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Original Case Report

A CASE OF LATE-ONSET DIAGNOSIS: GLYCOGEN STORAGE DISEASE TYPE IA (VON GIERKE'S DISEASE) IN A 28-YEAR-OLD FEMALE-CASE REPORT

Article History:

Name of Author:
V Suryanarayanan[1](Corresponding author) , S Yogeshwari[2], M Kavya[3], Dr. M.K. Sundar Sri[4], Dr. Karthickeyan krishnan[5] , Dr. Palani Shanmugasundaram[6]

Affiliation: [1]*Pharm.D Intern, School of Pharmaceutical Sciences, Vels Institute of Science, Technology and Advanced Studies, Pallavaram, Chennai – 600117, India*
[2]*Pharm.D Intern, School of Pharmaceutical Sciences, Vels Institute of Science, Technology and Advanced Studies, Pallavaram, Chennai – 600117, India*
[3]*Pharm.D Intern, School of Pharmaceutical Sciences, Vels Institute of Science, Technology and Advanced Studies, Pallavaram, Chennai – 600117, India*
[4]*Assistant Professor, Department of Pharmacy Practice, School of Pharmaceutical Sciences, Vels Institute of Science, Technology and Advanced Studies, Pallavaram, Chennai – 600117, India*
[5]*Professor and Head, Department of Pharmacy Practice, School of Pharmaceutical Sciences, Vels Institute of Science, Technology and Advanced Studies, Pallavaram, Chennai – 600117, India*
[6]*Dean, School of Pharmaceutical Sciences, Vels Institute of Science, Technology and Advanced Studies, Pallavaram, Chennai, TN-600117*

Corresponding Author:
V Suryanarayanan

Abstract: ***Introduction:*** Glycogen Storage Disease Type Ia (GSD Ia) is an autosomal recessive disorder with a rare incidence and an enzymatic deficiency of glucose-6-phosphatase, which results in gluconeogenesis and glycogenolysis impairment. The condition tends to present in infancy with acute fasting hypoglycemia and lactic acidosis. Diagnosis in adults is infrequent and often delayed because of non-specific clinical manifestations. ***Case Presentation:*** A case of a 28-year-old woman who came with altered sensorium and a history of chronic hypoglycemia. The clinical examination was notable for short stature, doll like face, and enormous hepatomegaly. Biochemical studies revealed severe hypoglycemia (32 mg/dL), lactic acidosis, hypertriglyceridemia, and hyperuricemia. There was a high suspicion of an inborn error of metabolism. Genetic testing revealed a homozygous pathogenic mutation in the G6PC gene, confirming GSD Ia. ***Management and Outcome:*** The histopathology report revealed hepatocytes with ballooned, pale cytoplasm consistent with massive glycogen storage, in line with a glycogen storage disease. The final diagnosis was confirmed by the hepatic tissue enzyme assay, where there was a near-total deficiency of glucose-6-phosphatase enzyme activity at <0.1 µmol/min/g of liver tissue (usual reference range: 1.5 - 3.5 µmol/min/g). This result is diagnostic of Glycogen Storage Disease Type Ia. After stabilizing, the patient was switched to an ordered dietary regimen. This consisted of regular, small feedings and bedtime administration of uncooked cornstarch. She was also started with Allopurinol for hyperuricemia and discharged stable after intensive dietary instruction. ***Conclusion:*** This case shows that GSD Ia may remain undiagnosed until adulthood. The detection by adult practitioners is important to *prevent metabolic crisis and allow proper long term management.*

Keywords: Glycogen Storage Disease Type Ia; Von Gierke's Disease; Glucose-6-phosphatase deficiency; Inborn errors of metabolism; Hypoglycemia; Hepatomegaly; Lactic acidosis; Hyperuricemia; Hypertriglyceridemia; Case report

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INTRODUCTION

Glycogen Storage Disease Type Ia (GSD Ia, Von Gierke's disease) is a rare autosomal recessive inborn error of metabolism with an estimated incidence of 1 in 100,000 live births [1]. It results from a deficiency in the glucose-6-phosphatase enzyme (G6Pase), which is critical for the final step of glycogenolysis and gluconeogenesis. This defect leads to the hallmark metabolic disturbances of fasting hypoglycemia, lactic acidosis, hyperuricemia, and hyperlipidemia, accompanied by hepatomegaly and growth retardation [2].

