Understanding the heterogeneity of "mitral atresia" with patent aortic root

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It is now more than 40 years since, together with my good friend and colleague Gaetano Thiene, we addressed the issue of the anatomical substrate of mitral atresia.[1] Shortly thereafter, with access to the excellent archive of congenitally malformed hearts held at Children's Hospital of Pittsburgh, my colleagues and I explored the anatomical heterogeneity that is encountered when such valvar atresia is found in the setting of a patent aortic root.[2] We encountered exactly the same problems that are now described by the team working at the All India Institute of Medical Sciences in New Delhi. The investigators working in New Delhi encountered an additional problem. They recognised that the problems existing in providing a simple classification for these lesions are greatly increased in the setting of isomeric, rather than lateralised, atrial appendages.[3,4] Hence, they have produced two excellent manuscripts, rather than seeking to combine the overall findings. Their reports now serve to show how analysis of computed tomographic datasets is rapidly establishing itself as the new "gold standard" for the description of the anatomy of the congenitally malformed heart. The technique has several advantages over gross dissection. In the first instance, it shows the anatomy of the heart within the chest. For too many years cardiac morphologists have described the heart as if removed from the chest and positioned on its apex, producing the so-called "Valentine arrangement". It is slowly becoming recognised that attitudinally appropriate description enhances the ability to make direct comparisons between the structures as now revealed by three-dimensional imaging and the names used to describe them.[5] Traditionalists can sometimes find it difficult to accept, for example, that the leaflet of the valve guarding the diaphragmatic aspect of the right atrioventricular junction is inferiorly located, and hence should not be described as being "posterior". It is the activities of groups such as those working in New Delhi, whose investigations are increasingly seen published in the pages of the Journal, which are demonstrating the advantages of describing cardiac structures in their attitudinally appropriate settings. A second great advantage of virtual dissection of computerised tomographic datasets is that the technique

shows the cardiac components in their correct orientation to each other, There is no need to open and stretch the heart, as is usually the case when performing gross cardiac dissection. And sections can be taken again and again, and in different planes, when assessing the three-dimensional dataset. A third advantage of computerised tomographic investigation is that it reveals the details not only of the cardiac structures, but also the arrangement of the remaining thoracic and abdominal organs. In the investigations now published in this issue of the journal, for example, the Indian investigators were able to demonstrate disharmony between the arrangement of the atrial, thoracic, and abdominal arrangements in three of their patients with lateralised atrial chambers, and in two individuals with isomeric atrial appendages.[3,4]Significantly, the disharmony in those with isomeric appendages was between the lungs, bronchuses, and abdominal organs, but still with the bronchial arrangement providing an accurate guide to the type of isomerism present. Even more significantly, when using their tomographic datasets, it proved possible in all instances to identify the presence of isomeric atrial appendages on the basis of the extent of their pectinated walls relative to the vestibules of the atrioventricular junctions.[6] This will now surely be the way forward to identifying and segregating those patients currently described in confusing fashion as having "heterotaxy". As the authors show, it is the abdominal organs that are "heterotaxic", whereas it is only the lungs, bronchuses, and atrial appendages which are able to show the features of isomerism.[7]

The major topic of the two investigations, however, was "mitral atresia" in the setting of a patent aortic root. As the investigators emphasise, atresia of the mitral valve is also one of the features of the hypoplastic left heart syndrome. In this setting, as in the hearts with patent aortic root, the atresia itself can be the consequence of presence of an imperforate leaflet, but more frequently is due to absence of the left atrioventricular connection, when the myocardial floor of the left atrial chamber is separated by a layer of fibroadipose tissue from the underlying crest of the ventricular mass. When mitral atresia is found in the setting of the hypoplastic left heart syndrome, it is usually found with co-existing aortic atresia, and the left ventricle is then often little more than a slit in the inferior wall of the ventricular mass. As the authors from New Delhi rightly emphasise, the main distinguishing feature between the hypoplastic left heart syndrome and the variants seen with patent aortic root is the integrity of the ventricular septum.[8] The cases with patent aortic root and ventricular septal defect, therefore, can be viewed as a different set of phenotypes. It is the categorisation of these phenotypes that then creates significant problems when seeking to describe them all as "mitral atresia". As the computerised tomographic datasets reveal, when the mitral valve is atretic in the setting of congenitally corrected transposition with usual atrial arrangement, then it is right-sided, rather than being left-sided. The Indian investigators, nonetheless, have rightly include some cases in their series in which the left atrioventricular valve is atretic, and is producing blockage of the direct outlet from the pulmonary venous atrium. And, as they discuss, had the left-sided atrioventricular connection been formed in this setting, it would have been guarded by a morphologically tricuspid valve. It is for this reason that the investigators correctly describe atresia of the left atrioventricular valve, rather than the mitral valve. This description then also holds good for the patients they identified with isomerism, since when the appendages are isomeric, it is not always possible to identify pulmonary and systemic venous atrial chambers. It does remain possible to distinguish between the right and left atrioventricular junctions. There is one problem remaining with their descriptions, which relates to the nature of the atresia when the atrial chambers are themselves mirror-imaged, as was the case in four of their patients with lateralised atrial chambers. If these patients truly had atresia of the left-sided atrioventricular valve, then this arrangement would be the equivalent of tricuspid atresia as seen when there is usual atrial arrangement. Since I was the referee of the manuscripts during the process of peer review, I should have asked for clarification of this point at an earlier stage. This small indiscretion on my part should not detract from the superb account the authors provide of cardiac anatomy in an unusual setting. Their figures and descriptions point to the quality of diagnosis now being achieved in the All India Institute. The nature of the material they describe also points to the numbers of patients now coming forward for treatment, and the variability in the combinations of lesions encountered. Above all, their account emphasises the great value of simple description of the overall findings.

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