The Clinical Recognition of Congenital Heart Disease

Second Edition

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Preface

The first edition of this book was based upon a simple premise—the interplay between clinical and laboratory information in congenital heart disease. If anatomic details are known, the pathophysiology can be comprehended, and if both the morphologic and functional derangements are understood, the clinical manifestations become intelligible. The converse is also the case—a careful, critical synthesis of information from the natural history, physical signs, electrocardiogram and chest x-rays generally permits relatively precise anatomic and physiologic inferences, i.e. the clinical recognition of congenital heart disease.

Since 1970, when the first edition appeared, progress has been vigorous, although not characterized by the quantum leaps of the fifties and sixties. The morphology and morphogenesis of many of the major malformations and their variations have been considerably refined or supplemented. We have witnessed an evolution of the concept of congenital defects as dynamic anomalies originating in fetal life and progressively changing during early and late postnatal development. Physiologic and structural modifications often continue for weeks, months, years or even decades, clarifying the variable clinical manifestations of malformations as they evolve with the passage of time. Much new data, especially in the neonate and young infant, stem from safer, simpler and more elegant techniques in the catheterization laboratory. Modern electrophysiology has begun to resolve the mechanisms of many time-honored electrocardiographic signs. M mode echocardiography has been a major step forward in providing a safe, painless, highly informative noninvasive laboratory tool that permits repeated study, and two-dimensional ultrasonography is gathering momentum.

Because of this healthy state of flux, most if not all chapters required revision, some relatively little, others considerable; still others were completely rewritten or added. Nevertheless, the basic premise was not changed; the point of view remains unaltered. This edition, like the first, is devoted to practical aspects of the clinical recognition of congenital heart disease in its natural (unoperated) state. It is not my purpose to deal with laboratory techniques as such or with therapeutics.

Large numbers of congenital cardiacs are being operated upon, and this desirable trend will persist, if not accelerate. The age range of these individuals is steadily increasing because of palliative or corrective surgery. Accordingly, we are confronted with a new and expanding patient population that requires, for proper care, knowledge of the surgical modifications as well as knowledge of the unoperated disorders. It is to the latter task—preoperative congenital heart disease in its clinical setting—that I again address myself.

JOSEPH K. PERLOFF

Acknowledgments

The second edition of this book was written while I was Chief of the Cardiovascular Section at the University of Pennsylvania School of Medicine. I am indebted to many colleagues—medical students, house officers, fellows and staff at the Hospital of the University of Pennsylvania, the Children's Hospital of Philadelphia, the Veterans Administration Hospital and the Philadelphia General Hospital—for abundant stimulation and support. I shall always cherish those on my staff from whom I learned so much and who assisted me so faithfully—particularly Drs. W. Bruce Dunkman, John W. Hirshfeld, Fred Holford, H. Edward Holling, Terry Langer, Joel Morganroth, Shirley Rubler and Arthur Whereat.

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To Marie Coscia, a special note of appreciation. It was she who beautifully coordinated the myriad of administrative details during the revision, and it was from her desk that the final typescript emerged.

It is a pleasure to acknowledge the assistance of personnel in Medical Photography and Illustration at the Hospital of the University of Pennsylvania. Joseph T. Rothrock gave advice that set the stage for handling the project, Deborah J. Olsen cheerfully supervised its execution, Steven T. Gigliotti was responsible for new artwork, and Eric Davidson patiently did and redid the final prints until we both were satisfied or at our wits' end.

Dennis C. Wood, Chief Technician in the Cardiac Catheterization Laboratory of the Children's Hospital of Philadelphia, diligently and enthusiastically undertook the angiographic and hemodynamic data search, always securing more and better information than I had anticipated. Kathleen M. Sundra, Chief Technician in

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It is still the convention to acknowledge one's wife for such services as typing the manuscript with fingers worn by housework, for protecting her husband from petty intrusions so he could devote himself to his magnum opus, for attending her knitting while patiently awaiting the return of her hero from an odyssey of scholarly effort. Marjorie did none of these things, but provided instead what I needed most—a genuine understanding of me and my work. My daughters Nancy and Carey, nurtured on academic diets, showed constant interest and fervor and shared with me the delight of completing the final chapter.

