Tetralogy of Fallot with Pulmonary Obstruction at the Level of the Conus Inlet

A CASE REPORT

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SUMMARY

A case of Fallot's tetralogy is described in a Black male who died of acute cardiac failure at the age of 17 years. The conus arteriosus was practically a separate chamber communicating with the right ventricle through a very small ostium. The embryology of the truncus arteriosus the bulbus cordis is discussed in the light of the anomalies described here. The question of maintenance of the pulmonary circulation in the absence of an open ductus arteriosus is discussed.

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CLINICAL HISTORY

The patient was sent to the H. F. Verwoerd Hospital in cardiac failure. Left heart catherisation was done and a congenital ventricular septal defect and pulmonary stenosis were diagnosed. Open-heart surgery was advised but the parents of the patient could not be traced. The patient suddenly collapsed and died of acute cardiac failure. Because no relatives could be located the body was sent to the Department of Anatomy, University of Pretoria.

During routine dissection of the thorax by medical students an enormously enlarged heart was found (Fig. 1). It weighed 950 g. The body was that of a Black male, aged 17 years, height approximately 1,625 m and mass 58 - 60 kg. These measurements and mass determinations were all done on the embalmed body.

DESCRIPTION OF THE ANATOMICAL FINDINGS

The heart was dissected *in situ* and showed an over-all enlargement. The ductus arteriosus was completely obliterated and no enlargement of the bronchial arteries was observed. The interior of the right atrium showed very prominent valves for the inferior vena cava and the coronary sinus. The interatrial wall of the fossa ovalis was extremely thin and a punched-out defect about 0,4 mm in diameter could be seen. The right ventricle was enlarged and the thickness of the wall was 13,5 mm, compared with the 12,5 mm thickness of the left ventricle. The right ventricle communicated with the left ventricle through a very large defect of the interventricular septum, which easily admitted 3 fingers and which was straddled by the aorta. The right ventricle was completely demarcated from the infundibulum or conus arteriosus, the only connection being an ostium of 7,5 mm in diameter.

The conus arteriosus was a well-developed entity, both externally and internally (Figs 1 and 2). The interior of the conus arteriosus adjoining the right ventricle showed trabeculae carneae, but the upper portion leading to the pulmonary valve was smooth. The pulmonary artery was reduced in size to half of that of the aorta, and had only 2 valves (Fig. 2). The left ventricle showed fewer trabeculae carneae than on the right side. The large interventricular defect in the region of the membranous interventricular septum was even more obvious when viewed from the left side. The enlarged aorta straddling the ventricular defect had a normal arrangement of the valves.

DISCUSSION

The tetralogy of Fallot is a well-known clinical entity with its four associated conditions: pulmonary stenosis; defect of the membranous part of the interventricular septum; the large aorta arising astride the septal defect; and an abnormally thick right ventricular wall.

Two aspects of this case, however, merit further discussion. Firstly, the rather unusual anatomical malformation of the conus arteriosus which was virtually a separate chamber. Secondly, the functional aspect as to how adequate pulmonary blood flow was maintained through the small ostium connecting the right ventricle with the conus arteriosus, associated with an abnormal pulmonary artery and no compensatory flow through an open ductus arteriosus. There was also no evidence of enlarged bronchial arteries as a source of collateral circulation.

The explanation of these anatomical anomalies are to be sought in the developmental history of the embryonic heart. Patten[1] points out that in dealing with pulmonary stenosis, either uncomplicated or as the primary factor in the tetralogy of Fallot, it should be emphasised that the narrowing of the pulmonary inlet is by no means always in the same location nor of the same developmental origin. In many cases it is the type described as being o[f]

