



Proceeding

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Neonatal Multiple Pituitary Hormone Deficiency: A unique presentation

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Introduction

Congenital multiple pituitary hormone deficiency (MPHD) can present with hypoglycaemia and rarely with neonatal cholestasis. Treatment with glucocorticoid and thyroxine can resolve MPDH related cholestasis.

Objective

To report an infant with MPHD, presenting with cholestatic jaundice, hypoglycaemia and high serum ferritin level suggesting neonatal hemochromatosis.

Methods

Comprehensive clinical and laboratory investigations were performed to establish the etiology of the presenting complaints. This included genetic, metabolic, infectious, as well as thorough hormonal profile (LH, FSH, PRL, TSH, FT4, ACTH and growth hormone stimulation tests).

Results

Hormonal evaluation revealed cortisol and growth hormone deficiency with central hypothyroidism. Other causes of

cholestasis were ruled out. In addition there was a high serum ferritin level of 2315ng/ml; but neonatal hemochromatosis was excluded by the absence of hemosiderin deposition in buccal mucosal biopsy. Treatment with cortisol and L-thyroxine resulted in dramatic improvement of the liver function tests, resolution of cholestatic jaundice and a significant reduction of serum ferritin level. These findings support the theory that thyroid hormone and cortisol affect the bile acid independent bile flow and deficiencies of these hormones can cause abnormalities of the biliary structure and the function of bile canaliculi essential for bile excretion.

Conclusion

To our knowledge, this is the first description of an infant with congenital MPHD presenting with cholestasis, hypoglycaemia and high serum ferritin. MPHD should be considered in any infant who presents with prolonged cholestasis and hypoglycaemia. The high serum ferritin level in this child may reflect acute phase reaction.



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