Birth Defects, Dysmorphology, Skeletal Dysplasia, Craniofacial Anomalies

AB046. Russell-Silver syndrome: a case report and brief review of the literature

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Background: Russell-Silver syndrome (RSS) is a rare genetic disease classified as an imprinting disorder. It is characterized by intrauterine growth retardation, body asymmetry dysmorphic facie including triangular face and clinodactyly of 5th digits, and increased risk of development delay. RSS is clinically recognizable but its etiology appears to be heterogeneous.

Methods: We report a 2-month-old baby who was brought to medical attention due to poor weight gain since birth and poor appetite. Clinical history was explored and laboratory investigations were performed.

Results: The baby was born at 39 weeks and 5 days of gestation, via vaginal delivery. Apgar scores were 7 and 9 at 1 and 5 minutes, respectively. Birth parameters included birth weight of 2,590 g, length of 48 cm, and head circumference of 32 cm (<3rd percentile). Newborn metabolic screening was reportedly normal. The baby had history of recurrent respiratory problems but no vomiting or cyanosis. Family history was benign. Physical examination revealed mild dysmorphic facie, normal cardiac, respiratory and abdominal examination and descended testes. Neurologically, the patient could partially follow object, socially smile. There was marked cervical and dorsal hypotonia. Complete blood counts showed mild anemia Hb 7.1 g/dL, Hct 20.2%, otherwise normal results. Blood glucose and serum electrolytes were within normal ranges. Serum IGF-I (Sm-C) was <25 (55–327 ng/mL). The patient's final diagnosis was RSS based on the established clinical criteria and normal biochemical and metabolic profiles. Molecular defects underlying RSS including alterations of imprinting genes in 11p15.5, uniparental disomy of chromosomes 7 and 14 was not performed in the present case.

Conclusions: Differential diagnosis of RSS can be made based on clinical findings, followed by molecular confirmation. However, the majority of cases are sporadic and molecular confirmation is shown positive in 30–40% of the patients; therefore, the diagnosis is currently based on established clinical criteria.

Keywords: Chromosomes; case report; Russell-Silver syndrome (RSS)

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