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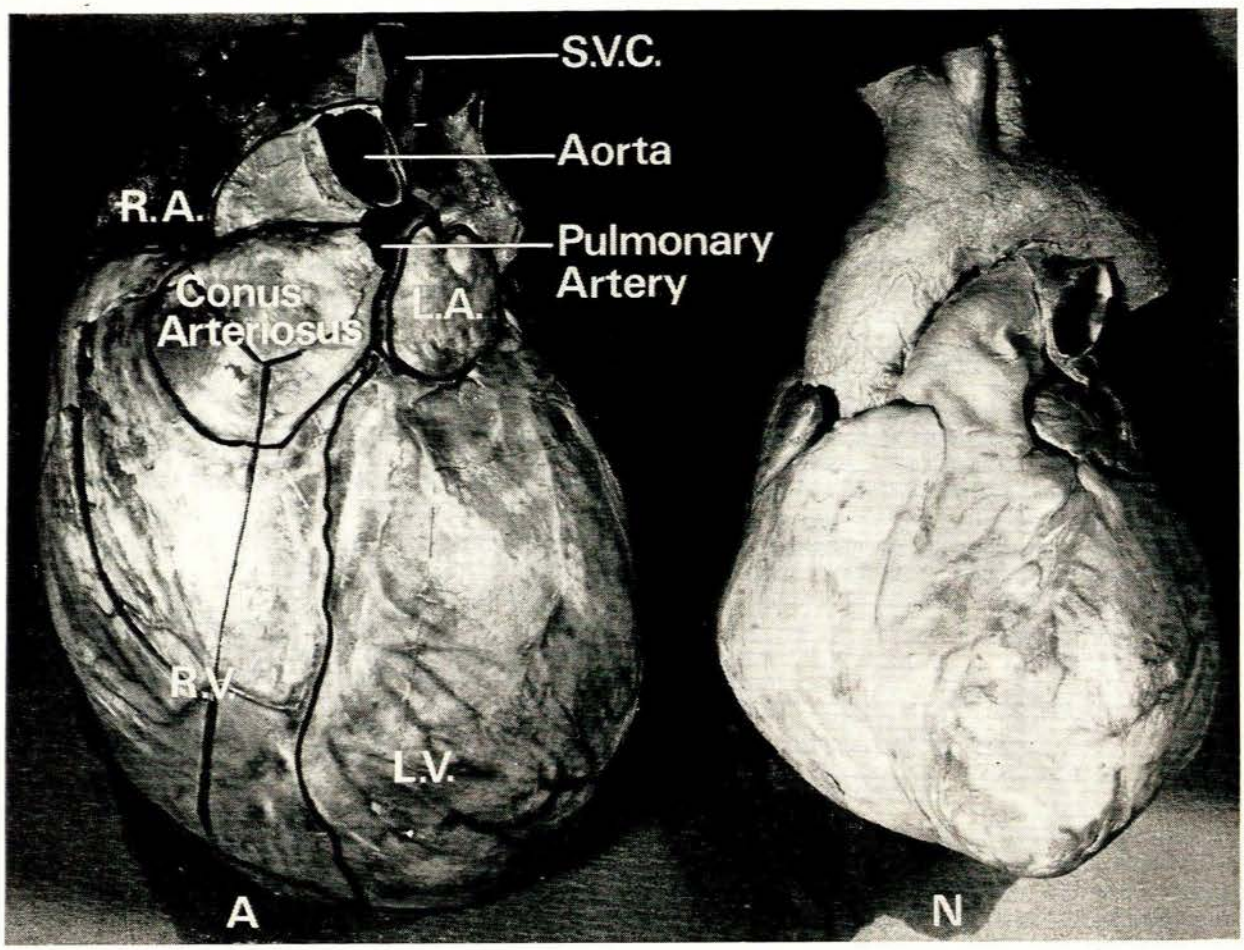

Fig. 1. Configuration of abnormal heart compared with normal heart. A—Fallot's tetralogy; N—normal heart; RV—right ventricle; LV—left ventricle; LA—left auricle; RA—right auricle; SVC—superior vena cava.

unequal truncus division. In other instances the narrowing is due to abnormal muscle bands encroaching on the pulmonary conus, so that the narrowing of the outlet is below valve level. Still another type of stenosis is that in which the valve leaflets are thickened and partially fused with one another, so that the orifice leading into the pulmonary trunk is seriously restricted.

In this particular case unequal division of the truncus arteriosus was present, as was evident in the reduced size of the pulmonary artery and an abnormal pulmonary valve with only two leaflets. Patten[1] postulates that stenosis from unequal truncus division is probably the first to become established. Stenosis at valve level must definitely be established later in development than any other types. The embryological story on obstruction of the conus outlet by abnormal configuration of its muscular walls is not yet entirely clear.

Kramer[2] made a study of the formation of the septal systems of the truncus and conus arteriosus and their union with the primary interventricular septum, and came to the conclusion that although they arise as more or less separate structures with locally accelerated growth rates, the truncus and conus ridges become moulded into a continuous spiral septum. In this process the sinistroventral truncus ridge becomes aligned with the dextrodorsal conus ridge, while the dextrodorsal truncus ridge is aligned with the sinistroventral conus ridge. The truncoconal septal system is brought into line with the interventricular septum at what is destined to be the interventricular septum membranaceum. This part of the process is brought about by the opposition and fusion of the parts of a ring of endocardial cushion tissue, contributed to by the right and left conus ridges, the endocardial cushion tissue contributed to by the right and left conus ridges, the endocardial cushion tissue at the crest of the interventricular septum, and the right tubercles of the atrioventricular canal cushions, to form the interventricular septum membranaceum (Fig. 3).

