



Ebstein's anomaly: natural and unnatural history

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■ Clinical features and natural history were analyzed in 30 patients with Ebstein's anomaly (mean age 26 years, range 1.5–58 years, 53% females). The main presenting symptoms were dyspnea and fatigue (83%). At presentation, there were six patients (20%) in New York Heart Association Functional Class (NYHA-FC) I, nine (30%) in NYHA-FC II, and 15 (50%) in NYHA-FC III or IV; 12 patients (40%) were cyanotic. Common auscultatory findings were widely split second heart sound in 21 (70%), third heart sound in 14 (47%), fourth heart sound in 16 (53%), and a systolic murmur in 22 (73%). Right bundle branch block was present in 21 (70%), documented supraventricular tachycardia in seven (23%), and Wolff-Parkinson-White syndrome in three (10%). Catheterization was performed in 93% without complications. Fourteen patients were treated surgically (12 [86%] in NYHA-FC III or IV, 10 [71%] with associated anomalies); tricuspid valve replacement was performed in eight, atrial septal defect repair in two, accessory pathway ablation in two, right atrial plication in one, and automatic cardioverter defibrillator implantation in one. Surgical treatment improved 10 patients from NYHA-FC III or IV to NYHA-FC I or II. Death occurred in nine patients (five treated surgically and four medically); four of these deaths were sudden. In the eight patients who had tricuspid valve replacement, there were one operative and two late deaths. The authors conclude that surgical therapy with tricuspid valve replacement improves the clinical status of patients who are severely ill. Risk of sudden death remains an important problem in patients with Ebstein's anomaly regardless of severity of the disease and mode of treatment.

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WILHELM EBSTEIN, in 1866, first described the anomaly that bears his name. He reported his autopsy findings in a 19-year-old patient who had had cyanosis, dyspnea, and palpitations since childhood.¹ The clinical presentation, management, and natural history of Eb-

■ See also the editorial by Mair (pp 626–627)

stein's anomaly have been reported.^{2,3} However, our understanding of the natural and the unnatural history of Ebstein's anomaly is still far from complete. We report our experience and long-term follow-up in 30 patients with Ebstein's anomaly.

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PATIENTS AND METHODS

Thirty-two patients at The Cleveland Clinic Founda-

tion were diagnosed between 1950 and 1986 as having Ebstein's anomaly. We excluded two patients for whom data were inadequate for analysis and analyzed the remaining 30 patients. Hospital records, electrocardiograms, chest radiographs, cardiac catheterization data, and surgical records were reviewed. All surviving patients were contacted by telephone and follow-up data were obtained on all patients.

RESULTS

Of the 30 patients, 16 were female and 14 were male. Mean age at presentation was 25.8 years, ranging from 1.5 to 58 years. Simple Ebstein's anomaly was present in 10 patients and 20 patients had other associated cardiac anomalies. Presenting symptoms were shortness of breath and/or fatigue in 25 patients (83%), palpitations in 15 patients (50%), and syncope or pre-syncope in eight patients (27%). Twelve patients (40%) were cyanotic. At presentation, there were six patients (20%) in New York Heart Association Functional Class (NYHA-FC) I, nine patients (30%) in NYHA-FC II, and 15 patients (50%) in NYHA-FC III or IV.

The first heart sound was loud in five patients (17%) and split in three patients (10%). The second heart sound was widely split in 21 patients (70%). A third heart sound was present in 14 patients (47%), and a fourth heart sound was also noted in 16 patients (53%). A systolic murmur was heard in 22 patients (73%), and a diastolic murmur was heard in nine patients (30%).

Supraventricular tachycardia was documented in seven patients (23%), two patients (7%) had complete atrioventricular block, one patient (3%) had atrioventricular dissociation, 21 patients (70%) had complete or incomplete right bundle branch block, and three patients (10%) had surface electrocardiograms that suggested Wolff-Parkinson-White syndrome (WPW). Radiographically, cardiomegaly was present in 28 patients (93%).

