

Congenital Heart Disease: Present Situation and Future Prospects

María T. Subirana

Sección Cardiopatías Congénitas, Hospital de la Santa Creu i Sant Pau, Barcelona, Spain.

We are all well aware of the important advances made in the realm of cardiology in recent years, but, undoubtedly, they have had a special impact in the realm of congenital heart disease.

Since 1938, when Robert Gross performed the first ligation of a patent ductus arteriosus; 1944, when William Blalock, pressured by Helen Taussig, created the first systemic pulmonary shunt in a child with cyanotic congenital heart disease (tetralogy of Fallot); and 1945, when Gross and Crafoord, working independently, repaired an aortic coarctation, the evolution of the treatment of patients with congenital heart disease has been spectacular. This has given birth to a new population of adolescents and adults with congenital heart defects that are more or less repaired (practically never cured), that requires specialized cardiological monitoring. We are facing a new challenge that does not appear to have an easy solution, especially if we take into account the fact that, in recent years, given that congenital heart disease was basically a pediatric problem, diseases of this type have been concentrated in pediatric services, and most adult cardiologists have forgotten they exist. It is necessary to carry out a far-reaching restructuring of the health care system, involving the creation of units and/or programs devoted to the monitoring and management of these patients, the training of specialists in the field who work closely with cardiologists and pediatric cardiac surgeons and, above all, the provision of cardiology services with the necessary resources to attend to this new task. But, how many cardiologists will it be

necessary to train in this subspecialty? What are the necessary resources and what amounts are needed? What areas should these resources be channeled into?

As became clear at the last Bethesda Conference (32nd Bethesda Conference: Care of the Adult with Congenital Heart Disease),¹ it is essential that we be aware of the extent of the problem and, to achieve this, before making any type of calculations, it is fundamental to determine the number of children with congenital heart disease born each year in a given population. We should know the incidence of congenital heart disease in our own population, not only the overall prevalence, but broken down according to the type and severity of the defect. This will enable us to determine the resources necessary and plan their distribution on the basis of something more than intuition. If we review the literature, we will realize how difficult it is to obtain these data. The values reported in different studies are extraordinarily variable. Early studies found values of four to five per thousand live births, rates that have increased progressively, reaching values of as high as 12 to 14 per thousand live births.² This circumstance can be easily understood, and there are a number of reasons to explain it:

1. Diagnostic capability. The introduction of ultrasound techniques, especially two-dimensional echocardiography, radically changed the diagnostic algorithm for congenital heart defects. These tomographic images made it possible for the first time to identify, noninvasively, anatomical abnormalities that previously required the performance of cardiac catheterization. This undoubtedly increased and improved our diagnostic capabilities, not only with respect to complex lesions, but in anomalies having minor symptomatology or none whatsoever. This is especially important in children, in whom these conditions can easily go undetected. The incidence rates reported up to that time had to be revised.

Now, with the introduction of fetal 2D Doppler echocardiography, we perceive a new horizon in the technological armamentarium for prenatal diagnostics: the detection of cardiac anomalies during the different stages of pregnancy, with the possible impact that this

SEE ARTICLE ON PAGES 1428-34

Correspondence: Dra. M.T. Subirana.
Sección Cardiopatías Congénitas, Hospital de la Santa Creu i Sant Pau.
Avda. S. Antoni M. Claret, 167. 08025 Barcelona. España.
E-mail: msubiranad@santpau.es

can have, both in obstetrics and in perinatal and neonatal monitoring and management, a subject we will address in the next section.

2. The population studied and the diagnostic techniques employed. Logically, if we want accurate incidence rates, the search to identify cases of congenital cardiac anomalies can not be limited to tertiary or referral centers, since that would imply the failure to include patients with the least severe defects.

On the other hand, if the information is obtained retrospectively from the clinical records of pediatric primary care centers, undetected cases will not be included, some because the diagnostic suspicion did not arise in early childhood and others because the proper diagnostic techniques had not been employed.

