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, Yoskikawa 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, Pilu 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)


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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


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normal ADP aggregation test. Liver function tests revealed total protein of 6.5 g/dl, with albumin 3.5 g/dl, alkaline phosphatease 200 IU/L, SGOT 250 IU/L, SGPT 150 IU/L and prothrombin time of 12.5 sec. Random blood sugar was 75 mg/dl, serum triglycerides 230 mg/dl and lactic acid 12.5 mg/dl. Serum concentrations of uric acid and cholesterol were normal.

Glucagon challenge test after an overnight fast resulted in sugar values: fasting –55 mg/l; 30 min–60 mg/dl; 60 min–65 mg/dl; 90 min-65 mg/dl; and 120 min–68 mg/dl. After meals, blood glucose was 75 mg/dl and 2 h after glucagon administration it rose to 115 mg/dl.

ECG. and *X*-ray chest revealed no abnormality. Ultrasonography of the abdomen showed grossly enlarged liver with no detectable focal parenchymal lesion and normal spleen and kidneys. Liver biposy specimen stained with PAS showed excessive accumulation of glycogen within hepatocytes both intracytoplasmic as well as intranuclear (*Fig. 1*). There was fibrosis around portal area but liver architecture was not lost (*Fig. 2*). The findings were consistent with Type I or Type III GSD.

Glucose-6-phosphatase estimated from liver tissue was 3.5 moles phosphate/g/min (normal 4.7 ± 1.9). a diagnosis of GSD Type III was, therefore, made.

