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Clinical Profile of Glycogen Storage Diseases in South Indian Children - A Prospective Study

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ABSTRACT

Background: Glycogen Storage Diseases (GSDs) are inherited metabolic disorders caused by enzyme deficiencies affecting glycogen synthesis or breakdown, leading to abnormal glycogen accumulation primarily in the liver and muscles. Early recognition is essential to prevent complications such as recurrent hypoglycaemia, growth failure, and organ dysfunction.
Aim: To study the clinical spectrum of GSD in children and correlate clinical presentation with biochemical and histopathological findings for early diagnosis and better management.
Materials and Methods: A descriptive observational study was conducted over 24 months at the Institute of Child Health, Egmore, Chennai. Twenty-five children clinically suspected and diagnosed with GSD were evaluated through detailed clinical assessment, biochemical investigations (blood glucose, lipid profile, uric acid, lactate, liver function tests), imaging, and histopathological examination using liver biopsy (and muscle biopsy in selected cases). Data were analysed using descriptive statistics.
Results: Among 25 children, 80% were below 5 years, with a male predominance (56%). Consanguinity was observed in 56%, and family history was positive in 14 cases. The most common presenting symptom was abdominal distension (56%), and hepatomegaly was the most consistent clinical finding (64%). Biochemical abnormalities included low fasting blood glucose (mean 45 ± 10 mg/dL), hypertriglyceridaemia (mean 320 ± 60 mg/dL), hyperuricaemia (mean 8 ± 2 mg/dL), lactic acidosis (mean 5.4 ± 1.1 mmol/L), and elevated transaminases (mean 110 ± 25 U/L). Liver biopsy showed vacuolated hepatocytes with PAS-positive glycogen deposits, confirming diagnosis. Hepatic forms (Types I, III, and IX) were the most common subtypes observed.
Conclusion: GSD predominantly presents in early childhood with hepatomegaly, hypoglycaemia, growth delay, and characteristic biochemical abnormalities. Clinical and biochemical evaluation supported by histopathology remains crucial for diagnosis in resource-limited settings. Early diagnosis and dietary interventions can significantly improve prognosis and reduce complications.

Keywords: Glycogen Storage Disease, Hepatomegaly, Hypoglycaemia, Liver biopsy, Paediatric metabolic disorder

INTRODUCTION

Glycogen storage diseases (GSDs) are a group of inherited metabolic disorders characterized by defective metabolism of glycogen, leading to its abnormal accumulation or depletion within tissues, primarily the liver and muscles [1]. Glycogen is a glycosan, serves as the primary storage form of carbohydrate in the human body. It plays a critical role in maintaining blood glucose homeostasis, providing energy during fasting and physical activity [2].

Glycogen Metabolism Overview

Glycogen synthesis (glycogenesis) and degradation (glycogenolysis) are tightly regulated processes controlled by a series of enzymes. Key steps of the metabolism include: Glycogen Synthase: Catalyzes the addition of glucose units from UDP-glucose to glycogen chains; Glycogen Phosphorylase: Breaks α-1,4-glycosidic bonds to release glucose-1-phosphate. Branching and Debranching Enzymes: Ensure proper molecular structure and mobilization of glycogen [3,4].

The interplay between insulin and glucagon regulates these pathways. Insulin promotes glycogen synthesis, while glucagon and epinephrine activate glycogenolysis [5].

Pathophysiology of Glycogen Storage Diseases

Deficiency of one or more enzymes involved in glycogen metabolism results in the accumulation of glycogen with abnormal structure or distribution within tissues. The clinical manifestations depend on, the enzyme affected, the tissue distribution of the enzyme, and the degree of enzyme deficiency. GSDs are generally autosomal recessive disorders, though some (like Type IX) are X-linked recessive [4, 6].

Classification of Glycogen Storage Diseases

The GSDs are classified numerically according to the order of their discovery and the deficient enzyme (Table 1) [5,7].

