

Reevaluation of Russell-Silver Syndrome

Russell-Silver syndrome is characterized by intrauterine growth retardation (IUGR) and postnatal growth retardation in association with asymmetry of the body, normal head size, triangular facies, and normal psychomotor development. In 1953, Silver first described two patients with IUGR, body asymmetry, and postnatal growth retardation. In 1984, Russell described another group of five patients who also had growth deficiency, triangular facies, and disproportionate shortening of the upper limbs. Only two of the five had limb asymmetry. Since these two patients appeared to have the same anomalies described by Silver, the clinical entity was named the "Russell-Silver syndrome."

Saal et al reevaluated 15 patients with Russell-Silver syndrome 2.9 to 13 years after their initial diagnosis. They observed great variability in each of the features of the syndrome, suggesting that Russell-Silver syndrome is not a discrete entity but a heterogeneous group of disorders. Most interesting are the data regarding eventual growth. Five of the 15 exhibited late catch-up growth and attained normal heights. Eight remained below the third percentile, but paralleled the growth curve. Six of the 15 had gross evidence of body asymmetry

at the time of diagnosis—four of the six continued to have a discrepancy in leg and/or arm length of more than 1 cm. One of the four had severe scoliosis.

Psychomotor development was abnormal in six of the 15. One of the six had seizures and six had café au lait spots, strongly suggesting neurofibromatosis. Psychomotor development in Russell-Silver syndrome had previously been thought to be normal, despite the frequent finding of gross motor delay in infancy. Eventual head circumference was normal in some and below the second percentile in others, unrelated to the degree of psychomotor delay. Russell-Silver patients are generally thought to have a normal head circumference. Eleven of the 14 patients originally described as having triangular facies were again so described on follow-up. Although hypogonadism and abnormal genital development had been described in several patients with Russell-Silver syndrome, all of these 15 patients had normal sexual development.

The authors conclude that the features of Russell-Silver syndrome are so diverse that it is highly probable the entity is a heterogeneous group of disorders. It is thus difficult to offer parents a clear prognosis. In addition, if the diagnosis is made too

loosely, the work-up for short stature may be prematurely terminated in some children; potentially correctable conditions could therefore be overlooked.

Saal HM, Pagon RA, Pepin MG: *J Peds* 1985;107:733.

Editor's comment—Although the Russell-Silver syndrome has been considered a well-defined form of IUGR, it is clear that patients with a variety of forms of prenatal growth retardation have been lumped under this term. Conflicting reports of responsiveness to growth hormone therapy probably reflect this heterogeneity, especially in view of the fact that one third of the patients in this series obtained normal height without therapy. In the absence of a specific laboratory diagnostic test, the delineation of heterogeneity within a syndrome is difficult, but must always be kept in mind when offering parents a prognosis. In addition, the authors are correct in emphasizing that Russell-Silver syndrome can occur concomitantly with hypopituitarism or other causes of growth retardation. Cassidy et al recently reported (*AJDC* 1986;140:155) the seventh case of growth hormone deficiency in association with Russell-Silver syndrome.