CLINICOPATHOLOGIC CORRELATIONS

Conditions Simulating the Tetralogy of Fallot

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SUMMARY
Cyanosis on the basis of a right-to-left shunt at the ventricular level results from pulmonary stenosis associated with a ventricular septal defect. While the tetralogy of Fallot is the most common anatomic state underlying this functional abnormality, there are a variety of anatomic states which may yield the same functional disturbance. These may be divided into three groups depending upon the state of the major arterial vessels. In each of the first two of the three groups, there are two arterial vessels. In one group, the great vessels are in normal relationship to each other, while in the second group the great arteries are transposed. In the third group, one arterial vessel leaves the heart and, from it, branches carry blood to the lungs.

Additional Indexing Words:
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Subpulmonary ventricular septal defect and pulmonary stenosis

In Patients cyanotic on the basis of pulmonary stenosis and a ventricular septal defect, the usual malformation is the tetralogy of Fallot. Yet there are exceptional conditions where the anatomic features of the tetralogy are absent, which have a defective ventricular septum in the presence of pulmonary stenosis, and a right-to-left shunt occurring at the ventricular level. These conditions, although uncommon, deserve attention since the operative steps for cure are different from those in the tetralogy.

The exceptional conditions may be divided into three major groups depending upon the state and relationship between the great arteries.

The first two groups are characterized by the presence of both the aorta and the pulmonary trunk. The first of these exhibits a normal relationship between the great vessels, as in the tetralogy, while the second shows a reversed anteroposterior relationship between these vessels (transposition). The third group is characterized by only one artery leaving the heart.

Normal Relationship of Two Great Arteries

In order to distinguish the tetralogy from other conditions, it is appropriate first to summarize the essential anatomic features of the tetralogy of Fallot.

Tetralogy of Fallot

In specimens with the tetralogy of Fallot in which an incision has been carried from the anterior wall of the right ventricle into the aorta (fig. 1a), all of the essentials of the condition may be demonstrated. The aorta arises from both ventricles above a ventricular septal defect. The ventricular septal defect is separated from the right ventricular infundibulum by a prominent vertical crista supraventricularis. The latter structure, by virtue of its mass and position, forms the important basis for the narrow infundibulum (subpulmonary stenosis) which is a characteristic of the tetralogy. Pulmonary valvular stenosis may be present additionally. The aorta, in arising from both ventricles, shows the normal feature of its valve making continuity with the mitral valve (fig. 1b).

Subpulmonary Ventricular Septal Defect with Pulmonary Stenosis

The subpulmonary ventricular septal defect (so-called supracristal defect) is a relatively uncommon
Tetralogy of Fallot. a. A section through the anterior wall of the right ventricle (R. V.) has been carried into the aorta (A.). This shows the latter vessel arising from both ventricles above a ventricular septal defect (probe). The narrow infundibulum (I.) is formed by a prominent vertical crista supraventricularis (C.) and the anterior wall of the right ventricle. b. Sagittal section showing the right and left ventricles (R. V. and L.V.), as well as the sectioned ventricular septum (V. S.). The aorta (A.) arises above the ventricular septum from both ventricles. Classically, as shown, the aortic valve (A. V.) makes continuity with the anterior mitral leaflet (A.M.).

Figure 2
Subpulmonary ventricular septal defect and pulmonary stenosis. The opened right ventricle (R. V.) leads to the pulmonary valve (P. V.) which was bicuspid and stenotic. The ventricular septal defect (D) lies immediately beneath the pulmonary valve and, through it, elements of the aortic valve (A. V.) are visible. From a 5-month-old patient.

Figure 3
Subpulmonary ventricular septal defect lies beneath the pulmonary valve (P. V.). Elements of the aortic valve (A. V.) are visible through the subpulmonary ventricular septal defect. An anomalous muscle bundle was responsible for pulmonary stenosis in this 8-year-old cyanotic patient.
type of ventricular septal defect but particularly uncommon in association with pulmonary stenosis.

We have observed two such cases, each in a cyanotic patient, one aged 5 months (fig. 2) and the other aged 8 years (fig. 3). In each, the ventricular septal defect lay immediately beneath the pulmonary valve. Through the defects, elements of the aortic valve joined the pulmonary valve leading to a situation in which the aortic valve presented in the upper part of the right ventricular infundibulum. In the younger patient, a stenotic bicuspid pulmonary valve accounted for pulmonary stenosis. In the older patient, an anomalous muscle bundle of the right ventricle and a stenotic bicuspid pulmonary valve caused pulmonary stenosis. In each, the clinical diagnosis was the tetralogy of Fallot. It is of note that in each the right ventricular infundibulum did not exhibit the characteristic deformity of the tetralogy of Fallot. Retrospective review of the angiocardiograms revealed changes suggesting the condition present.

It is of interest that the angiocardiographic features of the supracristal type of ventricular septal defect without pulmonary stenosis have been described and this may serve as a basis for identification of this type of defect in association with pulmonary stenosis.

Double Outlet Right Ventricle with Subpulmonary Stenosis

Double outlet right ventricle is characterized by both great vessels arising from the right ventricle without continuity between the aortic and mitral valves. The relationship between the great vessels at the cardiac junction is abnormal but at higher levels the relationship may either approach normal or show obvious transposition. If pulmonary stenosis is present, its basis is usually a malformation of the right ventricular infundibulum similar to that in the tetralogy of Fallot (fig. 4a).