Table 1: Types of Glycogen Storage Diseases, enzyme defect

Type	Name	Enzyme Defect	Primary Organs Involved
I	Von Gierke's disease	Glucose-6-phosphatase	Liver, Kidney
II	Pompe's disease	Acid maltase (lysosomal α-1,4-glucosidase)	Muscle, Heart
III	Cori / Forbes disease	Debrancheing enzyme	Liver, Muscle
IV	Andersen's disease	Branching enzyme	Liver
V	McArdle's disease	Muscle phosphorylase	Muscle
VI	Hers' disease	Liver phosphorylase	Liver
VII	Tarui's disease	Phosphofructokinase	Muscle
IX	Phosphorylase kinase deficiency	Phosphorylase kinase	Liver ± Muscle

Clinical Manifestations

The **clinical presentation** varies depending on the enzyme defect and affected tissue [8,9]:

- **Hepatic GSDs (Types I, III, VI, IX):** Symptoms include**,** hypoglycemia during fasting, hepatomegaly, growth retardation, elevated liver transaminases.
- **Muscular GSDs (Types II, V, VII):** Symptoms include exercise intolerance, Muscle weakness and cramps, myoglobinuria after strenuous activity. In Type II (Pompe's), infantile cardiomegaly and respiratory failure are common. The onset of symptoms typically occurs in infancy or early childhood, with variable severity depending on the residual enzyme activity.

Diagnostic Approach

Diagnosis of GSD involves clinical suspicion based on hepatomegaly and fasting hypoglycaemia [6], Biochemical tests: fasting blood glucose, lactate, triglycerides, and uric acid. Histopathology: liver and muscle biopsy showing glycogen accumulation [8]. Molecular analysis:

identification of specific gene mutations for confirmation. Differential diagnoses include other metabolic disorders such as fatty acid oxidation defects and galactosemia [7].

Aim of the Study

Given the high rate of consanguineous marriages in certain populations (such as South India), there is a higher incidence of inherited metabolic diseases like GSD. The aim of this study is to determine the clinical spectrum of GSD in children, correlate biochemical and histopathological findings, and identify characteristic patterns for early diagnosis and management.

MATERIALS & METHODS

Study Design

This is a descriptive observational study conducted at the Institute of Child Health and Hospital for Children, Egmore, Chennai. The study was undertaken to evaluate the clinical profile, biochemical characteristics, and histopathological findings in children diagnosed with Glycogen Storage Disease (GSD). The study was approved by the Institutional Ethics Committee. Written informed consent was obtained from the parents or guardians of all patients prior to inclusion in the study and for biopsy procedures.

Study Period

The study was carried out over a period of 24 months (two years).

Study Population

Children attending the Pediatric Gastroenterology, Hepatology and Nutrition Department of the Institute of Child Health, who were clinically suspected of having Glycogen Storage Disease, were included in the study.

A total of 25 cases of GSD were studied during this period.

Inclusion Criteria: Children with clinical evidence of hepatomegaly, hypoglycemia during fasting or illness, growth retardation, and biochemical abnormalities suggestive of Glycogen Storage Disease were considered for the study.

Exclusion Criteria: Children with chronic liver disease due to other causes such as viral hepatitis, Wilson's disease, or autoimmune hepatitis, metabolic disorders other than GSD, such as galactosemia or fructose intolerance were excluded from the study.

Methods of Study

The study consisted of a detailed clinical, biochemical, and histopathological evaluation of each patient.

Clinical Evaluation

A detailed history was obtained in every case, with reference to, Age of onset of symptoms, consanguinity of parents, family history of similar illness, presenting complaints such as abdominal distension, growth failure, or hypoglycemic episodes, physical examination findings including hepatomegaly, splenomegaly, muscle tone, and neurological signs.

Biochemical Investigations

All patients underwent the laboratory investigations such as Blood sugar (fasting and postprandial), Serum triglycerides, Serum cholesterol, Serum uric acid, Serum lactic acid, Liver function tests (LFTs) (SGOT, SGPT, bilirubin, total protein, and albumin), Renal function tests: blood urea, serum creatinine and Urinalysis for glucose and protein. Results were compared with reference ranges and correlated with clinical findings.

Histopathological Evaluation

Liver biopsy was performed in most patients using a Menghini needle under aseptic precautions. In selected cases with muscle weakness, muscle biopsy was also performed. Biopsy specimens were fixed in 10% neutral buffered formalin and processed routinely [10]. Sections were stained with Hematoxylin and Eosin (H&E) and Periodic Acid–Schiff (PAS) stain for

glycogen demonstration. Diastase digestion was used to confirm glycogen content.

