Early anatomical knowledge of cardiac malformation

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So many cardiologists in the past twenty-five years have been concerned with cardiac malformations that the following notes may be of interest.

Our anatomical knowledge of the common malformations of the heart and of many less common ones goes back a long time. Stenson (1672) is generally credited with the first description of what has become known as Fallot's tetralogy. Bonet (1679), among many good accounts of necropsies, gave a graphic description of the aortic stenosis that was probably congenital as he was 'not yet old'.

A Parisian tailor, not yet old, having dined and left his house had walked hardly 40 paces when he suddenly fell to the ground and expired. His body was opened and no disease was found except that the three semilunar cusps leading to the aorta were bony' (Bonet, 1679, translated by White, 1951).

In the 18th century Johannes Senac (1749) wrote a great book on the structure and action of the heart and its diseases, with a beautiful engraving of himself as a frontispiece. He described the closing of the 'trou oval' and the 'canal artériel' and realized that sometimes these failed to close. His short chapter on malformations included a case of complete absence of the ventricular septum, and he rightly explained the cyanosis as being due to an admixture of venous and arterial blood.

Morgagni (1761) described a patient with pulmonary stenosis and an atrial septal defect who had been cyanosed during life. He attributed the cyanosis to venous stasis and started a controversy that recurred for many years.

Edward Sandifort (1742–1814) was a professor of physiology and much else at Leyden, a near contemporary of the great Boerhaave who first incorporated scientific chemistry into medicine. In his Observations Anatomico-pathologicae (1777), under the heading 'Concerning a very unusual malady of the heart in a child blue from birth', he gave a clear description of Fallot's tetralogy with good pictures of the malformation. He wrote in Latin but it has been translated by Bennett (1946).

William Hunter (1784) described three examples of pulmonary stenosis or atresia with a ventricular septal defect. He gave a good account of attacks of cyanosis, dyspnoea, and unconsciousness. Coarctation of the aorta was described for the first time by Paris (1791).

Much more was added in the 19th century. It seems worth mentioning one of the first books on maladies of the heart by Corvisart (1806), Napoleon's chief physician; and another by Gintrac (1824) on cyanosis.

More important for our present purpose was Farre's small book Pathological Researches. Essay 1. On Malformations of the Human Heart (1814), the first to cover this field widely. He wrote about the foramen ovale that was still open or dilated. It might be associated with an open ductus arteriosus alone or with pulmonary atresia, or with stenosis of the 'ostium arteriae pulmonalis'.

He wrote also of ventricular septal defects and of transposition, of the pulmonary artery or the aorta being common to both ventricles, and of pulmonary stenosis as a lobe lesion. Finally he wrote of stenosis of the 'ostium aortae' and described the case of a woman aged 24 who died suddenly during pregnancy, and had a greatly narrowed aortic orifice with only two semilunar cusps that were severely ossified.

The importance of the collateral circulation in coarctation of the aorta in providing an adequate blood flow to the lower parts of the body through the intercostal arteries was recognized by Meckel (1827), Reynaud (1828), and Jordan (1830), only a generation after its first description by Paris (1791). Reynaud's
specimen came from a man who died at the age of 92.

From 1835 onwards coarctation was diagnosed in life from time to time. Craige (1841), who first put forward the theory that coarctation was caused by the obliteration of the ductus arteriosus, collected 10 reported cases with necropsies. Peacock (1860) added another 29, and with Rokitansky's cases Barié (1886) had increased the number to 88. Vierordt (1898) raised it to 126 and Bonnet (1903) to 160: he distinguished clearly between infantile (55) and adult (105) types, of which a quarter showed almost complete obliteration of the aorta with a good collateral circulation. No other cardiac malformation was so well documented at this time.

