The natural history of certain congenital cardiovascular malformations

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Information on the “natural history” of congenital cardiac malformations is difficult to obtain and may have very little meaning for the statistician. One is aware that the facts obtained may yield a profile only of those patients with heart malformations who have signs and symptoms significant enough to bring them to a cardiac center. The others, either not sick enough to come to a center, or too sick and even dying before they can be brought to the specialist, or living too far from the medical center, therefore, will not be included in any survey. The only way the entire congenital heart disease population may be caught in the net of the investigator is by following up carefully a large group of newborns.

Though information gathered may not be statistically significant it is none the less important as a frame of reference in making recommendations to patients and their families. Also it is of necessary historical import that we document the course of patients with malformations as yet unaltered by the hand of the surgeons. The effect of surgery on patients with certain malformations is far reaching and changes the course of their natural history. Another reason why we should try to find out what happens without operation to certain patients is that there will be a tremendous advance in cardiovascular surgical techniques for many lesions yet in the future. Nobody questions the fact that cardiac surgery, in many, if not most, areas will be better tomorrow than it is today.

The notable exceptions to improvement of surgical techniques include repair of patent ductus arteriosus, coarctation of the aorta, secundum atrial septal defect, and pulmonic stenosis. These defects are operable at an irreducible risk and the results of surgery have proven to be good. The length of followup after repair of patent ductus extends to 25 years, of coarctation of the aorta to over 15 years, and of secundum atrial defect or pulmonic stenosis (by closed technique) to over 10 years. Since in patients with these malformations operative mortality is negligible and certainly much lower than the risk of early death or prolonged morbidity, one should not really spend time discussing their natural history without surgery.

Congenital cardiac malformations which
Fig. 1. Speculations on the course of 100 newborns with large ventricular septal defect. PAH: pulmonary artery hypertension, FS: pulmonic stenosis, PVO: pulmonary vascular obstructive disease, Qp: pulmonary blood flow, AR: aortic regurgitation.

are operable at an acceptable, but not irreducible, risk include ventricular septal defect, endocardial cushion defect, aortic stenosis, and Tetralogy of Fallot. The present-day surgical mortality is such that in most institutions it would stand improvement or the long-term follow-up indicative of persistent good results in missing. It is in these situations that an assessment of the natural history without operation is so important. Among these lesions, however, one ought probably not to discuss Tetralogy of Fallot since surgical intervention of one kind or another certainly will be necessary for adequate functioning of the individual and this is in contradistinction to many, even all of the other members of this group. An accurate study of the natural history of the other three lesions may allow the physician and the surgeon to procrastinate with patients in whom there is no immediate danger and wait for the time when surgical risks have reached an irreducible minimum and restoration of a completely normal circulation will be approximated.

Regarding the natural history of patients with ventricular septal defect, it is apparent that the spectrum is broad, depending on the size of the defect and its physiologic consequences. I would speculate that small and large defects may be equally common, but there is no conclusive proof of this. One thing that is relatively clear from the data derived from the literature is that the so-called “Eisenmenger complex” with maximal pulmonary vascular disease and right-to-left shunt is rare and certainly no more, and may be considerably less than, 15% of the total ventricular defect population. All evidence at present points to the benign nature of small to moderate ventricular septal defects with the expectation of near normal life span in about 90% of them; some may succumb with bacterial endocarditis. In contrast, near normal life expectancy can be anticipated in only about 20% of those with large ventricular defects. Though death may occur in infancy due to congestive heart failure, comprising about 5% of this group with large defects, catastrophe in the survivors is unlikely to strike before the third or fourth decade. Approximately 50% of the group will succumb between the ages of 20 and 40 with congestive heart failure, pulmonary vascular obstructive disease, or bacterial endocarditis, and the remainder by 60 years. The natural history of the group up to 30 years of age can be summarized from the speculations on the courses of 100 newborn infants with large ventricular defects (see Fig. 1). That spontaneous closure of certain defects, even large ones, may occur is now well recognized. As noted in Figure 1, about 20% of the group will have normal pulmonary artery pressure at 1 year of age, and these patients for the most part do well and may be the only ones with open defects who survive to the fourth decade. About 10% of the group by 1 year of age will have developed right ventricular outflow tract hypertrophy resulting in pulmonic stenosis. Pulmonary vascular obstructive disease (PVO) is already present in 5% of the group by the end of the first year, and has markedly increased by the end of the first decade. It should be noted, however, that the majority of patients at 1 and 10 years are those with large pulmonary blood flow (Qp) and pulmonary artery hypertension (PAH). The presence of progressive pulmonary hy-
pertension, however, in ours and other people's experience is outweighed by the advantage of operative closure of the defect at a time optimal for the surgeon.

Data available on endocardial cushion defect are scanty indeed and profiles of natural history are not available. Of the entire group, it would seem that the defect is partial (ostium primum atrial septal defect with or without a cleft in the mitral or tricuspid valve) in 75% and complete (atrioventricular canal) in 25%. Near normal life expectancy can probably only be anticipated in those with small partial defects. Congestive heart failure is the usual cause of death and may occur under 2 years in those with complete canals. Bacterial endocarditis and pulmonary vascular obstructive disease are rarely encountered in patients with endocardial cushion defects.

The natural history of aortic stenosis is also incomplete. Operation on the aortic valve or the subvalvar area is indeed safe. However, we do not have any significant information on a large number of patients with any long-term postoperative follow-up. We have to know who are the patients in whom operation may be safely postponed to a time when the effect of surgery on those with maximal aortic obstruction may be assessed from the point of view of (1) reduction of gradient, (2) persistence of reduction, (3) reversibility of electrocardiographic changes, and (4) creation of aortic regurgitation. In reviewing statistics on mortality it is apparent that the nonsurgical group with aortic stenosis is probably an unnaturally low number since most of the severe cases are operated upon and they would have been the ones who would have died without operation. This is a good example of the difficulty in assessing problems of natural history accurately.

CURRENT CONCEPTS IN PEDIATRIC CARDIOVASCULAR SURGERY

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Among the many interesting congenital cardiac malformations amenable to surgery today, four will be discussed, namely, coarctation of the aorta, ventricular septal defect, Tetralogy of Fallot, and transposition of the great vessels. Recommendations for surgical therapy are based on the experience at the Mayo Clinic.

Coarctation of the aorta continues to be most satisfactorily treated by operation and the hospital mortality risk is extremely low. In general, the optimal age for surgery is about 10 to 12 years. Unusually severe systemic hypertension or cardiac enlargement constitute an indication for somewhat earlier surgery. When infants with isolated coarctation of the aorta show severe cardiac failure, and this certainly occurs although uncommonly, operation should be advised at that age. The risk even then is very low, but a few cases may require reoperation at age 12 because of failure of adequate growth of the anastomosis.

The indications for repair of ventricular septal defect must take into account the estimated size of the lesion and its hemodynamic effects. Patients with small ventricular septal defects who have normal-sized hearts and normal electrocardiograms, that is, those without clinical evidence of significant hemodynamic derangement, are not advised by us to undergo repair of the defect at present. Patients with ventricular septal defect and mild or moderate pulmonary artery hypertension (that is, patients whose pulmonary artery pressure is less than 75% of systemic artery pressure) can be operated upon at essentially no hospital risk. Operation is clearly advisable in all of these patients. Patients with ventricular septal defect who have severe pulmonary artery hypertension associated with a large
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