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Rare DCDC2 Mutation Causing Renal-Hepatic Ciliopathy

Authors : Atitallah Sofien, Bouyahia Olfa, Attar Souleima, Missaoui Nada, Ben Rabeh Rania, Yahyaoui Salem, Mazigh Sonia, Boukthir Samir

Abstract : Introduction: Ciliopathies are a spectrum of diseases that have in common a defect in the synthesis of ciliary proteins. It is a rare cause of neonatal cholestasis. Clinical presentation varies extremely, and the main affected organs are the kidneys, liver, and pancreas. Methodology: This is a descriptive case report of a newborn who was admitted for exploration of neonatal cholestasis in the Paediatric Department C at the Children's Hospital of Tunis, where the investigations concluded with a rare genetic mutation. Results: This is the case of a newborn male with no family history of hepatic and renal diseases, born to consanguineous parents, and from a well-monitored uneventful pregnancy. He developed jaundice on the second day of life, for which he received conventional phototherapy in the neonatal intensive care unit. He was admitted at 15 days for mild bronchiolitis. On clinical examination, intense jaundice was noted with normal stool and urine colour. Initial blood work showed an elevation in conjugated bilirubin and a high gamma-glutamyl transferase level. Transaminases and prothrombin time were normal. Abdominal sonography revealed hepatomegaly, splenomegaly, and undifferentiated renal cortex with bilateral medullar micro-cysts. Kidney function tests were normal. The infant received ursodeoxycholic acid and vitamin therapy. Genetic testing showed a homozygous mutation in the DCDC2 gene that hadn't been documented before confirming the diagnosis of renal-hepatic ciliopathy. The patient has regular follow-ups, and his conjugated bilirubin and gamma-glutamyl transferase levels have been decreasing. Conclusion: Genetic testing has revolutionized the approach to etiological diagnosis in pediatric cholestasis. It enables personalised treatment strategies to better enhance the quality of life of patients and prevent potential complications following adequate long-term monitoring.

Keywords : cholestasis, newborn, ciliopathy, DCDC2, genetic
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