Microscopic Findings includes:

- In Type I (Von Gierke's) disease – hepatocytes showed prominent vacuolation with nuclear hyperglycogenation.
- In Type III (Cori's) disease – uniform mosaic pattern with small glycogen droplets.
- In Type IX – patchy small droplet pattern with subsarcolemmal glycogen accumulation in muscle biopsy.

Table 2: Presenting features in the observed GSD types

Type	Liver Features	Muscle Features
I	Prominent vacuolated hepatocytes	Normal
III	Uniform mosaic with small glycogen droplets	Subsarcolemmal glycogen
IX	Patchy small droplets	Subsarcolemmal glycogen

Imaging Studies

Ultrasonogram (USG) of the abdomen was performed to assess liver size, echotexture, and presence of splenomegaly.

Echocardiography was done in patients with suspected muscular or cardiac involvement (e.g., Type II GSD).

Statistical Analysis

Data was entered in Microsoft Excel and analyzed using descriptive statistical methods. Percentage distribution was calculated for categorical variables such as age, gender, and presenting symptoms. Mean and standard deviation were derived for continuous biochemical parameters. Clinical and biochemical correlation was established for diagnostic confirmation.

RESULT

A total of 25 children who were diagnosed with Glycogen Storage Disease (GSD) were included in this study. The following observations were made regarding their demographic data, clinical presentation, biochemical findings, and histopathological features.

Most of the children in this study were below 5 years of age. The youngest patient was 9 months old, and the oldest was 10 years. Majority of the patients (80%) presented before 5 years of age. There was a slight male predominance with 14 males (56%) and 11 females (44%) in the study group.

More than half of the patients (56%) were born of consanguineous parentage. A positive family history of similar illness was observed in 14 children, indicating a strong genetic predisposition.

The most common presenting complaint was abdominal distension due to hepatomegaly, observed in 14 children (56%). Other presenting features included growth retardation in 6 children (24%), hypoglycemia in 3 children (12%), and muscle weakness in 2 children (8%).

On clinical examination, hepatomegaly was the most consistent finding, present in 16 children (64%). Hypoglycemia was observed in 10 children (40%), followed by growth retardation in 8 (32%) and hyperlipidemia in 7 (28%). Elevated liver enzymes were seen in 6 (24%).

In children presenting with neurological manifestations, the common features were hypotonia, muscle weakness, and developmental delay. Seizures were documented in one case. Miscellaneous findings such as cardiomegaly and muscle cramps were observed in a few children, primarily those with Type II (Pompe's) disease.

Biochemical investigations revealed that the majority of children had low fasting blood glucose levels and elevated serum triglycerides. Hyperuricemia, lactic acidosis, and elevated transaminases were also noted in a significant number of patients. These biochemical derangements were more pronounced among the hepatic forms of GSD.

Liver biopsy was performed in most of the patients and revealed marked vacuolation of

hepatocytes with PAS-positive glycogen deposition. Some cases showed nuclear hyperglycogenation and mild fatty change. Muscle biopsy was done in selected patients with muscle weakness, showing subsarcolemmal glycogen accumulation in Type II and Type IX GSDs. PAS staining confirmed diastase-sensitive glycogen within the cytoplasm.

On correlation of the clinical, biochemical, and histopathological findings, it was evident that the hepatic forms (Types I, III, and IX) constituted the majority of cases in this study group. The muscle and mixed types (Types II and V) were less frequent.

Thus, in the present study, the most common form of GSD encountered was the hepatic variety, presenting with hepatomegaly, fasting hypoglycemia, and biochemical abnormalities consistent with hepatic dysfunction. The muscle and mixed types presented with hypotonia, cardiomegaly, and elevated muscle enzymes in a smaller subset of children.