Cardiac catheterization was performed in 28 patients (93%) (Table 1). The remaining two patients were diagnosed by two-dimensional and Doppler echocardiography. There were no catheterization-related deaths. During cardiac catheterization, one patient required cardioversion for sustained ventricular tachycardia, and another patient developed ventricular fibrillation that was converted to sinus rhythm by cardiac massage. Cardiac catheterization revealed the following associated cardiac anomalies: atrial septal defect in 17 patients (57%), ventricular septal defect in three patients (10%), mitral regurgitation in four patients (13%), mitral steno-

sis in one patient (3%), and patent ductus arteriosus in two patients (7%).

Of 30 patients, 14 (47%) underwent 17 cardiac surgical procedures: tricuspid valve replacement alone or in combination with other procedures in seven patients, ablation of accessory pathway in two patients, and thoracotomy for implantation of an automatic implantable cardioverter defibrillator (AICD) in one patient. Second cardiac surgical procedures were needed in five patients. All of these five surgical procedures involved tricuspid valve replacement. Of 14 surgical patients, 10 had associated cardiac anomalies. In the surgical group 10 patients were in NYHA-FC III or IV before surgery; 10 patients improved to NYHA-FC I-II following surgery.

There were 16 patients who were treated medically. Of these 16 patients, nine had associated cardiac anomalies. In this group, only three patients were in NYHA-FC III or IV.

Follow-up was complete in 100% of patients; mean follow-up duration was 11.9 years (range 2–31 years). There were nine deaths, four of which were sudden. There were five deaths in the surgically treated group and four in the medically treated group.

Of five surgically treated patients who died, three died in the early postoperative period; a 7-year-old boy died of pulmonary edema 24 hours following atrioseptal defect repair, a 57-year-old man died in a low cardiac output state three days following right atrial plication, and a 49-year-old woman died, while she was still in the hospital, of ventricular fibrillation six weeks after surgery for tricuspid valve replacement and atrial septal defect repair. All three patients who died in the early postoperative period were in NYHA-FC III or IV before surgery.

There were two late deaths in the surgical group. A 27-year-old man, who was NYHA-FC I following surgery, died suddenly seven years after tricuspid valve replacement, right atrial plication, and atrial septal defect repair. Another patient in the surgical group died at age 28 because of prosthetic valve dysfunction 13 years following tricuspid valve replacement. Of the eight patients who underwent tricuspid valve replacement, only one patient died in the early postoperative period.

In the medically treated group, one patient who was in NYHA-FC IV died at age 18 months of congestive heart failure. Two patients who were in NYHA-FC II died suddenly, one while playing basketball and one while playing volleyball. Both of these patients had a history of syncope and palpitations. Another patient in this group died of cerebrovascular accident at age 39. Of

TABLE 1
EBSTEIN'S ANOMALY

Time of Treatment		Presentation		Surgery		Follow-up	
Patient No., Sex	Age (yr)	F.C.	Catheterization	Age (yr)	Type	Age* (yr)	F.C.
1 M	18 mo	IV	VSD, TR, ASD, MR	—	—	18 mo, E-CHF	IV
2 M	2	II	TR, ASD	—	—	19, E-SD	II
3 F	4	III	TR, ASD, PDA	17	TVR, RA _p , ASD _r , TVR	26	I
4 M	6	IV	PS, ASD, PDA, TR, MR	6	ASD _r , PDA _r , PV	38	III
5 F	7	III	TR, ASD	31	MVR, TVR	7 E-CHF	IV
6 M	8	II	ASD	7	ASD _r	24, E-SD	II
7 F	13	III	TR	15	TVR	21	I
8 F	38	III	—	39	AICD	41	I
9 M	17	I	—	17	WPW ablation	19	I
10 M	15	III	TR, ASD	15	TVR, RA _p , ASD _r	28, E-SBE	
11 F	26	III	TR	26	TVR	31	II
12 M	58	III	TR, MR, ASD	59	TVR, MVR, ASD _r	65	II
13 F	38	III-IV	TR, ASD	49	TVR, ASD _r	49, E-SD	I
14 M	19	III	TR	20	TVR, RA _p , ASD _r	27, E-SD	I
15 F	46	III	ASD	47	ASD _r	65	III
16 F	31	I	ASD, TR	—	—	54	I
17 F	18	II	VSD, TR	—	—	35	II
18 M	49	II	TR	—	—	56	II
19 F	37	III	ASD, TR	—	—	50	II
20 F	32	III	TR, MR, ASD	—	—	35	III
21 M	15	I	TR	—	—	26	II
22 M	42	II	TR	—	—	58	III
23 F	34	I	TR	—	—	51	I
24 F	16	II	ASD, TR	—	—	25	II
25 F	46	I	TS, TR	—	—	51	I
26 M	29	II	ASD, TR	—	—	39, E-CVA	II
27 F	17	II	ASD	45	ASD _r , WPW ablation	46	II
28 F	57	IV	TR	57	RA _p	57, E-CHF	IV
29 M	21	I	MVP	—	—	30	I
30 M	42	II	—	—	—	45	II