Shauer,[3] however, in his work on embryo pig hearts, came to the conclusion that it seemed more probable that the division of the primitive aorta is generally into approximately equal parts, so that the aorta and pulmonary artery start with equal calibres. Then some cardiac anomaly deflects blood from one vessel into the other. The slighted vessel lags behind in consequence, just as do many other embryonic vessels in the normal growth of the embryo.

Fig. 2. Interior of conus arteriosus. O—ostium leading to right ventricle; TC—trabeculae corneae; PV—two semilunar valves of pulmonary artery.

Stenosis and atresia should be regarded as the secondary results of some other cardiac disorder of structure and function. He points out that the fundamental defect in both aortic and pulmonary stenosis is the retention of the characteristics of the early conus beyond their time. Such a retention probably has several causes of which the defective development of the atrioventricular cushions is only one. But whatever the cause, the heart with such a conus shows three well-known characteristics: an overriding or right-sided aorta; a projecting superficial pulmonary inlet; and usually an open interventricular foramen.

The anomalies in this particular case seem to fit in better with the views expressed by Patten and Kramer[1,2] rather than that of Shauer,[3] although the theory expounded by him is very plausible. This brings one back to the embryology and abnormalities of the bulbus cordis. In this heart there was a very marked obstruction at the level of the conus inlet from the right ventricle rather than at the valve outlet. The conus was practically a separate chamber. Keith[4-6] gives an excellent description of the fate of the embryonic bulbus cordis of the human heart and its malformations, which fits in very well with the anomaly of the bulbus cordis in this case. He came to the conclusion that the infundibulum of the right ventricle must be regarded as a distinct constituent of the heart. Evidence to this effect is supplied by the comparative anatomy and embryology. The bulbus cordis in the vertebrate heart was originally a gill-pump. The bulbus is interposed between the gills and the ventricular pump as a safety mechanism. In fishes the lower orifice of the bulbus is usually provided with valves, and marks the separation of bulbus and ventricle. The upper orifice, which may be valvular or not, indicates the junction of bulbar and primitive aorta. Keith[4] points out that three great changes took place in the course of evolution from the primitive four-chambered heart of the fish to the mammalian heart. Firstly, the primitive auricle and ventricle have become completely divided into right and left chambers; then the sinus venosus has become partly, or as in man, almost completely, submerged in the musculature of the right auricle; and thirdly, the bulbus cordis has become separated from the left ventricle and aorta, and completely incorporated in the right ventricle as the infundibulum of the chamber.

Keith[4-6] regards the bulbis cordis as essentially a part of the respiratory system, and apparently chiefly concerned in regulating the flow of blood through the lungs. The

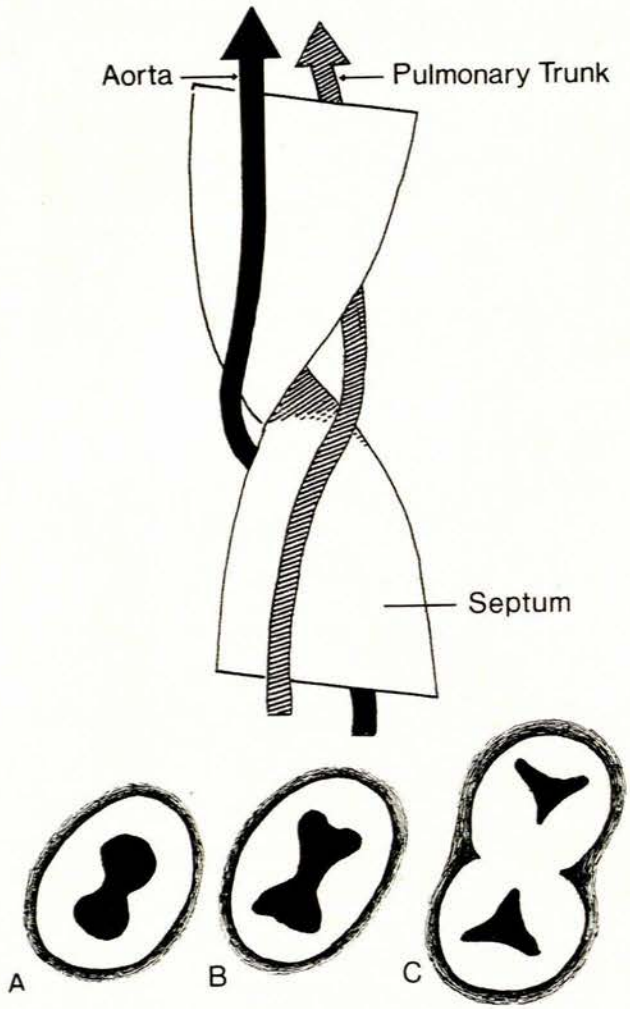

Fig. 3. Schematic representation of the spiral aortico-pulmonary septum at 5 weeks (top). **(Redrawn from Gardner *et al.*[3]** (Bottom) **A — truncus arteriosus; B and C — partitioning at 4½ weeks and 5 weeks of embryonic life.**

cavity of the bulbus cordis is incorporated in the right ventricle by an upgrowth of the ventricular musculature around it, the musculature of the bulbus being thus replaced by the musculature of the ventricle in the same way as the musculature of the auricle replaces a great part of that of the sinus venosus (Fig. 4). The submergence of the bulbus evidently constitutes a critical phase in the developmental metamorphosis of the heart, and it is during this critical phase that malformations are apt to occur.

