

EBSTEIN'S DISEASE

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A case of Ebstein's malformation of the tricuspid valve is described. The clinical diagnosis and the findings at angiocardiology and cardiac catheterization are reviewed in all the cases published since 1948.

CASE HISTORY

Linda B. was born in September, 1946. Delivery was normal and her mother had been well during pregnancy. She was first seen at St. Thomas's Hospital in January, 1947, when aged 4 months. There was a history of vomiting for one week attributed to a mild attack of gastro-enteritis. A systolic murmur was heard over the praecordium, but cyanosis was not noted. At the age of 2 years the child was an in-patient with severe stomatitis; comments at the time included that she had a red face and a red tongue, and one observer recorded "a reduplicated apical second sound." In January, 1953, when aged 6½ years she was re-admitted with the following history. She had had a cough, fever, and abdominal pain for the previous 24 hours. This settled rapidly. But she was seen to be cyanosed, and on direct questioning her father said that the child would never hurry and became blue if she tried to do so. He had noticed this for the previous two to three years. She also squatted when tired.

She was a red-haired, normally developed girl (weight, 2 st. 4 lb.; height, 3 ft 5 in.). The abnormalities were confined to the cardiovascular system. There was moderate central cyanosis and the fingers and toes were clubbed. The pulse was regular (90 per minute). The blood pressure was 100/75 mm. Hg. Jugular venous pulsation was visible in the neck, but the pressure was not raised and its form was normal. The *right* side of the chest was slightly prominent. The apex beat was in the fifth space in the anterior axillary line. There was no right ventricular impulse to the left of the sternum. The most obvious auscultatory finding was a triple rhythm. The added sound occurred at about the time of a third heart sound, and was loudest in the fourth left space parasternally where it was as loud as the first sound. There was a moderately loud systolic murmur maximal at the same site, accompanied, in full expiration, by a slight thrill. There was no diastolic murmur.

A radiograph showed that the heart shadow was considerably enlarged to the right and left, and the borders showed an even contour. On radioscopy there was a marked outward pulsation of the right border during systole. On the left border pulsation was confined to the lower third, and was inward during systole. In the left anterior oblique position (60°) the right auricle was enlarged and the posterior border of the heart overlapped the spine. The pulmonary vascular markings were diminished.

An electrocardiogram (Fig. 2) showed that the P.R. interval was 0.24 sec. There was a complete right bundle branch block, the form of which was unusual in that both primary and secondary R. waves were small in V₁, V₂, and V₃.

The blood count showed Hb 118% (17.6 g.), R.B.C.s 5,540,000, P.C.V. 54%.

The phonocardiogram is described under Fig. 12.

An A.P. and right lateral series of angiocardigrams were taken (Figs. 3–6). Injections of 25 ml. of 70% "pyelosil" were given and eight films exposed in five seconds on each occasion using the cassette changer

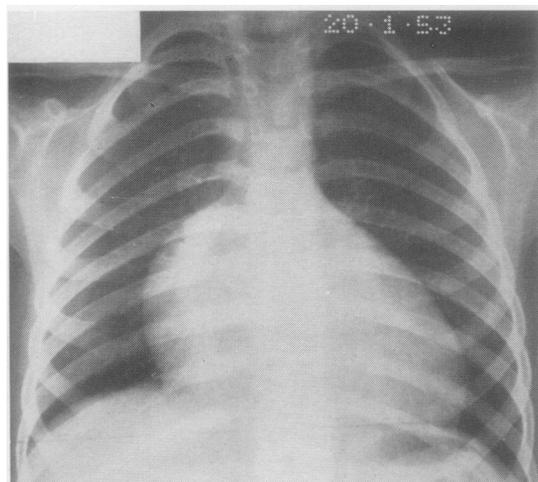


FIG. 1.—Postero-anterior chest radiograph showing typical outline and diminished lung vascular shadows.

devised by one of us (Thursfield, 1953). In the A.P. series the dye can be seen entering the right auricle in the first film (Fig. 3); 0.6 seconds later the heart shadow is largely opaque and the aorta and pulmonary artery contain dye (Fig. 4). In the right lateral series dye can be seen passing from the right to the left auricle in the first film (Fig. 5). In the second film the left auricle is opaque and the pulmonary artery and aorta contain dye (Fig. 6). As

no film was taken after five seconds delayed emptying of the right auricle could not be judged. No filling defect attributable to the tricuspid valve was seen.