Subsequent studies found hypertriglyceridemia, hyperuricemia, and suggestive chronic adaptation history of frequent feeding, which indicated a glycogen storage disorder. Genetic analysis finally diagnosed Glycogen Storage Disease Type Ia (GSD Ia), an uncommon autosomal recessive disease due to G6PC gene mutations leading to glucose-6-phosphatase deficiency.

The classic presentation occurs in infancy, but a subset of patients with milder phenotypes may evade diagnosis in childhood. These individuals often unconsciously adapt by frequent feeding, masking the severity of their metabolic defect [3]. We report a case of a 28-year-old female diagnosed with GSD Ia, highlighting the diagnostic challenges and management strategies for this rare adult presentation.

RESULTS

A 28 year old female presented to the emergency department with acute confusion and drowsiness. Her relatives reported a two-day history of reduced oral intake due to poor appetite, followed by worsening mental status. She had a history of frequent “fainting episodes” since childhood, particularly when meals were delayed, as well as abdominal fullness and poor growth.

Physical Examination:
Height: 145 cm (<3rd percentile), Weight: 48 kg
Facial characteristic : doll like facies, full cheeks

Table 1: Vital Signs on Admission

PARAMETER	VALUE	REFERENCE RANGE
Temperature	36.8°C	36.5–37.5°C
Pulse Rate	112 bpm	60–100 bpm
Blood Pressure	105/70 mmHg	90/60 – 120/80 mmHg
Respiratory Rate	28 breaths/min	12–20 breaths/min
Oxygen Saturation	Not documented*	≥ 95% on room air

Abdominal exam: Marked hepatomegaly (liver palpable 10 cm below the right costal margin), no splenomegaly
No signs of chronic liver disease or ascites

Table 2: Initial Laboratory Findings:

PARAMETER	RESULT	REFERENCE RANGE	REMARKS
Blood Glucose	32 mg/dL	70–100 mg/dL (fasting)	Severe hypoglycemia
pH	7.18	7.35–7.45	Acidemia
HCO_3^-	10 mmol/L	22–26 mmol/L	Low bicarbonate
Lactate	12 mmol/L	0.5–2.2 mmol/L	Markedly elevated
Liver Enzymes			
ALT	68 U/L	7–56 U/L	Mild elevation
AST	75 U/L	10–40 U/L	
Uric Acid	9.8 mg/dL	2.4–6.0 mg/dL (female)	Hyperuricemia
Lipid Profile			
Triglycerides	880 mg/dL	<150 mg/dL	Severe hypertriglyceridemia
- Total Cholesterol	280 mg/dL	<200 mg/dL	Hypercholesterolemia
Complete Blood Count	Mild normocytic anemia	Hemoglobin 12–16 g/dL (female)	Mild anemia
Renal Function	Within normal limits	BUN: 7–20 mg/dL; Creatinine: 0.6–1.2 mg/dL	Normal renal profile

MANAGEMENT AND OUTCOME

The patient was treated in the ICU with a maintenance 10% dextrose drip. She demonstrated quick recovery of consciousness and normalization of lactate and blood glucose concentrations.

According to the time-honored characteristics—fasting hypoglycemia, lactic acidosis, hepatomegaly, hypertriglyceridemia, and hyperuricemia—an inborn metabolic error, namely GSD Type I, was suspected. Ultrasound of the abdomen revealed hepatomegaly with normal renal morphology.

