Editorial

The challenge of risk stratification in Ebstein's anomaly

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ARTICLE INFO

Ebstein's anomaly (EA) is characterized by a disturbed right heart development due to an incomplete delineation of a variable degree of the septal, posterior and anterior leaflets of the tricuspid valve leading to a displacement of the valvular hinge points and the functional tricuspid annulus to the apex and right ventricular (RV) outflow tract. The consequence is an "atrialization" of the inlet portion of the RV. The myocardium of this atrialized part of the RV is thin, associated with a severely reduced or absent contractility. The tricuspid valve shows usually a varying degree of regurgitation (TR). In addition, tethering and perforation of leaflets as well as, but less frequent, tricuspid stenosis are observed. The hemodynamic consequences of this cardiac malformation are a progressive dilatation and dysfunction of the functional RV which is related to the degree of TR, leading to a diminished RV output and ultimately to a reduced left ventricular (LV) filling [1,2]. The reduced LV preload in addition to a RV/LV interaction results in a reduced LV ejection fraction.

Outcome of patients varies depending on the time of presentation and accompanying cardiac lesions like atrial septal defect or RV outflow tract obstruction [3]. Early, i.e. fetal or neonate presentation is often associated with more severe forms of EA.

Due to the rare incidence and the heterogeneity of the anatomic anomalies, establishment of risk factors predicting outcome and identifying patients requiring early surgical treatment is challenging. In addition, surgical repair techniques and medical management evolve over time, limiting the comparability of patient populations between different periods and centers. An early study by Celermajer et al. identified in neonates the echocardiographic grade of severity, fetal presentation and RV outflow obstruction as predictors of outcome [3]. The exercise capacity is related to the disease severity and predicts outcome in adult patients [4]. The volume of the atrialized RV and the ratio of the total right to left cardiac volumes as determined by cardiac magnetic resonance (CMR), appear to be related to exercise capacity and parameters of heart failure in EA patients [5].

Although CMR is a very promising imaging tool, its availability is limited, and it usually requires sedation in neonates and young children. Echocardiography remains therefore a very important imaging modality, particularly in the pediatric population [6]. It allows to follow the patients' evolution and evaluate the hemodynamic situation before and after surgical correction [2].

In the current issue, Prota et al. present a study evaluating in 50 consecutive pediatric EA patients the value of echocardiography in predicting outcome (ventricular tachyarrhythmia, heart failure, need for surgery and/or death) [7]. This study tries to identify from parameters grading the severity of the anatomic anomalies on the one hand and parameters describing the ventricular and valvular function on the other hand, those, who distinguish stable from patients with progressive disease using classic and advanced echocardiographic imaging techniques. Although univariate analyses identified both anatomic and functional parameters related to outcome, multivariate analysis revealed only right atrial (RA) peak systolic strain and RV fractional area change (RV-FAC) as predictors of outcome. It is interesting in this context that TR, which is believed to be the most important hemodynamic lesion in EA, was not independently related to outcome, probably due to the relatively small number of included patients with a severe TR.

The findings of the study are interesting from two perspectives. The first observation is that the impact of the anatomic anomalies on RA and RV function appears to be more relevant for the outcome of the patients than the severity of the underlying anatomic anomalies itself. The established scores evaluating EA severity might only incompletely describe the very heterogenous anatomic findings of EA patients, which could explain why they fail to identify patients at risk for progressive disease in this pediatric population. RA function, however, has recently gained interest in CHD patients. It is related in EA to markers of heart failure and RV function [8]. The remarkable new finding of the present study is that RA peak systolic strain, a marker of the RA reservoir function,

allows identification of patients at risk for progressive disease suggesting its use as an early predictor for an unfavorable outcome.

Less surprisingly, RV-FAC which is a measure of the "volumetric" RV function, is related to outcome in contrast to echocardiographic parameters describing only the longitudinal RV function. As previously shown, the latter parameters do not very well correlate to RV ejection fraction as determined by CMR contrary to RV-FAC [9,10].

The second interesting observation is that only LV global strain was related in univariate analysis to a progressive disease and none of the parameters describing the LV diastolic function or filling. As alluded to earlier, congenital heart disease lesions involving the RV like EA, lead to a reduced RV output followed by a diminished LV preload and finally an impaired LV systolic function. Surgical correction of EA improves RV output and consequently LV filling despite reduction of RV ejection fraction [2]. These important implications on hemodynamics of the LV can well be detected by Doppler echocardiography. However, the impaired LV filling is probably a marker of a more advanced disease stage, which could explain why none of these echocardiographic parameters was related to a progressive disease in the studied pediatric patient population.

In conclusion, echocardiography is an important tool for follow-up and risk stratification, particularly in pediatric EA patients. In addition to the disease severity, function of the RA and the functional RV are important parameters to follow. The routine use of RA strain imaging might not be possible in every center, however, determination of RV-FAC, a rather simple echocardiographic parameter, should probably be part of a routine follow-up in pediatric EA patients.

Disclosures

None.

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