Series Editor J.P. Shillingford

CONGENITAL HEART DISEASE

Series Editor J.P. Shillingford

CONGENITAL HEART DISEASE

Edited by
F.J. Macartney

Professor of Paediatric Cardiology
The Hospital for Sick Children
London

MTP PRESS LIMITED
a member of the KLUWER ACADEMIC PUBLISHERS GROUP
LANCASTER / BOSTON / THE HAGUE / DORDRECHT

Published in the UK and Europe by
MTP Press Limited
Falcon House
Lancaster, England

British Library Cataloguing in Publication Data

Congenital heart disease.——(Current status of clinical cardiology)
1. Heart——Abnormalities
I. Title II. Series
616.1'2043 RC687

ISBN-13: 978-94-010-8656-1 e-ISBN-13: 978-94-009-4872-3
DOI: 10.1007/978-94-009-4872-3

Published in the USA by
MTP Press
A division of Kluwer Boston Inc
190 Old Derby Street
Hingham, MA 02043, USA

Contents

Preface

After the allies landed on the Normandy beaches in 1944, the immediate sense of celebration was followed by an acrimonious dispute as to the overall strategy. Eisenhower favoured the advance of all the armies on a broad front. Montgomery wanted to concentrate the bulk of the allied front on a narrow front, a rapier thrust to the heart of Germany. The broad front was bound to be slow. The rapier thrust was likely to be risky, because of the attentuation of supply lines and the danger of an exposed flank.

These strategic issues were, as ever, complicated by personal ones. Montgomery's sense of his own destiny made the idea of a rapier thrust led by him particularly attractive: the idea of Patton's army advancing on a narrow front would almost certainly have seemed to him less appealing. Eisenhower, in his role as supreme allied commander, undoubtedly wanted to be seen to be fair to each of the ambitious generals under his command. If so, advance on a broad front was inevitable.

The parallels with scientific advance are striking. Inside each researcher battling to push back the frontiers in this own particular small patch is a Montgomery. Lurking within anyone involved in the distribution of rewards for scientific research is an Eisenhower, be he responsible for acceptance of papers, granting of research applications or nomination for prizes.

Where does editing a book come into this scheme of things? I suspect mainly at the Eisenhower end, though most doctors I know regard being asked to write a chapter as a penalty rather than a prize, and I certainly do not see myself as some kind of benevolent sponsor handing out laurel wreaths. In no way have I felt constrained to present the whole of a broad front; there are many exciting new areas of development, such as in non-invasive imaging, measurement and biochemical analysis, which do not appear here. My guiding principle has been to cover widely divergent *approaches* to research in paediatric cardiology, be they philosophical, as in questions of nomenclature, statistical, as in survival analysis, technological, as in interventional catheterization,

descriptive, as in fetal echocardiography, physiological, as in analysis of ventricular function and the Fontan circulation, or pharmacological, as in medical manipulation of the arterial duct. This book really belongs to the contributors, whom I can hardly thank enough for their diligence and promptness. I trust that every paediatric cardiologist will find at least one chapter which is of immediate importance and relevance.

Fergus J. Macartney

List of Contributors

L. D. Allan
British Heart Foundation
Perinatal Research Centre
Guy's Hospital
St Thomas Street
London SE1 9RT
UK

R. H. Anderson
Department of Paediatrics
Cardiothoracic Institute
Brompton Hospital
Fulham Road
London SW3 6HP
UK

E. H. Blackstone
Department of Surgery
University of Alabama at
Birmingham
Birmingham, AL 35294
USA

C. Bull
Department of Paediatric Cardiology
Institute of Child Health and
Hospital for Sick Children
Great Ormond Street
London WC1N 3JH
UK

K. E. Fellows
Department of Radiology
Children's Hospital
300 Longwood Avenue
Boston, MA 02155
USA

T. P. Graham
Department of Pediatric
Cardiology
Vanderbilt Medical Center
Nashville, TN 37232
USA

J. F. Keane
Department of Cardiology
Children's Hospital
300 Longwood Avenue
Boston, MA 02115
USA

J. E. Lock
Department of Cardiology
Children's Hospital
300 Longwood Ave
Boston, MA 02115
USA

Y. H. Siew
Department of Paediatrics
Cardiothoracic Institute
Brompton Hospital
Fulham Road
London SW3 6HP
UK

E. D. Silove
Department of Paediatric
Cardiology
Children's Hospital
Ladywood Middleway
Birmingham B16 8ET
UK

Series Editor's Note

The last few decades have seen an explosion in our knowledge of cardiovascular disease as a result of research in many disciplines. The tempo of research is ever increasing, so that it is becoming more and more difficult for one person to encompass the whole spectrum of the advances taking place on many fronts.

