CASE REPORTS

TWO CASES OF CONGENITAL DISEASE OF THE HEART WITH SUB-DIAPHRAGMATIC SITUS INVERSUS

BY

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The two rare cases to be described, which happened to be sent to this department within a few months, are worth recording both for their own interest and as an addition to the scanty literature relating to this unusual syndrome. It is also of interest that in both cases the diagnosis was not at once apparent, although all the necessary information was readily available on examination.

Case Histories

Case 1. A male, aged 9 months, had always been healthy and normal in development. During routine attendance at a child welfare centre the doctor noticed that he was cyanosed and sent him to a hospital, where a diagnosis of respiratory embarrassment due to an enlarged thymus was made. The child was sent to University College Hospital, where further investigations were carried out.

PHYSICAL EXAMINATION. A well developed, plethoric child showed cyanosis of lips, fingers, and toes, and moderate clubbing of fingers and toes. There was no dyspnoea or stridor. The weight was 17½ lb.

The heart was in the normal position, slightly enlarged to left, with a loud systolic murmur over the praecordium; the maximum murmur was at the pulmonary area. The lungs were normal. The liver was symmetrically palpable. All other systems were normal.

RADIOGRAPHY. The chest radiograph showed the heart enlarged to left with very prominent pulmonary conus. The liver shadow was symmetrical (fig. 1).

Barium swallow. Barium was seen in the oesophagus, passing to the right through the diaphragm and outlining a normal stomach and duodenum in the mirror-image position.

Barium enema. The sigmoid and descending colon were in the right iliac fossa.

ELECTROCARDIOGRAPH. This showed simple sinus tachycardia, abnormal Q.R.S. complex, and large S wave in all three leads, and large P and T waves in Lead II (fig. 2).

FAMILY HISTORY. The parents and their relatives are normal. There is one male sibling surviving;

he is normal and aged eleven years. The mother had had one stillbirth in 1933, a baby with bilateral talipes. There had been no post-mortem examination.

Case 2. A female, aged 17½ months, had been apparently normal and healthy up to the age of 12 months, when she ceased to gain weight. The parents had noticed blueness of the lips on several occasions, especially when the child cried or coughed. She was taken to a hospital, where a diagnosis of tuberculous hilar adenopathy with pulmonary congestion was made on the radiographic findings and history of four weeks' cough. She was then transferred to University College Hospital where further investigations were made. A history of pertussis contact was later elicited.

PHYSICAL EXAMINATION. The child was small, thin, and pale, with blue lips, and rapid pulse and respiration rates. She had repeated bouts of coughing. She was normally intelligent and co-operative. There was early clubbing of the fingers. The weight was 16 lb.

The heart was in the normal position, enlarged to right and left, with no murmurs.

There were widespread rhonchi and crepitations in the lungs, more on the right side than on the left. A slightly enlarged and soft liver was palpable in the left hypochondrium. All other systems were normal. The Mantoux reaction was negative.

RADIOGRAPHY. A radiograph of the chest showed the heart enlarged to both sides, a prominent pulmonary conus, and congestion of both lung fields spreading from the hilum. There was a stomach bubble under the right dome of the diaphragm, with the liver shadow under the left (fig. 3).

After a barium meal the position of the stomach was confirmed; the duodenum was not clearly outlined but was probably in the mirror-image position; the caecum was in the left iliac fossa, the descending colon and sigmoid in the right.

ELECTROCARDIOGRAPH. This showed simple sinus tachycardia. There were substantially normal curves with flat T wave in all leads and inverted P in Lead III (fig. 4).

FAMILY HISTORY. The parents are normal. There is one female sibling, normal, and aged four and a half years; and one male sibling, normal, and aged three months. A paternal aunt has a deformed hand, the fingers being only half an inch long, and a paternal cousin congenital absence of the right foot and deformity of one hand.

Review of the Literature

The literature on visceral heterotaxy is extensive, but particular reference must be made to Lichtmann's (1931) thorough review of the subject in its relation to heart disease. In brief, the incidence of situs inversus with this association may be summarized from the available literature of the last fifteen years as follows:

1. Total situs inversus with mirror-image dextrocardia giving rise to no abnormality is relatively common.

2. Total situs inversus with congenitally abnormal dextrocardia is not unduly rare. Six cases picked at random showed three instances of Eisenmenger's complex, one of Fallot's tetrad, and two of functionally bilocular heart with persistent conus arteriosus.

3. Partial situs inversus with normal laevocardia is uncommon; eight cases in all were recorded, mostly chance findings in otherwise healthy subjects.

4. Isolated dextrocardia is rare, but, when it occurs, is usually associated with congenital abnormality. In Lichtmann's series of 161 cases, only three showed a normal heart.