Moreover, as we mentioned above, we now have access to another diagnostic alternative, fetal echocardiography, which makes it possible to diagnose the cardiac anomaly earlier. Its systematic use should lead to a change in, or a clarification of, the definition of the term "incidence of congenital heart disease." The diagnosis of congenital heart disease in the fetus during the first 22 weeks of gestation, especially if the anomaly is complex, introduces the option of therapeutic abortion. If the "incidence of congenital heart disease" is calculated on the basis of the number of live-birth patients during a given period, it is quite possible that future studies report a decrease in said "incidence"; however, this does not necessarily mean that there are fewer fetuses with congenital heart defects.³ Moreover, the tendency of women to have children at a later age, as well as the progressive increase in the number of women with congenital heart disease seeking obstetrical care, women who, years ago, would have died during childhood, but now live to childbearing age, are factors that influence the rate of recurrence of the different congenital heart defects in their children. If we add the fact that these mothers are not always in the best hemodynamic condition, with the occasional persistence of cyanosis, development of arrhythmias, need for drug treatment with possible teratogenic effects, etc, all these circumstances can lead to the paradox of there being a greater prevalence. Therefore, it is necessary to clarify the terminology and to be aware of the information obtained, since it could be that resources that could be obviated in the treatment of congenital heart disease during childhood should not be eliminated, but be rechanneled toward systems of prevention or toward the fetal stage, in which the evaluation of invasive techniques for the treatment of certain heart defects has already begun.

3. Forms of congenital heart disease included. Not all the studies in which the incidence of congenital cardiac anomalies is assessed include the same types of heart disease. Some authors exclude bicuspid aortic valve with no hemodynamic impact, persistent left superior vena cava draining into coronary sinus, hypertrophic

cardiomyopathy or genetic diseases such as long Q-T syndrome, etc. Mitchell et al⁴ proposed the following definition for congenital heart disease: "an evident structural anomaly of the heart or thoracic great vessels with real or potential functional impact." As we can deduce, the expression "potential function impact" broadens the range of heart diseases to be included. Thus, if we choose to be extremely rigorous, we should not exclude from studies on the incidence of congenital heart disease in the pediatric population certain anomalies such as bicuspid aortic valve, despite the fact that, at the time of the study, they have no easily detectable impact on hemodynamic function. In any case, this is not always easy since it makes it necessary to perform well-designed prospective studies in the search for the diagnosis.

In this issue of REVISTA ESPAÑOLA DE CARDIOLOGÍA, Martínez Olorón et al⁵ publish a study on the incidence of congenital heart disease in Navarra, a Spanish community of 523 563 inhabitants, in which 47 783 infants were born between 1989 and 1998. The data provided by these authors are extremely important, since it may be possible to extrapolate them to other Spanish communities, serving as a reference point for calculating the needs. We wish to stress the important effort made by the authors in the attempt to identify "all" the patients diagnosed as having congenital heart disease. They performed an exhaustive search in all the public and private hospitals and primary care centers in the Community of Navarra and in the hospitals with cardiac surgery outside Navarra that are referral centers to which they send patients whom they consider susceptible to invasive treatment. The incidence they report (8.96 per thousand live births) may be quite close to the real incidence, with 90% of the anomalies detected corresponding to the most frequently observed cardiac malformations. However, we should point out that they have excluded lesions such as nonstenotic bicuspid aortic valve (in the pediatric age group), mitral valve prolapse, and cardiac arrhythmias, which have a clear tendency to evolve and strong possibilities of requiring proper cardiological monitoring as time goes by. Likewise, although this is a matter of little or no practical impact, the report being retrospective, it is highly possible that the study did not include certain anomalies with no evident impact on hemodynamic function, the diagnosis of which involves a detailed search, such as small ductus arteriosus, persistent left superior vena cava, some cases of partial anomalous pulmonary venous drainage, etc.