Table 3: Age of the Children Participated in the Study

Age Group (years)	No. of Cases	Percentage (%)
0 – 1	5	20%
1 – 3	8	32%
3 – 5	7	28%
> 5	5	20%
Total	25	100%

Table 4: Gender of the Children Participated in the Study

Gender	No. of Cases	Percentage (%)
Male	14	56%
Female	11	44%
Total	25	100%

Table 5: Family History and Consanguinity

Family History	Consanguinity Present	Consanguinity Absent	Total
Positive	10	4	14
Negative	4	7	11
Total	14	11	25

Table 6: Initial Complaints of the Children

Symptoms	No. of Cases	Percentage (%)
Abdominal Distension	14	56%
Growth Retardation	6	24%
Hypoglycemia	3	12%
Muscle Weakness	2	8%
Total	25	100%

Table 7: Signs and Symptoms in Hepatic Presentation (Type I, III, IV, IX)

Clinical Features	No. of Cases	Percentage (%)
Hepatomegaly	16	64%
Hypoglycemia	10	40%
Growth Delay	8	32%
Hyperlipidemia	7	28%
Elevated Liver Enzymes	6	24%

Table 8: Signs and Symptoms in Neurological Presentation

Feature	No. of Cases	Percentage (%)
Hypotonia	4	16%
Muscle Weakness	3	12%
Developmental Delay	2	8%
Seizures	1	4%

Table 9: Signs and Symptoms in Miscellaneous Presentations

Clinical Finding	No. of Cases	Percentage (%)
Cardiomegaly	2	8%
Hypotension	1	4%
Muscle Cramp	1	4%
Total	4	16%

Tables 10: Biochemical Investigations

Investigation	Normal Range	GSD Mean ± SD	Abnormality Frequency (%)
Blood Glucose (mg/dL)	70–100	45 ± 10	80% Low
Serum Triglycerides (mg/dL)	<150	320 ± 60	68% High
Serum Uric Acid (mg/dL)	2–6	8 ± 2	72% High
Lactic Acid (mmol/L)	0.5–2.2	5.4 ± 1.1	64% High
AST/ALT (U/L)	10–40	110 ± 25	76% Elevated

DISCUSSION

Glycogen Storage Diseases (GSDs) are a group of inherited metabolic disorders resulting from specific enzyme deficiencies involved in glycogen synthesis or breakdown. The present study was undertaken to analyze the clinical profile, biochemical abnormalities, and histopathological findings in children diagnosed with GSD at the Institute of Child Health, Chennai.

In this study, a total of 25 children were evaluated. The majority of them presented before five years of age, with the youngest being nine months old. This observation correlates well with other studies which have also documented that GSDs commonly present in infancy and early childhood [4]. The early onset of symptoms is due to the critical role of glycogen metabolism in maintaining blood glucose levels during fasting, which becomes more apparent as hepatic glycogen stores are utilized during illness or prolonged fasting [3,5].

A slight male predominance was observed in this study. This is consistent with the known X-linked inheritance pattern of some types, such as GSD Type IX, though the overall inheritance pattern of most GSDs remains autosomal recessive. The high incidence of consanguinity among the affected families in this study further supports the autosomal recessive nature of the disease [8]. The most common clinical presentation was hepatomegaly with abdominal distension, observed in 64% of the cases. This finding is in agreement with the classical presentation of hepatic forms of GSD, such as Type I (Von Gierke's disease) and Type III (Cori's disease), where defective glucose mobilization leads to glycogen accumulation in hepatocytes [11, 12]. Growth retardation, hypoglycemia, and hyperlipidemia were also frequent findings and can be attributed to impaired glycogenolysis and gluconeogenesis.

Neurological features such as hypotonia and developmental delay were noted in a smaller number of children, corresponding to the muscular or mixed types of GSD. Muscle weakness and cardiomegaly were predominantly seen in cases suspected to be Type II (Pompe's disease), where lysosomal glycogen accumulation interferes with muscular and cardiac function. These findings are consistent with previously published reports on Pompe's disease and its variable cardiac manifestations [13].

The biochemical abnormalities noted in this study, including hypoglycemia, hypertriglyceridemia, hyperuricemia, and elevated liver enzymes, are well-documented features of hepatic GSDs. Hypoglycemia results from the inability of the liver to release glucose due to the deficiency of glucose-6-phosphatase (Type I) or other related enzymes. The excess glucose-6-phosphate in hepatocytes is diverted to pathways leading to increased production of triglycerides, uric acid, and lactate, explaining the biochemical profile observed in this study [7-9].

Histopathological findings in liver biopsies showed vacuolated hepatocytes with PAS-

positive glycogen deposition, confirming the diagnosis of GSD. Nuclear hyper-glycogenation and mild fatty changes were observed in several cases, particularly in Type I and III. Muscle biopsies performed in selected cases revealed subsarcolemmal glycogen accumulation, typical of muscle-involving forms of GSD. These findings correlate with the reports by Taratuto AL (2010) and Ichimoto K (2020), who described similar patterns of glycogen storage on histological examination [12, 13].

