

Lipemic Serum and Severe Hypertriglyceridemia as Presenting Features of Glycogen Storage Disease Type Ia in an Infant: A Case Report

Dr. Chandni Krishnani,¹  Dr. Firoz Sheikh,^{2*}  Dr. Renu Kale,³ Dr. Kailash Kaiwartya ⁴ 

1. Associate Professor, Department of Pathology, Raipur Institute of Medical Sciences, Raipur, Chhattisgarh, India
2. Associate Professor, Department of Pathology, Raipur Institute of Medical Sciences, Raipur, Chhattisgarh, India.
3. Professor, Department of Pediatrics, Raipur Institute of Medical Sciences, Raipur, Chhattisgarh, India.
4. Postgraduate Resident, Department of Pathology, Raipur Institute of Medical Sciences, Raipur, Chhattisgarh, India.

*Corresponding Author:

Dr. Firoz Sheikh, Associate Professor, Department of Pathology, Raipur Institute of Medical Sciences, Raipur, Chhattisgarh, India. Email id: thirdpage1@gmail.com

Abstract:

Glycogen storage disease type Ia is a rare inherited metabolic disorder caused by glucose-6-phosphatase deficiency. This leads to impaired glycogenolysis and gluconeogenesis. Infants commonly present with hypoglycemia, hepatomegaly, and other metabolic abnormalities. We report the case of an eight-month-old female infant who presented with abdominal distention, vomiting, and respiratory distress. Clinical examination revealed doll-like facies and marked hepatomegaly. Laboratory evaluation revealed severe hypoglycemia and marked hypertriglyceridemia with visibly lipemic serum. Liver biopsy revealed glycogen accumulation within hepatocytes, confirmed by periodic acid–Schiff staining. The combined clinical, biochemical, and histopathological findings supported a diagnosis of GSD-Ia. This case highlights the diagnostic value of lipemic serum as an early clinical clue and emphasizes the importance of an integrated clinicopathological evaluation in preventing complications.

Keywords: Glycogen storage disease type Ia; Hepatomegaly; Hypertriglyceridemia; Infant; Lipemic serum; Von Gierke disease

Introduction:

Glycogen storage disease type Ia (GSD Ia), also known as Von Gierke disease, is an autosomal recessive metabolic disorder caused by a deficiency of glucose-6-phosphatase. This enzyme is essential for maintaining blood glucose levels during fasting. Its deficiency leads to glycogen accumulation in the liver and kidneys, resulting in hepatomegaly and metabolic disturbances.^[1,2] Patients typically present in infancy with hypoglycemia, lactic acidosis, hyperlipidemia, and growth retardation.^[3] Severe hypertriglyceridemia may produce visibly lipemic serum, which can serve as an early diagnostic clue. Early identification is essential to prevent complications, such as hepatic adenomas and renal dysfunction.^[4] We report a case of an infant with marked lipemia, emphasizing the diagnostic relevance of the biochemical and histopathological correlations.

Case Report

An eight-month-old female infant was brought to the pediatric department of Raipur Institute of Medical Sciences, Raipur, a tertiary care hospital in central India, with complaints of abdominal distension and recurrent vomiting for five months. The child also had fever for one month and rapid breathing for three to four days. The patient had no history of seizures.

On examination, the child had characteristic doll-like facies with puffy cheeks and thin extremities. Abdominal examination revealed significant hepatomegaly, with the liver palpable 10.5 cm below the costal margin. The liver span was approximately 14 cm (Figure 1).

Laboratory evaluation revealed severe hypoglycemia, with a blood glucose level of 37 mg/dL. Hematological analysis revealed microcytic and hypochromic anemia. Biochemical investigations revealed marked hyperlipidemia. The serum was milky and lipemic (Figure 2). Triglyceride levels were markedly elevated at 3520 mg/dL, along with increased total cholesterol and very-low-density lipoprotein levels. Repeat testing showed persistent hyperlipidemia, although it was reduced after the initial management (Table 1).