Even more difficult is to include the advances as they affect clinical practice in one textbook of cardiovascular disease. Fifty years ago all that was known about cardiology could be included in one textbook of moderate size and at that time there was little research so that a textbook remained up to date for several years. Today all this has changed, and books have to be updated at frequent intervals to keep up with the results of research and changing fashions.

The present series has been designed to cover the field of cardiovascular medicine in a series of, initially, eight volumes which can be updated at regular intervals and at the same time give a sound basis of practice for doctors looking after patients.

The volumes include the following subjects: heart muscle disease; congenital heart disease, invasive and non-invasive diagnosis; ischaemic heart disease; immunology and molecular biology of the heart in health and disease; irregularities of the heart beat; and each is edited by a distinguished British author with an international reputation, together with an international panel of contributors.

The series will be mainly designed for the consultant cardiologist as reference books to assist him in his day-to-day practice and keep him up to date in the various fields of cardiovascular medicine at the same time as being of manageable size.

J.P. Shillingford
British Heart Foundation

1
The diagnosis and naming of congenitally malformed hearts

R. H. ANDERSON AND SIEW YEN HO

INTRODUCTION

Some people become very irritated with the amount of time and journal space taken up by the gurus of cardiac morphology in polemics concerning the semantics of congenitally malformed hearts. At the same time, other people, perhaps even those who object to unseemly disputations, find certain aspects of congenital heart disease difficult to understand. This is particularly so in those so-called 'complex' cases where the arrangements and relationships of the cardiac segments are not as anticipated in the normal heart. These 'complex' cases are, in reality, no harder to understand than is the morphology of the normal heart.

A combination of circumstances conspires against the universal understanding of both normal and abnormal morphology at present. The basis for analysis in the past has often been presumed knowledge of cardiac morphogenesis which, for the most part, remains speculative. A plethora of Latin terms, often compounded by alphanumeric subcategorizations, has then all too regularly been used for descriptions and classification. Definitive terms have frequently been interposed between originator and receiver when a simple descriptive phrase would have been preferable. In short, there has been an unhealthy desire to create brief and cryptic nosologies suited for 'corridor talk'. It is true that brevity is the soul of wit. In most circumstances, understanding is much more important.

It is our belief that the morphology of congenitally malformed hearts is a simple topic. It can and should be understood by all concerned with diagnosis and treatment. This is as true for the intensive care nurse or the echocardiographic technician as for the paediatric cardiologist or surgeon. Indeed, it is possibly easier for the former presently to achieve this understanding. They are less likely to be constrained by the preconceived notions and received wisdom that

prevent, in some cases, the acquisition of new ideas by the latter. In this chapter we set out concepts, derived from studies with many collaborators, which might provide the required simplicity. We discuss at the same time a number of features which may in future ease the exchange of information. Any system for diagnosis can only be as good as the words used to describe it. We start, therefore, with a plea for the abandonment of Latin terms. Instead, we recommend the substitution (at the appropriate time and place) of the vernacular equivalents. We then discuss the role of knowledge and of normal anatomy in the understanding of the abnormal. In doing so, we show how certain basic features of anatomy are retained in all hearts no matter how malformed, yet other prominent features of malformed hearts have no counterpart in the normal. Finally, we summarize a system of sequential segmental analysis which permits any heart, no matter how severely malformed, to be described logically and succinctly, even if it has never been encountered before.

USE OF LATIN TERMS

In bygone years it could reasonably be argued that Latin provided a common language for scientific discourse. Nowadays there is little doubt that English has become the universal language. More and more journals of national societies are being published in English. It makes sense, therefore, to discontinue the use of Latin terms such as abound in the vocabulary used for congenital heart malformations. The rewards of this enterprise in terms of style and syntax can be great. How many know that the plural of 'ductus' is 'ductus' (a fourth declension Latin noun)? The plural of 'duct' is self-evidently 'ducts'! How many know the correct plural of 'vena cava'? When this term is used as an adjective, does the user stop to think whether flow (for example) is venous or hollow (cava = hollow)? In other words, are we more correct in describing 'caval venous' or 'vena caval' flow? Over and above this, there are good scientific reasons for discontinuing the use of Latin. The 'atrioventricularis communis' defect is more readily understood on the basis of the defective atrioventricular septation underscoring this group of lesions. Is not 'common arterial trunk' more readily understood than 'truncus arteriosus'? The conservative may find this approach unpalatable and argue that many Latin words have already become assimilated into the English language. True. But mostly they have been adopted in anglicized form. The beauty of the English language is its electicism. We can continue this process by substituting good Anglo-Saxon synonyms and translations in paediatric cardiology.

