Ebstein's anomaly

Clinical study of 32 patients in childhood

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Thirty-two patients with Ebstein's anomaly are described. A case is made to maintain a clinical diagnosis because of the potential danger of physiological investigation. It is also to be noted that in the absence of persistent cyanosis or paroxysmal tachycardia, the prognosis of this anomaly in childhood is good, and physiological investigation can be reserved for a preoperative measure. The established criteria for the clinical diagnosis are the presence of a 'scratchy diastolic murmur', and of P pulmonale in the absence of right ventricular dominance in the electrocardiogram.

It is over 100 years since Wilhelm Ebstein first described the anatomy of this condition (Ebstein, 1866), yet not until 1950 were Engle *et al.* and Reynolds able to suggest that a clinical syndrome was present which should lead to the diagnosis of this anomaly. It is still true that this anomaly presents difficulty in diagnosis, and this may in fact be due to its relative rarity. During the years 1954-1970, 32 patients with Ebstein's anomaly have been under the care of the Thoracic Unit of The Hospital for Sick Children, giving an incidence within the spectrum of congenital heart disease discovered in childhood of less than 0.03 per cent.

Subjects

There were 32 patients in all. Of these 10 were boys and 22 girls, a finding which, giving as it does a conspicuous female preponderance of more than 2:1, is not in keeping with the majority of other recorded series (Baker, Brinton, and Channell, 1950; Mayer, Nadas, and Ongley, 1957; Vacca, Bussmann, and Mudd, 1958; Schiebler *et al.*, 1959; Genton and Blount, 1967).

In this series a familial incidence of the anomaly was present in two pairs, in one case father and daughter, in the other two brothers. In a third pair the brother had a ventricular septal defect.

The age at diagnosis of these patients is set out in Table 1, and the age at death of the 7 who

died is given in Table 2. It is to be noted that the infants were more frequently investigated than the other children. This is because the infants presented with ill health and were more difficult to diagnose clinically.

History The variability of the severity of this lesion is such that the history of ill health can date from birth, one case being described in which the lesion was the probable cause of intrauterine death (Genton and Blount, 1967), and in this series cyanosis drew attention to the lesion in the neonatal period in 17 patients. The others were found later in life, the commonest cause for referral being an incidentally found systolic murmur. Effort intolerance at some time in childhood was found in 13 patients, and all those with persistent cyanosis had this. Palpitations are an uncommon complaint in childhood, but 6 of the 7 patients with paroxysmal atrial tachycardia complained of this symptom.

TABLE 1 Age at diagnosis in 32 children

4 dy.*	3 yr.*
5 dy.*	3 yr.
7 dy.*	3 yr.*
10 dy.*	3 yr. 3 mth.
20 dy.	4 yr.
30 dy.	5 yr.*
10 wk.*	5 yr.
10 wk.*	5 yr.
7 mth.*	6 yr.*
9 mth.*	8 yr.
9 mth.*	8 yr.*
1 yr.	9 yr.*
1 yr. 4 mth.	9 yr. 9 mth.
2 yr.*	10 yr.
2 yr.	10 yr.*
2 yr.	11 yr.*

* Investigated.

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TABLE 2 Age at death in 7 children

5 dy.
8 mth.
2 yr.
10 yr. 3 mth.
11 yr.
13 yr.
15 yr.

TABLE 3 Diagnostic criteria for Ebstein's anomaly in 32 children

Patient and sex	Scratchy type diastolic murmur	P pulmonale	Absence of right ventricular dominance	Right bundle-branch block
A.R., M	+	+	+	+
G.P., M	+	+	+	+
F.G., M	-	+	+	+
W.S., M	+	+	-	+
C.B., M	+	+	+	+
L.S., F	+	+	+	-
T.V., F	+	+	+	+
G.L., M	+	-	-	+
P.A., F	+	-	+	+
K.A., F	+	+	+	+
L.D., F	+	-	+	+
S.W., F	+	+	+	+
G.B., F	+	+	+	+
A.L., M	-	+	+	-
N.T., M	-	+	+	-
C.M., F	+	+	+	-
B.J., F	+	-	+	+
S.L., F	-	+	+	+
S.D., F	+	+	+	+
L.W., F	+	+	+	+
S.S., F	-	+	+	-
K.M., F	+	+	+	+
Y.B., F	+	+	+	+
A.D., F	-	-	+	-
C.A., F	-	+	+	-
L.M., M	-	+	+	+
S.B., F	+	+	+	+
K.G., F	+	+	+	+
C.M., F	+	+	+	-
M.C., M	+	-	+	+
J.L., F	+	+	+	+
M.R., F	+	+	+	+

Physical findings

Cyanosis at some time was a common complaint in this series, 17 patients out of 32 had cyanosis at birth with improvement occurring in 11. This finding is not very important, for peripheral and respiratory cyanosis at birth is so common, but in 4 babies cyanosis persisted beyond the neonatal period. Persistent cyanosis of later onset occurred in 4. Persistent cyanosis in this condition carried a poor outlook: of the 7 with persistent cyanosis, 4 died and only 1 was successfully operated upon.¹

¹ By Mr. Donald Ross at the National Heart Hospital.

Clubbing was found as expected only in those with persistent cyanosis.

Cardiac enlargement was found clinically or radiologically in 24, and in only 2 of these was it reported as slight. Obvious radiological right atrial enlargement was recorded in 11 patients.

