Heart Disease in Paediatrics

Second Edition

S. C. JORDAN

OLIVE SCOTT

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S. C. JORDAN
MD, FRCP
Consultant Cardiologist
Bristol Royal Hospital for Sick Children
and
OLIVE SCOTT
MD, FRCP
Consultant Paediatric Cardiologist
Leeds Regional Thoracic Centre
Killingbeck Hospital, Leeds

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POSTGRADUATE PAEDIATRICS SERIES

under the General Editorship of

JOHN APLEY
C.B.E., M.D., B.S., F.R.C.P., J.P.

Consultant Paediatrician, United Bristol Hospitals and Bath Clinical Area; Lecturer in Diseases of Children, University of Bristol

Preface

Since the first edition of this book in 1973, paediatric cardiology has expanded and developed rapidly. More consultant paediatric cardiologists have been appointed and centres are being developed throughout the country to ensure that all children with heart disease may benefit from advances in investigation and treatment. There have been an increasing number of meetings to share knowledge and the first World Congress of Paediatric Cardiology in 1980 established the speciality in its own right.

The purpose of this book has not changed but it has been completely revised and brought up to date. One important new development has been the use of non-invasive investigations and this new edition has a section on echocardiography and its value is indicated when individual lesions are discussed. The use of ambulatory electrocardiographic monitoring in the study and treatment of arrhythmias is also discussed.

There is a new chapter on the advances in cardiac surgery in which the various operations (many of them eponymous) are defined and the use of new materials and prostheses discussed.

Early diagnosis and early referral of newborn infants has improved the outlook for many of them and there are details of the best way of managing these babies in the special chapter on the newborn infant.

The aim for the future must be to correct anatomical abnormalities sufficiently early to keep the function of the heart as normal as possible enabling children not only to survive operation but to lead truly normal lives.

S.C.J.
O.S.

Preface to the First Edition

This book does not aim to be a comprehensive work on all aspects of paediatric cardiology. It is intended for the paediatrician and others who look after children to use as an introduction to paediatric cardiology and to provide up-to-date knowledge of the methods of diagnosis and treatment, particularly of congenital heart disease. The approach is essentially a practical one. It indicates to what extent the clinical signs and symptoms, assisted by radiological and electrocardiographic findings, can indicate the diagnosis, and when further investigation by cardiac catheterization and angiography will be required.

The two authors, one a paediatrician with cardiological training and one a cardiologist with special experience in paediatric cardiology, have slightly different views on some of the subjects, so that the text has been a result of the two approaches. In a book of this size it has not been possible or desirable to discuss in detail some of the more controversial aspects of paediatric cardiology, and indeed views on such topics as early corrective surgery in infancy are changing rapidly at the moment. This should not deter the paediatrician, as the same principles apply with regard to referral of patients for further investigation and treatment.

Few paediatricians will be directly concerned with cardiac catheterization, but decisions regarding surgery are frequently influenced by the results of this investigation and a section has therefore been included on the technique and interpretation.

One of the most striking changes in paediatric cardiology in recent years has been the increasing interest in the management of the newborn infant with heart disease and a special chapter has been included, which presents a scheme to simplify diagnosis in this difficult group of patients.

S.C.J.
O.S.

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GENERAL CARDIOLOGY

CHAPTER 1

Incidence and Aetiology of Congenital Heart Disease

INCIDENCE

Congenital heart disease is the commonest single group of congenital abnormalities, accounting for about 30 per cent of the total. The incidence is about 8/1000 live births. There are eight common lesions which account for 85 per cent of all cases. They are ventricular septal defect, patent ductus arteriosus, atrial septal defect, pulmonary valve stenosis, aortic stenosis, coarctation of the aorta, tetralogy of Fallot and transposition of the great arteries. The remaining 15 per cent is made up of a variety of more rare and complex lesions.

Congenital heart disease as a whole occurs with equal frequency in males and females, but some lesions such as aortic stenosis, coarctation of the aorta, transposition of the great arteries and tetralogy of Fallot are more common in males whereas patent ductus arteriosus and atrial septal defect are more common in females. About 13 per cent of patients who have one congenital heart defect will have an additional cardiac defect. Between 10 and 15 per cent of patients with cardiac defects will have another *non*-cardiac deformity (Campbell, 1965).