NORMAL VERSUS ABNORMAL CARDIAC ANATOMY

Study of malformations of the heart requires a thorough understanding of normal cardiac anatomy. At the same time, overzealous application

of principles derived from study of the normal can in some circumstances produce difficulties in the description of abnormality. As an example, take the group of lesions variously described as 'endocardial cushion defects' or 'atrioventricular canal malformations'. These hearts are unified because of a deficiency of atrioventricular septation[1]. To understand this, it is necessary to have precise knowledge of the extent of the normal atrioventricular septum. This points to the need for knowledge of the normal. Thereafter it is also necessary to consider the effects of abnormal septation on the rest of the heart. The usual anatomy of the atrioventricular septal defect itself must then be taken as the norm. Thus, the left valve in these hearts bears scant resemblance to a normal mitral valve beyond its residence in the morphologically left ventricle. Yet the substrate for regurgitation across this valve has almost universally been described in terms of a 'cleft mitral valve'. Only when it is appreciated that morphologically the structure is *not* a mitral valve can its function be understood and the valve be appropriately repaired[2]. The principle to be adduced from this example is that analysis should start with a thorough knowledge of normal anatomy. Thereafter, features of normality are used in descriptions of abnormal hearts as far as they can be applied with accuracy. Whenever this proves impossible, the abnormal hearts should be described in terms of their own intrinsic features rather than instituting Procrustean remedies.

The morphological method

The cornerstone of analysis of any malformed heart is the establishment of the nature, connections and relationships of the different chambers and structures within each of the cardiac segments[3]. This we describe in our section devoted to sequential analysis (see below). Fundamental to this approach is the principle introduced by Lev[4] and dubbed by Van Praagh[5] the 'morphological method'. In simple terms, structures must be analysed in terms of their own intrinsic morphology. This means that parts of a given chamber or artery which are inconstant cannot be used as criteria for identification. Inherent in the morphological method, therefore, is also the need to identify the segments or 'building blocks' of the heart.

The pioneering studies of Van Praagh and his colleagues[6] identified the three basic cardiac components, namely the atrial chambers, the ventricular mass and the arterial trunks (Figure 1.1). We follow this division precisely, but find it desirable to give a secure definition to the middle segment, the ventricular mass, which is lacking in the approach of Van Praagh *et al.*[6] We define this part of the heart as extending from the atrioventricular to the ventriculo-arterial junctions. The myocardial mass thus delimited is an anatomical whole and functions electrically as a single unit. It is separated from the atrial muscle mass by the fibro-fatty atrioventricular tissue planes at all points around the

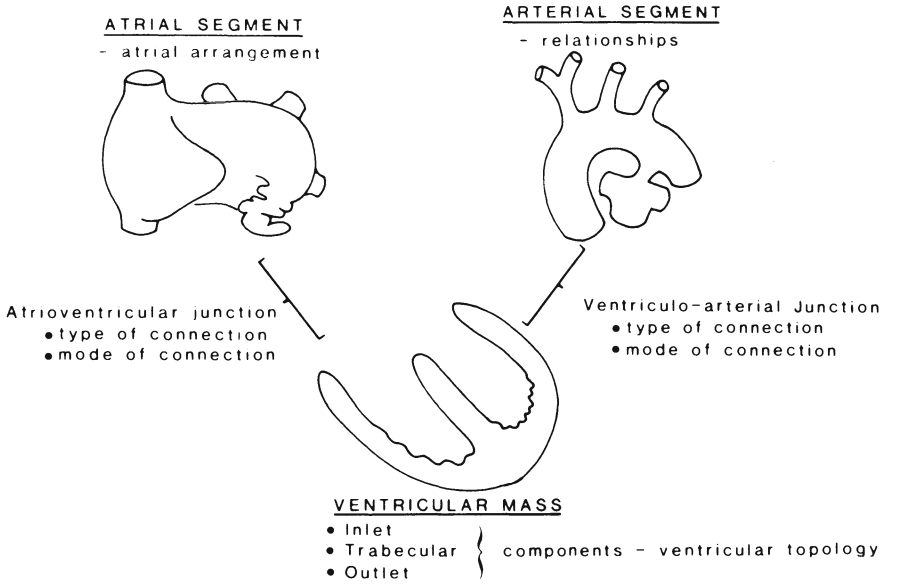

Figure 1.1 Diagram of the three basic cardiac segments

junction except at the site of penetration of the atrioventricular conduction axis. Its distal extent is limited by the semilunar attachments of the sinuses of the arterial valves (in part to ventricular muscle and in part to the fibrous cardiac skeleton). By defining the ventricular mass in this fashion we avoid the need to introduce further segments (the atrioventricular canal and conus) as suggested more recently by Van Praagh[7]. These additional parts are an integral part of the ventricular mass as here defined and can readily be described as such.

