
INFORMATION
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HISTORY

- 1 year female child
- 3 rd degree consanguineous marriage

- Fever – High grade, without localizing symptoms - 1 week

- Day 5 of fever, 1 episode of generalised seizures 10 min

- Treated with parenteral antibiotics for 5 days
  (Inj.Mero & Inj.Amikacin)

  hence referred for further evaluation.

- History of progressive abdominal distension – since last 4 months

- History of poor weight gain+ - appetite good

HISTORY

- Antenatal history : nil significant

- Birth History :

  Full term, Normal vaginal delivery,

  Birth weight : 3.7 kgs, Cried imm.after birth

- Neonatal history :

  Day 1 of life, admitted in NICU, for seizures.

  (? Hypoglycemic seizures)

  Was treated with Phenobarbitone till 3 months of life.

Past history

History of recurrent hospitalisation for infections since early infancy.

- At 2 months of age : Underwent treatment for umbilical sepsis (drainage of umbilical abscess)

- At 5 months of age : hospitalised for UTI

  Incidentally found to have Hepatomegaly and evaluated.

  In view of Positive Mantoux, she was treated with ATT for 6 months.

- Developmental history : Predominant motor delay present
- Family history: nil significant
- No h/o contact with TB

SUMMARY

- 1yr old, 3rd degree consanguineous marriage
- History of Neonatal seizures
- History of Motor Developmental delay
- Progressive abdominal distension
- Recurrent hospitalisation for infections

- Short febrile illness with seizures

DIAGNOSTIC POSSIBILITIES??

On examination

	Observed	Expected	Percentile
Length	64 cm	75 cm	< 3 rd Percentile
Weight	6.3 kg	10.5	< 3 rd percentile
Head circumference	44.5cm	45	50 th percentile

Chubby cheeks

- P/A – Distended, non tender, Liver 6 cm below Rt. Costal margin, Span : 11cm Firm in consistency.
- CNS – Mild Hypotonia, no focal deficit, Fundus – Normal
- CVS – S1,S2 Present, No murmur
- RS – NVBS+

DIAGNOSTIC POSSIBILITIES??

Course in the Hospital

- After a few hrs of admission,
- Developed Focal Seizures followed by secondary generalisation lasting for more than 20minutes
- Dx – 130 mg/dl
- Treated with lorazepam, Fosphenytoin and Intubated

- Shifted to PICU, commenced on Mechanical ventilation.
- Persisting seizures – required leviteracetam
- Documented hypoglycemia in PICU – treated with dextrose
- Started on broadspectrum antibiotics pending cultures

TC – 9500 P5 L95 (ANC – 495) **Hb – 10.2** **Platelet – 5.7 lakhs**	**RBS – 155** **Urea – 22, Creatinine – 0.36** **Na – 127, K – 3.6, HCO3 – 28**
Bilirubin – 0.3 SGOT – 621 SGPT – 137 SAP – 414 GGT – 184 Albumin – 3.7 gm/dl	
INR – 0.97	
Urine microscopy – normal	
Chest x-ray – normal	

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Blood culture	No growth
Urine culture	No growth
CSF (Glucose / Protein / Cell count)	113 / 15 / nil
CSF CULTURE	No growth
HIV ELFA	Non reactive
IMMUNOGLOBULINS (IgA / IgG / Ig M)	199,1040,185
FLOW CYTOMETRY - T / B /NK CELLS	low absolute counts in all subsets, normal %

Serum Triglycerides	577 mg/dl
Serum Uric acid	8.2 mg/dl (2 – 5.5 mg/dL)
Serum Lactate	8.6 mmol/L (0.63 – 2.4 mmol/ L)
Serum Ammonia	88 Umol /L (21 – 50 umol/L)

USG Abdomen – marked hepatomegaly (fatty change), mild bilateral nephromegaly, 4 mm calculus in right calyx.

