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 others endocrinopathies: tyroid involvement is as 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 girls.

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 eutocic delivery. There were no family pathologic history, neither parental blood relatives.

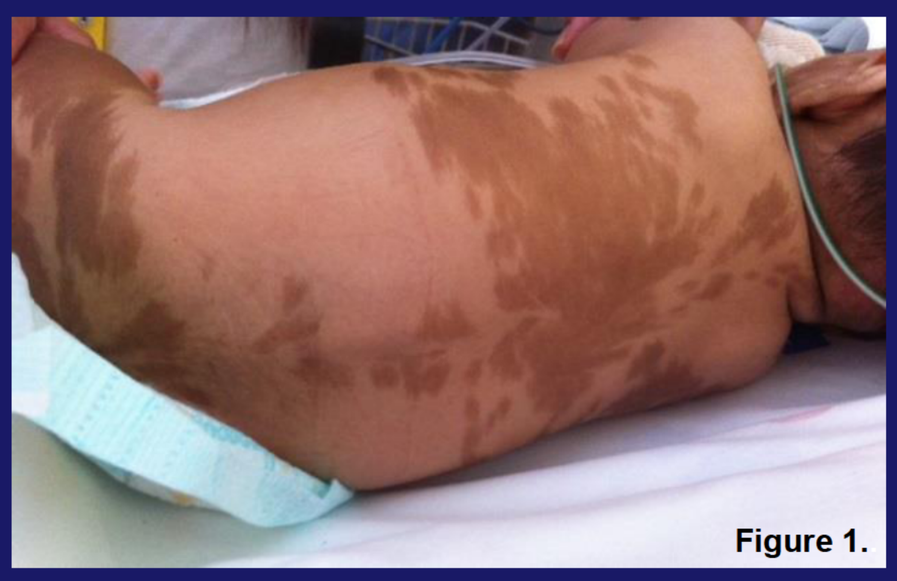


Figure 1.

Figure 2.

JAUNDICE AND CHOLESTASIS

On day of life (DOL) 5 he presented jaundice: direct bilirubin was in 7 mg/dl, without acholia, 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 GOT and GPT, and normal levels of GGT. In addition, he started usodeoxycholic acid and liposoluble vitamins.

CARDIOMEGALY

As a likely consequence of held tachycardia caused by hyperthyroid status, chest x-ray image showed cardiomegaly. The echocardiography exam revealed dilated left cardiac cavities and slight pericardial effusion, with preserved cardiac functionality. It was increased the dose of propranolol, administered at 2mg/kg per day. The evolution of the dilatation has been favorable in subsequent scans.

THYROID INVOLVEMENT

On DOL 10, we detected serum levels of thyroid stimulating hormone (TSH) in 0.1 uIU/L, thryroxine (T4) in 32.7 pg/ml and triiodothyronine (T3) in 6.44 pg/ml. Anti-thryroglobulin, anti-thryroperoxidase 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, lugol and methimazole. A week later, TSH was in 0.03 uIU/L and free T4 in 17.3 pg/ml, therefore, we optimize the dosage of antithyroid drugs.

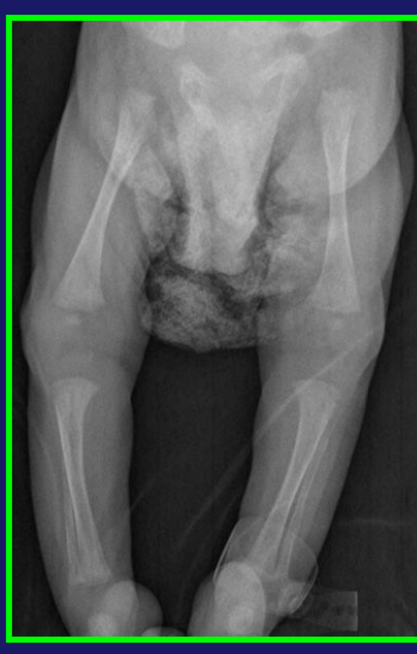
Thyroid hormone levels become normal in approximately 30 days from the beginning of the optimized treatment.

By controlling the thyroid function, bilirubin started to decline. However, GOT and GPT continued increasing up to a maximum of 280 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.

APLASTIC ANEMIA

On DOL 45, he showed erythrocytes in 2.62 x10^6/uL, haemoglobin in 6.8 g/dl and hematocrit of 21%. Perypheral blood smear and bone marrow were analized: Myeloid and megakaryocytic cell lines were normal. The diagnosis was selective erythoblastopenia, and the patient required red blood cells concentrate transfusions. It reverted at the end of the antithyroid treatment, and reappeared with the reintroduction.

FIBROUS DYSPLASIA



Radiological image showed ground glass density in the left fibula and in the right ischium: typical pattern of polyostotic fibrous dysplasia. There were not mutations in the gene GNAS1.

CONCLUSIONS

- MAS must be suspected in a newborn with fibrous displasia, café-au-lait macules and endocrinological disorders, despite the low incidence or male condition.
- We report a newborn with the spot, hyperthyroidism 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.

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Misc 2

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