In most hearts the ventricular mass possesses two cavities, which conventionally are described as 'ventricles'. These ventricles do not always possess all the parts found in the normal heart. The ventricular mass may rarely be composed of a solitary chamber. More frequently one or other of the ventricles is variously rudimentary or supernormal. A system of ventricular subdivision must therefore be devised which accounts for all known varieties of ventricles irrespective of whether they are or are not connected to the atrial chambers and the arterial trunks, respectively. Here the 'morphological method' comes into its own. The connections or non-connections of the ventricles to the adjacent segments cannot be taken into account when naming the ventricles, since they themselves are the most significant variables. Indeed, the time-honoured convention of dividing a ventricle into inflow and outflow is disqualified by the morphological method. An inflow tract, or 'sinus', is conventionally defined as the part of the ventricle containing the atrioventricular valve. The outflow tract is usually de-

scribed as the 'conus'[6]. Such a system founders irretrievably when applied to one particular variety of heart with two ventricles. This is the case with one ventricle connected to both atria and both arterial trunks and the other ventricle connected to neither an atrial chamber nor an arterial trunk (Figure 1.2). To cater for this and other abnormal arrangements, it is preferable to consider ventricles as having three

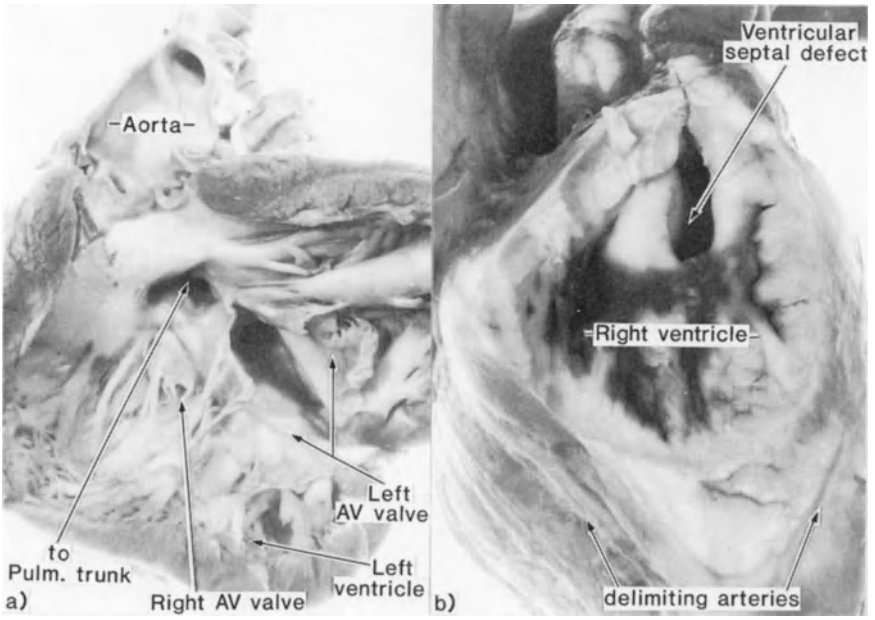

Figure 1.2 A specimen with both arterial trunks arising from a morphologically left ventricle which has a double inlet atrioventricular connection: (a) is a left ventricular view and (b) shows the rudimentary right ventricle which is a trabecular pouch

basic components. These are the inlet, apical trabecular and outlet portions (Figure 1.3). In the normal heart with two normal ventricles it is not possible to draw with precision the boundaries between these parts. In rough terms, nonetheless, it is possible to recognize that the inlet component extends from the atrioventricular junction to the distal attachments of the tension apparatus of the atrioventricular valve. Similarly, the outlet component can be recognized in broad terms as the region supporting the semilunar attachments of the arterial valve. The apical trabecular component is then recognized as the third part by exclusion of the other two.