The results of cardiac catheterization under rectal thiopentone are shown in Figs. 7-11 and Table II. The patient was horizontal; zero level, sternal angle. The A.P. chest diameter was 15 cm. The right auricle was very enlarged. The catheter repeatedly entered the right ventricle about half way up the left cardiac border. Here ventricular extrasystoles were provoked. Thus in the position shown in Fig. 9 the catheter tip was in the right auricle (Fig. 7), and in Fig. 10 it was in the right ventricle (Fig. 8). Between the two lines in Fig. 11 the catheter recorded a right ventricular pressure. Beyond the second line the catheter tip passed to the apex, on some occasions recording a right auricular, and on others a right ventricular pressure. It was therefore thought to be passing either behind, or in front, respectively, of the tricuspid sheet. The blood oxygen saturation was 55% in both the right auricle and the right ventricle.

DISCUSSION

Ebstein first described this malformation in 1866 (Ebstein, 1866). The post-mortem descriptions of the abnormality are remarkably constant. The anterior cusp of the tricuspid valve is a sheet, frequently with fenestrated edges, attached to the inferior and septal walls of the right ventricle, dividing an enlarged right auricle posteriorly from a reduced right ventricle anteriorly. The free margin of the sheet bounds the atrio-ventricular orifice just below the pulmonary valve; the blood flow from the right auricle is diverted upwards. The wall of the right ventricle is thin. There is frequently either a patent foramen ovale or a fenestrated fossa ovalis.

Walton and Spencer (1948) reported a case and listed the findings in the 18 previously recorded cases. Table I summarizes the clinical features of the cases published since then, and Table II the catheter findings.

From Table I a characteristic picture emerges. Thus, in those patients in whom the findings are recorded the incidence of certain features is cyanosis (17 out of 21 patients); systolic murmur (20 out of 24); diastolic murmur (13 out of 26); added sound (16 out of 19); cardiac outline resembling that of a pericardial effusion (19 out of 24); right bundle branch block (20 out of 25); peaked P waves (19 out of 25). Our case showed all these features except a diastolic murmur.

In nine patients a note is made of the presence or absence of pulsation to the left of the sternum in the area where a hypertrophied right ventricle may be felt. In four it was absent and in the

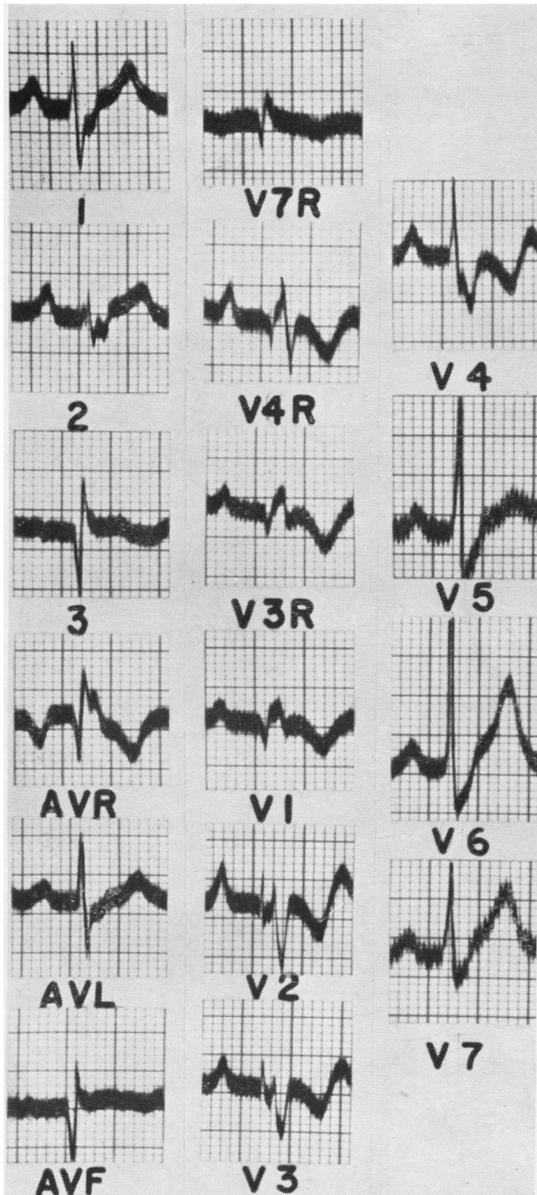


FIG. 2.—Electrocardiogram (QRS darkened).