Histopathological examination of the liver biopsy revealed a preserved lobular architecture. Hepatocytes were enlarged, with clear to pale eosinophilic cytoplasm. The nuclei were small, with occasional hyperglycogenation. Periodic acid–Schiff staining revealed strong positivity, consistent with glycogen accumulation. Mild periportal fibrosis was also noted (Figure 3). Diastase sensitivity confirmed glycogen deposition (Figure 4).

Based on the clinical presentation, biochemical abnormalities, and characteristic histopathological findings, a diagnosis of glycogen storage disease type Ia was established, and molecular genetic testing was advised for confirmatory diagnosis and subtype characterization.

Table 1: Lipid Profile of the Patient

Parameter	Initial Value (mg/dL)	Follow-up Value (mg/dL)	Reference Range (mg/dL)
Total Cholesterol	400	185	<200
Triglycerides	3520	968	<150
HDL	72	45	>40
LDL	376	53	<100
VLDL	704	193	5 to 40



Figure 1: Clinical photograph showing doll-like facies with puffy cheeks and a protuberant abdomen.



Figure 2: Lipemic serum sample demonstrating a milky appearance.

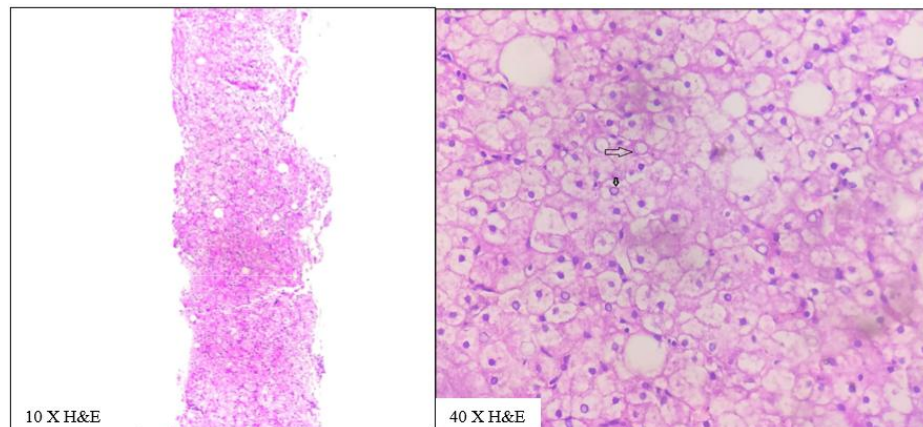


Figure 3: H&E staining (10× and 40×) showing enlarged hepatocytes with vacuolated cytoplasm due to glycogen accumulation. The large arrow indicates intracytoplasmic inclusions, and the small arrow indicates nuclear glycogen deposition.

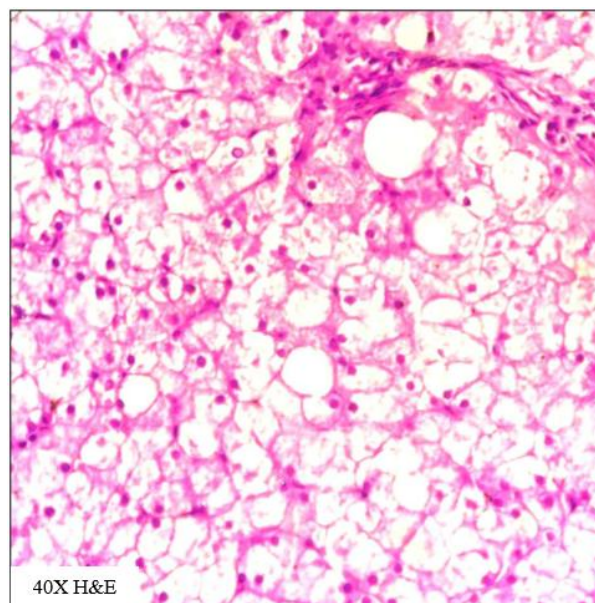


Figure 4: PAS staining (40×) showing glycogen accumulation in hepatocytes with diastase sensitivity, confirming glycogen deposition.

