Picture Story

Silver-Russell syndrome

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Sri Lanka Journal of Child Health, 2004; **34**: 62-4

(Key words: Silver Russell syndrome)

A baby boy weighing 1500g was delivered by caesarean section at 40 weeks of gestation. He was the 3rd sibling born to non-consanguineous parents with 2 other normal children. Prenatal ultrasonography at 31 weeks and 38 weeks of gestation showed asymmetrical intrauterine growth retardation (IUGR) with maturity compatible with 24 and 31 weeks respectively. The mother, a rhesus negative lady, had an uneventful antenatal period with no history of pyrexia or pregnancy induced hypertension and at routine screening Rh antibodies were not detected during this pregnancy. There was no family history of abnormal or short babies.

Apgar score was normal at delivery and baby was admitted to Special Care Baby Unit where blood sugar was monitored and a septic screen done. Baby was discharged with the onset of weight gain and was evaluated two weekly at neonatology clinic where the occipito-frontal circumference (OFC) and weight were monitored. As days passed baby developed features compatible with Silver-Russell syndrome (SRS).

According to Dr Marilyn Cowger1, every SRS individual has, almost without exception, eight features. All these features were found in this baby.

1. Low birth weight[1] - 1500g (<3rd centile)
2. Low birth length[2] - 46cm (<3rd centile)
3. Triangular shaped face (Figure 1)
4. Long narrow head at birth
5. OFC at birth (30cm) was greater compared to weight
6. Post natal growth retardation (Figures 2 & 3)
7. Fifth finger clinodactyly due to hypoplastic middle phalanx.(Figures 4 & 5)
8. Poor appetite in early years

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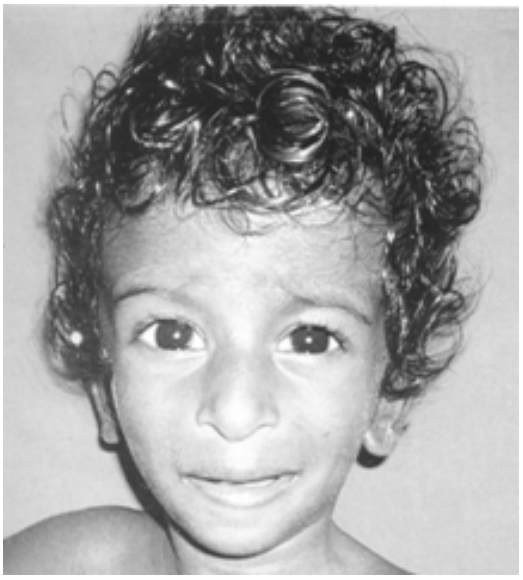

