GALACTOSAEMIA- CASE REPORT

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ABSTRACT

BACKGROUND
Galactosaemia is an autosomal recessive disease caused by deficiency of galactose-1-phosphate uridylytransferase deficiency (GALT) and presenting in newborns or young infants as jaundice, hepatomegaly, convulsions, lethargy, irritability, feeding difficulties, poor weight gain, aminoaciduria, cataracts or mental retardation.

CLINICAL HISTORY
A 1½ months old male child had been brought to the pediatric emergency ward with history of fever, failure to thrive and yellowish decolourisation of eyes and urine.

Clinical Findings- Baby icteric, hepatomegaly present. Patient was investigated and the results were- Liver function tests abnormal with high bilirubin and enzyme levels. Urine is positive for galactose tested by osazone test and mucic acid test. Urine chromatography showed high levels of galactose.

CONCLUSION
Early diagnosis and dietary restriction of galactose can prevent serious morbidity and mortality.

KEYWORDS
Galactose-1-Phosphate Uridylytransferase, Cataract, Jaundice, Liver Cell Failure, Mucic Acid, Galactokinase.


BACKGROUND
Galactosaemia is an autosomal recessive disorder due to deficiency of galactose-1-phosphate uridylytransferase deficiency of carbohydrate metabolism. Classical galactosaemia usually presents with life-threatening complications like jaundice, hepatosplenomegaly, hepatocellular insufficiency, food intolerance, hypoglycaemia, renal tubular dysfunction, muscle hypotonia, sepsis and cataract. Affected infants are normal at birth and clinical manifestations develop a few days after the infant has started to have milk feeds.

Case Report
A full-term 1½ months old male infant delivered per vaginum normally was brought to the paediatric casualty with fever, poor feeding, vomiting, abdominal swelling and failure to thrive. Family history- No history of any diseases running in families. On examination- Baby was lethargic, hypotonic, icteric and febrile. CVS and RS examination- Normal. Abdomen examination- Hepatomegaly present.

Investigations
1. CBC- Anaemia.
2. Liver function test- Total bilirubin- 12.7 mg/dL; direct bilirubin- 7.3 mg/dL; indirect bilirubin- 5.4 mg/dL; AST- 221 IU/L; ALT- 104 IU/L; ALP- 312 IU/L; total protein- 5.6 g/dL; albumin- 3 g/dL; globulin- 2.6 g/dL.
3. Tandem mass spectrometry done to rule out metabolic defects in amino acid metabolism, organic acids, fatty acids and urea cycle. TMS was normal.
4. Gas Analysis - Galactostix strip positive, mucic acid test positive.
5. Gas chromatography-Mass Spectrometry (GC-MS) showed increased galactose, galactitol levels.

Treatment
Baby was put on galactose-free diet. Baby’s general condition didn’t improve as liver cell failure had set in.

Mucic Acid Test for Galactose
DISCUSSION
Classical galactosaemia is caused by a deficiency of galactose-1-phosphate uridyltransferase (GALT). Other enzyme deficiencies causing galactosaemia are Galactokinase (GALK), UDP galactose 4-epimerase (GALE). Clinical presentation may vary from life-threatening complications to normality without any complications. GALT deficiency presents early and occurs within the first week after milk ingestion with life-threatening illness. Clinical presentation includes vomiting, jaundice, sepsis, poor feeding resulting in poor weight gain. Life-threatening complications are due to osmotic damage occurring as a result of galactitol accumulation. Lethargy and hypotonia indicate galactitol deposition in brain tissue. In GALT deficiency, babies present with hepatocellular disease-causing jaundice and clotting abnormalities, sepsis, renal tubular disease-causing albuminuria, aminoaciduria, galactosuria and rarely haemolytic anaemia. Cataracts are mild in the first few weeks of life. Long-term complication like defective cognition, ovarian failure and ataxia. In GALK deficiency, bilateral cataract maybe the clinical presentation. Also, children may present with deposits in the macula, seizures or pseudotumour cerebri. GALE deficiency have raised galactose-1-phosphate levels and the children may show normal growth and development without any renal disease and cataract. Classical galactosaemia is diagnosed based on suspicion on all infants presenting with liver disease, jaundice, recurrent vomiting and failure to thrive. Diagnosis is by raised galactose, galactitol levels in blood by GC-MS, (fluorescent spot test) Beutler test for GALT activity. Molecular diagnosis is confirmed by quantitation of galactose-1-phosphate uridyltransferase (GALT).

CONCLUSION
Our patient is a case of classical galactosaemia presented with sepsis, liver cell failure and failure to thrive. Measurement of galactose-1-phosphate uridyltransferase activity in erythrocytes is the standard diagnostic tool for classic galactosaemia. As the enzyme quantitation is not accessible, the diagnosis of classic galactosaemia in our case was based on clinical features and accessible laboratory results. Early diagnosis of galactosaemia as early as within 15 days of birth can prevent morbidity and mortality in most situations.

REFERENCES