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A 2-Month-old baby with typical dysmorphic facies

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Background: The Russell-Silver syndrome is a rare genetic disease classified as an imprinting disorder with epigenetic disorders, characterized by intrauterine growth retardation, body asymmetry and specific dysmorphic facies. It's clinically recognizable but its etiology appears to be heterogeneous and has a higher risk of development delay.

Case: The case study begins with two-month-old baby, who attends for low weight gain from birth, Scarce appetite. Mother refers to recurrent respiratory problems, No vomiting or cyanosis. Personal history: 39+5 weeks of gestation, CIR, Apgar 7/9, Weight: 2590 g and length: 48 cm Cephalic perimeter: 32 cm, P3 at birth. Physical examination: Good condition, Mild dysmorphic facies. Cardiopulmonary auscultation: normal and abdomen soft and depressible. Neurological exploration: partially fixes the gaze, social smile. Marked cervical and dorsal hypotonia.

Complementary explorations: With the initial diagnosis of failure of medro and hypotonia is referred to the principal hospital to complete relevant studies. Hemogram: Hemoglobin: 7.1g/dL, Hematocrit 20.2%, Leukocytes 10.54x103 mm3, Platelets 318x106. Biochemistry: Glucose 78 mg/dL, Urea 30 mg/dL, Creatinine 0.20 mg/dL, Ion Sodium 140 mmol/L, Potassium Ion 5.5 mmol/L, IGF-I (Sm-C) <25 ng/ml.

Conclusion: The initial diagnostic options in our case included from neonatal metabolic pathologies to diseases of genetic imprint, however with the results obtained in the initial analyzes, some diagnoses were ruled out and complementary tests continued. The patient was finally diagnosed as the Russell-Silver syndrome because has all the clinical criteria of the current literature and it was possible to confirm it at the molecular level, since the genetic study for this pathology is based on a methylation test using MPLA (analytical sensitivity 95-98%). Some abnormalities were detected in the 11p15.5 region and in chromosomes 7 and 14. Most of cases are sporadic and in approximately 30-40% of patients with compatible phenotype, molecular confirmation is not possible and the diagnosis is clinical.

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