JACC Vol. 16, No. 7
December 1990:1701–2

Editorial Comment

On the Birth Prevalence of Congenital Heart Disease*

CHARLOTTE FERENCZ, MD, MPH, FACC

Baltimore, Maryland

Congenital heart disease due to environmental teratogens. The thalidomide tragedy brought about the sudden realization that "the human embryo was not sequestered in an impervious maternal body where it was shielded from all but genetic harm" (1). Consequent anxiety regarding environmental teratogens triggered an international commitment to malformation surveillance with an organized alertness to possible similar occurrences (2). The malformations chosen as "sentinels" were those most reliably recognized at birth and, until recently (3,4), congenital heart disease was not among them. The reason for this is the difficulty in diagnostic ascertainment: neonatal indicators of a cardiovascular malformation may be very subtle, infants might die after birth without recognition of their heart disease and infants with neonatal distress require special studies in tertiary care centers to resolve the differential diagnosis of cardiac or pulmonary causes. Thus, the "view" from the pediatric cardiology center includes a very selected case group. Epidemiologists who are searching for causes seek a different view and must take into account every potential bias before making comparisons of regional birth prevalences. The study by Mayberry et al. (5) in this issue of the Journal illustrates the magnitude of this problem. The variations and potential changes in the observation time in only four categories of relevant factors indicate the necessary caution.

1. *Population factors*, such as migration, race/ethnicity with cultural socioeconomic and genetic differences including consanguinity may alter predisposition to anomalies, exposure likelihood, access to and use of medical care.

2. *Family factors*, such as maternal age, reproductive history (fetal loss, low birth weight, infant mortality) may alter the medical care sought and received.

3. *Community medical practice* varies in availability, access and cost, suspicion of an anomaly, screening by fetal ultrasound, amniocentesis and ultimately in a physician's decision to seek specialty advice for particular patients at various ages.

4. *Cardiology center practice* has also altered over time in diagnostic definitions, in the availability and use of invasive and noninvasive diagnostic studies and in the definition of the cardiac defects and associated noncardiac anomalies included in the prevalence estimates.

The present study. Although most of these issues were not considered in the study of Mayberry et al. (5), the intention of a pediatric cardiology center to evaluate regional referral differences is a commendable example for all specialty services because it could promote the search for ways to optimize access to the best possible care for all affected infants and children. The comparison of prevalence in the Yuma and Sierra Vista areas is handicapped by small numbers and by the introduction of two-dimensional echocardiographic diagnosis that virtually coincided with the onset of the study period (1983 to 1988). This change in diagnostic method may be responsible for the steeply rising prevalences in both areas shown in their Table 1. Without an assessment of this and other sources of ascertainment bias, a conclusion of different environmental factors could not be supported.

Comparison with other epidemiologic studies. The Baltimore-Washington Infant Study, an epidemiologic investigation of congenital heart disease (6), is referred to by Mayberry et al. (5) and has been quoted by others but the comparisons to other area prevalences were not always appropriate because of differences in ascertainment methods, case definitions including age of diagnosis, diagnostic exclusions and study years of different levels of pediatric cardiology expertise (8–10). Although modeled after the New England Regional Infant Cardiac Program (7), the birth prevalence of congenital heart disease in the Baltimore-Washington Infant Study's first report (6) (1981 to 1982) already exceeded that obtained in New England in 1969 to 1977. Recent studies of other authors that were similar in methodology and timing showed similar results (11,12) but the problems of differential ascertainment were emphasized in each.

With all methods constant in the Baltimore-Washington area, secular changes have yielded new insights into the fragility of prevalence-at-livebirth determinations based on diagnosis from infancy. The increase in congenital heart disease occurrence in 1983 to 1984 was due to improved detection techniques by two-dimensional and Doppler echocardiography (13) and in 1985 to 1986 still more healthy infants were referred for this diagnostic confirmation (14). However, the 6 year results also demonstrated the relative

*Editorials published in *Journal of the American College of Cardiology* reflect the views of the authors and do not necessarily represent the views of JACC or the American College of Cardiology.

From the University of Maryland School of Medicine and The Baltimore-Washington Infant Study, Baltimore, Maryland. This study was supported by the National Institutes of Health, National Heart, Lung, and Blood Institute (R37 HL 25629), Bethesda, Maryland.

Address for reprints: Charlotte Ferencz, MD, MPH, University of Maryland School of Medicine, Department of Epidemiology and Preventive Medicine, Howard Hall–Room 146-D, 660 West Redwood Street, Baltimore, Maryland 21201.