TABLE I
CLINICAL, X-RAY, E.C.G., AND POST-MORTEM FINDINGS IN EBSTEIN'S DISEASE

Walton and Spencer's (1948) Review of 18 Cases Plus Another	History		Physical Signs							Radiograph			E.C.G.			Operation	Necropsy			
	Age	Sex	Paroxysmal Tachycardia	Onset of Cyanosis	Central Cyanosis	Clubbing	Jugular Pulse	Right Ventricular Impulse	Systolic Murmur	Diastolic Murmur	Added Sounds	Cardiac Outline	Movement	Pulmonary Vascular Markings	P.R. Interval			O.R.S. Complex	P Wave	
Berber (1947) ...	No details given	F	+		0	0	-	-	0	-	-	T	F	N	0.24	RBBB	N	0	0	
Tourniaire and others (1949)	39	F	+	9	+	+	+	+	EDM	Gallop rhythm	T	<N	<N	<N	<N	RBBB	P	0	PFO	
Engle and others (1950) 1	10	F	+1	0	+	+	N	-	0	" "	T	-	<N	<N	RBBB	P	0	0	PFO	
Engle and others (1950) 2	5	M	0	0	+	+	-	-	0	" "	T	-	<N	<N	RBBB	P	0	0	PFO	
Baker and others (1950) 1	16	F	0	3	+	+	-	-	0	Third	C	-	<N	<N	RBBB	P	0	0	PFO	
Baker and others (1950) 2	17	M	0	0	+	+	-	-	0	Triple rhythm ⁴	D	-	<N	<N	RBBB	P	0	0	PFO	
Reynolds (1950)	11	F	0	0	+	+	N	-	0	Reduplicated 1st and 2nd	T	<N	<N	<N	RBBB	N	0	0	FFO ⁸	
Paul and others (1951)	33	F	0	0	+	+	-	-	0	0	T	<N	<N	<N	RBBB	N	0	0	0	
Barger and others (1951)	16	M	0	7	+	+	-	-	0	Third	G	<N	<N	<N	RBBB	P	0	0	PFO	
Solier and others (1951)	30	M	0	0	+	+	-	-	EDM	0	T	<N	<N	<N	RBBB	P	0	0	PFO	
Aegerter	34	F	0	0	+	+	-	-	0	0	T	<N	<N	<N	RBBB	P	0	0	0	
Mannheimer and others (1951)	3	F	0	0	+	+	-	-	0	Gallop rhythm ^{4,15}	T	-	<N	<N	RBBB	N	0	0	0	
Graux and Merlen (1951)	72	M	+	0	0	0	N	-	0	0	H	<N	<N	<N	RBBB	N	0	0	0	
van Lingen and others (1952)	22	F	0	0	0	0	N	-	MDM	0	T	<N	<N	<N	RBBB	P	0	0	0	
Blacket and others (1952)	20	F	0	6	+	+	N	-	MDM	0	T	<N	<N	<N	RBBB	P	0	0	0	
Blacket and others (1952)	16	F	0	0	+	+	N	-	0	0	T	<N	<N	<N	RBBB	P	0	0	0	
Blacket and others (1952)	2	M	0	0	0	0	N	-	0	Late diastolic	T	<N	<N	<N	RBBB	P	0	0	0	
Blacket and others (1952)	2	M	0	0	0	0	N	-	0	Mid and late diastolic	T	<N	<N	<N	RBBB	P	0	0	0	
Blacket and others (1952)	18	F	0	0	0	0	N	-	0	Third	T	<N	<N	<N	RBBB	P	0	0	0	
Goodwin and others (1953)	4	M	0	0	0	0	N	-	MDM	0	J	<N	<N	<N	RBBB	N	0	0	0	
Goodwin and others (1953)	13	M	0	9	+	+	V	-	0	0	T	<N	<N	<N	RBBB	P	0	0	0	
Goodwin and others (1953)	14	M	0	12	+	+	AV	-	0	0	T	<N	<N	<N	RBBB	P	0	0	PFO	
Goodwin and others (1953)	13	M	0	0	+	+	N	-	0	0	T	<N	<N	<N	RBBB	P	0	0	0	
Broadbent and others (1953)	6	M	0	0	+	+	-	-	0	0	G	<N	<N	<N	RBBB	P	0	0	0	
Broadbent and others (1953)	35	M	0	<1	+	+	-	-	EDM	0	T	<N	<N	<N	RBBB	P	0	0	0	
Broadbent and others (1953)	2	M	0	0	+	+	-	-	DM	0	T	<N	<N	<N	RBBB	P	0	0	0	
Henderson and others (1953)	29	M	0	0	+	+	-	-	EDM	0	T	<N	<N	<N	RBBB	P	0	0	0	
Henderson and others (1953)	26	M	0	0	+	+	-	-	EDM	0	T	<N	<N	<N	RBBB	P	0	0	0	
Present case	6	F	0	4	+	+	N	-	0	0	T	<N	<N	<N	RBBB	P	0	0	0	
Total	47																			

