Journal of Advanced Medical and Dental Sciences Research

@Society of Scientific Research and Studies

Journal home page: www.jamdsr.com doi: 10.21276/jamdsr ICV 2018= 82.06

(e) ISSN Online: 2321-9599; (p) ISSN Print: 2348-6805

Case Report

Glycogen Storage Disease (Type IA)

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***ABSTRACT*:**
Glycogen storage diseases (GSDs) are a group of metabolic disorders determined by the accumulation of glycogen in different tissues. GSDs are caused by the enzyme deficiencies effect on glycogen synthesis, glycogen breakdown or glycolysis (glucose breakdown), typically within muscles and/or liver cells. GSD are group of IEM characterized by accumulation of glycogen in various tissues. Accumulation is histological hallmark of these disorders although the phenotype shows various overlap. The disorders were numbered as they were discovered which classified chronologically by GSD type I (von Gierke disease) to GSD type XI. Hereby, we present a case report of a 2.5 years old male child with Type I Glycogen storage disease.
Keywords: Glycogen storage, metabolic disorder, G6PC gene.

Received: 28 August, 2019 Revised: 6 October, 2019 Accepted: 8 October, 2019

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This article may be cited as: Jain P, Thamke R, Gavhane JB. Glycogen Storage Disease (Type IA). J Adv Med Dent Scie Res 2019;7(11):121-124.

INTRODUCTION:
Glycogen storage diseases (GSDs) are a group of metabolic disorders determined by the accumulation of glycogen in different tissues. GSDs are caused by the enzyme deficiencies effect on glycogen synthesis, glycogen breakdown or glycolysis (glucose breakdown), typically within muscles and/or liver cells. GSD are group of IEM characterized by accumulation of glycogen in various tissues. Accumulation is histological hallmark of these disorders although the phenotype shows various overlap. The disorders were numbered as they were discovered which classified chronologically by GSD type I (von Gierke disease) to GSD type XI.[1] Glycogen storage disease type 1A is caused by the deficiency of Glucose-6-phosphatase catalytic activity which results from mutations in the G6PC gene.[2] This condition is inherited in an autosomal recessive pattern. The cumulative incidence of these diseases is approximately 1 in every 20,000 live births.[3] Although some newborns present with severe hypoglycemia, it is more common for infants with hepatomegaly, lactic acidosis, hyperuricemia and/or hypoglycemic seizures. Untreated children typically have doll-like faces with fat cheeks and relatively thin extremities.[4] Hereby, we present a case report of a 2.5 years old male child with Type I Glycogen storage disease.

CASE REPORT:
2.5 years old male child brought by parents with complaints of yellowish discoloration of eyes intermittently from 3 months of age, passing dark yellow urine with abdominal enlargement. Child was born to primi mother at FTND with no adverse perinatal factors. Given exclusive breastfeeding. No delayed passage of meconium or neonatal jaundice. No other active problem until developed yellowish discoloration of conjunctiva at 3 months of age. No clay colored stool

or staining of diapers. Initially took ayurvedic treatment for jaundice. Repeated hospitalization for fever, cough thereafter had 1 GTC convulsion at 1 year of age. Normal weight gain and development according to parents.

ON EXAMINATION

General Examination:

Conscious, active, Mild pallor +
HR-110/min Icterus +/-
RR-32/min. No cyanosis, clubbing, koilonychia
Temp: 37 0C CRT < 3secs.
Chubby, Puffy face+ Frontal bossing +

Anthropometry:

Weight: 15 kg
Height: 83 cm
OFC 48: cm

Systemic Examination:

No lymphadenopathy, rash, ecchymotic patch.
CVS- Normal
R/S – Normal
CNS – child was conscious, active
P/A: Distended (AG 56 cm), skin stretched; no visible veins.
Liver 10 cms BCM (Span 13Cm) firm, rounded margin.
Spleen 8 cm.
No fluid thrill / shifting dullness

INVESTIGATIONS

Hb – 11
PCV - 34
MCV - 96.6
MCH - 31
MCHC – 32.1
TLC – 10,800
N/L/M/E – P56, L38, E6, M0
Platelets – 4.08 lakhs
ESR – 118
PT – 12.6
INR – 0.86
Mx – Negative
Bilirubin (T) 5.42, (D- 4.3)
SGOT – 933.4
SGPT – 455.8
Protein(T) – 7.29
Albumin – 4.09
Globulin – 3.2
ALP – 280
GGT – 252
Uric acid – 6.3
Urea – 20
Creatinine – 0.19
Hep-A – Negative
HBsAg – Negative
HCV - Negative

SPECIAL INVESTIGATIONS

Cholesterol – 269
TGL – 380
HDL - 34
LDL - 159
VLDL – 76
Serum Lactate – 0.70
Fasting RBS – 43 mg/dl.
GGT: 232.80 (Normal value : 5-16)

RADIOLOGICAL INVESTIGATION

USG (Abdomen): Liver echotexture hyperechoeic as compared to renal cortex.
PV and CBD normal.

