

Natural history of disease shows that gestational timing may be prolonged with birth weight of 450 to 1900 g, birth length of 33-42 cm. The final height attained is about 90 to 105 cm. Patients show moderate to severe mental deficiency though early motor progress may be normal. Though our patient showed physical characteristics similar to those of Seckel's syndrome described previously, she had a normal intelligence for this age, despite having microcephaly. This was an unusual finding in this case. Majewski and McKusick *et al.*, reported a few cases with normal intelligence of minimal mental retardation(4,9).

By natural history, we can prognosticate final height or age in this patient. However, autopsy studies in these patients have shown the cerebrum to be small with simple, primitive convolutional pattern resembling that of chimpanzees but all these cases presented with mental deficiency right from the beginning(9). Therefore, the future mental status of this patient can not be predicted accurately.

REFERENCES

1. Mann TP, Russel A. Study of a microcephalic midget of extreme type. *Proc Soc Med* 1959, 52: 1024-1026.
2. Seckel HPG. In: Bird-Headed Dwarfs. Ed. Seckel HPG. Springfield Illinois, Charles C. Thomas 1960, pp 241-243.
3. Jones KL. Seckel's syndrome. In: Smith's Recognizable Patterns of Human Malformation. Philadelphia, Ed. Jones KL. W.B. Saunders Company, 1988, pp 100-101.
4. Majewski F, Goecke T. Studies of microcephalic primordial dwarfism I: Approach to a delineation of the Seckel syndrome. *Am J Med Genet* 1982, 12: 7-21.
5. Shanker V, Keyriwal NL. Seckel's syndrome. *J Indian Med Assoc* 1986, 84: 122-123.
6. Singh SD, Jethmalani N, Mukerjee DP. Bird headed dwarf (Seckel's syndrome). *Indian Pediatr* 1975, 12: 935-937.
7. Bhadran TR, Poullose KP, John KI. Bird-headed dwarf. *J Assoc Phys India* 1978, 26: 1189-1190.
8. Harper RG, Orti F, Baker RK. Bird-headed dwarfs (Seckel's syndrome). A familial pattern of developmental dental, skeletal, genital and central nervous system anomalies. *J Pediatr* 1967, 70: 799-804.
9. McKusick VA, Mahloudji M, Abbot MH, Lindberg R, Kepan D. Seckel's bird headed dwarfism. *New Engl J Med* 1967, 277: 279-285.

Antenatal Diagnosis of Ebstein's Anomaly

K. Balakumar

Major cardiac anomalies can be detected in the antenatal period by targetted imaging of the fetal heart. This unusual case reported here illustrates a fetus with Ebstein's anomaly born out of consanguinous marriage. There were

From the Department of Radiology, P.V.S. Cancer Research Centre, Calicut 2, Kerala 673 002.

*Received for publication August 8, 1990;
Accepted January 10, 1991*

associated skeletal anomalies and the history was suggestive of recurrence among the siblings.

Case Report

A 27-year-old fourth gravida was subjected to ultrasound scanning because of previous history of multiple skeletal and cardiac anomalies of one child noted after premature delivery (no details were available). She had an early abortion as well and has a normal male child. The present pregnancy was of 38 weeks duration.

The placental appearance was normal and there was moderate degree of oligohydramnios. The biometry showed a growth of 36 weeks with asymmetrical growth retardation. The long bones were poorly mineralised having a wavy contour and their length was in the upper normal range. The binocular and the interocular distances were normal. The teeth buds were unusually prominent. The external genitalia was ambiguous.

The targetted cardiac imaging showed a greatly dilated right atrium (*Fig. 1*). The walls of this chamber were thin and it was occupying a major portion of the right hemithorax (*Fig. 2*). The tricuspid valve was seen placed towards the apex. The right ventricle was less prominent. The interventricular septum showed no discontinuity. The pulmonary artery was not atretic. The aortic valve and its lumen were normal. No overriding was apparent. There was no undue enlargement of other chambers. The foramen ovale was normal. The mitral valve was also appearing normal. There were no evidences of hydrops.

This pregnancy ended with the intra-uterine death of the fetus at full term and

the postmortem appearance was consistent with the diagnosis.

Discussion

Ebstein described this condition as congenital downward displacement of the tricuspid valve with distortion of the leaflets. This rare anomaly can be detected in the antenatal period by ultrasound scanning; the true incidence of which is not documented in our country. A recent analysis abroad shows that the incidence of the congenital heart diseases has increased within the last 3 decades. The accepted old incidence rate of 3 to 5 per 1000 live births has gone even above 10 per 1000 live births(1).