AV, "a" and "v" waves. C, Impossible to assess; chest deformity. D, Typical except left auricular appendix prominent (angiogram film only published). E, R.A. pulsation > normal; R.V. < normal. F, R.A. pulsation > normal. G, Left heart border straighter than the typical. H, Atypical. I, RBBB, 2/Incomplete right bundle branch block. J, Atypical; almost boot shaped. LBBB, Atypical left bundle branch block. FFO, Fenestrated fossa ovals. MDM, Mid-diastolic murmur. N, Normal. P, Peaked. PFO, Patent foramen ovale. T, Typical biconvex outline, resembling that of a pericardial effusion. TV, Typical Ebstein-type abnormality tricuspid valve. >N, More than normal. <N, Less than normal. -, Not recorded. +, Recorded present. 0, Recorded absent. 1, Under "age at onset cyanosis" indicates present from birth. 2, Attacks abdominal pain; colour peculiar. 3, Systemic-pulmonary anastomosis; death. 4, Phonocardiogram. 5, Thoracotomy and cardiotomy; death. 6, Marginal fenestration of septum primum. 7, On exertion only. 8, Slight, 9, Weak, 10, Systolic impulse over outflow tract of right ventricle. 11, Parasternal systolic pulsation in second and third L.I.C.S. 12, Death followed induction of anaesthesia for systemic-pulmonary anastomosis. 13, Pulmonary systolic click. 14, (probably reduplicated first sound). 15, On admission in cardiac failure had auricular fibrillation. 16, On admission P.R. interval was 0.24 while the patient was taking digitalis (Jackson, 1953).

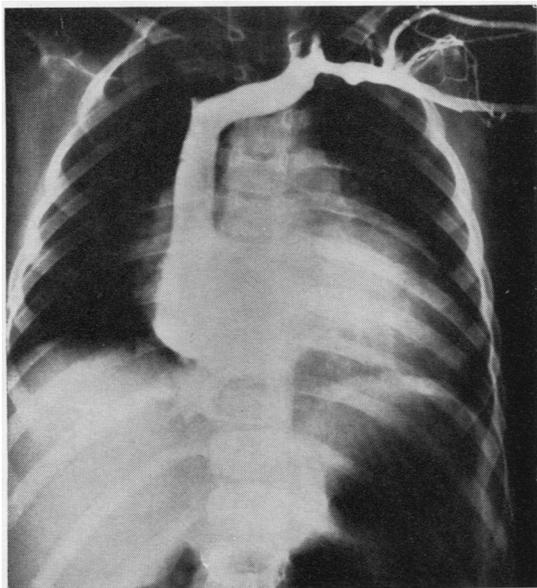


FIG. 3.—Antero-posterior angiogram.

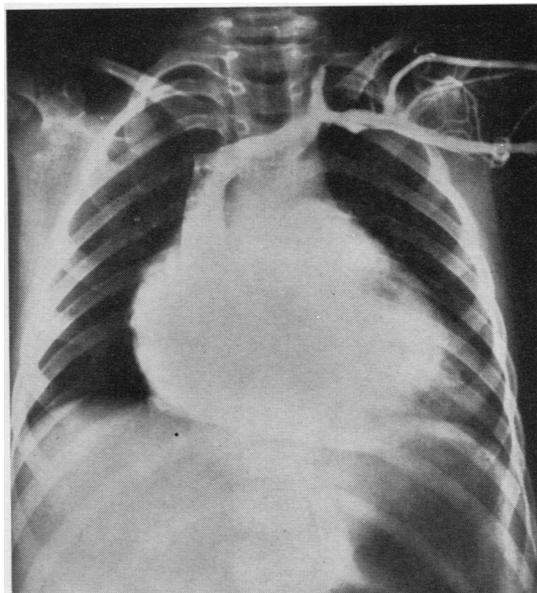


FIG. 4.—Antero-posterior angiogram taken 0.6 sec. after Fig. 3.



FIG. 5.—Right lateral angiogram. A jet of dye can be seen passing from the right auricle to the left.