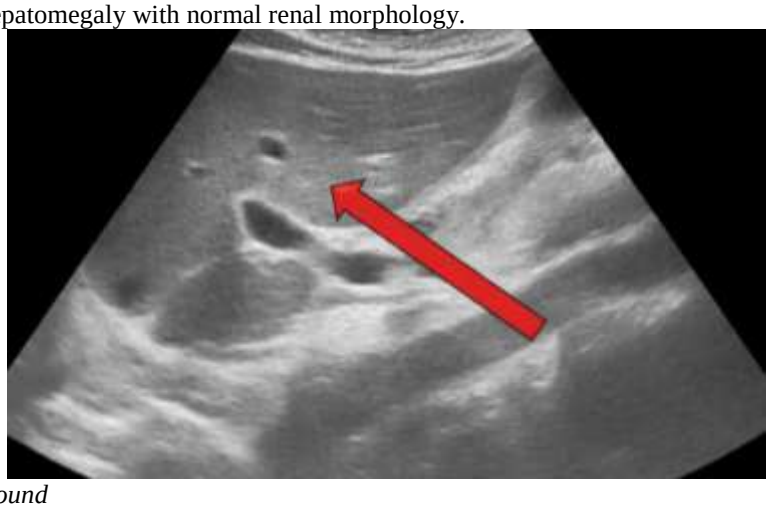

Fig1 : Abdominal ultrasound

An ultrasound-guided percutaneous liver biopsy was done. The gross examination of the specimen showed an enlarged pale greasy parenchyma of the liver. Histopathological examination established the diagnosis:

- Light Microscopy (H&E): Hepatocytes were significantly distended with pale, vacuolated cytoplasm secondary to extensive glycogen and fat accumulation, resulting in a diagnostic "plant cell" or "mosaic" appearance.
- Special Stains: Periodic Acid-Schiff (PAS) stain demonstrated strong cytoplasmic positivity, which was diastase-labile (PAS-D negative), thereby establishing the presence of glycogen. Fat stains (Oil Red O) demonstrated conspicuous mixed microvesicular and macrovesicular steatosis.

The final confirmation was received from the hepatic tissue enzyme assay, which quantitated the activity of glucose-6-phosphate conversion to inorganic phosphate and glucose. The assay revealed a very low level of glucose-6-phosphatase activity, i.e., <0.1 µmol/min/g of liver tissue (normal range: 1.5 - 3.5 µmol/min/g). This is diagnostic for Glycogen Storage Disease Type Ia.

Long-Term Management Plan:

- Frequent complex carbohydrate-rich meals every 2–3 hours
- Uncooked cornstarch bedtime dose (about 2 tablespoons in water)
- Fasting, galactose, and fructose avoidance
- Allopurinol 100 mg daily for hyperuricemia
- Planned follow-ups to watch for liver adenomas, kidney function, and bone health
- The family and patient were taught extensively regarding the condition and dietary compliance. She was discharged stable with referrals to a metabolic specialist and dietician for further follow-up.

DISCUSSION

As a pharmacy intern, this case reinforced the critical role of pharmacotherapeutic knowledge and interprofessional collaboration in managing rare metabolic diseases. The patient's long-standing symptoms—commonly dismissed or misattributed—were textbook for GSD Ia in hindsight.

Adult-onset diagnosis of GSD Ia, while rare, is increasingly recognized. Some patients adapt their lifestyle unknowingly (e.g., frequent snacking), which masks severe hypoglycemia until a stressor, like illness or fasting, unravels the underlying defect.

The biochemical features—especially the simultaneous presence of hypoglycemia, lactic acidosis, and hyperlipidemia—should immediately raise suspicion for a disorder in hepatic glucose metabolism. Pharmacologic support (like allopurinol for hyperuricemia) and dietary therapy remain cornerstones of management. Strict metabolic control reduces long-term risks like hepatic adenomas and renal dysfunction.

CONCLUSION

This case serves as a reminder that inborn errors of metabolism are not exclusive to pediatrics. Adult healthcare providers, including pharmacists, play a vital role in recognizing rare disorders like GSD Ia. Early diagnosis and intervention can significantly improve prognosis and quality of life. This experience highlighted the importance of clinical awareness, patient education, and interdisciplinary care in managing rare genetic disorders.

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