When considered in this fashion, the apical trabecular component (unrecognized when ventricles are divided into 'sinus' and 'conus') is the most constant part of a ventricle irrespective of how abnormal it may be. Furthermore, it is the nature of this apical component which

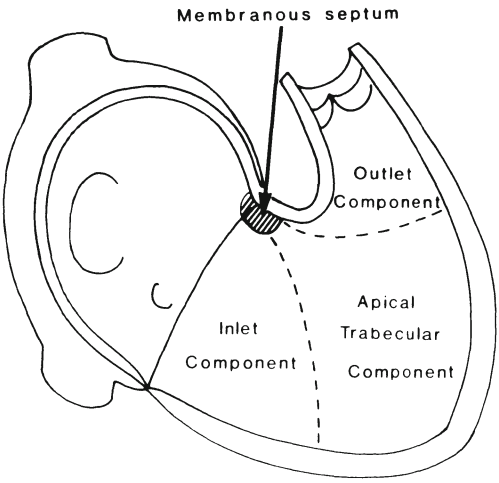

Figure 1.3 Diagram of the tripartite ventricular septum

most readily differentiates a morphologically right from a left ventricle, being coarsely trabeculated in one and fine in the other (Figure 1.4). All varieties of biventricular hearts (hearts with two chambers in their ventricular mass) are then readily described according to the way in which the inlet and outlet components are shared between the apical trabecular parts. In doing this, note should be taken that one or other inlet or outlet portion may be totally lacking in some hearts with so-called atrioventricular or arterial valve atresia (Figure 1.5). When considered in this light, it can readily be seen that the 'ventricle proper' of Van Praagh is not the 'sinus' but is the apical trabecular component. The 'atrioventricular canal' and 'conus' defined by Van Praagh[7] are directly comparable with the ventricular inlet and outlet components respectively. In short, by recognizing the extent of the ventricular mass anatomically and by dividing it according to the morphological method, it is possible to construct a simple tripartite template within the three segmental model of the heart[6] which accounts for all known types of abnormal ventricle.

Fully to describe such abnormal ventricles it is then necessary to account for their morphology according to the pattern of the apical trabecular component; for their component make-up; for their relationships and finally for their size. All of these are mutually independent features. In the context of morphology, it should be noted that almost always ventricles in congenitally malformed hearts are of right or left ventricular type and almost always they coexist. Very rarely, however, hearts are encountered having a solitary ventricle. Sometimes these hearts are of right or left ventricular morphology, the complementary ventricle being so tiny as to be unrecognizable. More fre-

