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A Case Report of Galactosaemia: Atypical Presentation.

Dinesh Kumar R*, Mythili SV, Manjuladevi AJ, Shanthi B, and Kalaiselvi VS

Department of Biochemistry, Sree Balaji Medical College and Hospital, Chrompet, Chennai-600044, Tamil Nadu, India.

ABSTRACT

Galactosemia is an autosomal recessive disorder caused by deficient or absent activities of any of three enzymes involved in galactose metabolic pathway. Classical galactosemia is an inborn error of metabolism caused by a deficiency of galactose-1 phosphate uridyl transferase. At the paediatrics department in sreebalajimedical college child was brought by mother for the complaint of low weight gain (body weight was 3.0 Kg)and occasional vomiting episodes present. On examination she had delay in motor functions and on abdominal palpation hepatomegaly was found. No complaints of any feeding difficulty. Follow up of the baby revealed the growth arrest and was further evaluated (body weight was 3.2 Kg). Child was on lactose-containing feeds. Deficiency of Galactose-1-Phosphate uridyltransferase activity and a significant elevation in red cell galactose- 1-phosphate was found out. These results were confirmatory to conclude with diagnosis of galactosaemia Galactose-1-Phosphate uridyltransferasedeficiency.molecular spectrum can also be used to confirm the diagnosis. Galactosaemia can present late with much milder clinical features than the classical form of the disease. Abnormalities in LFTs, particularly AST, were not as derranged as is usual in the classical form of the disorder. Hence it is essential for every clinician to assess the course of the disease named galactosemia so that we can reduce the children mortality by diagnosing and treating the condition.

Keywords: galactose-1-phosphate uridyltransferase, galactosemia

Corresponding author

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INTRODUCTION

Galactosemia is a rare genetic metabolic disorder that affects an individual's ability to metabolize the sugar galactose properly. Galactosemia occurs due to autosomal recessive mode of inheritance that confers a deficiency in an enzyme responsible for adequate galactose degradation.

Brief case history and biochemical measurements

A girl baby was full term delivery by means of lower abdomen Caesarean section and the indication is foetal distress, with (9/10) as Apgar score and birth weight is 2.7kg. She was on breast feeding and was on lactose-rich feeds from birth.

At 3 Months of birth

At the paediatrics department in Sree Balaji Medical College child was brought by mother for the complaint of low weight gain (body weight was 3.0 Kg) and occasional vomiting episodes present. On examination she had delay in motor functions and on abdominal palpation hepatomegaly was found. No complaints of any feeding difficulty. The preliminary laboratory investigations were as follows: Normal Renal function, Blood gases was normal. Normal liver function tests, no jaundice in total blood count Macrocytic anaemia was detected, in Urine examination is acidic.

At 7 Months of birth

Follow up of the baby revealed the growth arrest and was further evaluated (body weight was 3.2 Kg). Child was on lactose-containing feeds. On examination the baby was icteric mildly with hepatomegaly, hypotonia [2] with motor delay which was mild, and grade 3/6 systolic murmur. Cheeks swollen and facial features like mild dysmorphism, which are often associated with glycogen storage diseases, mucopolysaccharidoses or congenital defects of glycosylation.

Investigations

Haematology

Macrocytic anaemia present Red cell B12 &folate Normal Sickle cell disease analysis Negative Prothrombin time was Normal

Biochemistry

Blood

Sl.no	Investigations	Results
1.	Total bilirubin	21 µmol/L (<18)
2.	ALP	438 U/L (60-330)
3.	ALT	44 U/L (12-47)
4.	AST	92 U/L (20-65)
5.	Alpha-fetoprotein (AFP)	653 kU/L (0-10)
6.	Creat kinase	Normal

LDH values are Normal, Cortisol (9 am) and Thyroid functions were normal.

Urine examination

Albumin and sugar present,qualitative test for galactose positive, a generalised aminoaciduria (qualitative partition chromatography) and marked increased urinary excretion of calcium was detected.Sugar chromatography showed galactose>48 mmol/L .normal=(0 – 1)

Differential diagnosis

- Fanconi-Bickel
- Mucopolysaccharidoses (MPS)
- Congenital defects of glycosylation
- Galactosemia

Confirmatory tests

Red cell galactose-1-phosphate uridyltransferase activity = 1.8 µmol/hr/g Hb (normal 18-40; homozygotes 0-6) heterozygotes 9-15;methodology – (Liquid chromatography –tandem mass spectrometry)

Red cell galactokinase is normal.

Deficiency of Galactose-1-Phosphate uridyltransferase activity and a significant elevation in red cell galactose- 1-phosphate was found out. These results were confirmatory to conclude with diagnosis of galactosaemia Galactose-1-Phosphate uridyltransferase deficiency, molecular spectrum can also be used to confirm the diagnosis [1].

DISCUSSION

Classical galactosaemia, is caused by deficiency of Galactose-1-Phosphate uridyltransferase. Children with this disorder typically present with symptoms during the first week of life, with poor breast feeding, poor weight gain, vomiting episodes, diarrhoeal manifestations, hepatomegaly and jaundice and hypotonia. Rare symptoms include excessive bruising, Cataracts, encephalopathy. Treatment module can be with intravenous fluid infusion and a galactose-free diet. Hepatic dysfunction occurs typically and is characterized by abnormal clotting functions and LFTs, conjugated hyperbilirubinaemia, Galactosuria and glycosuria are present in most cases, with albuminuria or aminoaciduria due to impairment in glomerular and proximal tubule functions.

TREATMENT

The treatment is to advise a galactose-restricted diet [3]. Symptoms and signs usually regress in one to two weeks following commencement of the restricted diet. The children are monitored for dietary compliance by measurement of red cell galactose-1-phosphate. Female patients are followed up with appropriate endocrinological investigations during puberty to determine whether hormone replacement therapy is required to maintain normal pubertal development or, in later life, as treatment for infertility. Ophthalmologic evaluation are done regularly for the detection of ophthalmic diseases like cataract.

SUMMARY

Galactosaemia can present late with much milder clinical features than the classical form of the disease. Abnormalities in LFTs, particularly AST, were not as derranged as is usual in the classical form of the disorder.

CONCLUSION

Hence it is essential for every clinician to assess the course of the disease named galactosemia so that we can reduce the children mortality by diagnosing and treating the condition.

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