JOSEPH K. PERLOFF

Preface to the First Edition

For over a decade I have held a weekly conference on congenital heart disease at the National Heart Institute in Bethesda. I was encouraged by the response to these conferences and have attempted to translate the spirit of dialogue into this book.

I have tried to achieve a healthy balance between clinical cardiology and laboratory information, between man and his instruments, between practical considerations and intellectual curiosity. Without the anatomic and physiologic data supplied by laboratories we remain at best descriptive empiricists limited in our range; on the other hand, without clinical application of laboratory information, we miss a prime purpose of medicine. The indissoluble bond between clinical and laboratory cardiology is expressed in the relationship between the clinical manifestations of congenital heart disease and their underlying mechanisms. Students are entitled to ask, "Why?" and we are obliged to give the answers.

The book does not confine itself to congenital cardiac disease in either children *or* adults but includes *all* age groups in order to give, insofar as possible, a complete, unified and uninterrupted impression of each malformation. It is worth emphasizing that no existing book is devoted exclusively to the *clinical* manifestations of congenital heart disease in *all age groups;* one of my purposes lies in the attempt to provide this needed perspective. In so doing, I have deliberately emphasized pure or relatively pure forms of each anomaly as a means of pointing out the essence of the problem and of avoiding the hopeless feeling of getting lost in a morass of peripheral details. I hope to stimulate clinicians to use the tools at their disposal and to feel that many insights can be gained apart from the laboratory; I also hope to point out to those who are responsible for laboratory investigations that their missions will be more successfully accomplished after sound clinical assessment has shown the way. It is my sincere wish that the point of view employed in this book will prove simple enough to encourage practical use, yet comprehensive enough to interest the specialist.

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Contents

Chapter 1

Introduction: Formulation of the Problem

"The disaster that Hesiod sees threatening a community that disregards justice . . . is not an eternity of damnation but the failure of nature to work —of crops to grow, of herds to bear, of women to produce normal children."[1] Preclassical Greeks were not only aware of congenital malformations but were impressed by their importance. *Congenital* is a Latin derivative of *con,* together, and *genitus,* born. However, the simple implication that congenital heart disease merely means "present at birth" requires qualification. The natural history begins before birth, because most anomalies compatible with six months of intrauterine life permit live offspring at term. A "congenital" anomaly originating in the developing fetus is often considerably modified, at least physiologically, by the dramatic circulatory adjustments at birth.[2] Weeks, months, or even years may then elapse before the anomaly evolves into the "typical" clinical picture. Both physiologic and structural changes subsequently continue, or conversely, the malformation may "vanish."

The ductus in a premature infant sometimes remains widely patent for months, finally closing spontaneously, leaving the baby with a normal heart. A ventricular septal defect that delivers a large left to right shunt in infancy may gradually develop progressive infundibular pulmonic stenosis, so that years later the physiologic and the clinical picture resemble classic cyanotic Fallot's tetralogy. A congenital bicuspid aortic valve that is functionally normal at birth may take two, three or more decades to stiffen, calcify, and present as overt aortic stenosis. Accordingly, congenital heart disease should not be viewed narrowly as a fixed group of anatomic defects present at birth but as a dynamic group of anomalies that originate in fetal life and alter during postnatal development.[2]

Certain defects that are not "anatomic" in the gross morphologic sense are considered congenital, such as congenital complete heart block, whereas others that are "anatomic," such as the aortic root disease of Marfan's syndrome, are, by convention, not dealt with as congenital.