Keith[4-6] divides malformations of the bulbus into three categories. In the first class the infundibulum and body of the right ventricle have developed to a normal extent but they have never completely fused, a construction remaining between them representing the ventricular orifice of the bulbus. The second class shows arrest of development of the bulbus cordis. Usually the bulbus is only partly expanded, the pulmonary orifice may be constricted and a wide interventricular orifice is commonly present. In the third class the arrest of development is

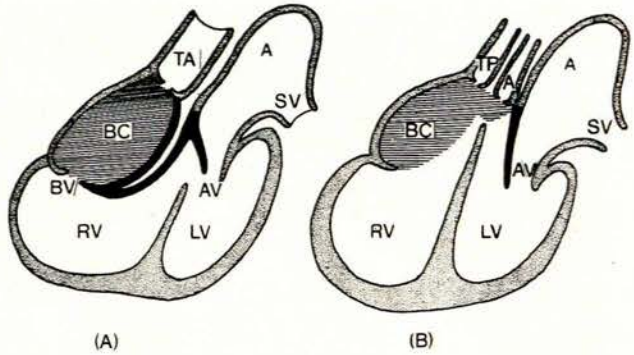

Fig. 4. (A) Diagrammatic section of the embryonic heart in the 4th week. SV — sinus venosus; A — atrium; AV — auriculoventricular junction; LV — left ventricle; RV — right ventricle; BV — bulboventricular junction; BC — bulbus cordis; TA — truncus arteriosus before division. (B) diagrammatic section of the fetal heart at the 3rd month (redrawn from Keith[4]). Bulbus cordis incorporated into right ventricle; division of the truncus arteriosus. TP — truncus pulmonalis; A — aorta.

more complete than in the second class, and the upper pulmonary opening is extremely narrow.

In this particular heart there was a very marked obstruction at the level of conus inlet from the right ventricle rather than at the valve outlet. The conus was practically a separate chamber. This fits in well with Keith's[4] first category, a well-developed infundibulum and the constriction between the right ventricle and infundibulum representing the ventricular opening of the bulbus. Associated with the congenital abnormality of the bulbus cordis there is the unequal division of the embryonic truncus arteriosus with the anatomical chain of events seen here. The punched-out hole in the interatrial septum reflects a congenital defect of the septum primum of the atrium.

Mulvihill[7] described some recent observations on domestic animals that have led to the identification of environmental teratogens, and have provided insight into the pathogenesis of some congenital defects and genetic diseases in man. Two common human birth defects, one of the heart and the other of the hip, seem to be polygenic traits. Comparative aspects of these diseases are strikingly similar in man and dog. For both species the over-all incidence of each defect is 2 - 7/1 000. Both defects occur more commonly in pure-bred dogs, and in some breeds the high incidence of heart malformations which might be considered breed-specific, for example in the Keeshond, had an excess frequency of ventricular septal defects as part of the tetralogy of Fallot.

Keith[4] examined 23 malformed human fetuses and infants presenting one or more of the following: anencephaly, hydrocephaly, spina bifida, umbilical hernia, atresia ani, cleft palate, hare-lip and stenosis of the oesophagus. Of his 23 cases, 14 showed a malformation of the heart, and 10 of these showed lesions of the bulbus cordis. In conclusion he says, 'The important point is that these lesions were similar to those I have just described in older hearts. It appears from the evidence, then, that the malformations of the bulbus are the result of the same

agent as produces the conditions of anencephaly, etc., with which lesions malformation of the bulbus is so often associated.'

Finally one comes back to the problem of how efficient pulmonary blood flow was maintained in this case for survival during childhood and up to the age of 17 years. The following possibilities can be postulated: that the ductus arteriosus took a longer time to close during infancy, thus allowing a period of aortic pulmonary shunt. In the fetal heart there is normally a right ventricular preponderance. The progressing hypertrophy of the right ventricle must have played an important role in forcing the blood through the ostium between the right ventricle and infundibulum. One can also visualise a gradual narrowing of this ostium as the heart grew in size. The infundibulum probably acted as a special pulmonary pump, according to Keith's[4] concept of the functional significance of the bulbus cordis. These compensatory mechanisms apparently prolonged life beyond normal expectation in the light of the anatomical anomalies described.

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