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(CASE REPORT)

# Unveiling an atypical presentation of galactosemia: A case report without hepatic involvement

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## Abstract

Galactosemia usually presents with hepatomegaly, jaundice, and liver dysfunction. Here, we describe a case of galactosemia in a neonate lacking hepatomegaly, leading to a delayed diagnosis. Our comprehensive analysis underscores the importance of considering atypical manifestations during the diagnostic process to facilitate early intervention and prevent complications. This case highlights the significance of vigilant assessment in recognizing uncommon presentations of galactosemia, ensuring timely management and improved outcomes for affected individuals.

**Case Report:** We present a case of a full-term male neonate, 3.4 kg in weight and 4 days of age, born to consanguineous parents with an uneventful prenatal history admitted to Paediatrics nursery. Patient was diagnosed with E. Coli sepsis then further workup confirmed diagnosis of classical galactosemia with an unusual absence of hepatomegaly, underscoring the diagnostic challenges encountered due to atypical clinical manifestations.

**Conclusion:** Atypical presentations of rare metabolic disorders such as galactosemia can confound diagnostic efforts. This case report emphasizes the significance of vigilance and a comprehensive metabolic workup in neonates presenting with unexplained symptoms even in the absence of classic clinical features. Early diagnosis and prompt initiation of therapy are imperative to mitigate the risk of long-term squealed associated with untreated galactosemia.

**Keywords:** Galactosemia; Autosomal Recessive Disorder; Atypical presentation; Absence of hepatic involvement; Hypoglycemia; Galactose-1-phosphate uridyltransferase (GALT).

## 1. Introduction

Galactosemia is an autosomal recessive disorder caused by the deficiency of enzymes involved in galactose metabolism, leading to the accumulation of galactose and its metabolites [1]. Classic galactosemia is due to deficiency of galactose-1-phosphate uridyltransferase (GALT) activity [3]. In classic galactosemia patient may have homozygous or compound heterozygous form of GALT gene [4]. Over 300 mutations have been identified to date [6]. The most prevalent pathogenic variant among individuals of European descent is c.563A>G (p.Gln188Arg), comprising 65% of all

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galactosemic alleles [6]. The next common variant is c.855G>T (p.Lys285Asn), both variants are associated with low GALT activity and a poor prognosis [6]. The characteristic clinical findings of classic galactosemia include poor feeding, jaundice, cataract, hypoglycemia, elevated liver enzymes, renal Fanconi syndrome and E coli sepsis (in some babies) [5]. Partial GALT deficiency presents a clinical spectrum that ranges from being clinically silent to milder cases with potential developmental deficits, rather than life-threatening complications [3]. We present a case of galactosemia with an unusual absence of hepatomegaly, underscoring the diagnostic challenges encountered due to atypical clinical manifestations.

## 2. Case Presentation

A full-term male neonate, 3.4 kg in weight and 4 days of age, born to consanguineous parents with an uneventful prenatal history admitted to Paediatrics nursery department at Naseer Teaching hospital, Peshawar, KP on 8 august 2023. Patient presented with fits, fever, vomiting and drowsiness for the last 2 days. Baby was born with normal vaginal delivery with delayed cry. Upon examination, the patient was febrile and exhibited poor feeding, lethargy, hypotonia, tachypnea, grunting and a normal liver span and no other positive systemic findings. Fontanelle was bulging with occipital frontal circumference was 33cm. Initial laboratory investigations revealed hypoglycemia, high C-reactive protein and normal serum transaminases, differential diagnosis of neonatal sepsis, hypoxic-ischemic encephalopathy (HIE) and Neonatal Seizure were made then on blood culture E coli growth was seen. The patient was treated accordingly for neonatal sepsis as a result the fever subsided but hypoglycemia, fits and drowsiness persisted. Patient had multiple episodes of hypoglycemia despite of appropriate intravenous fluid. Ultrasound abdomen shows liver span of 5.7cm with smooth margins and homogenous heterozygous parenchymal echo pattern. No evidence of focal solid or cystic mass lesion. Portal vein was normal. Common bile duct was normal. No evidence of intra-hepatic biliary channels dilatation. Skull ultrasound shows no positive findings. Serum albumin, Serum creatinine, Urea and Thyroid stimulating hormone (TSH) were in normal range. Then, further metabolic workup was done, including tandem mass spectrometry and urinalysis showing elevated galactose levels which pointed to a diagnosis of galactosemia. Genetic testing was not performed due to unavailability of the testing service. Immediate initiation of a galactose-restricted diet resulted in rapid clinical improvement with resolution of feeding difficulties and normalization of biochemical markers and fits. Long-term follow-up for developmental milestones and intellectual outcomes was commenced.