Congenital diseases of the heart are therefore not static in time but change anatomically and physiologically during the course of their natural histories. A given congenital cardiac defect may exist in harmony with the fetal circulation, but it is then confronted with dramatic circulatory changes at birth that alter this harmony to widely varying degrees. It is appropriate to examine briefly the major immediate and delayed circulatory alterations at birth and in the neonatal period. The *immediate* changes consist of: (1) a colossal decrease in pulmonary vascular resistance associated with expansion of the lungs; (2) a pronounced rise in systemic vascular resistance associated with elimination of the low resistance placental circulation; (3) a decrease in blood flow to the right atrium because of abolition of umbilical venous return; (4) an abrupt rise —as much as 10-fold—in

pulmonary blood flow, which is promptly translated into a rise in left atrial volume and pressure; (5) functional closure of the valve of the foramen ovale because of the rise in left atrial and the fall in right atrial pressure; and (6) constriction of the ductus arteriosus at about 12 hours after birth, chiefly in response to an increase in systemic arterial pO_2.

Several important *delayed* changes complete the picture. The thick-walled fetal pulmonary arterioles are designed to meet the full force of systemic right ventricular pressure the instant the lungs expand. After this need has been met, the fetal arterioles involute during the first few months of life. As respiration is established at birth, there is a marked increase in alveolar and systemic arterial oxygen tension to which pulmonary arterioles are exquisitely sensitive, setting the stage for dilatation and anatomic involution. In addition, the larger pulmonary arteries may also play a role, although much lesser, in determining the total drop in pressure across the lungs after birth. Maturational changes may affect both the neonatal disparity in size between the main and branch pulmonary arteries as well as the angulation at the origins of the right and left branches. Both of these factors have been held responsible for a physiologic drop in pressure distal to the pulmonary trunk. The third important delayed change relates to the fetal right ventricle, which slowly loses its relative thickness during the first year of life. Adaptive hypertrophy is an expected feature of the fetal right ventricle, which ejects at systemic pressure via the ductus arteriosus. After birth, with the stimulus of right ventricular afterload eliminated, there is a gradual reduction in its thickness relative to septum and left ventricle. The thick neonatal right ventricular wall does not undergo regression. It merely does not increase its thickness as rapidly as the left in the growing infant.

These physiologic adaptations of the normal heart to the events at birth are remarkable in their own right. It is no surprise that congenital defects of the heart or circulation will, to varying degrees, interact with or be modified by adaptations to extrauterine life. Three selected examples suffice. At one end of the spectrum is the ductus arteriosus, which is a normal part of fetal circulation. When the fetal ductus remains widely patent after birth, however, the neonatal decrease in pulmonary vascular resistance establishes a left to right shunt. Pulmonary blood flow increases, the left ventricle is volume overloaded and may fail under its burden. Thus, a normal structure in the fetus becomes a potentially hazardous congenital defect after birth.

At the other end of the spectrum, there is aortic atresia. This anomaly is characterized by an atretic aortic valve, a rudimentary left ventricular cavity and a rudimentary or atretic mitral valve. The left atrium has no effective exit, but in the fetus this is not a serious handicap because flow into the left atrium via the lungs is negligible and right to left flow across the foramen ovale is not vital. Accordingly, survival to term is the rule because systemic venous blood received by the fetal right heart is pumped into the systemic circulation via the ductus, bypassing the left heart. At birth, however, the lungs expand and pulmonary blood flow suddenly and dramatically increases, abruptly delivering a large volume of blood into a left atrium that has no effective outlet, because forward flow through the left ventricle is totally obstructed by the atretic mitral or aortic valves. Temporary survival depends upon decompression of the left atrium via a herniated valve of the foramen ovale. Death follows shortly.

An intermediate case is large ventricular septal defect, which, though abnormal, does not disturb the fetal circulation because it allows right ventricular blood to enter the aorta in a fashion analogous to the fetal ductus. After birth, however, a decrease in pulmonary vascular resistance establishes a left to right shunt that may significantly disturb the postnatal circulation. Subsequently, the pulmonary vascular resistance may rise again, and in a decade or so, reverse the shunt (Eisenmenger's complex), reestablishing a circulatory state similar to the intrauterine presence of the ventricular septal defect.