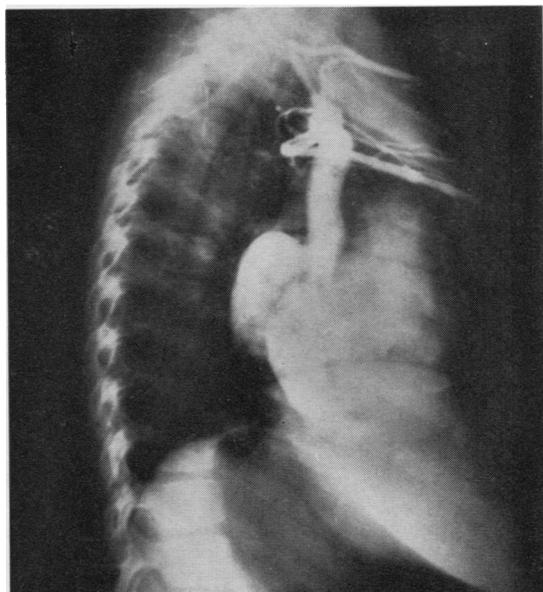


FIG. 6.—Right lateral angiogram taken 0.6 sec. after Fig. 5.

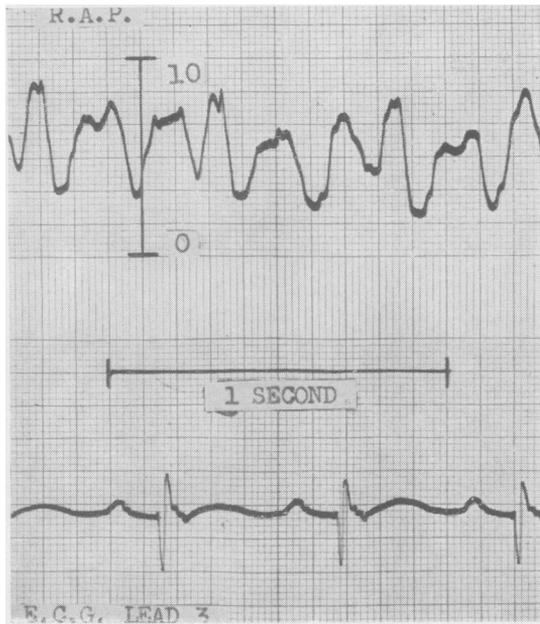


Fig. 7

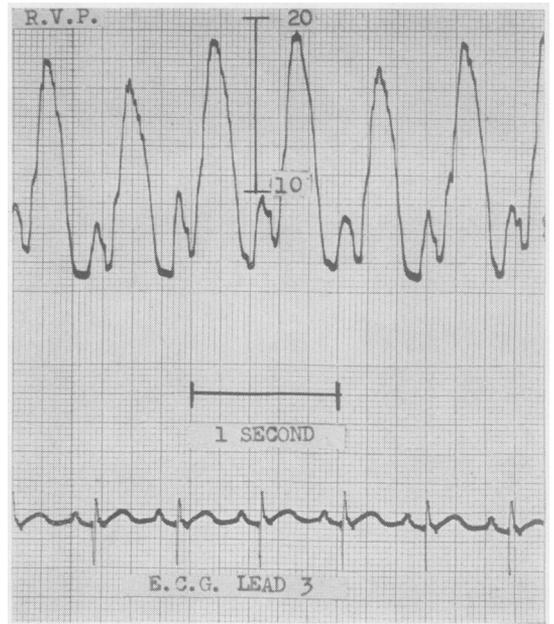


Fig. 8

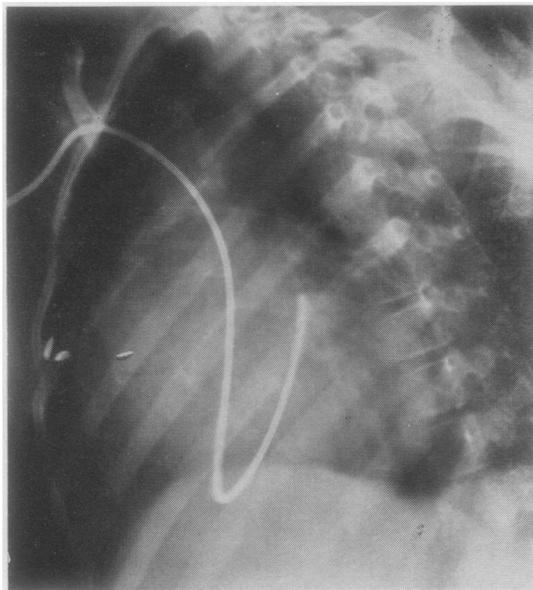


Fig. 10

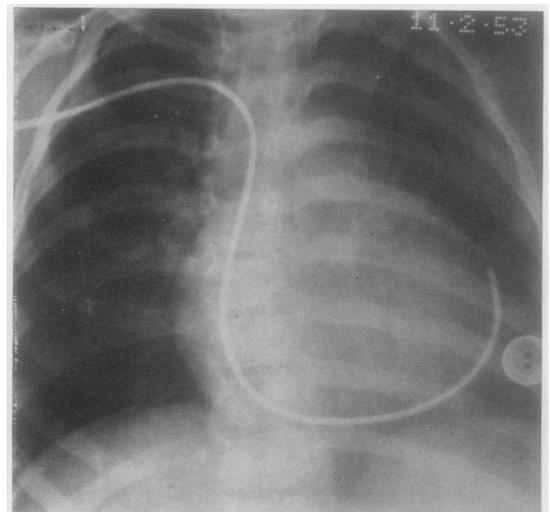


Fig. 9

FIG. 7.—Right auricular pressure (paper speed 50 min./sec.).

FIG. 8.—Right ventricular pressure. Note large “a” wave. (Paper speed 25 min./sec.)

FIG. 9.—Catheter tip recording right auricular pressure. Just above this point it entered the right ventricle (right-hand horizontal line, Fig. 11).

FIG. 10.—Left anterior oblique radiograph; the catheter tip has just entered the right ventricle.

FIG. 11.—When the catheter tip was above the horizontal lines a right ventricular pressure was recorded. As the tip passed from the left-hand line to the apex, the catheter pressure was either right auricular or right ventricular.

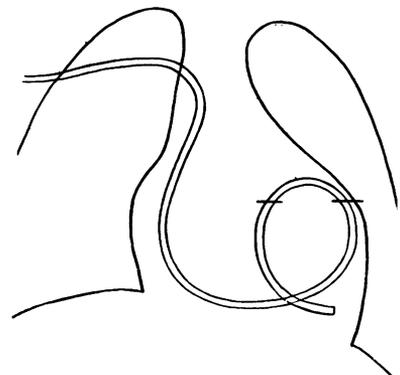


Fig. 11

TABLE II
CATHETER FINDINGS

	Reference Point	Large Right Atrium	Right "Auricular" Pressure*	Right Ventricular Pressure*	Pulmonary Arterial Pressure*	Entered Left Atrium	Arterial Saturation	Hazards	Other Features
Tournaire and others (1949)	-	-	13/10	16/13	16/13	-	-	-	Calcified tricuspid
Engle and others (1950)	-	-	8/1	21/7	-	+	66	-	-