The echo features in this case were diagnostic of Ebstein's anomaly as evidenced by the cardiomegaly due to atrialised right ventricle and the presence of apically displaced tricuspid valve with severe degree hypoplasia (the leaflets could not be assessed). In the differential diagnoses, the following conditions are to be excluded. First of all, the idiopathic giant right atrium was excluded as the tricuspid valve was considerably low placed. Fetal cardiac doppler studies may have to be done to assess tricuspid regurgitation as it is often incompetent(2). The septal papillary muscle of the tricuspid is usually prominent in normal cases whereas it was not so in this case.

Associated atrial septal defect is found in 30 to 78 percentage of cases of Ebstein's anomaly(3,4) though it was not evident in this case. Pulmonary atresia may also be noted in these cases(5). In atresia of pulmonary veins the right atrium may be larger, but there is right ventricular enlargement along with a smaller left atrium. The normal right ventricle to left

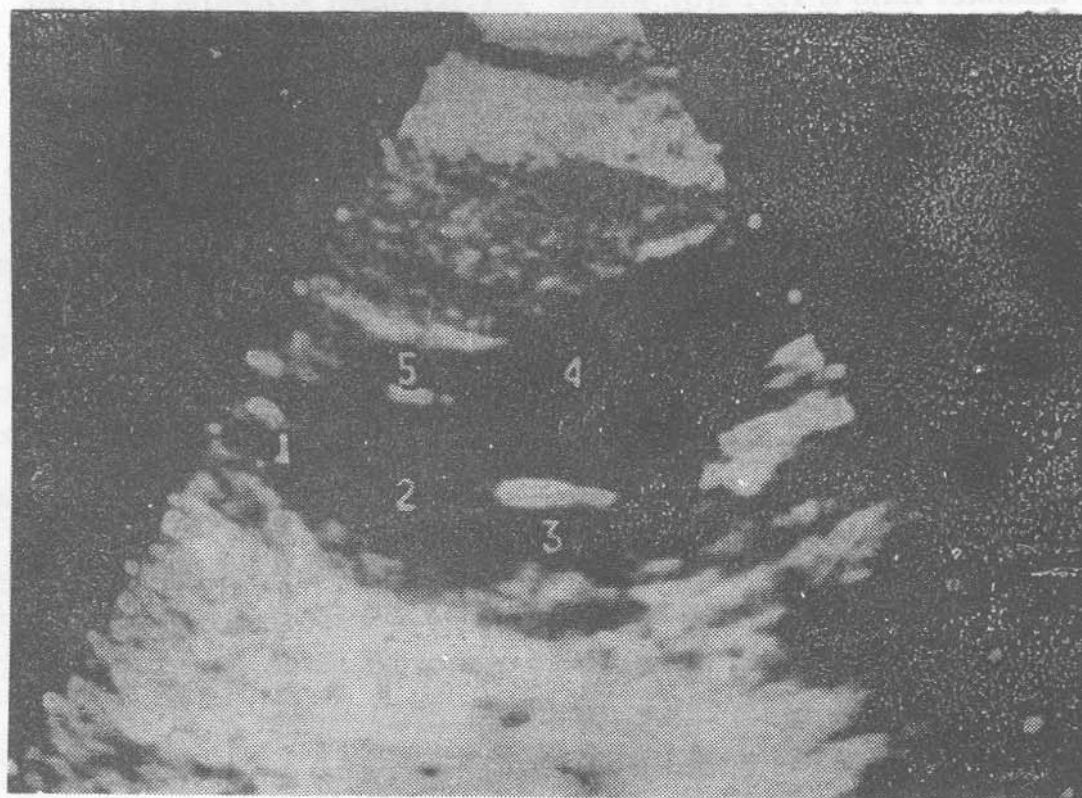


Fig. 1. The four chamber view of the fetal heart—(1) apex; (2) left ventricle; (3) left atrium; (4) right atrium; and right ventricle.