The principle to be extracted from these examples is clear. The anatomy and physiology of the heart and circulation in congenital heart disease change with the passage of time from the fetus to the dramatic changes at birth to further changes in the infant, child, adolescent and adult survivor. Some of these changes result in neonatal death; others express themselves gradually over weeks, months, years or decades. A satisfactory comprehension of the clinical manifestations of congenital heart disease requires that these patterns be taken into account and provides the background for clinical recognition.

The clinical diagnosis of congenital heart disease can represent the epitome of applied medical logic. When sound inferences are drawn from accurate observations, correct diagnoses can be made with gratifying frequency. Throughout this book the clinical expressions of congenital cardiac disease are dealt with in terms of the anatomic and physiologic mechanisms responsible for their production. Logical thought is encouraged and memorization minimized. In each chapter the pathologic anatomy is first clarified in order to shed light on the resulting physiologic derangements. The question can then be asked: What clinical manifestations result from these anatomic and physiologic derangements? The stage is then set for clinical diagnosis, which depends upon a synthesis of information derived from the history, the physical signs, the electrocardiogram, the x-ray, and the laboratories (noninvasive and cardiac catheterization). Similarly, the physical diagnosis consists of a synthesis of information from its own five sources, namely, physical appearance, arterial pulse, venous pulse, precordial movements and palpation, and auscultation. It is axiomatic that emphasis must be placed on the *relationship of the parts of the whole*, a relationship that ideally results in a complete, harmonious picture, devoid of internal contradictions, and *not* a loose confederation of unrelated observations. Maximum data should be extracted from each clinical source while relating information from one source to that of another. A simple principle emerges—on the one hand, depth, on the other hand, synthesis. Each step should advance our thinking and narrow the diagnostic possibilities. By the end of the clinical appraisal, untenable considerations should have been abandoned, diagnostic possibilities retained for due consideration and high priority probabilities brought into sharp focus. Conclusions should become more and more refined as the clinical evaluation progresses step by step. The essence of this thesis stems from Herophilus' ancient adage that the best physician is one who is able to distinguish between the possible and the impossible. A single additional word gives this often quoted expression modern relevance—distinguish between the *probable*, the possible and the impossible.

Once an impression is gained from an analysis of the first step in the clinical assessment, this impression necessarily influences the objectivity with which subsequent steps are appraised. Thus, if the same sequence of evaluation is always employed, one never objectively evaluates the latter steps. It is therefore useful to vary the order in which information is assembled. Begin on occasion with the physical examination, or with x-rays or with the electrocardiogram. In infants, it is often practical to take advantage of temporary periods of calm and start with the physical examination, which a short time later may be difficult or impossible. It is not the *sequence* that counts, but rather the depth and synthesis. Irrespective of how the order of assembling information is arranged, two questions must always be asked: How does one step relate to the next? How do all parts relate to the whole?

Diagnostic thinking benefits from the devices of *anticipation* and *supposition*. Anticipate what the next step might reveal and less will be missed. Having drawn tentative conclusions from the history, it is useful to pause momentarily and ask: If these assumptions are correct, what can I expect the physical examination to show? What specific points might I anticipate in the electrocardiogram or x-ray in order to support or refute the conclusions based on the history? The device of anticipation not only helps achieve a synthesis of each step with the next but also serves to heighten interest as the clinical assessment progresses. As a result, confirmations come as sources of satisfaction and errors in judgment stand out in bold relief. Nor should we be afraid of an occasional error. The truth will emerge sooner from error than from confusion. The device of supposition lends itself to the clinical classification of congenital heart disease proposed in Table 1–1. As clinical information becomes available, it can be directly related to this classification so that orderly thinking begins apace. For example, we can ask: Suppose this were a congenital cardiac defect in an acyanotic patient with a left to right shunt, which, if any, of the malformations in this category are appropriate to the information thus far at hand? By simply asking: Suppose this were so, what is likely to follow? one is permitted the dual advantages of thoughtful consideration without inflexible commitment.