MRI Brain – Posterior aspect of the pons, splenium of corpus collasum, right thalamus and periventricular white matter of both cerebral hemispheres show diffusion restriction without any contrast enhancement

ECHO - normal

Doll like facies,

Marked hepatomegaly

Hypoglycemia

Hyperuricemia

Hypertriglyceridemia

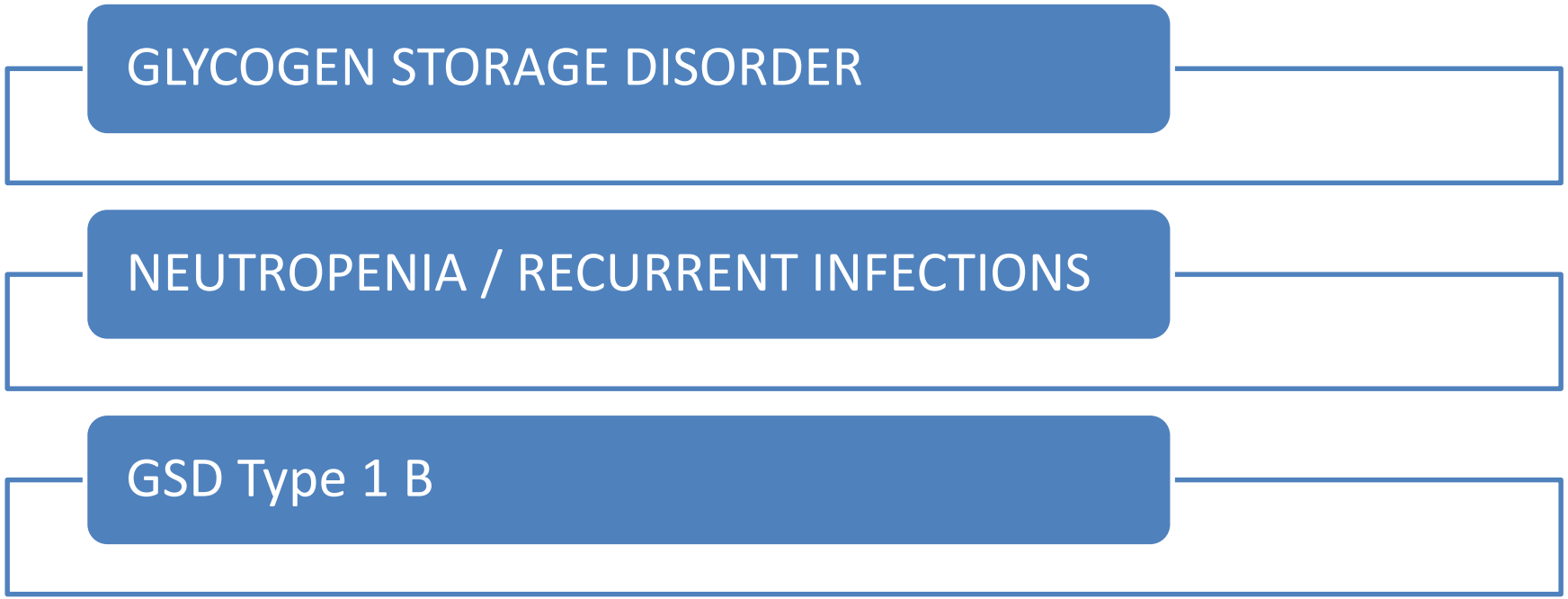
Increased lactate

Elevated liver enzymes

USG – fatty liver, b/l nephromegaly

Recurrent infections

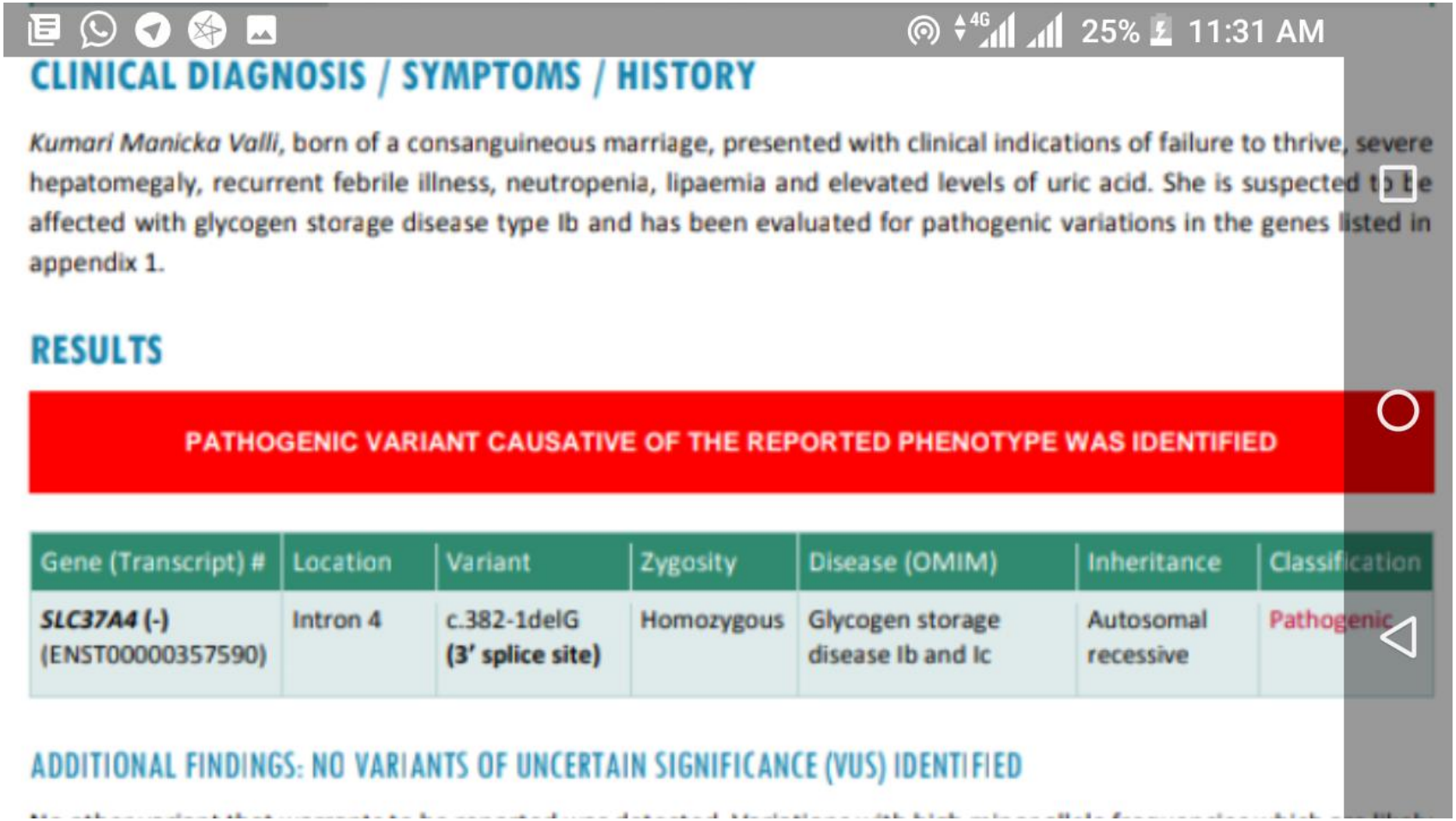
Neutropenia


GLYCOGEN STORAGE DISORDER
NEUTROPENIA / RECURRENT INFECTIONS
GSD Type 1 B

- Treated with G-CSF
- Started on corn starch diet
- Genetic counselling done & sample sent for molecular analysis

- Readmitted with fever, lethargy, hypoglycemia
- Adhesive intestinal obstruction/abdominal sepsis/pneumonia
- Succumbed

Confirmed GSD type 1 b



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CLINICAL DIAGNOSIS / SYMPTOMS / HISTORY

Kumari Manicka Valli, born of a consanguineous marriage, presented with clinical indications of failure to thrive, severe hepatomegaly, recurrent febrile illness, neutropenia, lipaemia and elevated levels of uric acid. She is suspected to be affected with glycogen storage disease type Ib and has been evaluated for pathogenic variations in the genes listed in appendix 1.

