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*Article Commentary*

# Understanding Ebstein's malformation

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In the current issue of the Journal, our colleagues from Seoul, South Korea,[1] present a detailed account of their search for accurate diagnostic markers that reveal the presence of Ebstein's malformation of the tricuspid valve in the setting of pulmonary atresia and intact ventricular septum. Subsequent to my review of their submission, the authors made substantial changes to their draft, which in my opinion, have significantly improved its presentation. It did not prove possible for them to respond to my most pressing request, namely, to assess the location of the hinge point of the inferior as opposed to the septal leaflet of the malformed tricuspid valve. This, in many ways, reflects their sharing of current "conventional wisdom" concerning the anatomy of Ebstein's malformation. If we are to take full advantage of the excellent study from our Korean colleagues, nonetheless, we should take heed of the problems that will continue if future investigators also concentrate their attention exclusively, during clinical investigation, on the septal as opposed to the inferior leaflet of the valve.

The very association between Ebstein's malformation and pulmonary atresia has itself been the source of previous controversy. My old friend and colleague Anton Becker, with whom I collaborated for the larger part of my career, took exception to a study in which we described hearts with pulmonary atresia and intact ventricular septum as also having Ebstein's malformation.[2] In his opinion, the eponym would better have been restricted to hearts, reflecting the "pure" version of the deformed tricuspid valve, as described by Ebstein himself. The fact remains, nonetheless, that pulmonary atresia is now well accepted as one of the associated lesions that can further complicate the presentation of patients having Ebstein's malformation. Indeed, in a website summarising the findings relative to Ebstein's malformation, George Hartnell provides an excellent overview of the techniques available for imaging the clinical findings.[3] He includes pulmonary atresia as one of the anticipated associated lesions. Like most other current authors, nonetheless, and like myself at the time I responded to Becker's editorial comment,[4] Hartnell describes the tricuspid valve as having septal, anterior, and posterior leaflets. He correctly points out that the alleged posterior leaflet is also termed the mural leaflet. Along with most other current investigators, he fails to observe that, when the heart is considered as it lies within the body, the mural leaflet is positioned inferiorly and not posteriorly. This feature is readily appreciated in his sixth image, a magnetic resonance angiogram in the coronal plane, which reveals the location of the deformed valve within the heart as seen within the body.[3]

I would suggest that it is the assessment of the malformed tricuspid valve using attitudinally appropriate nomenclature[5] that serves to clarify the abnormal anatomy. Such assessment also points to the most reliable means of distinguishing between minimal degrees of Ebstein's malformation and the normally attached tricuspid valve. As was pointed out by Kenneth Anderson in another study in which I was involved, the displacement of the hinges of the malformed tricuspid valve is greatest at the crux of the heart, which is where we normally find the zone of apposition between the septal and inferior leaflets.[6] When assessing the displacement of the septal leaflet, the landmark usually taken to diagnose Ebstein's malformation is the distance between the hinges of the septal leaflet of the tricuspid valve in the right ventricle, and that of the mitral valve in the left ventricle. This ratio, proposed for clinical use by the group from the Mayo Clinic spearheaded by Shiina,[7] was the one explored by our Korean colleagues in their study. As they have pointed out in their discussion, its value had already been called into question by another study that appeared almost contemporaneously with that emanating from the Mayo Clinic.[8] My old friend Becker was the guiding morphological light in this second study. As is also pointed out by our colleagues in Seoul, in another of our

Correspondence to: R. H. Anderson, 60 Earlsfield Road, SW18 3DN, London, United Kingdom. Tel: 00-44-20-8870-4368; E-mail: sejjran@ucl.ac.uk

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own subsequent studies we had pointed out that, if attention is directed to the inferior, rather than the septal, leaflet of the malformed valve, there is no need to rely on such subtle ratios so as to distinguish the presence or absence of Ebstein's malformation.[9] This is because any distal displacement of the inferior leaflet will move its hinge away from the atrioventricular junction. Therefore, attention to the inferior, as opposed to the septal, leaflets provides an "all-or-none" criterion for the diagnosis of Ebstein's malformation.[9]

Attention to the location of the hinges of the leaflets in an attitudinally appropriate manner then provides another insight into the pathology of Ebstein's malformation. Throughout the account of Hartnell, the leaflets of the valve are described as being "downwardly displaced". This was also the description used in the editorial commentary of Becker,[2] and in my own response.[4] It is then paradoxical that, although Hartnell describes such "downward" displacement, all of his echo images provided in a four-chamber projection reveal upward displacement.[3] This, of course, reflects the ongoing practice of most adult echocardiographers and some paediatric echocardiographers of showing the four-chamber echocardiographic images in an attitudinally upside-down manner. Such illogical usage, based on historical precedent, further complicates the problems emerging from non-attitudinal display. When considered in an attitudinally appropriate manner, as shown in another of our morphological studies,[10] the hinges of the deformed septal and inferior leaflets are displaced in rotational, rather than inferior, manner. Indeed, the drawings of the initial heart described by Ebstein, reproduced in Becker's editorial comment,[2] show that the effective valvar orifice is displaced almost into the subpulmonary infundibulum, where it takes on a decidedly bifoliate appearance. In such situations, assessment using the four-chamber projection, and relying on identification of the hinge of the septal leaflet, can give the spurious impression of unguarded tricuspid valvar orifice. Directing attention to the hinge of the inferior, or mural, leaflet will clarify the situation.

It is easy, of course, for anatomists such as myself to argue that sonographers should concentrate on the inferior, rather than the septal, leaflets when seeking to diagnose Ebstein's malformation. Whether this can be achieved in practice remains to be seen. Nonetheless, appreciation of the need to assess the deformed valve in an attitudinally appropriate manner will make it easier for one and all to understand that the essence of the malformation is rotational, as opposed to "downward" displacement of the hinges of the leaflets away from the atrioventricular junction.

*Robert H. Anderson*
*Institute of Genetic Medicine*
*Newcastle University*

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