

Persistent Jaundice and Multiple Fractures in a Newborn

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CASE DESCRIPTION

A 32-year-old African American mother brought her 1-month-old baby boy to the pediatrician's office, concerned that the baby had yellow eyes that appeared to be getting worse. The mother also mentioned that the baby had irregular sleeping patterns and had suffered from a broken leg. On physical examination, the baby appeared irritable and his liver and spleen felt enlarged. However, his temperature, respirations, and heart rate were all within normal limits. The baby was born after a full-term uncomplicated pregnancy and weighed 6 pounds 8 oz at birth. The baby had 1 older sibling who was healthy. Family history showed that hypertension and diabetes were prevalent in the family.

The pediatrician noted the neonate's bilirubin concentrations at his 2-week checkup were slightly increased, but the parents were told at that time not to be concerned because increased bilirubin concentration is common in African American babies and should resolve without intervention. Results for a lipid panel at that time were within reference limits, and results for blood cultures and serology tests were all negative. Abnormal complete blood count findings included an increased red cell distribution width of 15.9% (reference interval, 11.1%–14.3%) with abnormal RBC morphology, target cells, helmet cells, hypochromasia, and anisocytes observed on the blood smear.

Palpable enlargement of the liver and spleen in conjunction with the baby's increased concentrations of liver enzymes and direct bilirubin prompted the pediatrician to order an ultrasonography examination of the liver. Underdeveloped bile ducts were noted on the ultrasonography. A liver biopsy was performed to further assess the damage and enlargement of the liver and spleen. The abnormal biopsy results led to the performance of a diagnostic intraoperative cholangiogram.

During his first few months of life, the infant suffered 2 broken legs and a broken arm that occurred during routine handling of the infant. Although he was under the care of a pediatric hepatologist, medical staff were alarmed and contacted the Department of Children's Services (DCS).

QUESTIONS TO CONSIDER
• Discuss possible etiologies for neonatal jaundice.
• What is the cause of the low vitamin D concentration?
• If the total bilirubin is increased in an infant, what testing should always be performed?

Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the December 2019 issue of *Clinical Chemistry*. To view the case and comments online, go to <http://www.clinchem.org/content/vol65/issue12> and follow the link to the Clinical Case Study and Commentaries.

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