The clinical diagnosis of congenital heart disease seems complex when first considered, but the principles are simple and readily understood when information is handled within the framework of an orderly classification. The classification system in Table 1–1 is employed because it is both *practical* and *clinical* and can be used effectively, irrespective of which of the five sources of infor-

Table 1–1 A CLINICAL CLASSIFICATION OF CONGENITAL HEART DISEASE

General

Innocent or normal murmurs
Congenitally corrected transposition of the great arteries
Congenital positional anomalies of the heart—cardiac malpositions
Congenital complete heart block

Acyanotic without a Shunt

Malformations originating in the left heart

1. Aortic stenosis
   a. Valvular
   b. Discrete subvalvular
   c. Supravalvular
2. Congenital aortic regurgitation
3. Coarctation of the aorta
4. Congenital mitral regurgitation
   a. Endocardial cushion defect
   b. Congenitally corrected transposition of the great arteries
   c. Primary endocardial fibroelastosis (dilated)
   d. Anomalous origin of the left coronary artery from the pulmonary trunk
   e. Miscellaneous (double orifice mitral valve, congenital perforations, accessory commissures with anomalous chordal insertion, congenitally short or absent chordae, cleft posterior leaflet, parachute mitral valve, etc.)
5. Primary endocardial fibroelastosis
6. Congenital obstruction to left atrial flow
   a. Cor triatriatum
   b. Mitral stenosis
   c. Pulmonary vein stenosis

Malformations originating in the right heart

1. Pulmonic stenosis
   a. Valvular
   b. Infundibular
   c. Supravalvular (stenosis of the pulmonary artery and its branches)
   d. Subinfundibular
2. Idiopathic dilatation of the pulmonary trunk
3. Congenital pulmonary valve regurgitation
4. Primary pulmonary hypertension
5. Ebstein's anomaly of the tricuspid valve

Acyanotic with a Shunt (Left to Right)

Shunt at atrial level

1. Atrial septal defect (isolated)
   a. Ostium secundum
   b. Ostium primum
   c. Sinus venosus
2. Atrial septal defect with mild pulmonic stenosis
3. Total anomalous pulmonary venous connection with low pulmonary vascular resistance
4. Partial anomalous pulmonary venous connection with intact atrial septum
5. Atrial septal defect with mitral stenosis (Lutembacher's syndrome)

Shunt at ventricular level

1. Ventricular septal defect (isolated)
   a. Infracristal
   b. Supracristal
   c. Muscular
   d. Endocardial cushion location
2. Ventricular septal defect with mild pulmonic stenosis
3. Ventricular septal defect with right ventricular origin of both great arteries
4. Ventricular septal defect (infracristal) with congenitally corrected transposition of the great arteries
5. Ventricular septal defect with aortic regurgitation
6. Ventricular septal defect with left ventricular to right atrial shunt

Shunt between aortic root and right heart

1. Coronary arteriovenous fistula
2. Ruptured sinus of Valsalva aneurysm
3. Anomalous origin of the left coronary artery from the pulmonary trunk

Shunt at aorticopulmonary level

1. Patent ductus arteriosus
2. Aorticopulmonary septal defect
3. Truncus arteriosus with large pulmonary arteries and low pulmonary vascular resistance

Shunts at more than one level

1. Complete endocardial cushion defect (complete persistent common atrioventricular canal)
2. Ventricular septal defect with patent ductus arteriosus
3. Ventricular septal defect with atrial septal defect

Cyanotic

Increased pulmonary blood flow

1. Complete transposition of the great arteries
2. The Taussig-Bing anomaly (right ventricular origin of both great arteries with supracristal ventricular septal defect or right ventricular aorta with biventricular pulmonary trunk)
3. Truncus arteriosus with large pulmonary arteries
4. Total anomalous pulmonary venous connection
5. Single ventricle with low pulmonary resistance and absent or mild pulmonic stenosis
6. Common atrium
7. Fallot's tetralogy with pulmonary atresia and increased collateral arterial flow
8. Tricuspid atresia with large ventricular septal defect and no pulmonic stenosis
9. Atrial septal defect with caval connection to left atrium

