Crossed asymmetry in Russell–Silver syndrome

Since the initial report by Silver et al (1953), more than 50 examples of the Russell–Silver syndrome have been reported. Unilateral congenital asymmetry of the extremities has been considered one of the major features of this disorder (Silver, 1964). We recently observed a child with otherwise typical features of the Russell–Silver syndrome who had enlargement of the right hand and of the left lower extremity. We know of no other recorded example of crossed asymmetry in this clinical entity.

Case report

A black boy who is now 6 years 9 months of age, has been followed at the State University Hospital for the past 4 years. Details of pregnancy, delivery, and early development are incompletely known. His mother was 28 and his father 29 years of age at the time of his birth. Both parents are chronic alcoholics. The patient is supposedly the last of their 5 children. Their first child, a boy, died soon after birth. The patient and the other children, 2 boys and a girl, are living with the same foster parents.

At 3 months of age the boy was admitted to hospital elsewhere for failure to thrive. Admission weight was 2.9 kg and the birthweight was recorded as 3 kg. Enlargement of the left leg and foot was noted. He was discharged with the diagnosis of failure to thrive resulting from maternal deprivation.

He was admitted to State University Hospital at 27 months and again at 54 months of age, for evaluation of skeletal asymmetry and a large phallus. At 54 months of age his height (98 cm) and weight (18.1 kg) were in the 3rd and 50th centiles, respectively. His head was dolichocephalic and measured 50 cm in circumference. The face was triangular and the corners of the mouth were turned down (Fig. 1). The palate was high-arched and narrow. There was minimal diastasis recti. No abdominal masses were palpated. The penis was 8 cm long; both testes were in the scrotum. The left testis measured $2 \times 1$ cm and the right $2.5 \times 1.5$ cm. Café-au-lait spots with smooth margins were present on each forearm; the one on the left measured $1 \times 2$ cm and the one on the right $1 \times 1$ cm.

There was obvious asymmetry of the extremities. The right hand was larger than the left; the entire left lower extremity was larger than the right (Fig. 2). Radiographs showed enlargement of the bones and soft tissues of the affected parts (Fig. 3). The left lower extremity was 2 cm longer than the right; the arms and forearms were symmetrical. There was soft tissue syndactyly of the 2nd and 3rd toes of both feet and clinodactyly of the left 5th finger. Cardiovascular, respiratory, and neurological systems were normal. He was judged to have an average intelligence.

Urinalysis, haemogram, protein-bound iodine, and survey of the blood chemical values were normal. The 24-hour urinary excretion of 17-ketosteroids, 17-ketogenic steroids, gonadotropins, and vanillylmandelic acid were normal. There was narrowing of the L3-L4 intervertebral space and increased height of the bodies of L3 and L4 vertebrae (congenital block vertebrae). The bone age was 42 months. Chromosomal study of cultured leucocytes showed a normal 46,XY pattern. The dermatoglyphs were within normal limits and almost identical on two sides.
Case reports

Intravenous urograms were done at 27, 54, and 80 months of age, respectively. At each examination, the size of each kidney was at the upper limits of normal. A bulge of the lower lateral margin of the left kidney was first observed at 27 months of age and did not change; the collecting systems remained normal (Fig. 4). A sonogram showed that the entire left kidney had acoustic properties of normal renal parenchyma and a scan with $^{197}$Hg-chlormedodrin confirmed that the bulge represented a variant of renal lobaration rather than a solid tumour.

The patient's 3 living sibs have been examined. Their growth was appropriate for their ages and none had asymmetry or other stigmata of the Russell-Silver syndrome.

Discussion

To our knowledge, crossed asymmetry, a striking finding in our patient, has not been previously reported in the Russell-Silver syndrome. In addition, our patient presented with several major and minor manifestations of the syndrome, such as small stature, aberrant sexual maturation, immature osseous development, characteristic facial appearance, café-au-lait spots, syndactyly and clinodactyly (Silver, 1964; Smith, 1970). A great degree of clinical variability is known to exist in this syndrome. Growth retardation and congenital asymmetry, two of the major features of the syndrome, are present in 93 and 79 per cent of the affected individuals, respectively (Smith, 1970). Our patient was noted to have normal birthweight; however, this could not be confirmed.

Crossed asymmetry is not unusual in isolated congenital hemihypertrophy (Fraumeni et al., 1967). In addition, there is an increased incidence of neoplasms of kidney, liver, and adrenal cortex in individuals with this condition (Fraumeni et al., 1967; Fraumeni, 1972), which contrasts with no reports of such tumours in the Russell-Silver syndrome (Silver, 1972). Though the slight deformity of our patient's left kidney was a source of some concern, lack of change on serial intravenous urograms and demonstration of normal renal parenchyma by sonography and radionuclide imaging have provided reassuring evidence that the focal renal enlargement is an anomaly of lobaration.

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References