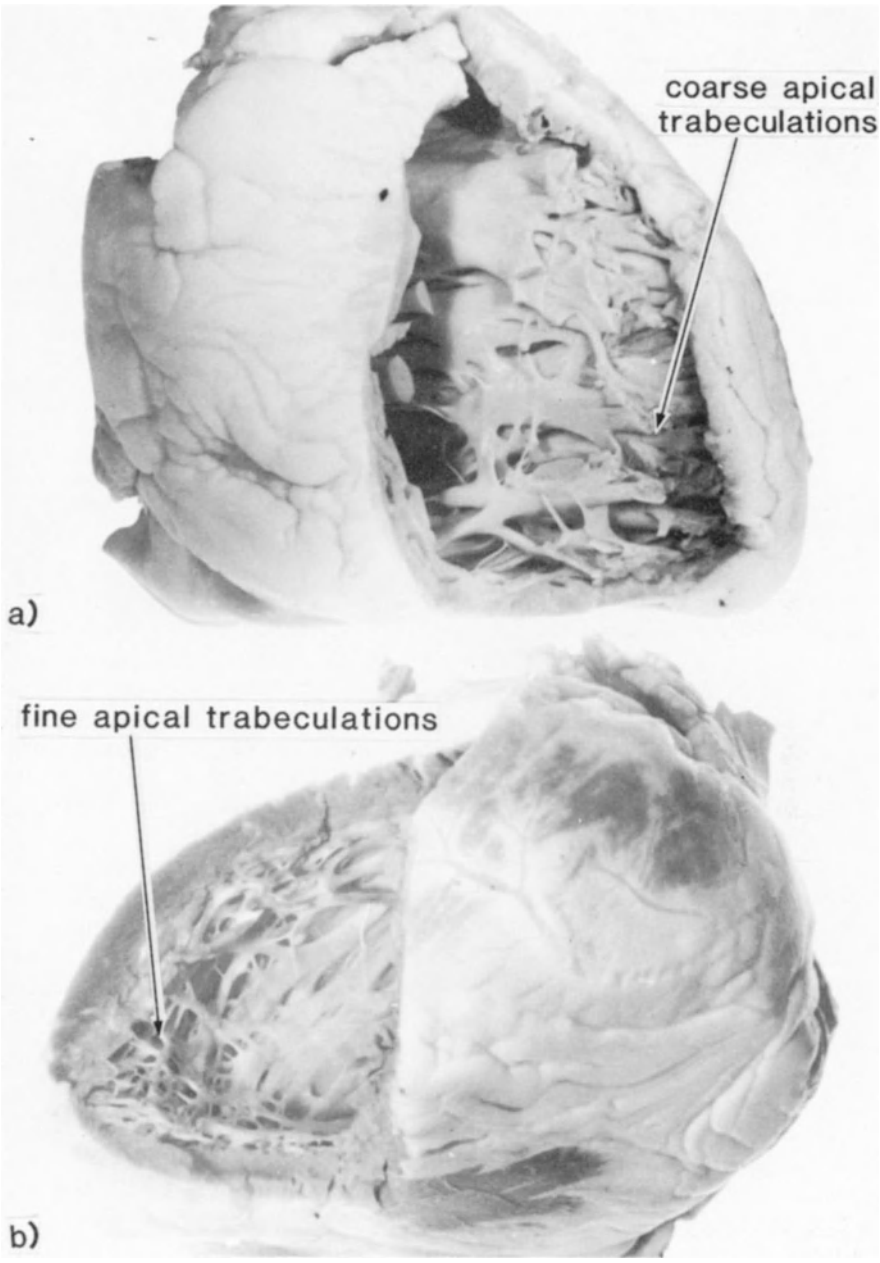

Figure 1.4 Dissections of the ventricular apices: (a) shows the coarse apical trabeculations of the right ventricle in contrast to the fine trabeculations of the left ventricle (b)

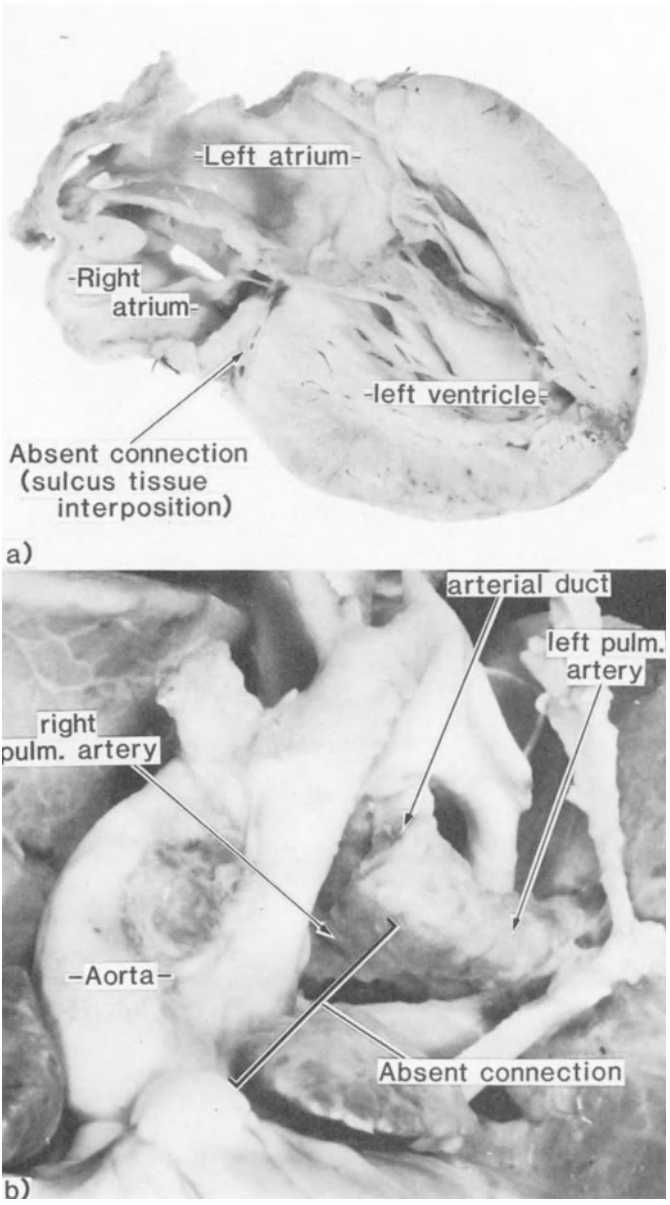

Figure 1.5 A heart with 'tricuspid atresia' and another with 'pulmonary atresia' showing the absent connection in each: (a) is a long axis section illustrating the muscular floor of the right atrium: (b) shows the pulmonary arteries supplied via a duct from the aorta and absence of the intrapericardial pulmonary trunk

quently the solitary ventricle is of neither right nor left ventricular morphology and possesses no apical septum. The apical trabecular component of these ventricles is most distinct, being coarser than a morphologically right ventricle and being criss-crossed by one or more large trabeculations. Such solitary ventricles are of indeterminate morphology (Figure 1.6).