Normal or decreased pulmonary blood flow

1. Dominant left ventricle
   a. Tricuspid atresia
   b. Ebstein's anomaly with right to left interatrial shunt (mechanical dominance)
   c. Pulmonary atresia with intact ventricular septum and diminutive right ventricle
   d. Congenital vena caval to left atrial communication
   e. Single ventricle with pulmonic stenosis and noninversion of the infundibulum
   f. Large pulmonary arteriovenous fistula in infancy
2. Dominant right ventricle
   Normal or low pulmonary arterial pressure
   a. Pulmonic stenosis or atresia with ventricular septal defect and right to left shunt (cyanotic Fallot's tetralogy)
   b. Pulmonic stenosis with right to left interatrial shunt
   c. Complete transposition of the great arteries with severe pulmonic stenosis

Table 1–1 A CLINICAL CLASSIFICATION OF CONGENITAL HEART DISEASE (*Continued*)

d. Pulmonic stenosis with right ventricular origin of both great arteries
e. Pulmonic stenosis with single ventricle and inversion of the infundibulum (electrical dominance)
f. Truncus arteriosus with hypoplastic or absent pulmonary arteries

Elevated pulmonary arterial pressure (pulmonary hypertension)

a. Atrial septal defect with reversed shunt
b. Ventricular septal defect with reversed shunt (Eisenmenger's complex)
c. Patent ductus arteriosus or aorticopulmonary septal defect with reversed shunt
d. Right ventricular origin of both great arteries with high pulmonary vascular resistance
e. Hypoplastic left heart (aortic atresia, mitral atresia, complete interruption of the aortic arch)
f. Complete transposition of the great arteries with high pulmonary vascular resistance
g. Single ventricle with high pulmonary vascular resistance
h. Total anomalous pulmonary venous connection with high pulmonary vascular resistance

3. Normal or nearly normal ventricles
   a. Pulmonary arteriovenous fistula
   b. Congenital vena caval to left atrial communication

mation one is dealing with. There are shortcomings in any classification, but these shortcomings, provided they are recognized and minimized, should not obscure the value of a practical, orderly grouping. *The fact that a number of defects are listed in more than one category simply emphasizes the variability of their clinical expressions.* The classification, first proposed by Paul Wood more than two decades ago, is based essentially on the answers to the five simple questions found in Table 1–2. It is not even necessary to ask all five questions for each patient, although the first two questions are obligatory. If the answer to question 1 is acyanotic, only two additional questions need be posed, namely: Is a shunt absent or present? (i.e., is pulmonary arterial flow increased or not), and second, if a shunt is absent, on what side of the heart, left or right, does the malformation originate (Fig. 1–1)? On the other hand, if the answer to question 1 is cyanotic, additional questions must be asked. First: Is pulmonary arterial flow increased or not? If the answer is that flow is normal or decreased we need to ask: Which is the dominant ventricle? and Is pulmonary hypertension present or absent (Fig. 1–2)?

Let us illustrate by dealing first with an *acyanotic* patient (Fig. 1–1). If the patient is acyanotic and the answer to question 2 is negative (acyanotic without shunt), we must then

Table 1–2 FIVE BASIC QUESTIONS

1. Is the patient acyanotic or cyanotic?
2. Is pulmonary arterial flow increased or not?
3. Does the malformation originate in the left or right heart?
4. Which is the dominant ventricle?
5. Is pulmonary hypertension present or absent?