Discussion

Glycogen storage disease type Ia (GSD Ia) is a rare autosomal recessive metabolic disorder with an estimated incidence of approximately 1 in 100,000 live births.^[3] It results from a deficiency of glucose-6-phosphatase, leading to impaired glycogenolysis and gluconeogenesis, with the accumulation of glycogen in hepatocytes and renal tissue.^[1]

The disease typically presents within the first six months of life. The common clinical features include hypoglycemia, hepatomegaly, hyperlipidemia, and lactic acidosis.^[4] The present case

showed a classical early presentation with marked hepatomegaly and severe metabolic derangement, consistent with previously reported cases.

Severe hypertriglyceridemia is a hallmark biochemical abnormality in GSD Ia, resulting from increased hepatic lipogenesis and reduced lipid clearance. In some patients, triglyceride levels may exceed 2000–3000 mg/dL, producing visibly lipemic or milky serum. Although this finding is well-recognized biochemically, it is less frequently highlighted as a visual diagnostic clue. Carvalho et al. reported a series of patients with GSD Ia showing marked hyperlipidemia, with triglyceride levels comparable to those observed in the present case.^[3] Similarly, Raza et al. described a pediatric case of severe hypertriglyceridemia requiring dietary management, emphasizing its diagnostic relevance.^[2] The present case highlights the importance of lipemic serum as an easily identifiable but underreported early diagnostic clue in infants with GSD Ia.

Reports specifically highlighting lipemic serum as an initial laboratory clue are scarce. However, visually turbid serum during routine sampling may prompt an early evaluation of underlying metabolic disorders. The present case supports this observation and underscores the importance of simple laboratory findings in guiding diagnosis, particularly in resource-limited settings.

The histopathological findings in GSD Ia are well established. Enlarged hepatocytes with pale cytoplasm due to glycogen accumulation and preserved lobular architecture are characteristic features. Periodic acid–Schiff positivity with diastase sensitivity confirms glycogen deposition.^[1] Similar findings have been reported previously by Howell et al.^[5]

Differential diagnoses include other glycogen storage disorders, fatty liver disease, and inherited metabolic conditions that present with hepatomegaly. However, the combination of severe fasting hypoglycemia, marked hypertriglyceridemia, and characteristic histopathological findings strongly supports the diagnosis of GSD Ia. Molecular genetic testing may be considered for confirmatory diagnosis and subtype characterization of this disease.

Early diagnosis is essential to prevent complications such as growth retardation, hepatic adenoma, renal dysfunction, and metabolic instability. Management focuses on maintaining euglycemia through frequent feeding and uncooked cornstarch therapy, which improves metabolic control and long-term outcomes.^[2]

Conclusion

This case highlights the importance of lipemic serum as an early diagnostic clue in infants with metabolic disorders. The integration of clinical findings with biochemical and histopathological evaluations is essential for an accurate diagnosis. Early recognition and timely management can prevent serious complications and improve the long-term outcomes.

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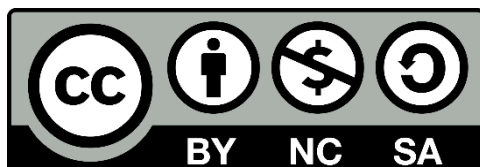
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Conflict of interest: Nil

Funding: Nil

Journal Homepage: <https://journalofmedicalanddentalfrontiers.com/>

PURLs: <https://archive.org/details/2-cr-jmdf-jun-2026-1-6>



Submitted: 05/04/2026

Revised: 29/05/2026

Accepted: 20/06/2026

Published: 30/06/2026

Cite this article:

Dr. Chandni Krishnani, Dr. Firoz Sheikh, Dr. Renu Kale, Dr. Kailash Kaiwartya. Lipemic Serum and Severe Hypertriglyceridemia as Presenting Features of Glycogen Storage Disease Type Ia in an Infant: A Case Report. *Jour Med Dent Fron* 2026;3(1):1-6