




Letter: Tyrosinemia Type 1 with Cytomegalovirus Infection in Infancy

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J Gastrointest Infect 2024;14:35–36.

Chronic liver disease in infancy is characterized by changes in hepatic architecture that may lead to the development of cirrhosis and portal hypertension.¹ The etiologies of chronic liver diseases in this age group include infections, biliary atresia, metabolic diseases, and neoplastic diseases. Detailed assessment is needed to determine the prognosis and treatment of chronic liver disease.²

A 6-month-old male child, born from a third-degree consanguineous marriage, presented to the pediatric outpatient clinic with abdominal distension, poor feeding, and intermittent fever for the last 3 months. The child was born at 37 weeks of gestation following spontaneous onset of labor and had birth weight of 2.8 kg. Antenatal ultrasound showed evidence of polyhydramnios. However, there was no history of maternal infection or fever. On examination, the child was weighing 6 kg and was irritable. The liver was palpable 3 cm below costal margin and free fluid was noted. Complete blood count revealed hemoglobin of 8.9 g/dL, total leucocyte count of 14,300/cu.mm, and platelets of 1.33 lakh per cu.mm. Liver function tests showed serum bilirubin of 2.7 g/dL, alanine aminotransferase of 68 IU/L, aspartate aminotransferase of 57 IU/L, serum albumin of 3.2 g/dL, serum globulin of 2.8 g/dL, and international normalized ratio of 2.4. Ultrasound abdomen showed liver size of 7 cm with coarse echotexture and multiple nodules measuring up to 8 mm in both lobes of liver. Spleen was enlarged (8 cm) and moderate ascites were noted. Computed tomography scan showed chronic liver disease with moderate ascites, and no space-occupying lesions in liver. Serological tests revealed

immunoglobulin G antibodies against cytomegalovirus (CMV) to be positive and CMV DNA polymerase chain reaction in urine showed more than 50 lakh copies/mL. Serum alpha fetoprotein levels (AFP) were more than 1 lakh. Other serological tests for hepatitis B, hepatitis C, autoimmune liver disease, and Wilson disease were negative. In view of extremely high AFP levels, tyrosinemia was suspected. Urine test (gas chromatography-mass spectrometry) showed elevated levels of succinyl acetone (test: 170.93, reference: 0%) and 4-hydroxyphenyllactic acid (test: 2,147.71, reference range: 1.8%) suggestive of tyrosinemia type 1.

The parents were explained regarding need to start valganciclovir therapy for CMV infection and genetic counseling was advised for tyrosinemia. Liver biopsy was planned after correction of coagulation profile. Since the baby had been started on top feeds, mother was advised to give a low-protein diet. She was instructed to avoid meat, eggs, cheese, milk, dals, dried beans, nuts, and soya. Hearing and ophthalmology examination was advised. In view of financial constraints, the parents did not pursue further tests and left against medical advice.

Occurrence of any congenitally acquired infection with tyrosinemia is rare. Association of tyrosinemia type 1 with CMV infection is extremely uncommon and was first described in 1978.³ Few other case reports have highlighted this rare entity.^{4,5} Detection of tyrosinemia is common in the West due to newborn screening programs for metabolic diseases. Our patient had tyrosinemia and CMV infection with very high viral load. Liver biopsy would have helped to diagnose presence of CMV hepatitis by documenting viral

received

August 20, 2023

accepted

August 24, 2023

article published online

November 9, 2023

DOI <https://doi.org/10.1055/s-0043-1776059>.

ISSN 2277-5862.

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inclusion bodies. However, this could not be done at presentation as coagulation parameters were deranged.

This case highlights an uncommon diagnostic conundrum—tyrosinemia with CMV infection.

Patient Consent

Consent obtained from the patient's parents, no patient identifiers used.

Ethical Clearance

Informed to ethics board of the hospital. Clearance waived off in view of no use of photographs/ images/ patient identifiers.

Authors' Contributions

All authors contributed equally to the article.

Data Availability

Data can be shared by the author on a reasonable request.

Funding

None.

Conflict of interest

None declared.

Acknowledgments

None.

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