A CASE OF HEPATOSPLENOMEGALY

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Presentor : R.Srivijayalakshmi -PG

- 9 months old male baby
- 2nd born to 3 degree consanguinity
- Abscess - left eyebrow for 10 days
- Fever for 1 day
- Abdominal distension – 3 months of age

PAST HISTORY

- H/o loose stools on and off since birth
- No h/o hematemesis /malena / jaundice
- No h/o seizures
- No H/o loss of appetite/loss of weight
- No h/o cough/fever/breathlessness
- ?CONTACT h/o Tuberculosis present.

- Alert
- Awake
- Afebrile
- pallor
- no jaundice
- no clubbing
- no cyanosis

- Abscess over left eyebrow
- Cervical lymphadenopathy1.5*1.5 size tender
- VITALS normal

- ANTHROPOMETRY
- Height 68cm - -2--3SD
- Weight 7kg - 0-2SD
- Weight for length - 0-2SD

SYSTEM EXAMINATION

- CVS & RS – normal
- P/A – distended
- Liver span – 9.5cm(normal – 6cm)
- Spleen tip palpable.
- Kidneys not palpable
- CNS – clinically normal

SUMMARY

- 3 deg consanguinity
- H/o loose stools since birth
- Abscess once
- Hepatosplenomegaly
- Developmentally normal
- Previous sibling normal
- Contact: h/o tuberculosis +

DIFFERENTIAL DIAGNOSIS

- TUBERCULOSIS
- IMMUNODEFICIENCY
- STORAGE DISORDER

- CBC – HB 7.8
- Total counts 9000/ Cu mm
- P36L64
- Platelets count – 3 lakhs
- Bleeding time – 2 min
- Serum ferritin – low
- Sr.iron binding capacity normal

TUBERCULOSIS WORKUP

- X RAY Chest – normal and mantoux - negative
- RGJ for AFB , Gene Xpert - negative
- HIV - not reactive
- Lymph node biopsy : granulomatous lesion.no caseation necrosis seen , Gene Xpert - negative

IMMUNODEFICIENCY WORKUP

- CBC – normal
- PS – Severe anisopoikilocytosis platelets in single and clumps
- NBT - normal
- IG PROFILE - normal

- Fasting CBG – 90 / 80 / 110 / 100 – normal
- SGOT – 103IU
- SGPT - 61IU
- Total proteins 6.7G/DL
- Blood urea 27 mg/dl
- Serum creatinine 0.8mg/dl

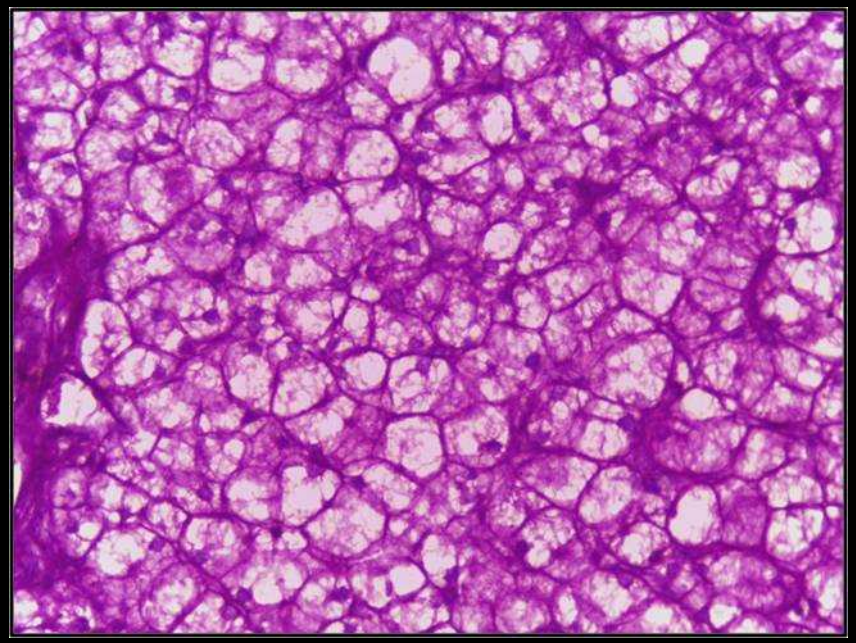
- Total cholesterol 104mg/dl
- Sr.TGL 321 mg/dl .
- Sr.lactate 40 mg/dl elevated
- Sr.uric acid 3.3 mg/dl
- Urine metabolic screening : normal

- Sr.cpk – 115mg/dl normal
- Viral markers – negative
- Usg abdomen –Massive hepatomegaly with mild splenomegaly both kidneys normal no free fluid seen
- Xray skeletal survey –normal
- ECHO – normal
- OPTHALMIC EVALUATION – no cherry red spots seen
- UGI SCOPY – lax LES

LIVER BIOPSY

- No significant portal inflammation
- Portal tract and central venules relationship was preserved.
- Microvascular steatosis.
- No significant fibrosis
- PAS positive
- Diastase sensitive
- IMP – storage disorder most consistent with glycogen storage disorger
- HPE – enlarged hepatocytes with rarified cytoplasmic glycogen stained with PAS and DIASTASE – s/o glycogenosis

GLYCOGEN STORAGE DISORDER



SUMMARY

- Doll like facies
- Massive hepatomegaly
- Hypertriglyceridemia
- Raised lactate
- Normal CPK
- Echo and opthalmic evaluation – N
- Liver biopsy – glycogenosis

GSD

Hepatic

- 1 – Von Gierkes
- 111-Coris
- 1V -Anderson
- V1-Phosphorylase
- 1X- Phosphorylase kinase

Muscle

- 11 – Pompes
- V - Mcardles
- V11 –Tauris

TYPES	Hypoglycemia	Uric acid	lactate	hyperlipidemia	others	
I	+++	Raised	Raised	++	Diarrhoea Neutropenia Platelet dysfunction	Renomegaly No splenomegaly
III	+	normal	normal	++++	Raised liver enzymes	splenomegaly
IV	no	normal	normal	normal	Cirrhosis/ PHT	HSM FTT
VI	+/-	normal	normal	++	hepatomegaly	
IX	+/-	normal	N/raised	-/+	Hepatomegaly	

type	Glycogen	PAS	Diastase	Fibrosis	fat
I	+	+	Sensitive	-	+
III	+	+	Sensitive	+++	-
IV	+(basophilic inc	+	Partial resistant	In overt cirrhosis	-
VI	+	+	sensitive	-	+/-
IX	+	+	sensitive	-	-

TYPE VI GSD

TYPE VI GSD

- Liver phosphorylase enzyme def
- Benign course
- Asymptomatic Hepatomegaly growth retardation
- Improves with age
- Symptomatic treatment

TYPE IX GSD

- Phosphorylase kinase def
- PHKA1 ,PHKA2,PHKG2
- PHKB
- Short stature, abdominal distension
- Improves with age

- TREATMENT
- No hypoglycemia – no management
- Hypoglycemia in type I – cornstarch supplementation
- Hypoglycemia in type III – protein supplementation
- Hypoglycemia – type VI&IX– carbohydrate, protein supplementation
- Hyperlipidemia – HMG CoA reductase inhibitors and fibrates
- Liver transplantaion - type 1 and type 111

TAKEHOME MESSAGE

- Asymptomatic hepatomegaly.....GSD
- Liver biopsy - Diagnosis and classification of types of GSD

THANK YOU