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Case Report

Glycogen Storage Disorder Type IV with Chorioretinitis

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ABSTRACT

We report a girl child of two years old who presented with progressive distension of the abdomen and failure to thrive. Clinical features and laboratory results and histopathological analysis led us to the diagnosis of type IV GSD. The child was treated with small frequent carbohydrate feedings throughout the day to maintain normal blood glucose levels.

Key Words: Child, Glycogen storage disorder, Chorioretinitis.

INTRODUCTION

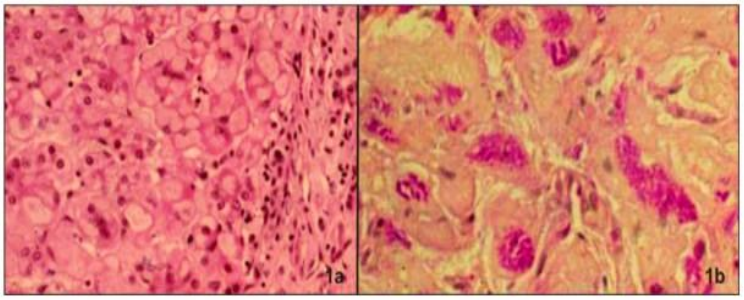
Glycogen, the storage form of glucose in animal cells is composed of glucose residues joined in straight chains by alfa 1-4 linkages and branched at intervals of 4-10 residues at alfa 1-6 linkages. Defects in glycogen metabolism causes accumulation of glycogen in tissues.[1] Glycogen is converted into the simple sugar glucose for the body's use as energy. The glycogen storage diseases (GSD) or glycogenoses are a group of rare inherited genetic heterogenous disorders of inborn errors of carbohydrate metabolism that lead to abnormal concentrations or structure of glycogen. They have a buildup of abnormal amounts or types of glycogen in their tissues. Several well defined disorders of glycogen metabolism have been described based on the identified enzymatic defects or sometimes the distinctive features. We present here a case of two year old female child with this rare form of disorder.

CASE REPORT

A two -year-old girl child born out of non-consanguineous parents was brought to our tertiary care centre. The baby was delivered vaginally with a birth weight of 2850 gm, post natal period was uneventful. The child was irritable with delayed milestones. Her height was 72 cms and weight was 6.7 kg. The child had presented with complaints of failure to thrive and progressive distension of abdomen since 1 year of age. Two months back the child had received two units of blood transfusion for hepatosplenomegaly. There was no history of jaundice, convulsion or similar disorder in the family. Clinical examination revealed that here was no evidence of any oedema feet. The liver was enlarged 5 cm below the right costal margin, with a liver span of 10.5

cm firm in consistency with well defined margin and smooth surface. Spleen and kidneys were not palpable. The rest of the systemic examination was non-contributory. Investigations showed a abnormal hemogram, Haemoglobin of 9.9 gm/dl, Total count of 8900/cumm. Liver function tests revealed Total Bilirubin 1.32 mg/dl, Direct Bilirubin of 0.44 mg/dl, Total Protein of 6.0 g/dl with Albumin 3.99 g/dl, Alkaline phosphates 64 IU/L, Aspartate Tranaminase (SGOT) 390 IU/L (normal upto 46 IU/L), Alanine Tranaminase (SGPT) 146 IU/L (normal upto 46 IU/L) and Prothrombin time 13.5 sec against the control time of 12.5 sec INR was 1.04 against 1.00 of the control. Fasting blood sugar was 54 mg/dl, where as Post meal it was 91mg/dl. Serum Triglycerides 205 mg/dl, Cholesterol 139 mg/dl, Serum CPK (Total) 66 IU/L , Serum CPK(Cardiac) 30 IU/L and Uric acid 4.6 mg/dl. Serum electrolytes were Serum Sodium 134 meq/L, Serum Potassium 4.6 meq/L where as Serum Chloride 105 meq/L. Fundus examination revealed old healed chorioretinitis.ECG and X-ray chest revealed no abnormality. Ultrasonography of the abdomen showed grossly enlarged liver with spleen with coarse echotexture. And minimal free fluid in the abdomen. Kidneys were of normal size and shape. Histopathological examination of liver tissue showed early changes of liver cirrhosis. Architecture was partially effaced. Hepatocytes were enlarged and showed eosinophillic hyaline material. Perivascular fibrosis was also seen. Special stain PAS (Periodic Acid Schiffs) was done which showed positivity while diastase was negative. Histological findings were consistent with storage disease. (images 1a and 1b) In view of these features a diagnosis of GSD was made. The child was subsequently discharged with nutritional supplements and was kept on regular follow up for a watch on progressive liver function tests as well as any evidence of Portal Hypertension. It needs to be emphasized that most cases of type IV GSD are characterized by huge hepatomegaly with splenomegaly and with cirrhosis[2-4] but the disorder should be borne in mind especially where there is family history of a similar disorder or where parental consanguinity exists.[5]

IMAGES:



1a) Liver section stained with hematoxylin and eosin. (40 x) Liver architecture is distorted with diffuse interstitial fibrosis that is micronodular cirrhosis. Hepatocytes are enlarged with deposition of eoninophlic granular material.