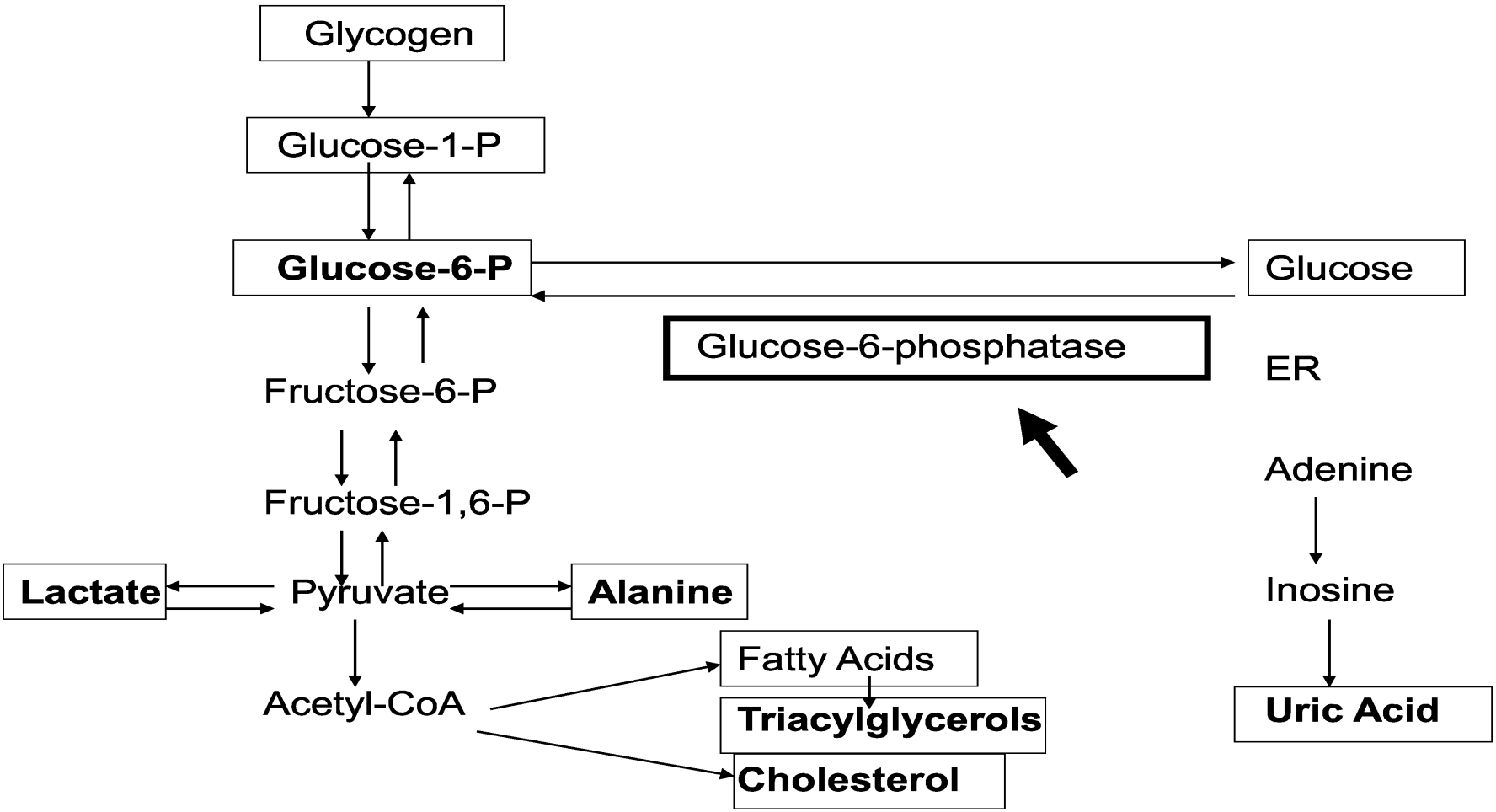
RESULTS

PATHOGENIC VARIANT CAUSATIVE OF THE REPORTED PHENOTYPE WAS IDENTIFIED

Gene (Transcript) #	Location	Variant	Zygosity	Disease (OMIM)	Inheritance	Classification
***SLC37A4* (-)** (ENST00000357590)	Intron 4	c.382-1delG **(3' splice site)**	Homozygous	Glycogen storage disease Ib and Ic	Autosomal recessive	Pathogenic

ADDITIONAL FINDINGS: NO VARIANTS OF UNCERTAIN SIGNIFICANCE (VUS) IDENTIFIED

Hepatic Glycogen Metabolism in GSD 1



GSD I a	GSD I b	GSD III
HYPOGLYCEMIA	HYPOGLYCEMIA	HYPO GLYCEMIA
LACTIC ACIDOSIS	LACTIC ACIDOSIS	NORMAL LACTATE
HYPER URICEMIA	HYPER URICEMIA	NORMAL URIC ACID LEVELS
HYPERLIPIDEMIA	HYPER LIPIDEMIA	-
NEUTROPENIA RARE	NEUTROPENIA COMMON	-
HEPATIC ADENOMA	HEPATIC ADENOMA	-
OSTEOPENIA	OSTEOPENIA	-
RENAL DISEASE	RENAL DISEASE	-
MYOPATHY - RARE	MYOPTHY - RARE	SKELETAL MYOPATHY
HEPATOMEGALY	HEPATOMEGALY	HEPATOMEGALY
SHORT STATURE	SHORT STATURE	SHORT STATURE


Thank You