A systolic thrill was felt at the left sternal edge in 7 patients, but was in no sense diagnostic.

Systolic murmurs were very common, occurring in 28 of 32 patients, but as in other series the condition was found to be murmurless in 4, and in 2 more the murmurs were noted to develop after the first examination. The form of the murmur was usually ejection, but in 5 patients it was so long as to be recorded as pansystolic.

Diastolic 'murmurs' were heard in 26 patients, but whether these noises should truly be called murmurs or not is in doubt. In small children with fast heart rates and in consequence a short diastole, the summation of gallop sounds may be indistinguishable from short diastolic murmurs. These 'murmurs' were found in 26 patients.

Ejection clicks were a surprising finding in this series in 4 patients; in one it was found on catheterization to be due to an associated stenosis of the pulmonary valve, and it is probable in the others also that there may be some associated defect of a semilunar valve.

Pulmonary valve closure—fixed splitting of the pulmonary second sound was recorded in 8 patients.

Electrocardiographic findings

P pulmonale was found in 26 patients; abnormalities of the PR interval in 5 patients, namely first degree block in 2, and 2:1 block in 1, and the WPW phenomenon in 2. Both the first degree block, and the 2:1 block were inconstant. There was right bundle-branch block in 24, and left bundle-branch block in 2, neither of the 2 latter having the WPW phenomenon. Right ventricular dominance, which in any case is to be expected under the age of 5 years, was absent in 24, and there was left ventricular dominance in 6 patients. If the absence of the expected right ventricular dominance is added to those who have left ventricular dominance then 30 of 32 patients show this.

Seven patients had paroxysmal atrial tachycardia, including the 2 with the WPW phenomenon.

Mortality

Seven died, 3 with paroxysmal atrial tachycardia, 1 of whom also had congestive heart failure, and the other 2 suddenly; 5 died with congestive heart failure, and of these 4 had persistent cyanosis.

Discussion

Though it is tempting to think the sex difference within this series is attributable to the fact that this report refers to children less than 15 years of age, it is unlikely that this is the explanation, for other series have also included infants and children. That this finding is probably due to chance is borne out by the analysis of 108 patients reviewed by Vacca *et al.* (1958) which shows a slight male predominance.

If the familial incidence in this series is a representative finding, then the familial incidence of Ebstein's anomaly is higher than in many other congenital malformations of the heart (Campbell and Polani, 1961).

It was Paul Wood who first drew the attention of one of us (R.E.B.-C.) to the value of the 'scratchy diastolic murmur' which he believed was important in the clinical diagnosis. Systolic ejection murmurs, though the rule in this condition, are of little diagnostic significance for they occur normally in childhood, and in the majority of congenital malformations also (Benson, Bonham-Carter, and Smellie, 1961). The problem of this 'diastolic murmur' is to know when it is to be called a murmur and when it is a summation of individual gallop sounds. We have regarded recognizable noises in diastole other than single gallop sounds as 'murmurs'. This 'diastolic murmur' has had ascribed to it a number of causes; its origin is as yet uncertain. The movement of the deformed displaced tricuspid valve in the right ventricular cavity has been proposed (Blacket *et al.*, 1952), and tricuspid stenosis has also been suggested (Schiebler *et al.*, 1959) as the cause.

It is to be expected, in a condition in which part of the right ventricle is 'atrialized', that right ventricular dominance will not be found, and in contrast that the prominent P wave of P pulmonale will be found; further, the frequent coexistence of an atrial septal defect in this anomaly leads one to suppose that right bundle-branch block would also be found.

Eighteen of these patients were subjected to physiological study by cardiac catheterization; in 6 the attempt was abandoned because of the development of a worrying arrhythmia but none of these was fatal. This points to the importance of using the established clinical criteria for the diagnosis of this anomaly.

Taking as an hypothesis for clinical diagnosis the previously established triad of the presence of a 'scratchy, diastolic murmur', P pulmonale without right ventricular dominance but with bundle-branch block on the right or the left, in this series we find: a 'scratchy diastolic murmur' present in 24 of 32 patients, P pulmonale in 26 patients, right bundle-branch block in 24 patients, and an absence of right ventricular dominance, which is more significant in childhood than in adult life, in 30. This figure of 30 includes 6 patients who showed left ventricular dominance abnormal for age (Cassels and Ziegler, 1966).

The fact that left bundle-branch block was found in 2 patients makes it unlikely that the finding of bundle-branch block is necessarily associated with the presence of a significant atrial septal defect, but more probably that the finding is associated with an anomaly of the bundle itself, which is likely when the anatomy of this anomaly is considered, and particularly when it is noted that in this series out of 7 recorded arrhythmias there were 2 with the WPW phenomenon, and the WPW phenomenon has been recorded in other series (Lev, Gibson, and Miller, 1955).

Conclusion

It is, therefore, suggested because of the potential danger of arrhythmia during cardiac catheterization (6 in 18 such investigations) that this should be reserved as a preoperative measure, and that a clinical diagnosis should be maintained until that time. The indications for operation in childhood are difficult because of the possibility of having to replace the tricuspid valve at a time when growth is incomplete. However, persistent cyanosis, which occurred in 4 out of the 7 who died, would in the older child be an indication for considering an operation.

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