Baker and others (1950)	1 Mid-axillary line	+	10	-	-	+	75	Popliteal embolism 9 days later	-
	2	-	-	-	-	+	93	-	-
Reynolds (1950)	-	-	-	-	-	+	79	-	-
Mannheimer and others (1951)	-	+	4.5/2.5	14/4.5	12.5/6.5	-	81	-	-
van Lingen and others (1952)	1 5 cm. post. angle of Louis	-	7.4	22/4	22/10	-	87	-	-
	2	-	9.5	22/8	22/10	-	69	-	-
Blacket and others (1952)	1	+	7/1	13.4	13.4	-	97	-	R.V. infundibular wall thin
	2	-	-	-	-	-	-	Cardiac arrest while catheter in S.V.C. Fatal	-
Goodwin and others (1953)	Sternal angle	+	3	-	-	-	88	-	-
Broadbent and others (1953)	1	-	11/4	18/3	18/10	-	65	Suprav. tachycardia Controlled "cedalinid"	-
	2	-	10/4	19/4	20/10	-	89	-†	-
	3	-	25/13	33/13	34/22	-	84	-†	C.F. Tricuspid incompetence
Henderson and others (1953)	Right atrium	+	7	9-10	14	-	91	-	-
Present case . . .	Sternal angle	+	8.4	18.4	-	-	-	-	See text

* All pressures converted to mm. Hg.

† Burchell, Helmholtz, and Wood (1953) report that one patient with Ebstein's disease had "a cerebral embolus three weeks after the procedure [catheterization] and the onset of the [auricular] fibrillation." Another had a "transient confusional state and hemiparasthesia lasting about twenty minutes." These two patients are not from among the three described by Broadbent and others (1953) in the same number of the *Proc. Staff Meet. Mayo Clinic* (Burchell, 1953).