IMPRESSION: Hepatomegaly with grade-1 fatty liver.

HISTOPATHOLOGICAL INVESTIGATIONS

Liver Biopsy done and suggestive of following features of Glycogen Storage Disease.

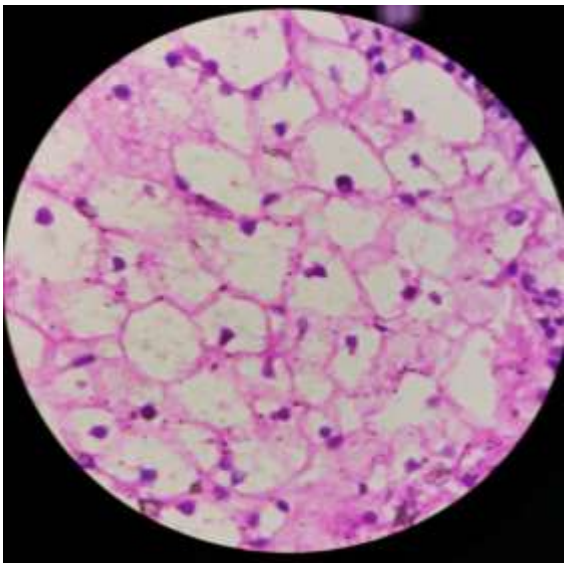

Figure 1: PAS-Diastase
Balooning of hepatocytes by glycogen and lipid, mosaicism

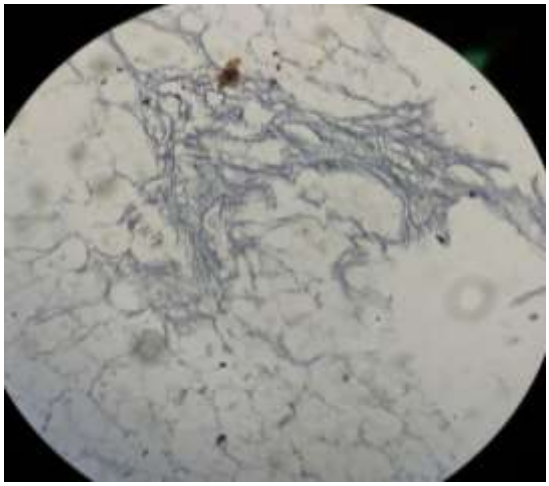

Figure 2: Masson's Trichrome Stain
Minimal fibrosis, absence of fat, large prominent vacuoles present.

On further evaluation in history, we also found similar features like doll like facies, hepatomegaly and hypoglycemia but liver biopsy could not be performed.
Patient treated as GSD & started on corn starch diet, fat soluble vitamins & other nutritional supplements. Child is on regular follow up.

DISCUSSION:
Glycogen storage disease type 1 is an inherited disorder caused by the buildup of a complex sugar called glycogen in the body's cells.

Figure 3: Younger sibling

The accumulation of glycogen in certain organs and tissues, especially the liver, kidneys, and small intestines, impairs their ability to function normally. [2] Glycogen storage disease type 1A is characterized by growth retardation leading to short stature and accumulation of glycogen and fat in the liver and kidneys. Although some newborns present with severe hypoglycemia, it is more common for infants to present at age three to four months with hepatomegaly, lactic acidosis, hyperuricemia, hyperlipidemia, and/or hypoglycemic seizures. Untreated children typically have doll-like faces with fat cheeks and relatively thin extremities. Xanthoma and diarrhea may be present.[4] Glycogen storage disease type 1A is caused by the deficiency of glucose-6-phosphatase (G6Pase) catalytic activity which results from mutations in the G6PC gene.[1] [2] This condition is inherited in an autosomal recessive pattern.[2] Liver and kidney functions tests, biochemical tests for estimating serum levels of sugar, lipid, uric acid and lactic acid are required to support the diagnosis. Radiological imaging especially ultrasound abdomen is used to evaluate liver span and renal size.
Histological confirmation of glycogen accumulation in hepatocytes in liver biopsy is diagnostic along with estimation of functional levels of Glucose-6-phosphatase.[5]

TREATMENT:
To maintain normal blood glucose level; achieved by NG infusion of glucose or uncooked corn starch. Uncooked corn starch provides slow release glucose (Dose: 1.6 gm/kg Q4 hrs for infants <2 yrs of age; older children 1.75 gm/kg Q6 hrly). Fructose & galactose

restricted. Calcium & vitamin supplementation required.

CONCLUSION:

GSD is a rare condition that changes the way body uses and stores glycogen, a form of sugar. Passed down from parents to children (inherited). Each parent must pass on one abnormal copy of the same gene. Most parents do not show any signs of GSD. So, the early and prompt diagnosis of GSD IA allows advances in therapy mainly dietary therapy, with improvement of survival and quality of life. Although hyperlipidemia, almost always present, should be treated to reduce risk of pancreatitis and cholelithiasis.

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