**Table 1** Lab investigations of patient

| Date of Tests | Reference range | 8/8/23 | 9/8/23 | 11/8/23 | 16/8/23 |
|---|---|---|---|---|---|
| Hemoglobin (g/dl) | 12.3 – 16.6 | 15.2 | 15.3 | - | 14.0 |
| RBC (10.e3/uL) | 4.25 - 6.02 | 4.30 | 4.33 | - | 4.13 |
| Haematocrit (%) | 38 - 50.7 | 42.5 | 43.1 | - | 41.3 |
| MCV (fL) | 78.7 - 96.3 | 98.8 | 99.5 | - | 100.0 |
| MCHC (g/dL) | 30 - 35.5 | 35.8 | 35.5 | - | 33.9 |
| TLC (10.e3/uL) | 4.8 – 11.3 | 11.99 | 8.74 | - | 7.39 |
| Platelets (10.e3/uL) | 154 – 433 | 48 | 200 | - | 179 |
| SCA+ (mg/dl) | 8.5 - 10.5 | 8.1 | - | 9.0 | 9.4 |
| CRP(ng/L) | <5 | 122.32 | - | - | - |
| Bilirubin (mg/dL) | Up to 1.0 | - | 1.2 | - | - |
| Serum ALP (Units/L) | Children 1200 | - | 138 | - | - |
| Serum ALT (Units/L) | up to 40 | - | 22 | - | - |
| Sodium (mmol/L) | 135 – 148 | 144.0 | - | - | 140.4 |
| Potassium (mmol/L) | 3.5 - 5.5 | 4.16 | - | - | 5.46 |
| Chloride (mmol/L) | 98 – 107 | 108.7 | - | - | 110.7 |

**Table 2** Random Blood Sugar (RBS) of patient

| Date and time of RBS | 9-10/8/2023 | 11/08/2023 | 15/08/2023 |
|---|---|---|---|
| 04:00 pm | 38 mg/dl | - | - |
| 06:00 pm | 61 mg/dl | - | - |
| 08:00 pm | - | 65 mg/dl | 99 mg/dl |
| 10:00 pm | 97 mg/dl | 49 mg/dl | - |
| 12:00 am | 34 mg/dl | - | - |
| 02:00 am | 45 mg/dl | 100 mg/dl | - |
| 04:00 am | 68 mg/dl | - | 72 mg/dl |
| 07:00 am | 97mg/dl | 44 mg/dl | 72 mg/dl |
| 09:00 am | 188 mg/dl | 34 mg/dl | 57 mg/dl |
| 12:00 pm | 27 mg/dl | - | 132 mg/dl |
| 02:00 pm | - | 285 mg/dl | 102 mg/dl |

## 3. Discussion

Galactosemia is a rare autosomal recessive disorder caused by the deficiency of galactose-1-phosphate uridyltransferase (GALT) leading to impaired galactose metabolism [1]. Classic galactosemia is a severe and life-threatening condition that can lead to serious morbidity and death if not diagnosed and treated early in life [6]. Typically, galactosemia manifests with hepatic involvement presenting as hepatomegaly, jaundice, and liver dysfunction [2]. However, rare cases can deviate from this classical presentation posing diagnostic challenges. This case report sheds light on an unusual presentation of galactosemia without hepatic involvement emphasizing the importance of considering atypical manifestations in diagnosis and management. This case highlights the diagnostic challenge posed by an atypical presentation of galactosemia without hepatomegaly, jaundice and cataract. The absence of hepatomegaly and jaundice in the neonate underscores the importance of considering galactosemia in the differential diagnosis of neonates presenting with nonspecific symptoms. The delayed diagnosis in this case underscores the need for heightened awareness among healthcare professionals to prevent potential complications.

## 4. Conclusion

Atypical presentations of rare metabolic disorders such as galactosemia can confound diagnostic efforts. This case report emphasizes the significance of vigilance and a comprehensive metabolic workup in neonates presenting with unexplained symptoms even in the absence of classic clinical features. Early diagnosis and prompt initiation of therapy are imperative to mitigate the risk of long-term squealed associated with untreated galactosemia.

## Compliance with ethical standards

### *Disclosure of conflict of interest*

No conflict of interest to be disclosed.

### *Statement of ethical approval*

The local Institutional Review Board deemed the study exempt from review.

### *Statement of informed consent*

Written informed consent was obtained from the patient's father for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-chief of this journal.

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