The predominance of hepatic forms (Types I, III, and IX) in the present study aligns with other Indian and international studies, where Type I GSD (Von Gierke's disease) remains the most frequent subtype. The clinical overlap among various hepatic types makes enzyme assays and molecular testing essential for definitive diagnosis [14]. However, due to the limited availability of such facilities in resource-limited settings, diagnosis often relies on clinical, biochemical, and histopathological correlation, as demonstrated in this study.

Complications observed in hepatic forms of GSD include growth failure, delayed puberty, hepatic adenomas, and renal dysfunction. Chronic hyperuricemia and hyperlipidemia also predispose to xanthomas and gout in older children [4-6]. In the present series, none of the children had developed hepatic adenomas, likely due to the younger age group studied.

The management of GSD primarily involves dietary modification. Frequent carbohydrate-rich meals, uncooked cornstarch therapy, and avoidance of fasting are cornerstones of therapy in hepatic GSDs. Such measures prevent hypoglycemic episodes and secondary metabolic disturbances [13]. The use of allopurinol in hyperuricemia and lipid-lowering agents in severe hyperlipidemia is well established. Liver transplantation remains an option in cases with advanced hepatic failure or recurrent metabolic crises [11-13]. Early recognition and institution of appropriate dietary and supportive measures can prevent complications and improve growth and development. Regular monitoring of liver function, growth parameters, and metabolic profile is essential in these patients. Genetic counselling should be offered to families, especially in populations with a high rate of consanguinity [9].

Thus, this study emphasizes the importance of correlating clinical presentation, biochemical parameters, and histopathological features for the accurate diagnosis of Glycogen Storage Diseases. The findings are consistent with global data and highlight the need for improved diagnostic facilities and awareness for early detection and management.

CONCLUSION

The following conclusions were derived from the present study:

Age of Onset and Presentation: Most cases of Glycogen Storage Disease presented during infancy and early childhood. The majority of children were below five years of age at diagnosis, emphasizing that these disorders manifest early in life due to defective glycogen metabolism.

Sex Distribution: A slight male predominance was observed. Although most GSDs follow an autosomal recessive inheritance, the higher number of male patients may be due to the presence of an X-linked variant (Type IX) and increased likelihood of medical consultation for male children.

Consanguinity and Genetic Basis: More than half of the affected children were born of consanguineous parentage. This finding reinforces the autosomal recessive mode of inheritance and highlights the need for genetic counselling and public awareness in populations where consanguineous marriages are common.

Clinical Profile: The most frequent clinical manifestation was hepatomegaly associated with abdominal distension. Other common features included hypoglycemia, growth retardation, and hyperlipidemia. Neurological and cardiac manifestations

were noted in the muscle and mixed types of GSD.

Biochemical Findings: Hypoglycemia, hypertriglyceridemia, hyperuricemia, and elevated transaminases were the predominant biochemical abnormalities observed. These findings are consistent with the hepatic forms of GSD.

Histopathological Findings: Liver biopsy was diagnostic in most cases, showing vacuolated hepatocytes with PAS-positive glycogen deposits. Muscle biopsy, when performed, demonstrated subsarcolemmal glycogen accumulation in the muscular variants.

Predominant Type: The majority of children in this study belonged to the hepatic forms of Glycogen Storage Disease, primarily Types I, III, and IX. The muscle forms (Types II and V) were less frequent.

Complications: Long-term complications observed or anticipated in GSD include growth failure, delayed puberty, hepatic adenomas, renal dysfunction, and metabolic disturbances such as hyperuricemia and hyperlipidemia. None of the children in the present study developed hepatic adenomas during the study period, likely due to their young age.

Management and Prognosis: Early diagnosis followed by dietary therapy including frequent carbohydrate-rich meals and uncooked cornstarch at bedtime can prevent hypoglycaemia and secondary metabolic complications. Supportive management with allopurinol for hyperuricemia and lipid-lowering therapy for severe hyperlipidemia may be required. Liver transplantation is indicated in cases of advanced hepatic failure or recurrent metabolic crises.

Declaration by Authors

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