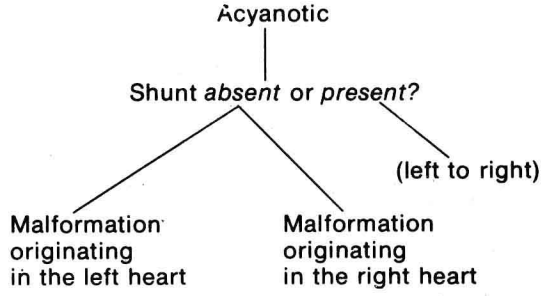

Figure 1–1

ask: Does the malformation originate in the left or right heart (Table 1–2, Fig. 1–1)? Now move step by step through the heart in the direction of blood flow (Table 1–3). If the malformation originates in the right heart, is it at the level of the vena cava (left superior cava), right atrium, tricuspid valve, right ventricular inflow (Ebstein's anomaly), right ventricular outflow, pulmonary artery and its branches (pulmonic stenosis) or pulmonary arterioles (primary pulmonary hypertension)? Similarly, if the malformation originates in the left heart (Table 1–4), is it at the level of the pulmonary veins (pulmonary vein stenosis), left atrium (cor triatriatum), mitral valve (mitral stenosis), left ventricular inflow (endocardial fibroelastosis), left ventricular outflow (aortic stenosis) or thoracic aorta (coarctation of the aorta)? If the answer to question 2 (Table 1–2) is that pulmonary arterial blood flow is increased in an acyanotic patient, then the shunt by definition is left to right, because cyanosis is absent. We then can consider methodically the origin of the shunt and the chamber or vessel that receives it. Again, move step by step through the heart in the direction of blood flow (Table 1–5), i.e., shunts at atrial level (atrial septal defect), ventricular level (ventricular septal defect), from

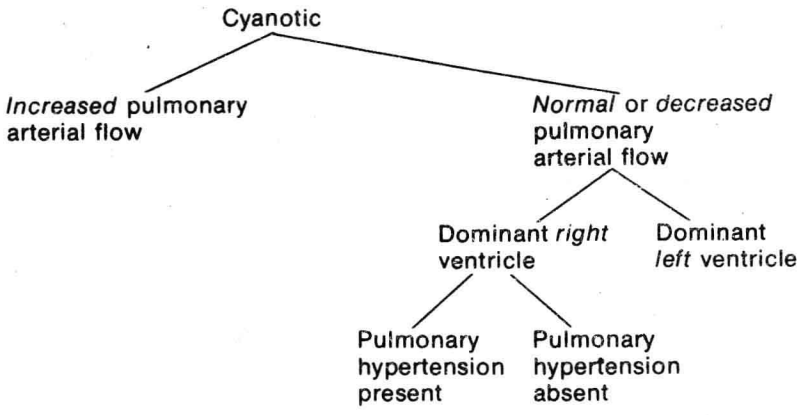

Figure 1–2

Table 1–3 ACYANOTIC WITHOUT SHUNT, MALFORMATION ORIGINATING IN RIGHT HEART

Start proximally (vena cavae) and end distally (pulmonary arteries)
Vena cavae
Right atrium
Tricuspid valve
Right ventricular inflow
Right ventricular outflow
Pulmonary artery and branches
Pulmonary arterioles

Table 1–4 ACYANOTIC WITHOUT SHUNT, MALFORMATION ORIGINATING IN LEFT HEART

Start proximally (pulmonary veins) and end distally (aorta)
Pulmonary veins
Left atrium
Mitral valve
Left ventricular inflow
Left ventricular outflow
Thoracic aorta

Table 1–5 ACYANOTIC WITH SHUNT

Where is the left to right communication, i.e., where does it originate and what chamber or vessel receives the shunt?
Start proximally and end distally
Atrial level
Ventricular level
Great artery level (aortic root, aortic arch)

the aortic root to right heart (Table 1–1) or from aortic arch to pulmonary artery (patent ductus) (Tables 1–1 and 1–5).

Now, let us deal with cyanotic patients (Figs. 1–2 and 1–3). If pulmonary arterial blood flow is *increased*, there are, for all practical purposes, about eight or nine possibilities (Table 1–1, cyanotic with increased pulmonary arterial blood flow), only three of which are relatively common: complete transposition of the great arteries, truncus arteriosus and total anomalous pulmonary venous connection. If pulmonary arterial flow is *normal or decreased* and the *left* ventricle is dominant, there are six possibilities, only two of which are likely, i.e., tricuspid atresia or Ebstein's anomaly of the tricuspid valve (Table 1–1). If the *right* ventricle is dominant, question 5 must be asked: Is pulmonary hypertension present of absent? If *pulmonary hypertension* is present, again move step by step through the heart in the direction of blood flow (Table 1–6). Is one dealing with a pulmonary hypertensive right to left shunt at atrial, ventricular or great artery level, i.e., reversed shunt through a pulmonary hypertensive atrial septal defect, ventricular septal defect or

