severe CHD. This study provides important data documenting the growing number of adults with CHD in the general population. These findings underscore the need for increasing healthcare resource allocation to this emerging population.
Congenital Heart Disease in the General Population: Changing Prevalence and Age Distribution
Ariane J. Marelli, Andrew S. Mackie, Raluca Ionescu-Ittu, Elham Rahme and Louise Pilote

Circulation. 2007;115:163-172; originally published online January 8, 2007;
doi: 10.1161/CIRCULATIONAHA.106.627224
Circulation is published by the American Heart Association, 7272 Greenville Avenue, Dallas, TX 75231
Copyright © 2007 American Heart Association, Inc. All rights reserved.
Print ISSN: 0009-7322. Online ISSN: 1524-4539

The online version of this article, along with updated information and services, is located on the
World Wide Web at:
http://circ.ahajournals.org/content/115/2/163

Data Supplement (unedited) at:
http://circ.ahajournals.org/content/suppl/2007/01/16/CIRCULATIONAHA.106.627224.DC1

Permissions: Requests for permissions to reproduce figures, tables, or portions of articles originally published
in Circulation can be obtained via RightsLink, a service of the Copyright Clearance Center, not the Editorial
Office. Once the online version of the published article for which permission is being requested is located,
click Request Permissions in the middle column of the Web page under Services. Further information about
this process is available in the Permissions and Rights Question and Answer document.

Reprints: Information about reprints can be found online at:
http://www.lww.com/reprints

Subscriptions: Information about subscribing to Circulation is online at:
http://circ.ahajournals.org/subscriptions/