One finding that should be pointed out, because of its practical implication, is the prevalence of prematurity (15.9%) and low birth weight (22%) recorded among children with congenital heart disease, with the resulting increase in morbidity and in the use of resources allocated to neonatology, as well as the difficulty or impossibility of performing cardiac

surgery, especially corrective surgery or that involving cardiopulmonary bypass, under these circumstances.

Another aspect that, given the current expansion of fetal echocardiography, especially in obstetric high-risk units, we feel that it is worthwhile to mention the percentage of cases, in the population of Navarra studied, in which the use of this technique led to a diagnosis of congenital heart disease. As the authors themselves indicate, in comparison with the findings in other studies,⁶ the percentage of cases detected is low (1.27%), but we feel that it is not possible to draw any conclusions on the basis of this value. The work is retrospective and the data obtained are the fruit of systematic examinations in which factors that can modify, to a greater or lesser extent, the diagnostic capability of ultrasound examination have not been taken into account. There is no doubt that, in the very near future, this technique is going to play an important role in the early diagnosis of congenital heart disease and will make it possible to better plan the perinatal and postnatal monitoring and management of infants with heart disease, with the corresponding improvement in the prognosis.⁶

Once the incidence of congenital heart disease is known, in order to address the major challenge of the management and monitoring of patients who reach adolescence and adulthood, it is important to study the progression of this population over the years. As we mentioned above, since the first surgical intervention was carried out in 1939 or since 1966, when Rashkind and Miller performed the first interventional catheterization (balloon atrial septostomy) in a patient with congenital heart disease, the advances in both techniques has been spectacular. Thus, studies should be carried out to periodically update the data, not only on mortality in these patients, but on morbidity as well. The findings would help us to establish priorities in the distribution of resources.

Wren et al⁷ studied 1942 children with congenital heart disease, born in England between 1985 and 1994, in a population of 377 310 live births (5.2 per thousand). The survival at one year was 82% (1582 children) and, on the basis of data from other studies, they predicted that the survival at 16 years would be 78% (1514 adolescents), possibly lower than that expected now. Undoubtedly, this trend toward a clear improvement in the survival of these patients is substantiated in the study carried out in the United States by Boneva et al,⁸ who observed a reduction of 39% in the mortality related to congenital heart disease during the period between 1979 and 1997 (2.5 to 1.5 per 100 000 population) and demonstrated that death usually occurs at an increasingly advanced age. Likewise, more recently, Hoffman et al⁹ reported that, in the United States, between 1940 and 2002, nearly 1.2 million infants were born with congenital heart disease, between 500 000 and 600 000 with moderate

lesions and nearly half a million with complex lesions. They made specific calculations of the expected survival for the different types of anomalies (from patent ductus arteriosus to single ventricle or double-outlet right ventricle), without treatment and with treatment. If we consider the data for treated patients, the percentage of survival was between 75% and 80% for children with a simple or moderate congenital heart disease and 40% for those with a complex anomaly. In any case, given that these values are broken down into five-year periods, the calculated percentage of survivors, especially with complex lesions, increased progressively and clearly from 1975 on, coinciding with the development of diagnostic and therapeutic techniques. Their calculations only go as far as the year 1997 and, therefore, it could be that the results they report are poorer than those being recorded now, in which, according to some authors, the overall survival of children born with congenital heart disease is around 85%.

In Spain, the situation clearly overlaps. This fact is evidenced by the study of Martínez Olorán et al,⁵ in which data collection was completed on 1 January 2003, meaning that the maximum follow-up period was 14 years. However, as can be seen in Figure 2 of their article (percentages of death related to surgery and catheterization between 1998 and 2000), that is time enough to demonstrate the progressive reduction in postoperative mortality ($P=.018$). Similar findings were published by Guía et al¹⁰ in 2001, after evaluating the course of 1216 children in the Spanish Autonomous Community of Murcia, born over a 13-year period and diagnosed as having congenital heart disease. While those authors confirmed that congenital cardiac anomalies were the cause of a high percentage of deaths in infants, they also observed that the mortality was lower in those born in the period from 1984 to 1990 as compared to those born between 1973 and 1983, especially among children with ventricular septal defect or patent ductus arteriosus.