Paget (1844) wrote 'in the majority of cases in which only two valves have been found in the aorta, these valves have been diseased'. Peacock (1853, 1865, and 1868) thought that bicuspid aortic valves were likely to become inflamed and thickened and later osseous so that there was increasing stenosis and regurgitation. He reported several cases including a man, aged 45, with very severe stenosis and bony valves (heart weight, 567 g.); and a boy aged 18, with stenosis and more regurgitation and a heart weighing 794 g. There is still one of Peacock’s beautiful specimens of calcific bicuspid aortic stenosis in the museum of St. Thomas’ Hospital (Smith and Matthews, 1955, Fig. 1). Clearly Peacock saw many examples and thought that bicuspid aortic valves were a not uncommon cause of aortic stenosis. It is strange how little attention was paid to this view for nearly a hundred years. He was familiar with the frequent association of bicuspid aortic valves with coarctation of the aorta.

Thomas Peacock (1812–82), physician to St. Thomas’ Hospital, wrote widely on the morbid anatomy of cardiac malformations. He described simple pulmonary stenosis and summarized his results in *On Malformations of the Human Heart* (2nd ed., 1866) with excellent illustrations. When in 1946 I was first interested in Fallot’s tetralogy, I read his book and thought the descriptions of valvar and infundibular stenosis, and of the shunt from the right ventricle through the ventricular septal defect to the aorta, could hardly be bettered.

Though Peacock was a good physician, he was also like many hospital physicians of his day a good morbid anatomist. His book, like most early work on cardiac malformations, was written primarily from this point of view. There are good case histories but not much discussion of the clinical aspects. James Hope (1801–41) had correctly diagnosed Fallot’s tetralogy in life in one of Peacock’s necropsy cases.

Henriette, Ferber, and Hiffe in the 1860’s are credited with the earliest descriptions of tricuspid atresia by Laubry and Pezzi (1921). Their reference to the famous Holmes heart cannot be accepted because it was an example of cor bia trium at triloculare (Abbot, 1936, p. vii).

About the same time, Ebstein (1866) described the curious and rather uncommon anomaly named after him. The tricuspid valve is grossly misplaced and malformed and sometimes the foramen ovale is patent and the patient cyanotic.

Carl Rokitansky (1804–78), professor of pathological anatomy at Vienna, will always be associated with atrial and ventricular septal defects. His great book *Die Defekte der Scheidewände des Herzens* (1875), with its handsome and dignified portrait of him as rector of the university, contains beautiful drawings of most types of septal defects. He described clearly the distinction between the ostium primum and ostium secundum types of atrial septal defect. There are many case histories but not much clinical discussion.

The blind acceptance of the view of Roger (1879) that all lone ventricular septal defects produced striking physical signs but no symptoms is one of the most curious medical errors. It was based on a few patients Roger had seen and his memory of a necropsy about 18 years before on a boy. It was 12 years before there was necropsy proof of such a case that had been diagnosed in life by Dupré (1891), who suggested the unfortunate name 'maladie de Roger'. As regards defects of small size, it was a lucky inspiration, but it is hard to understand what diagnosis was made of the larger defects that are more common.

Osler (1886) emphasized the frequency of bicuspid aortic valves and how often they were attacked by bacterial endocarditis, a view that was widely accepted, unlike that of Peacock. He put forward some evidence that they were true malformations and not due to foetal endocarditis. He found 18 examples in 800 necropsies, at least 4 times as many as are usually found, though bicuspid aortic valves are the most common malformation of the heart. Only 3 died from unconnected causes, 6 dying from bacterial endocarditis, 6 from congestive heart failure, 2 suddenly, and 1 from rupture of an intracranial aneurysm.

No reason was found for the two sudden deaths—in a woman of 25, in whom heart disease had been diagnosed for some years, and in a blacksmith aged 45. Nor is it obvious...
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why many others died with hypertrophied left ventricles and heart failure, for at several necropsies Osler demonstrated the absence of aortic regurgitation and did not mention aortic stenosis. Is it possible that he did not speak of aortic stenosis because of the fairly long opening, the obstruction being due to the thickening and rigidity of the cusps?

