A Baby Boy with Hypothyroidism and Hemangioendothelioma

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CASE DESCRIPTION

A full-term baby boy underwent an ultrasound scan of the abdomen at 6 weeks of age for the follow-up of an antenatal diagnosis of left-sided hydronephrosis. Although there was no hydronephrosis, the abdominal ultrasound revealed an enlarged liver with multiple hypoechoic lesions measuring up to 25 mm in diameter. He was admitted to the hospital for further evaluation of the hepatic lesions.

Examination revealed multiple small external hemangiomas on the scalp and both wrists and axilla. The infant's abdomen was distended with hepatomegaly 6–7 cm below the costal margin. There was no evidence of cardiac failure, an observation that was confirmed by a cardiology consult. His electrolytes, creatinine, urine catecholamine metabolites, α-fetoprotein, and liver enzymes were all within reference intervals, except for an increased γ-glutamyl transferase of 257 IU/L (reference interval, 7–64 IU/L). Thyroid function tests (TFT) revealed an increased thyroid-stimulating hormone (TSH) concentration of 37.7 mU/L (age-specific reference interval, 0.30 –5.00 mU/L), free thyroxine (fT4) within the reference interval at 17.9 pmol/L (age-specific reference interval, 12.0 –30.0 pmol/L), and low free triiodotyronine (fT3) of 3.3 pmol/L (age-specific reference interval, 3.8–6.0 pmol/L) (Beckman DxI, Beckman Coulter). Thyroid peroxidase and thyroglobulin antibodies were negative. The newborn TSH screening test results were within reference intervals. MRI scan of the abdomen showed multiple liver lesions consistent with hemangioendothelioma.

Questions to Consider
• What are the possible causes for discrepant TSH and fT4 results?
• What additional biochemical testing would be helpful to elucidate the cause of the discrepancy in the TSH and fT4?
• What is the most probable diagnosis?

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Final Publication and Comments

The final published version with discussion and comments from the experts will appear in the June 2014 issue of *Clinical Chemistry*. To view the case and comments online, go to http://www.clinchem.org/content/vol60/issue6 and follow the link to the Clinical Case Study and Commentaries.

Educational Centers

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