0735-1097/90/$3.50

JACC Vol. 16, No. 7
December 1990:1701–2

stability of prevalence for uniformly diagnosed severe malformations such as transposition of the great arteries and total anomalous pulmonary venous return and a slight downward trend in the prevalence of hypoplastic left heart syndrome and double outlet right ventricle probably due to expanding use of fetal ultrasonography and pregnancy termination. Moreover, the impact of these evolving medical practices was not uniformly distributed. An evaluation of white-black differentials in infant heart disease by socioeconomic factors, soon to be reported by Correa-Villasenor, has revealed the impact of several societal differences that could totally overshadow the subtle effects of teratogens.

Implications. With all these caveats one might ask: "Why measure congenital heart disease prevalence?" As cardiologists, we wish to know of each infant with a cardiovascular malformation. Information that could improve maternal and child health in our communities would constitute adequate justification and heighten awareness of many issues that had previously escaped our attention. Detailed evaluations of birth defect monitoring programs (2,15) provide compelling arguments for such systematized accounting, with periodic evaluations of changes within and across monitoring systems. At the least, they would facilitate the planning and effective conduct of case-control investigations, some based on possible clustering of cases, increases or decreases or stimulated by changes in the environment, such as exposure to nuclear radiation or water contamination (16). The surveillance system implies a state of readiness and a clearer comprehension of community issues than we have ever had in the past. Epidemiologic assessment of cardiovascular malformations, in contrast with that of adult heart disease, is still very underdeveloped. The ongoing improvement and refinement of studies such as that of Mayberry et al. (5) could eventually establish a national network of powerful capability in improving patient care and in recognizing risk factors that, in turn, could lead to the prevention of some cardiovascular malformations.

References

1. Wilson JG. The evolution of teratologic testing. Teratology 1979;20:205–12.
2. Källén B. Epidemiology of Human Reproduction. Boca Raton, Florida: CRC, 1988:74–105.
3. Edmonds LD, Layde PM, James LM, et al. Congenital malformations surveillance: two American systems. Int J Epidemiol 1981;10:247–52.
4. California Birth Defects Monitoring Program. Procedure Manual, vol. 1. Berkeley, CA: California Department of Health Services, 1986:14,28.
5. Mayberry JC, Scott WA, Goldberg SJ. Increased birth prevalence of cardiac defects in Yuma, Arizona. J Am Coll Cardiol 1990;16:1696–700.
6. Ferencz C, Rubin JD, McCarter RJ, et al. Congenital heart disease: prevalence at livebirth. Am J Epidemiol 1985;121:31–6.
7. Fyler DC. Report of the New England Regional Infant Cardiac Program. Pediatrics 1980;65(suppl):375–461.
8. Borman B, Chapman C, Howard K, Buckfield P, Findlay J. Using a national register for the epidemiological study of congenital heart defects. NZ Med J 1987;100:404–6.
9. Stoll C, Alembik Y, Roth MP, Dott B, De Geeter B. Eur J Epidemiol 1989;5:382–91.
10. Fixler DE, Pastor P, Chamberlin M, Sigman E, Eifler CW. Trends in congenital heart disease in Dallas County births, 1971–1984. Circulation 1990;81:137–42.
11. Carlgren LE, Ericson A, Källén B. Monitoring of congenital cardiac defects. Pediatr Cardiol 1987;8:247–56.
12. Grabitz RG, Joffre MR, Collins-Nakai RL. Congenital heart disease: incidence in the first year of life. Am J Epidemiol 1988;128:381–8.
13. Martin GR, Perry LW, Ferencz C. Increased prevalence of ventricular septal defect: epidemic or improved diagnosis. Pediatrics 1989;83:200–3.
14. Ferencz C, Neill CA. Cardiovascular malformations: prevalence at livebirth. In: Freedom RM, Benson LN, Smallhorn JR, eds. Neonatal Heart Disease, chapter 2. New York: Springer-Verlag, 1991.
15. Kallen B, Hay S, Klingberg M. Birth defects monitoring systems. In: Kalter H, ed. Issues and Reviews in Teratology, vol. 2. New York: Plenum, 1984:1–22.
16. Swan SH, Shaw G, Harris JA, Neutra RR. Congenital cardiac anomalies in relation to water contamination, Santa Clara County, California, 1981–1983. Am J Epidemiol 1989;129:885–93.