AICD = automatic implantable cardioverter defibrillator; ASD = atrial septal defect; CVA = cerebrovascular accident; CHF = congestive heart failure; E- = expired of; F.C. = New York Heart Association functional class; MR = mitral regurgitation; p = plasty; PDA = patent ductus arteriosus; PS = pulmonary stenosis; PV = pulmonary valvotomy; r = repair; RA = right atrium; SBE = subacute bacterial endocarditis; SD = sudden death; TR = tricuspid regurgitation; TS = tricuspid stenosis; TVR = tricuspid valve replacement; WPW = Wolff-Parkinson White syndrome.

*Age and cause of death were listed when appropriate.

the 12 patients in this group who are alive, 10 are in NYHA-FC I or II. Five patients in the medically treated group and two in the surgically treated group had successful pregnancies.

DISCUSSION

The clinical presentation, physical findings, chest radiographs, and electrocardiograms in our patients were similar to those in previously reported series.^{2,3} A typical patient with Ebstein's anomaly presents with dyspnea, fatigue, and palpitations. On physical examination, a systolic murmur of tricuspid regurgitation, widely split second heart sound, and third and fourth heart sounds are audible. The electrocardiogram reveals right bundle

branch block and right atrial enlargement, and the chest radiograph shows cardiomegaly.

Cardiac rhythm abnormalities are common in Ebstein's anomaly. Atrial fibrillation/flutter, ventricular tachycardia, and reentrant arrhythmias associated with WPW have been previously reported.^{4,5} At presentation, 23% of patients had documented supraventricular tachycardia and an additional 40% had a history of palpitations. Only three of our patients had delta waves on their electrocardiograms that suggested WPW. Two of them underwent surgery for ablation of the accessory pathway. WPW has been reported in 10%–25% of patients with Ebstein's anomaly.⁵ A recent report⁶ described the development of the right atrioventricular annulus with a hypoplastic central heart skeleton favor-

ing the persistence of fetal atrioventricular communications of high arrhythmogenic potential in patients with Ebstein's anomaly.

In addition to WPW-related ventricular arrhythmias, several investigators have reported episodes of unexplained ventricular fibrillation in patients with Ebstein's anomaly.^{4,7,8} Sudden death has also been reported in the early and late postoperative period in these patients.⁹ There were four sudden deaths in our patient population and one patient received an AICD for aborted sudden death. Two patients who were treated medically died suddenly during strenuous exercise; both were in their 20s and were in NYHA-FC I. They both had had recurrent syncopal episodes and palpitations in the past. Two patients in the surgical group died suddenly, one in the early and the other in the late postoperative phase. These patients both had a history of palpitations preoperatively; however, they did not have any syncopal episodes. Continuous electrocardiographic monitoring was available in only one of these four patients and it did not reveal any significant ventricular arrhythmias. The data from our series and observations of previous investigators⁹ show that some patients with Ebstein's anomaly are at significant risk for sudden death irrespective of clinical symptoms and mode of treatment. As most of our cases were diagnosed and managed before continuous electrocardiographic monitoring became available, we do not have enough information to assess the value of continuous electrocardiographic monitoring in predicting sudden death. The predictive value of programmed electrical stimulation in Ebstein's anomaly needs to be determined. Considering that two of our patients who experienced sudden death were involved in strenuous exercise at the time of their death and the other two were engaged in mild to moderate exercise, it seems prudent to perform exercise stress testing in all patients with Ebstein's anomaly.