Campbell (1953) reports one death in six patients catheterized, and reports Dr. Paul Wood's experience of one death during catheterization out of four or five. Götzsche and Falholt (1954) catheterized six without incident.

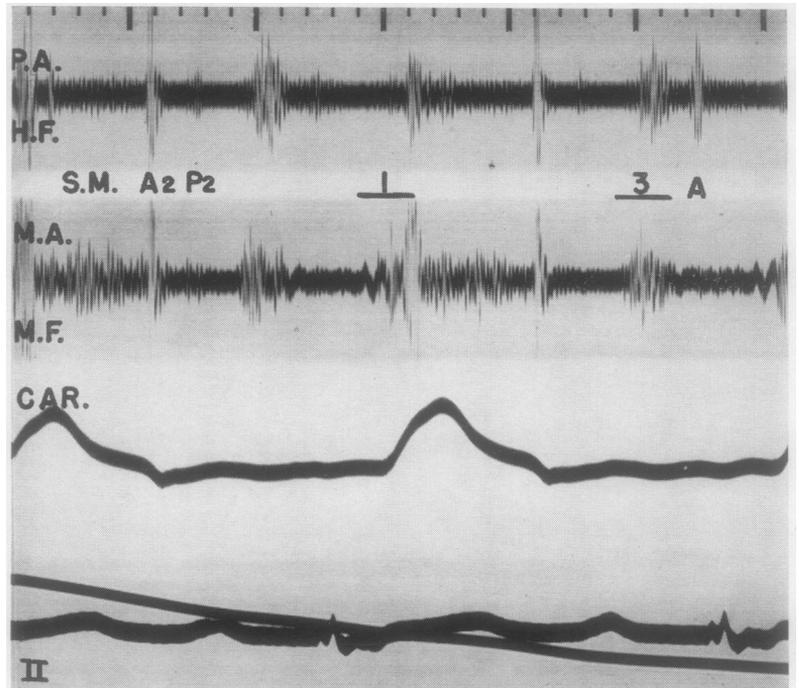
remainder slight. Special attention to this sign in future cases may show that absence of a marked pulsation is a useful sign in differentiating Ebstein's disease from forms of cyanotic congenital heart disease in which the right ventricle is hypertrophied.

Reference from the phonocardiogram (Fig. 12) and the pressure records (Figs. 7 and 8) to the electrocardiogram in each shows that the louder and later element of the first heart sound coincides with the onset of right ventricular systole. The added sound (3 in Fig. 12) occurs in early diastole when the right ventricular falls below the right auricular pressure. The louder element of the first sound therefore coincides with the tri-

cuspid valve closing, and the added sound with the tricuspid valve opening. Right ventricular systole (Fig. 8) and left ventricular systole (carotid tracing Fig. 12) are of the same duration, but the right ventricular systolic period is delayed 0.08 sec., presumably by the right bundle branch block. Another sound (A, Fig. 12) occurs at the time of auricular contraction (Figs. 7 and 8).

Of the 10 patients in whom an angiocardiogram was reported, early filling of the left auricle was noted in all except that of Reynolds (1950), the first of Goodwin's (Goodwin, Wynn, and Steiner, 1953), and Henderson's (Henderson, Jackson, and Swan, 1953). Only in the first of Baker's (Baker, Brinton, and Channell, 1950) patients was there

FIG. 12.—Synchronous phonocardiograms; high frequency (HF) from pulmonary area (P.A.) and medium frequency (MF) from mitral area (M.A.), carotid tracing (CAR.), and electrocardiogram (II). First heart sound split into several components, the first probably associated with mitral valve closure because it precedes the carotid upstroke; later components probably associated with tricuspid valve closure delayed by right bundle branch block. Long systolic murmur (SM). Second heart sound split into earlier aortic component, shown by synchrony with dicrotic notch of carotid tracing followed 0.07 sec. later by pulmonary component. Diastolic filling sounds accentuated and possibly separated into left- and right-sided third sounds, the latter delayed by right bundle branch block, and a later auricular sound (A). (Time marker, 0.2 sec. and 0.04 sec.) (The phonocardiogram and report were kindly provided by Dr. A. Leatham.)



no delay in the emptying of the right auricle. A filling defect thought to be due to the misplaced tricuspid valve was described by Soloff, Stauffer, and Zatuchni (1951) and Goodwin, Wynn, and Steiner (1953).