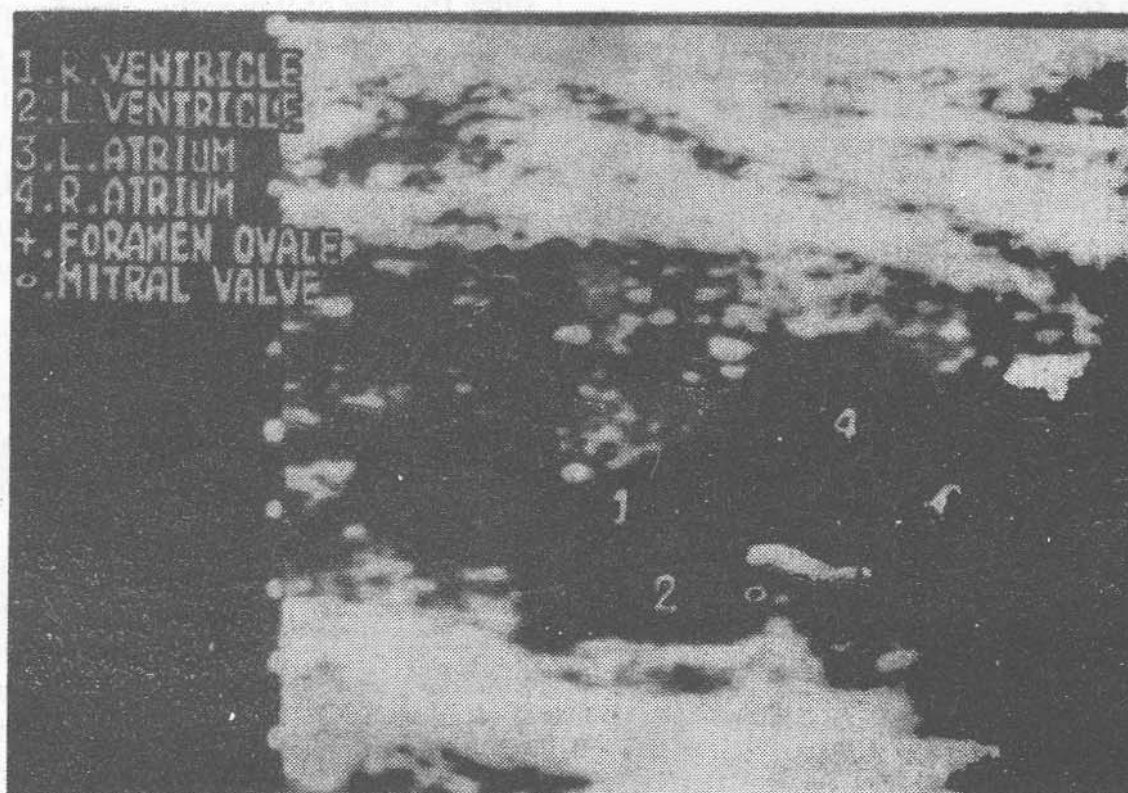


Fig. 2. The four chamber view at a different angle showing the grossly dilated 'R' atrium.

ventricle diameter ratio is 1.8 : 1(6), where as the right ventricular size was smaller here. Other associated major cardiac anomalies can also occur(7).

The parents were warned about the possible risk of recurrence; the reported incidence of which among the siblings and offsprings is 1 and 5, respectively(8).

REFERENCES

1. Hoffman JIE. Congenital heart disease: Incidence and inheritance. *Pediatr Clin North Am* 1990, 37: 25-43.
2. Yoshida K, Yosikawa J, Shakudo M, *et al.* Color Doppler evaluation of valvular regurgitation in normal subjects. *Circulation* 1988, 78: 840-844.
3. Anderson K, Zuberbuhler J, Anderson R, *et al.* Morphologic spectrum of Ebstein's anomaly of the heart: A review. *Mayo Clin Proc* 1979, 54: 174-180.
4. Danielson G, Fuster V. Surgical repair of Ebstein's anomaly. *Ann Surg* 1982, 196: 499-503.
5. Sharf M, Abinader EG, Shapiro I, *et al.* Prenatal echocardiographic diagnosis of Ebstein's anomaly with pulmonary atresia. *Am J Obstet Gynecol* 1983, 147: 300-307.
6. Sahn DJ, Lange LW, Allen WD, *et al.* Quantitative real time cross sectional echocardiography in the developing normal human fetus and newborn. *Circulation* 1980, 62: 588-597.
7. Romero R, Pihu G, Jeanty P, Ghidini A, Hobbins JC. The heart. In: *Prenatal Diagnosis of Congenital Anomalies*. Ed. Romero R. California, Appleton and Lange, 1988, pp 149-151.
8. Burn J. The etiology of congenital heart disease. In: *Pediatric Cardiology*. Eds Anderson RH, Macartney FJ, Shinebourne EA, *et al.* London, Churchill Livingstone, 1987, pp 15-63.

Glycogen Storage Disease (Type-III)

A.K. Sarkar
T. Ghosh
T. Choudhury
G. Saha
R. Danda

The glycogen storage diseases (GSD) or glycogenoses are the result of metabolic errors that lead to abnormal concentrations or structure of glycogen. Several well defined disorders of glycogen metabolism have been described. We wish to report our experience with one such case of Type III GSD.

Case Report

A 2-year-old male child was brought with progressive distension of abdomen since the age of 9 months, and a convulsion one month prior to hospitalization. Clinical examination revealed an alert child with hugely distended abdomen. The liver was enlarged 10 cm below the right costal margin, firm in consistency with rounded margin and smooth surface. Spleen and kidneys were not palpable. Rest of the systemic examination was normal. Family history revealed that his father had suffered in childhood from hepatomegaly which ultimately receded. Developmental history was normal.

Investigations showed a normal hemeogram, including platelet count and

From the Department of Pediatrics, Institute of Post Graduate Medical Education and Research and S.S.K.M. Hospital, Calcutta.

Received for publication October 3, 1989;

Accepted February 12, 1991