5. Partial situs inversus with congenitally abnormal laevocardia (the condition under discussion) is also rare. Diligent search has brought records of seven cases to light, the description of one of which (Clemente, 1931) is not available in this country. The case recorded by Shaw and Blake (1924) is not felt to be a true instance of this syndrome because they obtained the electrocardiograph typical of dextrocardia, while the clinical position of the heart appears from their evidence to have been at least central.

Developmental Aspects

Cockayne (1938) suggests that situs inversus is a Mendelian factor carried by a recessive autosomal gene, and has therefore a definite familial incidence in families where consanguineous matches have occurred. He cites the frequent association of other congenital abnormalities, particularly heart disease, and mentions the association of congenital bronchiectasis, and paranasal sinusitis with situs inversus—Kartagener's triad (Kartagener, 1933; Kartagener and Ulrich, 1935). He also suggests that in partial situs inversus the frequency of congenital morbidity is higher than in complete. He explains the partial type on the basis of an 'allelomorphic' gene.

The embryological account of the matter refers to the failure of completion of the normal 'embryonic spiral' in which the abdominal and thoracic

organs migrate from their early median positions to their normal adult-situations. Abbott (1936) refers to the arrest of development of the heart at a stage bearing an atavistic resemblance to reptilian or amphibian types, as in the case of bilocular and trilocular hearts with persistent right-sided aorta.

In an article which is the only critical review of this subject Forgacs (1947) goes into the embryological aspect of the question more fully and gives a most convincing account of the condition's development. For practical purposes, he believes, when the heart is on the same side as the liver it is never free from congenital morbidity.

In the two cases here described there is no suggestion of consanguineous marriage for three generations and no evidence of situs inversus in any near relations. The incidence of the other types of congenital deformity in relatives of Case 2 is, however, of some interest.

Diagnosis

Although it is almost impossible to draw any conclusions, it is perhaps relevant to mention the six other cases of which records are available. Of the three who came to post-mortem examination all had transposition of the aorta and pulmonary artery with left-sided aortic arch; all three had patent interventricular septa—two with small openings and loud systolic murmurs, one with almost complete absence of the septum and no murmur; two had atresia of the pulmonary artery with persistent ductus arteriosus; all were cyanotic and dyspnoeic on effort.

Added to these facts, Abbott has a radiograph in her atlas of a case of transposition of the great vessels with defective interventricular septum; there is enlargement of both ventricles and prominence of the pulmonary conus remarkably similar to that shown in our two cases.

Neither of Forgacs's cases had come to autopsy.

Prognosis

It is possible to say only that one child looks fit and the other far from it. Of the six similar cases, one was alive at nine years (Girod and Sarasin, 1932) and another died at nineteen years (Pernkopf, 1928). The others lived only for ten weeks (Miller, 1925) and seven months (Hu, 1929). Of Forgacs's cases, one was alive and fit at 25 years, the other less fit at five years. Abbott's case of transposition of the great vessels with defective septum also had complete heart block and lived until the age of twenty years.

Conclusion

It seems fair to infer from the rather scanty evidence that partial situs inversus tends to be associated with gross abnormalities of the heart of a type which can be accounted for theoretically by disorders of the 'embryonic spiral.' It is not

FIG. 1.—Case 1: radiograph showing heart in normal position, with enlarged pulmonary conus. A stomach bubble can be seen overlying the right lobe of the symmetrical liver.

FIG. 2.—Case 1: electrocardiograph. (For description see text, p. 132.)

PLATE I

FIG. 3.—Case 2: radiograph showing heart in normal position with enlarged pulmonary conus, a stomach bubble under the right dome of the diaphragm, and post-pertussis hilar adenopathy on the right.

FIG. 4.—Case 2: electrocardiograph. (For description see text, p. 132.)

PLATE II

unnatural to expect that transposition of the vessels in some degree should be a feature, and defective septum is evidently associated with it. For the rest, it must be said that in all three autopsied cases there were numerous minor cardiac defects.

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REFERENCES

- Abbott, M. E. (1936). *Atlas of Congenital Heart Diseases*. American Heart Association. New York.
- Clemente, G. (1931). *Folia med., Napoli*, 17, 347, 494, 521.
- Cockayne, E. A. (1938). *Quart. J. Med.*, n.s. 7, 479.
- Forgacs, P. (1947). *Brit. Heart J.*, 9, 27.
- Girod, R., and Sarasin, R. (1932). *Schweiz. med. Wschr.*, 62, 1035.
- Hu, C. H. (1929). *Amer. J. Path.*, 5, 389.
- Kartagener, M. (1933). *Beitr. Klin. Tuberk.*, 83, 489.
- , and Ulrich, K. (1935). *Ibid.*, 86, 349.
- Lichtman, S. S. (1931). *Arch. intern. Med.*, 48, 683.
- Miller, J. (1925). *Amer. J. Path.*, 1, 467.
- Pernkopf, E. (1928). *Z. Anat. Ewick lungsgesichte*, 87, 661.
- Shaw, E., and Blake, H. K. (1924). *Amer. J. Dis. Child.*, 28, 204.