Case Report

Russell Silver Syndrome

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Abstract
Russell-Silver syndrome (RSS) is a heterogeneous condition characterized by intrauterine and postnatal growth retardation, craniofacial disproportion, syndactyly, webbed fingers, feeding difficulties, asymmetry in the size of the two halves or other parts of the body. We present a case of a 4 year old child presented in OPD of Social Security Hospital with upper respiratory tract infection and typical phenotype of Russell-Silver Syndrome.

Key Words: Russell-Silver syndrome, fetal growth retardation

Introduction
Russell-Silver syndrome (SRS) is a clinically and genetically heterogeneous condition characterized by intrauterine and postnatal growth retardation, craniofacial disproportion, downward curvature of the corner of the mouth, syndactyly, webbed fingers, feeding difficulties, and asymmetry in the size of the two halves or other parts of the body. Hypomethylation of the paternal 11p15 imprinting control region 1 (ICR1) and maternal uniparental disomy of chromosome 7 are found in 50-60% and in 5-10% of SRS patients, respectively. The estimated number of people who develop this condition is about 1 in 3,000 people. Males and females are equally affected.

Case Report
A 4 year old child presented in February 2013 in OPD of Social Security Hospital with upper respiratory tract infection. On examination he had wide forehead with a small triangle-shaped face, small narrow chin and thin, wide mouth. He was having short arms with clinodactyly and short, stubby fingers and toes. There were multiple café-au-lait spots on body with body asymmetry. He had short stature i.e.74 cm (below 5th percentile), weight of 12 kg (below 5th percentile) and head circumference 37 cm (below 5th percentile).

Discussion
In 1953 and 1954, Silver and Russell independently described groups of small-for-gestational-age [SGA] children whose pregnancies had been complicated by intrauterine growth retardation [IUGR]. Their common findings were short stature without catch-up growth, normal head size for age, a distinctive triangular face, low-set ears and incurving fifth fingers. These two groups of patients are now considered to have had variations of the same disorder that we now call Russell-Silver Syndrome. One interesting and important aspect of the Russell-Silver syndrome is its variation in phenotype. Some individuals with RSS have many traits, thus a severe phenotype, while others have very few traits, thus a mild phenotype. In general, the most noticeable symptom of Russell Silver syndrome is a child’s failure to grow and this may suggest the diagnosis. The infant is born small and does not achieve normal length/height for his/her age. The distinctive facial features may be identified in infants and children, but may be harder to recognize in teenagers and adults.

There are no specific laboratory tests to diagnose Russell-Silver syndrome. Diagnosis is usually based on the clinical judgment of the child. However, the following tests may be done:
- Blood sugar (some children may have low blood glucose)
- Bone age testing (bone age is often younger than the child's actual age)
- Chromosome testing (may detect a chromosomal problem)
- Growth hormone (some children may have a deficiency)
- Skeletal survey (to rule out other conditions that may mimic Russell-Silver syndrome)

Currently for RSS and non-RSS/SGA patients, the prospect for a normal life with a normal adult height is closer than ever before. By understanding the importance of balanced diet of these children, malnutrition and low blood sugar can be avoided, that in the past has negatively affected their growth and development. With the recent U. S. Food and Drug Administration's approval of growth hormone for the treatment of the growth failure, these young children can be treated to attain normal height (if treated early). By taking medications to postpone puberty, called LHRH (luteinizing hormone releasing hormone) analogues, the older children can recover growth potential lost in-utero, in infancy and in early childhood. By continuing growth hormone until...
growth is complete, the teenagers have a better growth spurt during puberty.

References