Potential dangers of cardiac catheterization in patients with Ebstein's anomaly have been reported. In an international cooperative study of 505 cases of Ebstein's anomaly, 360 cardiac catheterizations were done.⁷ There were 19 cardiac arrests, which resulted in 13 deaths. There were no cardiac catheterization-related deaths in our study and there was only one patient with sustained ventricular tachycardia who required cardioversion and one with ventricular fibrillation that was converted by external massage. Our data suggest that cardiac catheterization can be performed safely in patients with Ebstein's anomaly. However, with the advent of two-dimensional and Doppler echocardiography, most cases of Ebstein's anomaly can be diagnosed

without catheterization.

The majority of patients who were mildly symptomatic were treated medically. Only one patient in this group progressed from NYHA-FC I to III. In this group, five patients had one or more pregnancies and delivered without major complications.

The surgical treatment of Ebstein's anomaly remains controversial. Mair et al⁹ and Danielson et al¹⁰ have reported good results utilizing plication of the free wall of the atrialized portion of the right ventricle, posterior tricuspid annuloplasty, and right atrial reduction, a procedure that they have used since 1972. This technique was favored because of reported poor outcome of tricuspid valve replacement. McFaul et al¹¹ published data showing poor results of tricuspid valve replacement in Ebstein's anomaly. However, other authors have demonstrated good short- and long-term results utilizing valve replacement in the tricuspid position with and without right atrial plication.¹²⁻¹⁴ Our results are in agreement with these investigators; we have been impressed with the good results of tricuspid valve replacement in these patients. One of eight patients who had tricuspid valve replacement died in the early postoperative period. This patient's death was due to a documented ventricular fibrillation at a time when the patient was in an excellent hemodynamic condition. Another patient with tricuspid valve replacement who was in NYHA-FC I died seven years after surgery. One patient who had tricuspid valve replacement developed prosthetic valve dysfunction and endocarditis and died 13 years after surgery. All seven patients with tricuspid valve replacement who survived hospitalization improved from NYHA-FC III or IV to I or II. Currently, all our surviving patients with tricuspid valve replacement are in NYHA-FC I or II.

Our observations over 38 years show that Ebstein's anomaly is a continuous rather than discrete abnormality. Patients with mild forms of disease whose functional capacity are mildly limited do quite well by medical management. On the other hand, patients with a more severe form of disease who have severely limited functional capacity improve by surgical treatment. Tricuspid valve replacement, and in some cases, mild right atrial plication, remain our surgical treatment of choice for these patients.

The risk of sudden death is an important problem in patients with Ebstein's anomaly regardless of severity of disease or mode of treatment. New antiarrhythmic drugs and AICD devices offer hope for patients with life-threatening arrhythmias. However, identifying patients who are at risk for sudden death remains a challenge.

Patients with mild forms of Ebstein's anomaly who are

asymptomatic or mildly symptomatic are followed medically at our institution. Evaluations include chest radiographs, electrocardiography, two-dimensional echocardiography, and Doppler and color flow Doppler studies. In addition, patients undergo periodic exercise tests and continuous electrocardiographic monitoring. If any serious arrhythmias are elicited, an electrophysiologic study

is performed. Patients who are symptomatic or who have progressive cardiomegaly, cyanosis, serious arrhythmias, or congestive heart failure undergo operation. Our procedure of choice is tricuspid valve replacement. They are followed in their postoperative course similarly to the medically managed patients, with specific attention to arrhythmias.

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