Russell-Silver Syndrome
MH Kariminejad*, MD, B. Bozorgmehr MD.

This syndrome originally was described by Silver and colleagues in 1953 and soon afterwards by Russell in 1954. The first reports were in children with characteristic facies, low birth weight, asymmetry, and growth retardation (1,2). The incidence is as high as 1 in 3000 to as low as 1 in 100000. Male to female ratio is equal (3).

At birth affected infants are unusually small for gestational age. Their pattern of growth usually parallels the normal growth curve but remains below the 3rd percentile (4). Clinical features are easier to identify in infants and younger children, particularly the small triangular facies. These findings are more difficult to recognize in adults (3).

One of the main features of this syndrome is asymmetry which is present at birth but can be quite variable in extent and degree. In some; one entire side of the body may be significantly larger than the other, while in others the asymmetry may be limited and involve only the skull, spine, and all or part of a limb (4). However, intelligence is usually normal. Because of the small facies, the upper head may appear large, though head circumference is within the normal range. This appearance, plus the relatively large fontanels in early infancy, may give rise to a false impression of hydrocephalus which they do not have (5).

Physical Findings
-Growth and skeletal: Short stature (prenatal onset), immature osseous development in infancy and early childhood, with late closure of anterior fontanel, asymmetry usually of the limbs, hemihypertrophy, clinodactyly, camptodactyly, syndactyly of second and third toes, Sprengel deformity (Congenital elevation of the scapula, due to failure of descent of the scapula to its normal thoracic position during fetal life).
-Facies: Small, triangular facies, a high forehead that tapers to a small jaw, micrognathia, prominent nasal bridge, down-turned corners of mouth, the sclera may be bluish in early infancy.
-Occasional findings: Cardiac defects, hypospadiasis, posterior urethral valves, renal anomalies, café au lait spots on skin, tendency

* Mohammad Hassan Kariminejad
1143 Med Bldg, Sanat Sq, Shahrek Ghods (Gharb) P.O.box 14665/154
Tehran-Iran/Tel:+982188363952/E-mail: mhkariminejad@mavara.com
toward excess sweating especially on the head and upper trunk during infancy,
Endocrine: -Liability to fasting hypoglycemia from about 10 months until 2 to 3 years of age.
-Malignancy: For example craniopharyngioma, testicular seminoma, hepatocellular carcinoma and Wilms tumor (4-6).

**Etiology**
The etiology of this disorder is unknown. The majority of cases are sporadic. A few cases of autosomal dominant transmission are described. Approximately 10% of patients have proven uniparental disomy (UPD). Imprinting plays a role in the clinical phenotype of Russell-Silver in these patients (3-5).
UPD is the inheritance of both alleles from the same parent and none from the other. Approximately 10% of patients may have maternal heterodisomy (inheritance of both alleles from one parent) and isodisomy (inheritance of 2 copies of a single allele from one parent) both are demonstrated (3-5).

**Medical Care**
A multidisciplinary approach is needed. Patients should have a nutritional evaluation to provide optimal calories for growth.
Growth hormone improves growth in some patients; however the final height is still less than normal. Periodic examination is indicated to rule out the presence of a tumor of the kidney, liver and testes (3,4).

**Genetic Counseling**
Most studies demonstrate that the siblings of children with Russell-Silver syndrome do not have the disorder. Therefore, risk of recurrence is expected to be minimal (3).

**References**