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Case Report

Glycogen storage disease type VI presenting with growth failure and hepatomegaly in a young child: a case report

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ABSTRACT

Glycogen storage disease (GSD) comprises a group of inherited disorders of carbohydrate metabolism caused by defects in glycogen synthesis or breakdown. We report a 2-year-2-month-old girl who presented with failure to gain height and weight for one year and progressive abdominal distension. She was born to second-degree consanguineous parents and had normal development. Examination showed pallor, short stature, low weight, frontal bossing, a flattened nasal bridge and marked hepatomegaly, with the liver palpable 8 cm below the right costal margin and a liver span of 13 cm. Her height was 76 cm and weight 8 kg, both approximately −3 to −4 SD. Investigations showed macrocytosis with a few microcytes, low haemoglobin, elevated transaminases, low serum iron and vitamin B12, and vitamin D insufficiency. Serum lactate and ammonia were elevated, while fasting glucose was 92 mg/dL. Abdominal ultrasonography demonstrated hepatomegaly with normal liver echotexture and normal portal Doppler. Whole exome sequencing established the diagnosis of glycogen storage disease type VI (Hers disease). The child was additionally diagnosed with dimorphic anaemia and vitamin D insufficiency. This case highlights the importance of considering hepatic glycogen storage disorders in children presenting with growth failure and hepatomegaly, particularly in the setting of consanguinity, and demonstrates the role of genetic testing in establishing the diagnosis.

Keywords: Glycogen storage disease, GSD VI, Hers disease, Hepatomegaly, Growth failure, Whole exome sequencing

INTRODUCTION

Glycogen storage diseases (GSDs) are inherited inborn errors of carbohydrate metabolism resulting from deficiency of specific enzymes involved in glycogen synthesis or breakdown. Depending on the enzyme affected, glycogen accumulates predominantly in the liver, skeletal muscle, or both. Hepatic forms may present during childhood with hepatomegaly, growth failure and abnormalities of glucose and liver metabolism. GSD type VI, also known as Hers disease, is a hepatic glycogen storage disorder. In children with growth failure and hepatomegaly, storage and metabolic disorders form an important part of the differential diagnosis. This case describes a young child with marked growth failure and hepatomegaly in whom whole exome sequencing established GSD type VI.[1,2]

CASE REPORT

A 2-year-2-month-old girl from Vikarabad was brought with complaints of not gaining height and weight for one year and progressive abdominal distension for one year. The abdominal distension was insidious in onset, gradually progressive and more prominent in the upper abdomen. The child was born to second-degree consanguineous parents and was fourth in birth order. She was a term baby delivered by lower-segment

caesarean section for previous caesarean section, with a birth weight of 3 kg, and cried immediately after birth. There was no neonatal intensive care admission. Developmental milestones were appropriate for age, with a developmental quotient of 100%.

Nutritional assessment showed a predominantly low-calorie and low-protein diet. Her estimated requirement was 1100 kcal/day and 18 g protein/day, while intake was approximately 600 kcal/day and 13 g protein/day. On examination, she was conscious and comfortable, with pallor. She had frontal bossing and a flattened nasal bridge. The height was 76 cm (−3 to −4 SD) and weight was 8 kg (−3 to −4 SD), with a weight-for-height Z-score of −1 to −2 SD. Her chronological age was 2 years 2 months, height age 1 year 2 months and weight age 8 months. The upper segment-to-lower segment ratio was 1.5:1 and arm span was 74 cm.



Figure 1: Clinical photograph showing child with abdominal distension.

The abdomen was distended with downward displacement of the umbilicus. A well-defined light-brown hyperpigmented patch measuring approximately 1.5 × 1 cm was present on the right side of the abdomen below the umbilicus, with multiple flat bluish-grey patches over the back. The liver was firm, smooth and non-tender and palpable 8 cm below the right costal margin; the liver span was 13 cm. The spleen was not palpable. Cardiovascular, respiratory and neurological examinations were otherwise unremarkable.

