# Spectrum of GSDs: A Single Center Experience From India

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## INTRODUCTION

Glycogen storage disorders represent a group of about 14 genetically distinct, biochemically related, inherited diseases affecting the metabolism and storage of glycogen. Data on prevalence of these disorders in Indian population is limited.

## OBJECTIVE

We present herewith the molecular spectrum of GSD seen in patients referred to our center from 2015 to 2018.

## MATERIAL & METHOD

This is a retrospective analysis of the data of patients with GSDs, for the period from 2015 to 2018. Patients clinically and biochemically suspected to have any GSD were selected for this study. Clinical features included hepatomegaly, hypoglycemia, developmental delay, growth retardation and myopathy. We suspected 46 patients clinically & biochemically whose samples were sent to Centogene AG, Rostock for molecular confirmation and 22 patients were confirmed by molecular analysis.

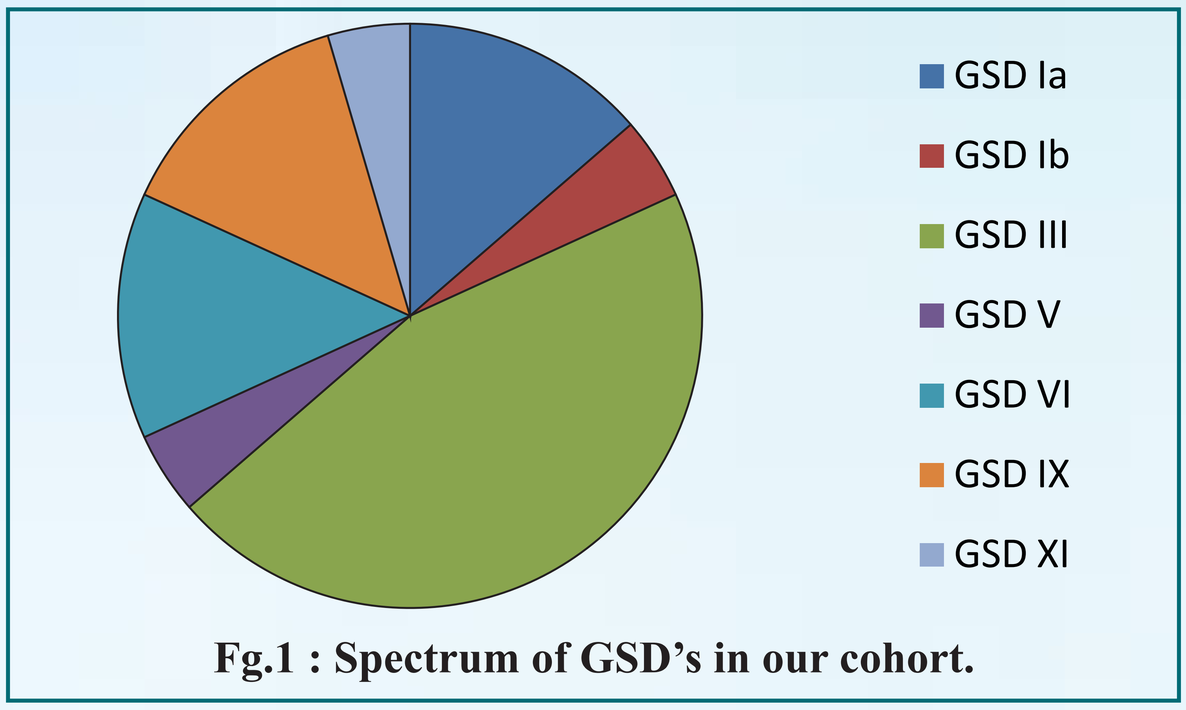
## RESULT

In the period 2015 through 2018, 46 individuals suspected to have GSD, based on clinical features or biochemical parameters were referred to our center. The most common presenting features included hepatomegaly, hypoglycaemia, developmental delay, growth retardation, and myopathy. Of these 22 were confirmed to have particular GSD by means of molecular analysis. GSD III was the commonest in our cohort with 10 (45%) confirmed cases. Other GSDs in our cohort included GSD Ia (n=3), GSD I b/c (n=1), GSD V (n=1), GSD VI (n=3), GSD IX (n=3) and GSD XI (n=1).

| GSD type | Gene Mutation | n= |
|---|---|---|
| **GSD Ia (von Gierke's)** | G6PC | 3 |
| **GSD Ib/c (von Gierke's disease)** | SLC37A4 | 1 |
| **GSD III(a, b,c,d) (Cori/Forbe's)** | AGL gene | 10 |
| **GSD V (McArdle's) & GSD VII (Ta rui's)** | PYGM gene | 1 |
| **GSD VI (Her's Disease)** | PYGL gene | 3 |
| **GSD IX** | PHKA1, PHKA2, PHKB/PHKG2 gene | 3 |
| **GSD XI (FBS)** | SLC2A2 | 1 |

## DISCUSSION

The overall GSD incidence is estimated 1 case per 20000-43000 live births and the most common one is types I and IX. However in our cohort GSD III was more common than the other types. Hepatomegaly, hypoglycemia, short stature, dyslipidemia were seen in most of these patients with GSD III in our cohort. Treatment for these patients is aimed at maintaining normoglycemia by frequent feeds and uncooked corn starch. Liver transplantation has not been tried in any of these patients.




**Fg.1 : Spectrum of GSD's in our cohort.**

Other common GSDs in our cohort were GSD Ia, GSD VI and GSD IX. All 3 patients with GSD 1a are under treatment with uncooked corn starch in the dose of 2g/kg/day in 3 divided doses. However we did not see any improvement nor was there any significant deterioration so far. Overnight UCCS drip was not possible as parents refused any such therapy. One of these also received Glycosade in combination with UCCS and parents felt that there is improvement after introduction of glycosade. One of our GSD VI patients has shown good improvement clinically with uncooked corn starch.

So far none of our GSD patients have developed any malignancy or any other complication and we are monitoring them regularly.

| GSD type | Clinical features |
|---|---|
| GSD 1 | Hepatomegaly (n=4), doll - like facies (n=3), delayed milestones(n=2), short stature (n=4 ), hypoglycemia, hyperuricemia,hypercholesterolemia, hypertriglyceridemia and abnormal LFTs (n-=4) |
| GSD III | Hepatosplenomegaly (n=3) , hepatomegaly (n=7), hypoglycemia (n=10), abnormal LFTs (n=10) |
| GSD V | Exercise intolerance, ataxic gait no rhabdomyolysis, elevated CK |
| GSD VI | Hepatomegaly (n=3), hypoglycemia (n=3), growth retardation (n=3), lactic acidosis (n=3), ketosis (n=3) |
| GSD IX | Hepatomegaly (n=4), hypoglycemia (n=4), growth retardation (n=3), fanconi syndrome (n=1) |

## CONCLUSION

Overall, the study demonstrates that GSD III followed by GSD Ia, VI and IX are the most common GSDs in our cohort. All our patients are managed with uncooked corn starch with no futher deterioration of the symptoms. None of the patients has undergone a liver transplant as per our knowledge.

**Conflict of Interest :** None

## REFERENCES:

"Hasan Özen. Glycogen storage diseases: New perspectives. World J Gastroenterol. 2007 May 14; 13(18): 2541-2553



*Poster presented at SSIEM, 2019, Rotterdam. Available on:* www.metabolicerrors.com