Recurrence in family

The risks of a sibling being affected by congenital heart disease is between 2 and 4 per cent. There is a high degree of concordance in that the sibling usually has the same lesion or one of its components

(e.g. pulmonary stenosis or ventricular septal defect occurs in siblings of patients with tetralogy of Fallot). The studies of Nora, McGill and McNamara (1970) give recurrences in siblings (*Table 1.1*). Now that children with congenital heart disease survive to have children

TABLE 1.1

Proportion of siblings and offspring of index patients with congenital heart disease who were also affected

	Per cent risks to siblings	*Per cent risks to offspring*
Patent ductus arteriosus	3.4	4.3
Ventricular septal defect	4.4	4.0
Atrial septal defect	3.2	2.5
Pulmonary stenosis	2.9	3.6
Coarctation of aorta	1.8	2.7
Transposition of the great arteries	1.9	—
Tetralogy of Fallot	2.7	4.2
Aortic stenosis		3.9

of their own, it is important to know the risks of their children being affected. Nora and Nora (1976) have determined these (*Table 1.2*). Again the majority of children had the same lesion as the affected parent. The results of this study are within the range one would expect in multifactorial inheritance. The recurrence rate is worrying but the data is still insufficient for us to know whether there will be an absolute increase in affected children. Nora's data suggests that individuals with congenital heart disease marry later and restrict their families more than the population as a whole. If another member of the family has congenital heart disease, recurrence rates increase two or threefold (Nora and Nora, 1978).

Recently Child and Dennis (1977) have calculated the sibling recurrence risk of endocardial fibroelastosis to be 5.4 per cent and hypoplastic left heart to be only 0.5 per cent.

Parents who have had one child with congenital heart disease should if they wish be advised by a genetic counsellor.

AETIOLOGY

The first question parents ask when they realize their child has a heart defect is 'what caused it?' Unfortunately we are still unable to answer this question precisely in the majority of patients.

Inheritance

A few families are reported in which a defect, especially atrial septal defect, follows a dominant pattern of inheritance.

Chromosomal abnormalities

Some chromosomal abnormalities are associated with congenital heart disease (*Table 1.2*) but these account for about 5 per cent of patients with congenital heart disease. The cardiovascular abnormality in Turner's syndrome is most commonly coarctation of the

TABLE 1.2

Incidence of congenital heart disease in children with chromosomal abnormalities

Chromosome abnormality	*Name of syndrome*	*Incidence of CHD (%)*	*Type of defect*
Partial deletion of 5	Cri du chat	50	VSD
21 Trisomy	Mongolism (Down's syndrome)	60	A-V defects VSD: ASD: Fallot
18 Trisomy	—	90	VSD; PDA; single umbilical artery; DORV; coarctation
13 Trisomy	—	90	VSD; ASD; single umbilical artery; dextrocardia
XO	Turner's syndrome	40	Coarctation, AS

CHD Congenital heart disease
VSD Ventricular septal defect
A-V Atrioventricular defect
ASD Atrial septal defect
PDA Patent ductus arteriosus
DORV Double outlet right ventricle
AS Aortic stenosis

aorta but in the XX and XY phenotype (Noonan's syndrome) pulmonary stenosis with dysplastic pulmonary valve and hypertrophic cardiomyopathy occur. About two-thirds of patients with Down's syndrome have heart lesions and the majority have atrioventricular defects. Tetralogy of Fallot, patent ductus arteriosus, atrial septal defect and ventricular septal defects also occur.

Multifactorial inheritance

The most readily acceptable theory for the genetic basis of congenital heart disease is the multifactorial hypothesis. The hypothesis suggests that the genetic material concerned with the normal development of the heart is carried on a number of genes and that abnormalities of these genes may not in themselves be enough to cause cardiac abnormalities, but in the presence of other environmental factors the genes predispose to the occurrence of such defects. In other words there is a genetic susceptibility to develop congenital heart disease if the appropriate environmental hazard occurs.