Haemoglobin was 8.3 g/dl with RBC count 2.8 million/cmm, MCV 109 fl and RDW 25%. Peripheral smear showed macrocytes with a few microcytes, consistent with a dimorphic pattern. Serum iron was 29.7 µg/dl, TIBC 486 µg/dl and transferrin saturation 6%; vitamin B12 was 148 pg/ml and vitamin D3 was 13.49 ng/ml. Liver function testing showed elevated AST of 274.1 U/l and ALT of 184.3 U/l with normal bilirubin, total protein and albumin. Serum lactate was 29.2 mg/dl and ammonia 147 µg/dl. Early morning fasting glucose was 92 mg/dl. Renal function, thyroid profile and urine metabolic profile were unremarkable.



Figure 2: Clinical photograph showing child with abdominal distension.

Ultrasonography of the abdomen showed hepatomegaly with a liver span of 11.5 cm and normal echotexture; portal Doppler was normal. Bone age was 2 years and the infantogram was normal. Ophthalmological examination showed a normal fundus. Two-dimensional echocardiography showed a small patent foramen ovale with left-to-right shunt. In view of growth failure with hepatomegaly, storage disorders including glycogen storage disorders, Gaucher disease, Niemann-Pick disease and tyrosinaemia type 1 were considered. Whole exome sequencing was performed and established the diagnosis of glycogen storage disease type VI (Hers disease), with associated dimorphic anaemia and vitamin D insufficiency.

DISCUSSION

The combination of growth failure and hepatomegaly in a young child raises consideration of hepatic storage and metabolic disorders. In the present case, the child had significant short stature and underweight, progressive abdominal distension and marked hepatomegaly. The normal bilirubin with elevated transaminases, together with the clinical presentation, supported evaluation for a metabolic or storage disorder. The absence of developmental delay and the normal neurological examination further characterized the clinical presentation.[1-3]

GSDs result from defects in enzymes involved in glycogen metabolism and may predominantly affect the liver or skeletal muscle. Hepatic forms include types 0, I, III, IV, VI and IX, among others. The presentation of failure to thrive with hepatomegaly can occur in several GSDs and overlaps with other metabolic disorders,

including Gaucher disease, Niemann-Pick disease and tyrosinaemia. In this child, whole exome sequencing provided the definitive diagnosis of GSD type VI (Hers disease).[1,2,4]

The child also had anaemia with macrocytosis and a few microcytes on peripheral smear. Low serum iron, low transferrin saturation and low vitamin B12 supported associated nutritional deficiencies, while vitamin D3 was in the insufficiency range. These findings emphasize the importance of nutritional assessment in children presenting with chronic growth failure. Dietary management is the mainstay described for GSD type VI, with small frequent meals containing complex carbohydrates such as uncooked or modified cornstarch and adequate dietary protein used to reduce the risk of hypoglycaemia.

CONCLUSION

Glycogen storage disease type VI should be considered in children presenting with growth failure and hepatomegaly. Careful clinical assessment, nutritional evaluation and appropriate metabolic investigation, followed by genetic testing when indicated, can establish the diagnosis and guide dietary management.

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REFERENCES

1. Brown LM, Corrado MM, van der Ende RM, Derks TGJ, Chen MA, Siegel S, et al. Evaluation of glycogen storage disease as a cause of ketotic hypoglycemia in children. J Inherit Metab Dis. 2015;38(3):489-93.
2. Kishnani PS, Goldstein J, Austin SL, Arn P, Bachrach B, Bali DS, et al. Diagnosis and management of glycogen storage diseases type VI and IX: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). Genet Med. 2019;21(4):772-89.
3. Massese M, Tagliaferri F, Dionisi-Vici C, Maiorana A. Glycogen storage diseases with liver involvement: a literature review of GSD type 0, IV, VI, IX and XI. Orphanet J Rare Dis. 2022;17(1):241.
4. Lu SQ, Feng JY, Liu J, Xie XB, Lu Y, Abuduxikuer K. Glycogen storage disease type VI can progress to cirrhosis: ten Chinese patients with GSD VI and a literature review. J Pediatr Endocrinol Metab. 2020;33(10):1321-33.
5. Kasapkara ÇS, Aycan Z, Açoğlu E, Senel S, Oguz MM, Ceylaner S. The variable clinical phenotype of three patients with hepatic glycogen synthase deficiency. J Pediatr Endocrinol Metab. 2017;30(4):459-62.

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