Table 1–6 CYANOTIC WITH NORMAL OR DECREASED PULMONARY ARTERIAL FLOW, DOMINANT RIGHT VENTRICLE, AND PULMONARY HYPERTENSION

Where is the right to left shunt?
Start proximally and end distally
Atrial level
Ventricular level
Great artery level

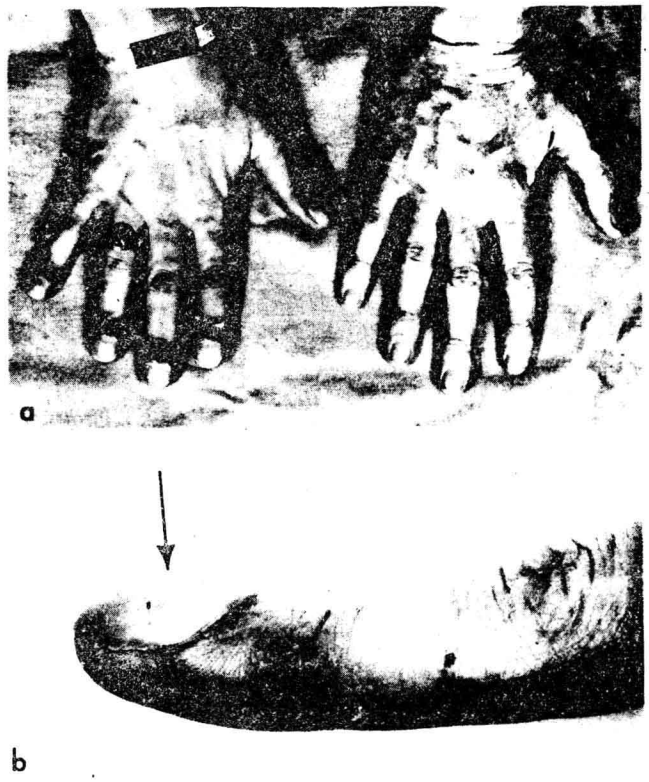

Figure 1–3 *a*, Typical cyanosis and clubbing of the fingers in a young adult (left) compared to normal (right). *b*, Close-up profile of clubbing (arrow).

patent ductus arteriosus? *Absent pulmonary hypertension* (Table 1–7) necessarily implies obstruction to outflow into the pulmonary bed, i.e., normal or low pulmonary arterial pressure with high right ventricular pressure. In practical terms, the probabilities are two: pulmonic stenosis with right to left shunt at either atrial or ventricular level, i.e., pulmonic stenosis with reversed flow through a foramen ovale or atrial septal defect, or through a ventricular septal defect (Fallot's tetralogy).

When approached according to the principles espoused in this introduction, the *clinical recognition of congenital heart disease* becomes at once a stimulating challenge, a satisfying discipline in logical thinking and a constant source of self-education.

Two other points need be made. First, no specific commentary on prognosis is made in this book. Prognosis simply refers to the expected natural history once a diagnosis is made. Viewed in this light, prognosis is simply a part of the natural history and is treated as such. Second, traditional catalogs or listings of differential diagnoses are not employed. Instead, each chapter ends with a summary or synthesis that states concisely the essence of the clinical manifestations of the congenital anomaly just covered in detail. The summaries are designed to bring together the highlights of the clinical recognition of each malformation and serve as concise references to which the reader can conveniently return.

Table 1–7 CYANOTIC WITH NORMAL OR DECREASED PULMONARY ARTERIAL FLOW, DOMINANT RIGHT VENTRICLE, AND NO PULMONARY HYPERTENSION

Pulmonic stenosis or atresia with right to left shunt at ventricular level
Pulmonic stenosis with right to left shunt at atrial level

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