In the tetralogy there is variation as to the proportion of the aorta which arises from the right ventricle. There are exceptional cases of the tetralogy in which the aorta arises almost entirely above the right ventricle (fig. 4b). While in such cases the surgical challenges are similar to those in double outlet right ventricle, continuity of the aortic

Figure 4

a. Origin of both great vessels from right ventricle with subpulmonary stenosis. The aorta (A.) arises exclusively from the right ventricle (R. V.). The probe shows the position of the ventricular septal defect which was the only outlet for the left ventricle. Subpulmonary stenosis is the result of a narrow right ventricular infundibulum (I.). b. An example of the tetralogy of Fallot in which the aorta (A.) arises more from the right ventricle than average. Although the origin of the aorta is dominantly from the right ventricle (R. V.), the aortic and mitral valves were in continuity. I. = right ventricular infundibulum; P. T. = pulmonary trunk.
Figure 5
Usual ventricular septal defect associated with stenosis of the pulmonary valve. a. View of the right ventricle (R. V.) and opened pulmonary trunk (P. T.). The pulmonary valvular cusps are short and fibrotic, yielding pulmonary stenosis. Beneath the crista supraventricularis is the position of the ventricular septal defect (arrow). b. View of left ventricle (L. V.) and aorta (A.). The ventricular septal defect (D.) lies in a common position and is overhung by the septal leaflet of the tricuspid valve (T.) which is seen in the left ventricle. A. M. = anterior leaflet of mitral valve.

and mitral valves serves, for classification purposes, to label such cases as examples of the tetralogy of Fallot.

Tetralogy and Endocardial Cushion Defect
Among some patients, often but not universally with Down's syndrome, the same heart may show both the tetralogy of Fallot and the endocardial cushion defect (persistent common atrioventricular canal). The usual clinical presentation resembles classical tetralogy of Fallot. That the endocardial cushion defect is associated may be suspected when the electrocardiographic and/or angiocardiographic features of the latter condition are also shown.4

Usual Ventricular Septal Defect and Pulmonary Stenosis
A relatively uncommon condition which yields a functional state like that in the tetralogy is that in

Figure 6
Stenosis of origins of major pulmonary arteries associated with ventricular septal defect. When the pulmonary arterial stenosis is severe, a right-to-left shunt may occur through the ventricular septal defect.
which the basic structure of the right ventricle is normal. The ventricular septal defect lies in a common position for isolated ventricular septal defect, that is, beneath the normally positioned crista supraventricularis and overhung by the septal leaflet of the tricuspid valve. Pulmonary stenosis resulting from a valvular malformation such as hypoplasia or dysplasia of the valve or a dome-shaped stenosis accounts for obstruction to pulmonary flow (fig. 5).

**Ventricular Septal Defect and Pulmonary Arterial Stenosis**

Stenosis of pulmonary arteries may involve one or several foci. The lesions may occur either in the major arterial trunks or in peripheral branches. The ventricular septum may be intact or show a defect. When a defect is present and the pulmonary arterial obstruction is of such a degree as to offer greater resistance to flow than does the systemic arterial bed, a right-to-left shunt occurs (fig. 6). The result may yield cyanosis.

**Single Ventricle**

Single ventricle with normally related great vessels and subpulmonary stenosis may yield clinical and roentgenologic states strikingly like those in the tetralogy. The basis for pulmonary stenosis is usually a stenotic tract at the base of the ventricle which leads to the pulmonary trunk (fig. 7a). Of essence in the clinical diagnosis is to demonstrate absence of a ventricular septum.

**Transposition of the Two Great Arteries**

When pulmonary stenosis on the basis of a right-to-left ventricular shunt is associated with transposition of the great vessels (excluding complete transposition), the conditions to be considered are single ventricle and corrected transposition, each with pulmonary stenosis. One should also bear in

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**Figure 7**

Single ventricle with pulmonary stenosis. a. Normally related great vessels. The pulmonary stenosis is at the subpulmonary level. b. Single ventricle associated with transposition of the great vessels. The obstruction to pulmonary flow is in the subaortic region.

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mind that certain cases of double outlet right ventricle exhibit an anteriorly-lying aorta and posterior pulmonary trunk.

**Single Ventricle**

The single ventricle associated with transposition need not exhibit pulmonary stenosis. When pulmonary stenosis is present, cyanosis is usually evident. The zone of stenosis is usually below the level of the pulmonary valve (fig. 7b), although valvular stenosis may be present either as the sole basis for obstruction to pulmonary flow or associated with subpulmonary stenosis. Mitral or tricuspid atresia may be associated and the former condition may be particularly occult clinically.

**Corrected Transposition**

The basic anatomic state of corrected transposition has been amply described. In this condition, the association of a ventricular septal defect and obstruction to pulmonary flow may account for a right-to-left shunt at the ventricular level. The obstruction to pulmonary flow may take the form of atresia at the valvular level or stenosis beneath the valve. In our experience, subpulmonary stenosis in corrected transposition may result from accessory tissue of the right atrioventricular valve which obstructs the related ventricular outflow tract (fig. 8).8

**Single Arterial Vessel**

A variety of anatomic states exists in which a ventricular septal defect is associated with only one artery arising from the heart. The pulmonary arterial supply is derived from that vessel. Bidirectional shunts are universal. Cyanosis, which is commonly present, is related to impaired flow through the arteries carrying blood into the lungs. The various anatomic states which pertain have recently been reviewed by Edwards and McGoon.9

**References**


*Figure 8*

Subpulmonary stenosis in corrected transposition. View of the opened pulmonary trunk (P. T.) and the base of the venous ventricle. Attached to the anterior leaflet of the anatomic mitral valve ("M. V.") is accessory tissue (A. T.) which causes obstruction of the subpulmonary region of the ventricle. A ventricular septal defect (D.) is present through which a right-to-left shunt has occurred.
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