At catheterization the pressures recorded were similar to the reported cases in Table II (with the exception of Broadbent's (Broadbent, Wood, Burchell, and Parker, 1953) case 3; this patient was in cardiac failure). The right auricular pressure was raised and the right ventricular pressure normal. In the absence of tricuspid stenosis this combination does not occur in any other form of congenital cyanotic heart disease. The high right auricular pressure is an indication of the inefficiency of the right ventricle, and is responsible for the right-to-left shunt. In none of the recorded cases has there been evidence of tricuspid stenosis as judged by a diastolic pressure difference across the valve. Only in the case of Broadbent's which was in cardiac failure was there evidence of tricuspid incompetence, namely a large "V" wave in the right auricular tracing. That there is a danger in catheterizing these patients is shown by the complications reported (Table II). However, anastomotic operations have been invariably

lethal in the reported cases (Table I) so that where there is doubt catheterization is justifiable.

SUMMARY

A case of Ebstein's disease is described. A finding at cardiac catheterization which we consider as diagnostic is recorded. Cases published since 1948 are summarized. The possibility of making a clinical diagnosis and the danger of operation are stressed.

The patient was under the care of Dr. J. Forest Smith, who kindly gave permission for publication. We would also like to thank Miss D. Holdway and Mrs. N. Thompson for secretarial, and Mr. J. O. L. Wrigley and Mr. K. G. Moreman for technical and photographic, assistance respectively.

REFERENCES

- Aegerter, E. E., quoted Soloff, L. A., *et al.* (1951).
 Baker, C., Brinton, W. D., and Channell, G. D. (1950). *Guy's Hosp. Rep.*, **99**, 247.
 Barger, J. D., Henderson, C. E., and Edwards, J. E. (1951). *Amer. J. clin. Path.*, **21**, 576.
 Berber, S. G. (1947). *Cuore e Circol.*, **31**, 54.
 Blacket, R. B., Sinclair-Smith, B. C., Palmer, A. J., Halliday, J. H., and Maddox, J. K. (1952). *Aust. Ann. Med.*, **1**, 26.
 Broadbent, J. C., Wood, E. H., Burchell, H. B., and Parker, R. L. (1953). *Proc. Mayo Clin.*, **28**, 79.
 Burchell, H. B. (1953). Personal communication.
 — Helmholz, H. F., and Wood, E. H. (1953). *Proc. Mayo Clin.*, **28**, 50.

- Campbell, M. (1953). *Brit. Heart J.*, **15**, 363. Editorial note.
- Ebstein, W. (1866). *Arch. Anat. Physiol., Lpz.*, p. 238.
- Engle, M. A., Payne, T. P. B., Bruins, C., and Taussig, H. B. (1950). *Circulation*, **1**, 1246.
- Goodwin, J. F., Wynn,*A., and Steiner, R. E. (1953). *Amer. Heart J.*, **45**, 144.
- Götzsche, H., and Falholt, W. (1954). *Ibid.*, in the press.
- Graux, P., and Merlen, J.-F. (1951). *Arch. Mal. Coeur*, **44**, 263.
- Henderson, C. B., Jackson, F., and Swan, W. G. A. (1953). *Brit. Heart J.*, **15**, 360.
- Jackson, F. (1953). Personal communication.
- Lingen, B. van, McGregor, M., Kaye, J., Meyer, M. J., Jacobs, H. D., Braudo, J. L., Bothwell, T. H., and Elliott, G. A. (1952). *Amer. Heart J.*, **43**, 77.
- Mannheimer, E., Bengtsson, E., Krämer, R., Warth, J., Wade, G., Wegelius, C., and Winberg, J. (1951). Proc. of the Tenth Northern Paediatric Congress, Stockholm, 1951. *Acta paediat., Uppsala*, **40**, Suppl. 83, p. 194. (Unpublished data kindly provided by Dr. Mannheimer.)
- Paul, O., Myers, G. S., and Campbell, J. A. (1951). *Circulation*, **3**, 564.
- Reynolds, G. (1950). *Guy's Hosp. Rep.*, **99**, 276.
- Soloff, L. A., Stauffer, H. M., and Zatuchni, J. (1951). *Amer. J. med. Sci.*, **222**, 554.
- Thursfield, W. R. R. (1953). *Brit. J. Radiol.*, **26**, 383.
- Tourniaire, A., Deyrieux, F., and Tartulier, M. (1949). *Arch. Mal. Coeur*, **42**, 1211.
- Walton, K., and Spencer, A. G. (1948). *J. Path. Bact.*, **60**, 387.