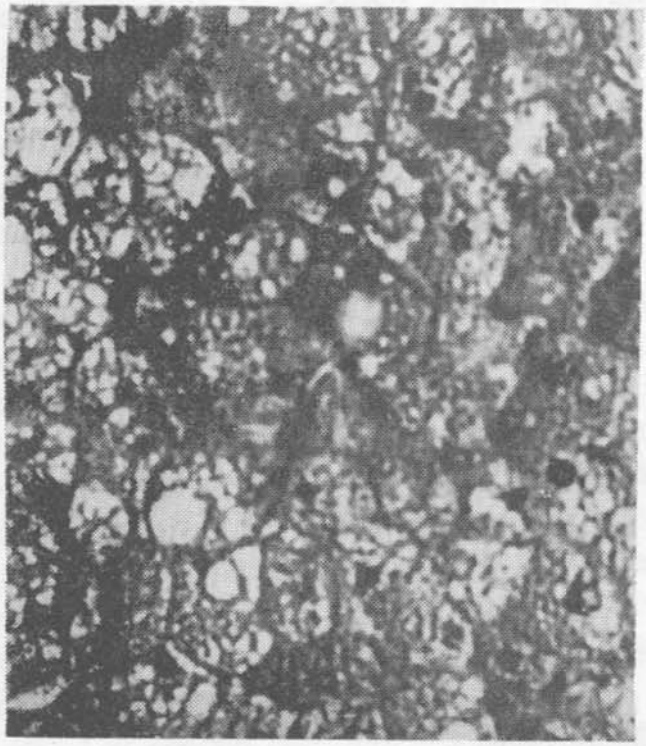

*Fig. 1. P.A.S. stain of liver biopsy specimen.*

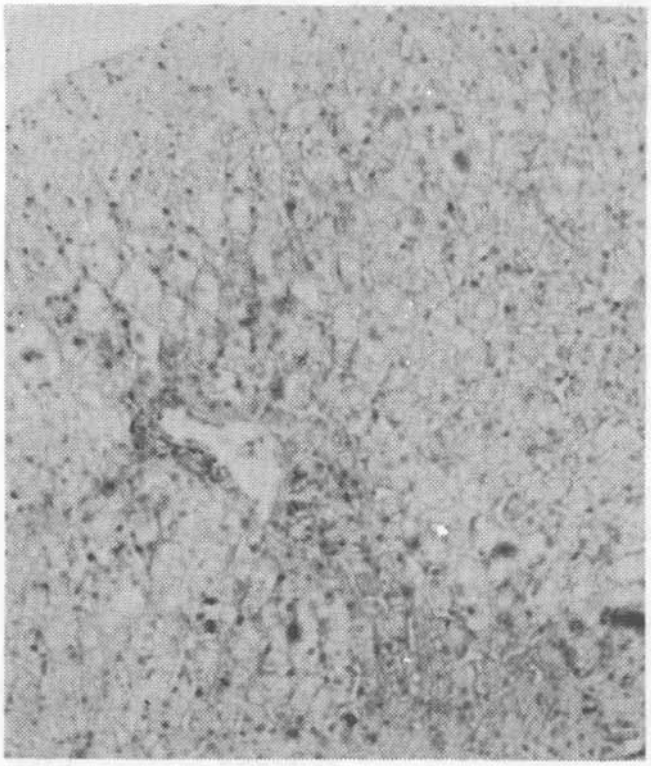

*Fig. 2. H/E stain of liver biopsy specimen.*

## Discussion

Gradual abdominal distension from infancy, history of convulsion and huge hepatomegaly pointed to a metabolic disorder. Hepatomegaly without significant splenomegaly may occur in GSD Type I, III, VI, VIII, IX and XI and conditions like fatty infiltration, parasitic infestation or congestive causes, all of which were easily

6. Fernandes J, Huizing F, VandeKamer JH. A screening method for liver glycogen diseases. Arch Dis Child 1969, 44: 311-317.

7. George H. Inborn error of metabolism. *In:* Nelson Text Book of Pediatrics, 13th edn. Eds Behrman RE, Vaughan VC, Nelson WE. Philadelphia, WB Saunders Co, 1987, pp 313-322.

8. Hutchison JH. Glycogen storage disease. *In:* Practical Pediatric Problems. London, Lloyd-Luke Ltd, 1975, pp 142-146.

9. Borowitz SM, Greene HL. Cornstarch therapy in a patient with type III glycogen storage disease. J Pediatr Gastroenterol Nutr 1987, 6: 481-486.

## Porencephaly: A Possible Complication of Chorion Villus Sampling ?

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Chorion villus sampling (CVS) has been a major advance in the field of prenatal diagnosis. Many studies have shown that the technique is safe with minimal immediate risks(1-3) but the long term effects on the fetus are not well documented. It is, therefore, necessary to take cognisance of all adverse outcomes to establish the long term safety of this procedure.

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### Case Report

The patient a 38-year-old G3 P2 Muslim lady with a non-consanguinous marriage underwent CVS at 9 weeks gestation because of her age. The procedure was carried out transcervically at another centre and therefore the details of number of insertions and weight of tissue obtained were not available. She had spotting for two days following the procedure but the pregnancy remained uneventful. Cytogenetic study of the aspirated tissue revealed a normal karyotype.

The patient had not taken any drugs during pregnancy other than vitamins and hematenics, her previous pregnancies and deliveries were normal and there was no family history of congenital malformations. At 28 weeks of gestation a GTT was performed which indicated gestational diabetes (F-110 mg/dl, 1 h- 146 mg/dl, 2 h-180 mg/dl). She was controlled on diet and crystalline insulin 5 I.U. twice a day.

An ultrasound performed at 37 weeks revealed an intracranial, unilaternal cystic area occupying most of one hemisphere. There was a marked mid line shift (*Fig.*). Biparietal diameter was 11 cm and head circumference was 38 cm. A provisional diagnosis of porencephaly was made. The patient had an elective ceserean section. The neonate weighted 3.7 kg, had an Apgar score of 8 and had no other apparent congenital malformation. The head circumference was 38.5 cm. CT scan confirmed the diagnosis of a porencephalic cyst.

### Discussion

Porencephaly is an extremely rare