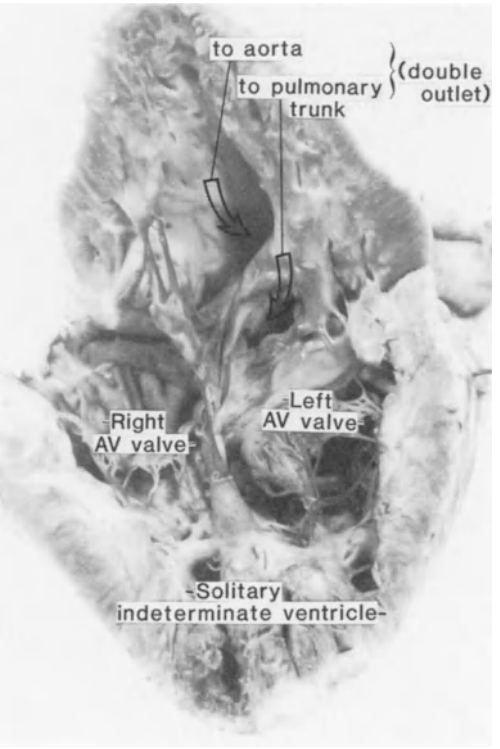

Figure 1.6 A specimen with double inlet solitary and indeterminate ventricle opened like a clam

When the atrial segment of the heart is considered in a fashion analogous to that described above for the ventricles, it is immediately evident that variable components are themselves ruled out as hallmarks for identification. In this way we exclude the great veins (frequently anomalously connected) and the atrial septum (frequently absent) as markers of morphological rightness or leftness. All that remains are the atrial appendages. These are the most constant parts of the atria. Their morphology readily permits atrial differentiation (Figure 1.7). The morphologically right atrium has a characteristic triangular shape with a broad junction with the venous component. The junction is

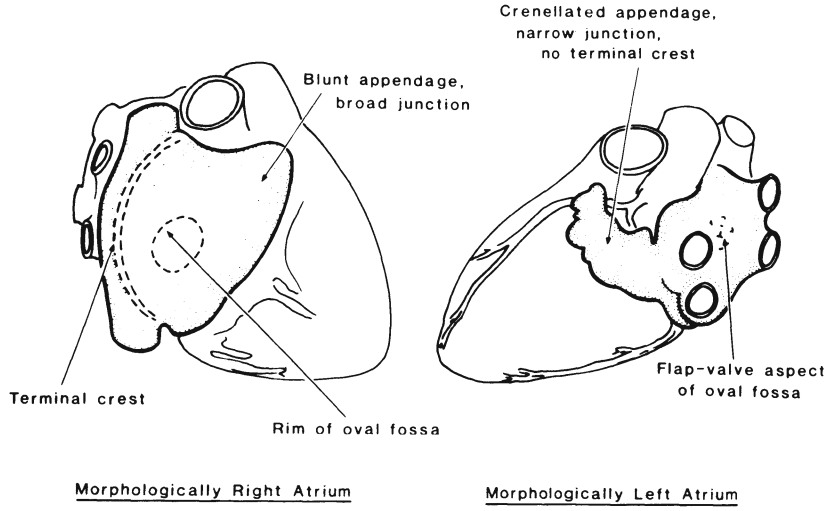

Figure 1.7 Diagrams showing the different morphological features between right atrium and left atrium

marked internally by the terminal crest and externally by the terminal groove. The morphologically left atrium has a narrow and convoluted shape with a constricted junction with the venous component which is not marked by a terminal crest or groove.

When we turn our attention to the third cardiac segment, the arterial trunks, the morphological method is found to be of less help in distinguishing the types of trunk (Figure 1.8). This is because the great arteries have no constant intrinsic differentiating feature. Rather they are recognized according to their branching pattern which is more-or-less variable. Fortunately the varieties of branching are rarely (if ever) severe enough to prevent distinction of an aorta from a pulmonary trunk from a common arterial trunk. In simple terms, the aorta gives off the coronary arteries followed by the systemic arteries. The pulmonary trunk does not normally give rise to coronary arteries and usually branches into its two major components. A common arterial trunk exits from the heart through a single arterial valve and branches immediately into coronary, pulmonary and systemic arteries. There are certain lesions which conspire to defeat these criteria, such as anomalous origin of the coronary arteries from the pulmonary trunk or origin of one pulmonary artery from the aorta. These are hardly, if ever, found in combinations such as to make differentiation impossible. One arrangement does rule out positive identification of an arterial trunk. This is when a solitary great artery supplies the coronary and systemic arteries in absence of any intrapericardial pulmonary arteries (pulmonary blood supply then almost always being supplied through major systemic–pulmonary collateral arteries but rarely through bi-