1b) Liver section stained with periodic acid-Schiff (PAS) after diastase treatment. (40x) Coarsely clumped PAS positive cytoplasmic material which represents abnormal glycogen which is resistant to diastase treatment.

DISCUSSION

In children one of the possibilities of GSD has to be kept in mind in a child who is presenting with unexplained hepatosplenomegaly. Glycogen is a branched polymer of glucose which serves

as a reservoir of glucose units. GSD can be genetic or acquired and are characterised by abnormal inherited glycogen metabolism in liver, muscle and brain. Genetic GSD are caused by inborn errors of metabolism and involve genetically defective enzymes. The acquired is due to intoxication with alkaloids.[6] There are at least 10 different types of GSD . The types are put into groups based on the enzyme that is missing. The most common types are I (one) III (three) and IV (four). About one in 20,000 can have GSD. People with glycogen storage diseases have a buildup of abnormal amounts or types of glycogen in their tissues. glycogen-storage disease type IV is named after investigator (DH Andersen) it result from defects in the gene that encodes for the glycogen-branching enzyme (*GBE1*) located on chromosome band 3p12. The function of this enzyme is to increase the number of branch points during glycogen synthesis. The branched nature of the glycogen molecule is important for its compact nature and solubility within the cell. The absence of this branching enzyme activity results in abnormal glycogen with long, unbranched outer chains that resemble amylopectin, which is a glucose polymer that is a major storage polysaccharide in legumes. Definitive biochemical diagnosis of glycogen-storage disease type IV relies on demonstration of deficient glycogen-branching enzyme activity in the liver or in the muscle tissue. The clinical entity presents itself with muscle cramps, hypoglycaemia, hepatomegaly, abdominal distension. The age of presentation and severity depends upon the type of GSD.Our case had features of involvement of liver and spleen, with failure to thrive. The other features of this entity include involvement of heart and muscles and development of liver cirrhosis by the age of 3 to 5 years. Clinically the child may have hypoglycaemia, hyperurecemia, hyperlipidemia lactic acidosis though initially the liver function tests may be normal. There is involvement of reticuloendothelial system which may have impaired platelets leading to abnormal haematological parameters.

The goal of treatment is to maintain normal blood glucose levels. This may be done with: A nasogastric infusion of glucose in infants and children under age two. In children over age two, frequent small carbohydrate feedings are given throughout the day. This may include uncooked cornstarch. Uncooked cornstarch provides a steady slow-release form of glucose. Allopurinol may be prescribed to reduce uric acid levels in the blood. This is done to prevent gout and kidney stones. Liver transplant is only for the patients who are with progressive hepatic failure. Caution must be taken in selecting type IV GSD patients for liver transplant because these patients may have variable phenotype, which includes a non progressive form of the liver disease.

CONCLUSION

Finally to conclude one should always keep the possibility of GSD, though its rare, in case of a child who is presenting with failure to thrive and unexplained hepatosplenomegaly and abnormal liver function tests with haematological abnormality. There is no way to prevent glycogen storage diseases. However, early treatment can help control the disease once a person has it.

REFERENCES

1. Hug G. Inborn Errors of Metabolism: Defects in Metabolism of Carbohydrates. *In:* Nelson Text Book of Pediatrics, 13th edn. Eds. Behrman RE, Vaughan VC, Nelson WE,Phi'adelphia, W.B. Saunders Co, 1987, pp 277-357.

2. Singh M, Fazal MI, Tana I, Arya LS, Goel 236 RG. Glycogen storage disease. Indian Pediatr 1983, 20: 208-213.
3. Sarkar AK, Ghosh T, Chowdhury T, Saha G, Sanda R. Glycogen storage disease (Type III). Indian Pediatr 1991, 28: 1058-1061.
4. Kalta V, Arya LS, Nayak NC. Glycogen storage disease (Type IV): A familial cirrhosis diagnosed by electron microscopy. Indian Pediatr 1980, 17: 625-627.
5. Singh M, Fazal MI, Tana I, Arya LS, Goel 236 RG. Glycogen storage disease. Indian Pediatr 1983, 20: 208-213.
6. Mingyi Chen, Glycogen Storage Diseases, Molecular Pathology of Liver Diseases Molecular Pathology Library. Volume 5, 2011, pp 677-681.

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