McCune–Albright Syndrome in a Male Newborn with Hyperthiroydism

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INTRODUCTION

McCune-Albright syndrome (MAS) is a rare disorder; the cardinal features include polyostotic fibrous dysplasia, precocious puberty, and large café-au-lait spots. It could be associated with other endocrinopathies: thyroid involvement is a common feature.

The prevalence is very low, being outstanding the neonatal diagnosis, especially in males. The mean age of diagnosis for MAS is 5 years, and precocious puberty is the most common presentation. It’s significantly more frequent in females.

CASE PRESENTATION

A male newborn with a café-au-lait extensive spot involving the back, the backside of arms and legs and scrotal area (Figures 1 and 2), is hospitalized in our neonatal unit at twelve hours of life with respiratory distress, which disappeared after 48 hours.

This male infant was born at 39 weeks of gestation with normal anthropometric measures, through eutenic delivery. There were no family pathologic history, neither parental blood relatives.

On day of life (DOL) 5 he presented jaundice: direct bilirubin was in 7 mg/dL, without acholicia, chyluria, hepatomegaly, or abdominal sonographic findings. Conjugated bilirubin levels were increasing up to 15.95 mg/dL on DOL 17, and total bilirubin was in 20.2 mg/dL.

On DOL 10, he presented cholestasis pattern in study of the hepatic function, with increased GGT and GPT, and normal levels of GGT. In addition, he started usodexoxycholic acid and liposoluble vitamins.

On DOL 10, we detected serum levels of thyroid-stimulating hormone (TSH) in 0.1 uIU/mL, thyroxine (T4) in 32.7 pg/ml and triiodothyronine (T3) in 6.44 ng/dL. Anti-thyroglobulin, anti-thyroxoperoxidase and anti-TSH receptor antibodies were negatives, and thyroid sonography was normal. On DOL 12, the patient started respiratory distress, increasing breathing and cardiac frequencies up to 200 bpm. Due that thyroid storm, he initiated propranolol, furosemide, and methimazole. A week later, TSH was in 0.03 uIU/mL and free T4 in 17.2 pg/ml, therefore, we optimize the dosage of antithyroid drugs.

Thyroid hormone levels become normal in approximately 30 days from the beginning of the optimized treatment.

By controlling the thyroid function, bilirubin started to decline. However, GGT and GPT continued increasing up to a maximum of 200 U/L and 499 U/L respectively after a month from the starting of antithyroid drugs, being probably consequence of the treatment. After his retirement, once normalize thyroid function, transaminases values began to decrease.

CONCLUSIONS

- MAS must be suspected in a newborn with fibrous dysplasia, café-au-lait macules and endocrinological disorders, despite the low incidence or male condition.
- We report a newborn with the spot, hyperthiroydism and polyostotic fibrous dysplasia: these items confirm the clinical diagnosis of MAS, although the genetical study of the gene GNAS1 was normal.
- Early treatment is essential for the stability of the patient; for a correct and complete management of the case it’s not enough to diagnose the entities which comprises, but the possible side effects of the drugs and their impact on children’s development.