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ORIGINAL ARTICLE

Ebstein's Anomaly in Those Surviving to Adult Life – A Single Centre Experience

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Background Ebstein's anomaly (EA) occurs in about one to five per 200 000 live births. Long-term follow-up data of adults with EA is scarce due to the relatively low frequency of the disease and the variation of its anatomic and haemodynamic severity.

Methods Since 1995, in our adult congenital heart disease (ACHD) centre, we have practised a uniform approach to management of adults with EA, with surgery reserved for those with refractory arrhythmia (failed medical and/or catheter-based treatment) or worsening symptoms of breathlessness. A retrospective review of medical records of all such patients with EA and normal cardiac connections was performed.

Results Fifty-one EA patients (17 males) were identified. Mean age at diagnosis was 21+/-21 years and mean follow-up time at our centre was 21 ± 14 years. During this time, 18 patients (35%) had documented supraventricular arrhythmia. Sixteen patients (30%) underwent ablation therapy with long-term relief from arrhythmia in nine (56%). Nine patients (18%) underwent tricuspid valve (TV) surgery (four repair and five replacement), with seven patients having undergone a tricuspid valve surgery prior to referral to our unit. Three patients died, one of cardiogenic shock after redo surgery (58 years), one of progressive heart failure (45 years) and one with malignancy. Overall survival was 100% to age 40 years, 95% to age 50 years and 81% to age 60 years.

Conclusions Ebstein's Anomaly in adulthood often has severe morphological abnormalities but is compatible with good medium-term survival, with a generally symptom driven approach to the indications for interventions.

Keywords Tricuspid Valve • Congenital Heart Disease • Arrhythmia

Introduction

Ebstein's anomaly (EA) is a rare form of CHD, occurring in approximately one to five per 200 000 live births, thus accounting for less than 1% of all congenital heart disease [1]. It occurs when the septal leaflet of the tricuspid valve joins to the septal surface below the valve annulus, into the body of the right ventricle, often with marked displacement of the tricuspid valve, displacing the coaptation point towards the right ventricle apex and/or outflow tract [2].

It is commonly associated with other cardiac anomalies such as atrial septal defect (ASD) (prevalence 80 – 94%[3]), ventricular septal defects (VSD) and ventricular pre-excitation (prevalence 15%) [4]. The structural tricuspid valve anomaly may lead to tricuspid regurgitation (TR) and increased right atrial volume. Age at clinical presentation varies depending on the severity of TR and on the presence of associated heart diseases. [5]. Repair of the tricuspid valve and/or ablation therapy are often indicated[1]. Optimal timing of intervention is often challenging, requiring careful

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consideration of patient symptoms, perioperative risk and the potential need for late re-operation [4].

Since the establishment of our Adult Congenital Heart Disease (ACHD) service, we have adopted a uniform approach of ''watchful waiting'' to the management of EA, with surgery reserved either for those with refractory arrhythmia (failed medical and/or catheter based treatment) or worsening cardiac symptoms. A lower threshold for surgery was applied in cases when the tricuspid valve was judged likely reparable (compared to requiring replacement) and was more often undertaken in those judged to have an adequate left ventricle and functional right ventricle.

Long-term follow up data of adults with EA are scarce due to the relatively low frequency of the disease and the remarkable variation of its anatomic and haemodynamic severity [5]. Therefore, we aimed to evaluate the long-term clinical outcomes of our adult patients (aged over 16) with EA and normal cardiac connections.

Methods

The study was approved by our Institutional Ethics Committee.

Study Population

Patients were identified from our ACHD database, which contains 3682 patients aged over 16 years, seen at least once between 2000 and 2013. The median age when first seen at our service was 30 years (IQR 21-44 years). The median age at the time of last follow up was 39 years (IQR 30-53 years).

Patients who had EA and concordant cardiac connections were included in the study. Those with EA in the setting of congenitally corrected transposition of the great arteries (n=10 in our database) were excluded.

Clinical data were retrospectively obtained from case notes, 12 lead electrocardiograms, echocardiograms and surgical reports. The date of last follow-up was recorded as the last contact with the patient and survival data was censored at this point.

Vital status was obtained for all EA patients from the Australian National Death Index Registry, with an ascertainment date at the end of 2013.

The first transthoracic echocardiogram (TTE) after the age of 16 years was used to assess for a ''small'' left ventricle, defined as end diastolic diameter less than 40 mm. Echocardiography data was unavailable for six patients. For the remaining patients, echocardiographic grading of right atrial systolic size and tricuspid regurgitation into mild, moderate and severe was based on visual assessment by experienced congenital heart disease specialists. New York Heart Association (NYHA) Functional Classification was used to assess patient's severity of heart failure symptoms at follow-up.

Management decisions were determined by the patient's ACHD cardiologist +/- cardiothoracic team in each case, as part of routine clinical care.

Statistical Methods

Analysis was performed using SPSS version 20 (IBM, Armonk, New York). Data for categorical variables are reported as frequency and percentage (%) and data for continuous variables are summarised using mean ± standard deviation. Continuous variables that were not normally distributed were reported as median and range (minimum and maximum). Kaplan-Meier curves were used to estimate survival, freedom from surgical intervention and freedom from ablation intervention.

Results

Patient Characteristics

Fifty-one adults were identified with EA and concordant cardiac connections. Seventeen (33%) were men. The mean age at diagnosis was 15 years (IQR 1-35 years). Sixteen patients (31%) were first diagnosed at ≥ 16 years of age, at our ACHD centre, with the remaining 35 patients diagnosed in childhood and 'transitioned' to our ACHD service.

Thirty-four (67%) patients had an associated congenital heart lesion, with eight patients (16%) having more than one associated congenital heart lesion. (Table 1)

Fourteen patients (27%) had undergone cardiac surgery prior to first review at our centre. Prior surgery for other congenital heart diseases included ASD and/or VSD repair (n=9), pulmonary valvotomy (n=3), pulmonary valve replacement (n=1), septoplasty (n=1) and septostomy (n=1). Seven patients had had tricuspid valve surgery including tricuspid valve repair in five and bioprosthetic valve replacement in two patients.

Echocardiographic Features at Baseline

At first TTE at our centre, mean right atrial (RA) area was 44.3 cm^2 ± 22 cm^2 (14 – 100 cm^2, median 38 cm^2). Four

Table 1 Associated congenital heart disease in patients with Ebstein's anomaly.

Associated Congenital Heart Lesions	Frequency in patients (n=51)	
	Number	% (of overall EA patients)
ASD	25	49
Pulmonary Stenosis	6	12
VSD	5	10
Mitral Valve Prolapse	4	8
Cleft Mitral Valve	1	2
Persistent ductus arteriosus	1	2

Associated congenital heart disease in patients with Ebstein's anomaly. Note, some patients have more than one associated congenital lesion. ASD= atrial septal defect, PFO= patent foramen ovale, VSD= ventricular septal defect.