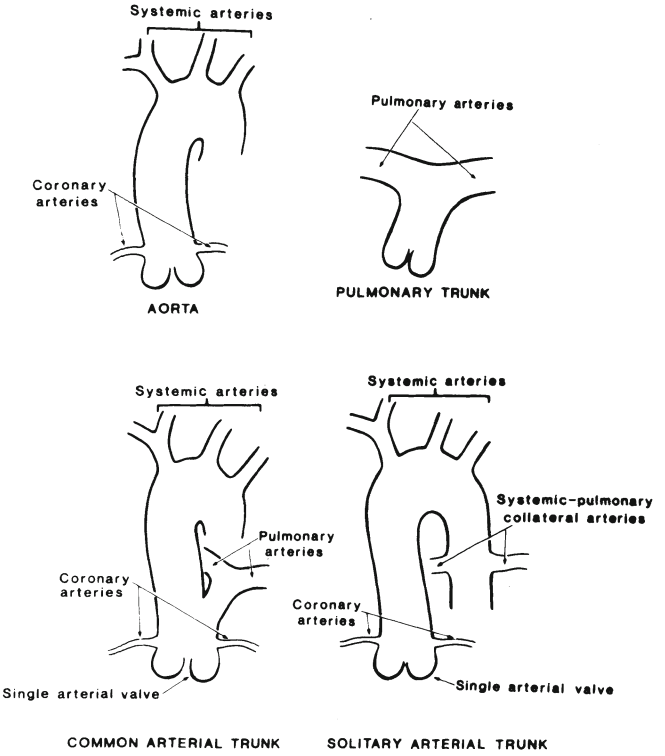

Figure 1.8 A diagram showing the four types of arterial trunk

lateral ducts). In this circumstance the only solution which is both simple and accurate is to label the trunk as a solitary arterial trunk.

Cardiac septation

Most of the simple cardiac malformations, which themselves account for the majority of congenital cardiac lesions, are either communications between the cardiac segments ('septal defects') or else stenoses along the systemic or pulmonary flow pathways. A sound knowledge of the normal septal structures is a prerequisite for complete understanding and accurate naming of septal defects. Surprisingly few textbooks of either anatomy or paediatric cardiology give the necessary information.

The atrial and ventricular chambers are separated by three rather than two septal structures. As may be anticipated, there is an atrial and a ventricular septum. The third septal structure is the atrioventricular septum. This has two components in the normal heart, the muscular and membranous portions. They exist because of two important

features of normal anatomy. These are the off-setting of the proximal septal attachments of the atrioventricular valves (Figure 1.9) and the deeply-wedged position of the subaortic outflow tract (Figure 1.10). Because of the valvar off-setting, the distal portion of the right atrial myocardium is separated from the left ventricular inlet to give the muscular atrioventricular septum. Because of the wedged position of the subaortic outflow tract, the supratricuspid component of the membranous part of the septum is separated from the left ventricular outlet component by the atrioventricular membranous septum. Deficiencies of these parts of the normal septum give rise to a particular group of lesions variously termed 'atrioventricular canal malformations' or 'endocardial cushion defects'. It makes more sense to label them for what they are – atrioventricular septal defects[1].

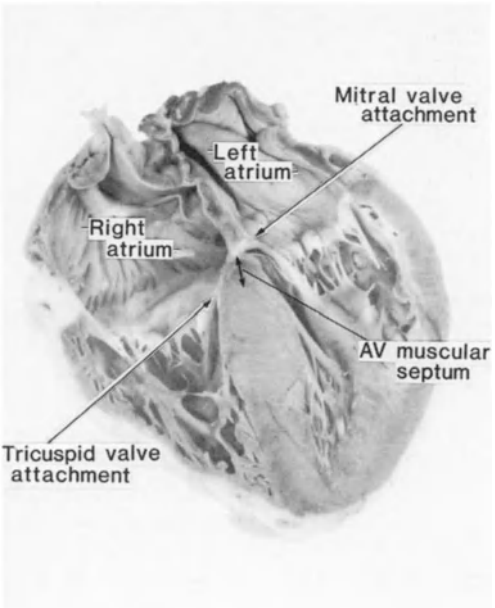

Figure 1.9 A long axis four chamber section showing the off-setting of the septal attachments of the tricuspid and mitral valves

When the adjacent parts of the atrial chambers are dissected, it is found that the interatrial septum is much less extensive than it seems at first sight (Figure 1.11). The area of true septum is confined to the oval fossa and its immediate environs. True atrial septal defects can exist only within this area, and are best termed oval fossa defects. Various other lesions can produce the facility for interatrial shunting of blood even though they are not defects within the atrial septum