Environmental factors

Maternal vitamin deficiency (Wilson and Warkany, 1949; Smithells, 1977) may cause cardiac defects. In 1 per cent of congenital heart disease cases rubella in the first trimester is associated with patent ductus arteriosus, ventricular septal defect, tetralogy of Fallot and peripheral pulmonary stenosis. Drugs taken during pregnancy are being increasingly suspected since thalidomide was found to be associated with peripheral pulmonary stenosis. Phenytoin is associated with an increased incidence of heart disease and warfarin is teratogenic. Pregnancy testing drugs are also under suspicion.

Congenital heart block is commonly seen in infants of mothers who have systemic lupus erythematosis. There is an increased incidence of congenital heart disease in infants of mothers who have diabetes. Smoking in the mother is associated with a higher incidence of congenital heart disease, and alcoholism produces abnormal babies half of whom have heart lesions, most commonly septal defects.

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CHAPTER 2

The Fetal Circulation and the Changes at Birth in Relation to Congenital Heart Disease

THE FETAL CIRCULATION

To appreciate the haemodynamic effects of congenital heart lesions, there must be clear understanding of the fetal circulation and the changes which take place after birth to establish the normal, independent circulation. These changes influence the circulation in normal infants but are of profound importance in infants with congenital heart disease. *A congenital heart lesion is not a static condition; changes continue to take place throughout the patient's life, but the most significant of these occur at birth.*

In the fetus, blood comes from the placenta via the umbilical vein and is relatively well oxygenated ($Po_2 = 30$ mmHg). Half of this blood passes through the liver and the remainder bypasses the liver through the ductus venosus and continues up the inferior vena cava, which receives blood leaving the liver by the hepatic veins and blood returning from the lower half of the body of the fetus. Most of the inferior vena caval blood passes through the foramen ovale to the left atrium and so to the left ventricle, ascending aorta and coronary circulation. This ensures that blood of a high Po_2 enters the cerebral and coronary circulations. A small amount of inferior vena caval blood passes through the tricuspid valve into the right ventricle. Blood returning from the head and neck of the fetus enters the right atrium by the superior vena cava, is joined by coronary sinus blood and then enters the right ventricle and pulmonary artery (*Figure 2.1*). In the fetus only about 15 per cent of the right ventricular blood

enters the lungs, and the rest passes through the ductus arteriosus into the descending aorta where it is joined by blood from the ascending aorta. In the fetus the ductus arteriosus is as large as the aorta itself and pressures in the pulmonary artery and aorta are equal.

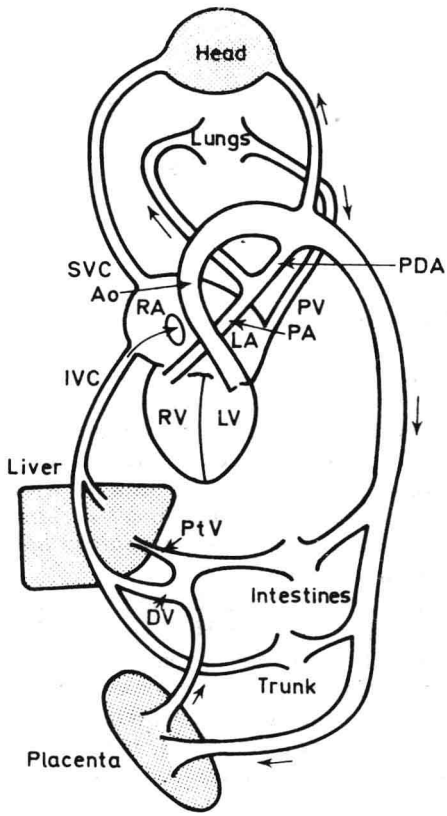

Figure 2.1. The fetal circulation. Ao: aorta; DV: ductus venosus; IVC: inferior vena cava; LA: left atrium; LV: left ventricle; PtV: portal vein; PV: pulmonary vein; RA: right atrium; RV: right ventricle; PA: pulmonary artery; PDA: patent ductus arteriosus; SVC: superior vena cava