Many pathologists and physicians had described pulmonary stenosis with a ventricular septal defect, allowing venous blood to pass into the aorta and thus to cause clinical cyanosis, since 1672. It seems strange, therefore, that the condition should be called after Antoine Fallot (1888), professor of pathological anatomy at Marseilles, who wrote about 55 reported examples and included three of his own cases. Perhaps his best claim to this distinction is the following abbreviated abstracts from his conclusions.

Physicians have until now considered the precise anatomical diagnosis of ‘la maladie bleue’ as almost impossible. From the assembled observations, it appears that it is the result of a small number of conditions, especially if infants are excluded. In three-quarters of the cases it is due to the following tetralogy: (1) stenosis of the pulmonary artery or of the infundibulum, (2) ventricular septal defect, (3) deviation of the origin of the aorta to the right, and (4) right ventricular hypertrophy, almost always concentric.

In the fourth condition is a secondary consequence of the high pressure in the right ventricle, and the third condition may be a secondary effect of the large flow of blood from the left ventricle to the aorta at first indirectly through the right ventricle. But certainly it was a masterly summary. It is necessary to exclude infants because in them transposition of the great trunks is about an equally common cause of cyanosis. But so many of these subjects die in the first year that afterwards Fallot’s tetralogy is much the commonest cause, with tricuspid atresia and Eisenmenger’s syndrome a long way behind.

Persistent ductus arteriosus had been known at necropsies for a long time. More than a century ago Wilkinson King ‘correctly diagnosticated patencies of the ductus’ in a patient of Addison, whose heart in the Guy’s Hospital Museum has long been familiar to me, and Chevers wrote about it in 1845. Bernitz (1849) also made the diagnosis in life.

Gerhardt (1867) gave an account of the murmur and Rokitansky (1875), in describing the condition, included clinical observations of Joseph Skoda (1805–81), one of the pioneers of percussion and auscultation. But the credit for making the continuous murmur more widely known must go to Gibson (1898, 1900) of Edinburgh, who wrote so clearly about its unique continuous quality.

Thérémin (1895) produced a large atlas of good drawings of most cardiac malformations. He had obtained his material for it when working at ‘la maison des enfants trouvés’ at St. Petersburg.

Eisenmenger (1897) described another condition that caused cyanosis – a ventricular septal defect accompanied by a right-to-left (reversed) shunt and right ventricular hypertrophy without pulmonary stenosis. Generally the reversed shunt was attributed to a dextroposed aorta, but Eisenmenger (1898) thought himself that this could be the result rather than the cause of the reversed shunt. It is, of course, merely a variety of ventricular septal defect with enough pulmonary hypertension to reverse the shunt.

Carpenter (1894), a physician at the Evelina Hospital for Children, wrote what was perhaps the first purely clinical book on Congenital Affections of the Heart. He was often content with this general diagnosis, though there are some specific diagnoses. The book is mainly sensible advice about what should be said to the parents of such children, a state of affairs that lasted much longer among many doctors and physicians.

It is appropriate to end this section with Professor Hermann Vierordt of Tübingen. His Die Angeborenen Herzkrankheiten (1898) gives the most complete picture of all that was known with extensive references to previous work. Truncus arteriosus, transposition of the great vessels, infundibular and valvar aortic stenosis, and tricuspid atresia are included. He mentioned patients with a persistent ductus not dying till they were 60, 58, 51, and 47 years of age, suggesting that most of them died much earlier. He pointed out that other malformations were unduly common in those with cardiac malformations.

It would be fair to say that the morbid anatomy of these malformations received more attention than the clinical diagnosis. Coarctation of the aorta and pulmonary stenosis with septal defects occupied a more prominent place than the equally common simple lesions, persistent ductus, atrial and ventricular septal defects, and simple pulmonary and aortic stenosis, though Peacock had emphasized the frequency of the last condition.

References


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