The data are convincing and lead us to foresee a progressive increase, over the coming years, in the number of adolescents and adults with congenital heart defects, often repaired but seldom "totally cured." Survival has increased and, as a consequence, there are a greater number of adolescents and adults with a congenital anomaly, often with the capacity to deteriorate. The report by Martínez Olorán et al⁵ makes it clear that, in a considerable proportion of patients, despite surgery or interventional catheterization, the results obtained are fair (10.8%) or poor (9%). Moreover, even in the remaining 62%, the outcome is described as "satisfactory," not "excellent." This is because there may be a residual lesion that, during the first few postoperative years, might have little functional impact or none whatsoever, but can evolve to the point that, sooner or later, medical or surgical treatment or interventional catheterization is necessary.

They are not fully cured, which means that, periodically, they should have access to more or less specialized monitoring and, at least, the same level of technical, scientific and humanitarian assistance they received as children, a threshold that pediatric cardiologists have raised to considerable heights.

A new population, with new problems and new challenges, has been born. We should replace the expression "pediatric cardiology" with "cardiology of congenital heart disease," which demands fresh knowledge and an innovative structure and planning. The patient with heart disease is ill prior to birth and remains so until his or her death. Thus, their monitoring should involve obstetricians, neonatologists, intensive care physicians, anesthesiologists, pediatric cardiologists, adult cardiologists, pediatric heart surgeons, adult heart surgeons, etc, while not forgetting molecular biologists, geneticists and physiologists, who should work in close collaboration with a common aim, that is, "the prevention, diagnosis, monitoring and treatment of congenital cardiac anomalies, regardless of the age of the patient." This is not easy, and the resources required have not been determined. Thus, reports that provide data on the real extent of the problem are of great importance, and are fundamental to a new approach to health care planning and organization in this field.

REFERENCES

1. Weeb GD, Williams RG. 32nd Bethesda Conference: Care of the adult with congenital heart disease. *J Am Coll Cardiol.* 2001;37:1161-98.
2. Hoffman JI, Kaplan S. The incidence of congenital heart disease. *J Am Coll Cardiol.* 2002;39:1890-900.
3. Hoffman JI. Incidence of congenital heart disease: II. Prenatal incidence. *Pediatr Cardiol.* 1995;16:155-65.
4. Mitchell SC, Korones SB, Berendes HW. Congenital heart disease in 56,109 births. Incidence and natural history. *Circulation.* 1971;43:323-32.
5. Martínez Olorón P, Romero Ibarra C, Alzina de Aguilar V. Incidencia de las cardiopatías congénitas en Navarra (1989-1998). *Rev Esp Cardiol.* 2005;58:1428-34.
6. Khoshnood B, de Vigan C, Vodovar V, Goujard J, Lhomme A, Bonnet D, et al. Trends in prenatal diagnosis, pregnancy termination, and perinatal mortality of newborns with congenital heart disease in France, 1983-2000: a population-based evaluation. *Pediatrics.* 2005;115:95-101.
7. Wren C, O'Sullivan JJ. Survival with congenital heart disease and need for follow up in adult life. *Heart.* 2001;85:438-43.
8. Boneva RS, Botto LD, Moore CA, Yang Q, Correa A, Erickson JD. Mortality associated with congenital heart defects in the United States. Trends and racial disparities, 1979-1997. *Circulation.* 2001;103:2376-81.
9. Hoffman JI, Kaplan S, Liberthson RR. Prevalence of congenital heart disease. *Am Heart J.* 2004;147:425-39.
10. Guía JM, Bosch V, Castro FJ, Tellez C, Mercader B, Gracian M. Factores influyentes en la evolución de la mortalidad de las cardiopatías congénitas. Estudio sobre 1.216 niños en la Comunidad Autónoma de Murcia (1978-1990). *Rev Esp Cardiol.* 2001;54:299-306.