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Hand anomalies in Russell Silver syndrome

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Summary Russell Silver syndrome (RSS) is a genetic disorder of unknown aetiology. The disorder is clinically and genetically heterogeneous, and various modes of inheritance and genetic abnormalities have been described. A large number of clinical features are associated with this condition. Growth retardation, typical facies, limb asymmetry, delayed bony growth and clinodactyly are some of the most constant features of RSS. We report a small series of patients presenting with a range of hand anomalies, some of which have not been previously reported in association with this condition.

Russell Silver syndrome (RSS, also known as Silver-Russell syndrome, Silver syndrome and Russell syndrome) is a genetic disorder of unknown aetiology. It was described independently by two physicians, Drs Silver and Russell, at about the same time. In 1953 Silver described two children with congenital hemihypertrophy, low birth weight, short stature, and elevated urinary gonadotrophins. In 1954 Russell described five patients with intrauterine growth retardation and characteristic triangular-shaped face with a broad forehead and pointed, small chin. Their independent and varied findings were eventually determined to be a variation of the same syndrome and came to be known as Russell Silver syndrome.[1,2]

RSS is considered a genetic disorder. The disorder is clinically and genetically heterogeneous, and various modes of inheritance and abnormalities involving chromosomes 1, 7, 8, 15, 17, 18 and the X chromosome have been associated with RSS. However, only chromosomes 7 and 17 have been consistently implicated in patients with a strict clinical diagnosis of RSS.[3] Both dominant and recessive inheritance has been described. It is now believed that the RSS phenotype is linked to more than one genotype, which explains the variability of the characteristics that RSS children display.

The genetic anomalies seem to exert their effects through growth hormone. Low levels of growth hormones and/or a relative resistance to growth hormone have been found in children who are small for gestational age, particularly in children with Russell Silver syndrome.[4,5] Growth hormone supplements can improve the growth velocity in these children.

No genetic or biochemical marker is currently available for diagnosis and the diagnosis is based on clinical criteria introduced by Price: (1) birth weight below or equal to -2 SD from the mean; (2) poor postnatal growth below or equal to -2 SD from the mean at diagnosis; (3) preservation of

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occipitofrontal head circumference; (4) classic facial phenotype; and (5) asymmetry.[1]

Clinical features

A large number of clinical features for Russell Silver syndrome have been described. They are loosely classified into major and minor features and the more frequently observed ones are summarised in Table 1.[3]

Hand anomalies in RSS

Asymmetry of limbs and delayed bony maturation are common features of RSS. Among the specific upper limb anomalies, clinodactyly of the fifth digit is one of the most constant features. This is mentioned in most case reports and larger series.[1] Abrahams described clinodactyly in 19 of the 25 patients he reviewed.[6] A few other abnormalities of the upper limb have been described infrequently. They include syndactyly,[6,7] camptodactyly,[3] absence of fingers,[7] a cleft hand[8] and a hypoplastic thumb.[9]

The senior author has treated a small series of three patients with this rare disorder who presented with a number of hand anomalies, some of which have not been reported previously. These include swan neck deformities in two patients, triggering of multiple fingers in one patient and a patient with a complex syndactyly and a cleft in adjacent webs. All three patients had some degree of clinodactyly.

Table 1 Clinical features observed in patients with Russell Silver syndrome

MAJOR
Low birth weight (<-2 SD)
Short stature (<-2 SD)
Typical facies (triangular face, broad forehead and pointed chin)
MINOR
Clinodactyly of little finger
Limb asymmetry
Relative macrocephaly (because of sparing of cranial growth)
Motor/neuropsychological delay
Delayed bone age
Crowding of teeth and microdontia
Muscular hypotrophy/hypotonia
Hypoglycaemia (low blood sugar) in infancy and early childhood (2—3 years)
Feeding difficulties
High-arched palate
Syndactyly of 2nd and 3rd toes
Urogenital anomalies including urethral valves and horseshoe kidneys
Hypospadias
Cryptorchidism
Café au lait naevi
Early or precocious puberty

Patient one

This patient, now 11 years old, was born after 38 weeks of gestation and was small for gestational age. He subsequently failed to thrive. Though his developmental milestones were more or less normal, at the age of 15 months his bone age was less than 3 months. At this point a diagnosis of RSS was made as he displayed many of the characteristic features of Russell Silver syndrome including short stature, failure to gain weight, a triangular face with broad forehead and bilateral clinodactyly. He was later started on growth hormone treatment at the age of 3 years.

He was referred to the department of Plastic Surgery at the age of 4 years for management of flexion deformities of his fingers. He was noted to have bilateral clinodactyly of little fingers (more pronounced on the left side), mild syndactyly of 2nd to 4th webs on both hands and pronounced swan neck deformities of all eight fingers excluding the thumbs (Figure 1). No active treatment has been undertaken so far, as he manages to maintain a reasonable degree of function in both his hands and has full range of active and passive flexion in his fingers. He remains under regular review in the congenital hand clinic.

Patient two

This young girl, now eight, was delivered at 37 weeks of gestation by elective caesarean section with intrauterine growth retardation (birth weight of 1.71 kg) and a breech presentation. She had problems with early poor feeding and needed nasogastric feeding from the age of 2½ months and later had a feeding gastrostomy at the age of 2½ years. Her hormonal profile was characteristic of a partial growth hormone resistance and she was started on growth hormone injections with good response. She also developed precocious puberty at the age of 7½ years and was started on gonadotrophin-releasing hormone (GnRH) agonists in an attempt to delay this. In addition to the growth retardation and typical facial features, she also had a high arched palate and speech difficulties and was receiving regular speech therapy.

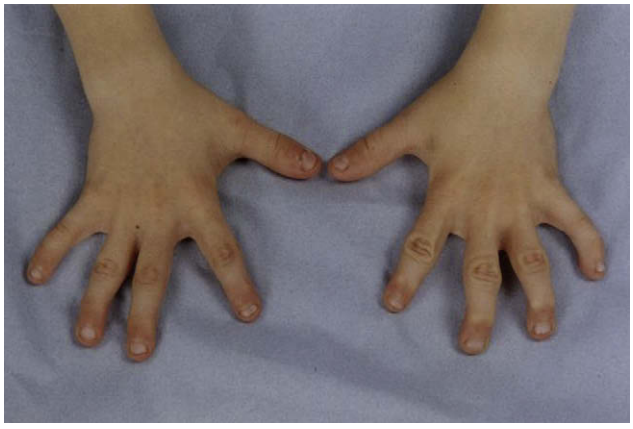

Figure 1 Bilateral clinodactyly with swan neck deformities of index to little fingers of both hands in patient one.