Figure 1 *Facial appearance*

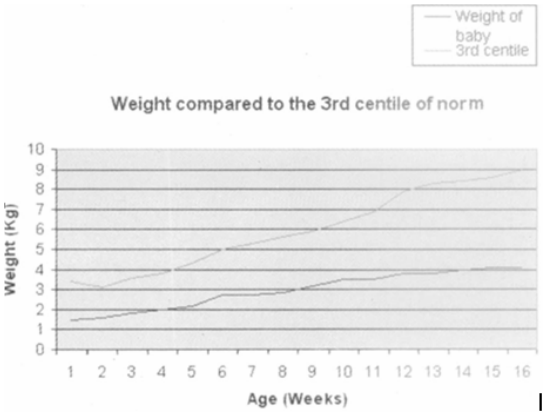

Figure 2 *Growth chart*

Figure 3 *Comparison with normal same age baby*

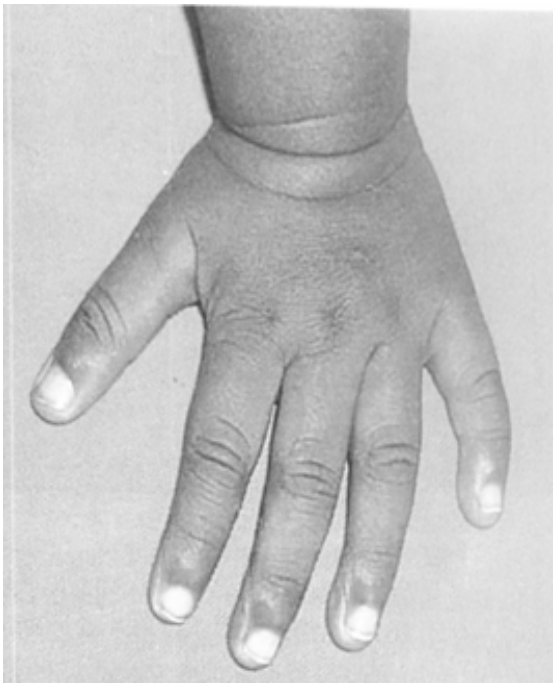

Figure 4 *Hand showing clinodactyly*

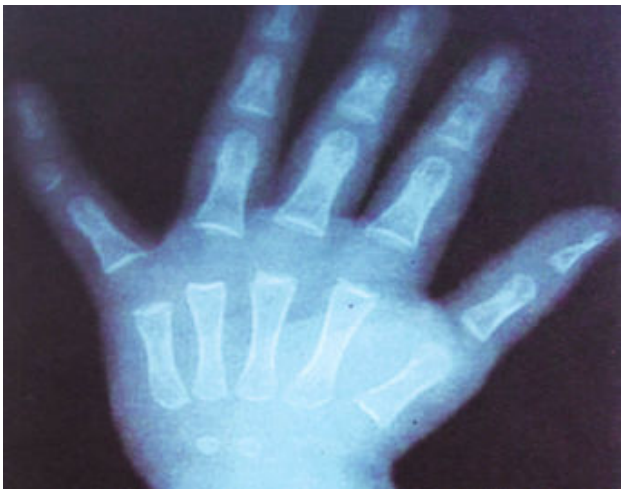

Figure 5 *X-ray of hand*

Most other common characteristics' in SRS, found in this baby, were

- Asymmetry in limb length (Figure 6)
- Broad forehead (Figure 1)
- Hypoplastic mandible/small chin (Figure 1)
- Thin upper lip (Figure 1)
- Abnormal (low set) ears
- Cryptorchidism-one undescended testis and the other ultasonographically missing
- Delayed bone age3,4 (Figure 5 – at 10 months compatible with 6 months)
- Excessive sweating on head and upper trunk

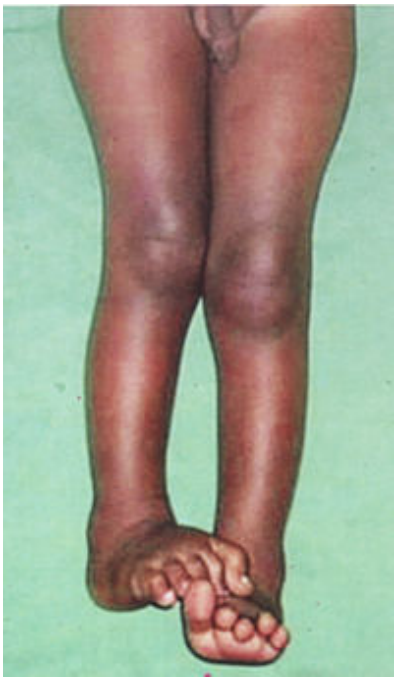

Figure 6 *Asymmetry in limb lengths*

Other common characteristics of SRS, difficult to elicit in this baby, are crowding of teeth and high pitched voice. Syndactyly of the toes and hypospadius were not found. Rare trait of SRS found in this baby was café-au-lait spots (Figure 7). Growth hormone deficiency could not be excluded due to lack of facilities.

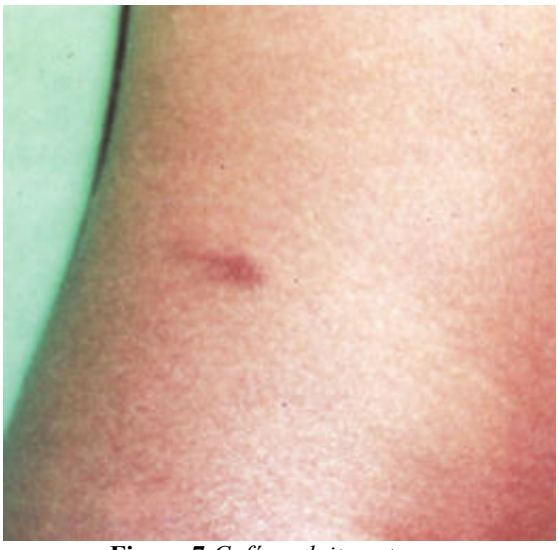

Figure 7 *Café-au-lait spot*

Now the baby is walking without support having some difficulty of asymmetry. Although the weight gain is poor, his developmental milestones are not affected. Baby is energetic but appetite is poor and he is having excessive sweating episodes that are compatible with hypoglycaemia.

Discussion

SRS is a rare cause of IUGR. Its aetiology is not fully identified. Males and females are equally affected3,4. A few cases of autosomal dominant transmission are described. Approximately 10% of patients have proven uniparental disomy and methylation in long arm of chromosome 75. Failure to thrive, feeding difficulties and fasting hypoglycaemia are the main problems in infancy6. Prognosis is relatively good. Although short stature and asymmetry persist, facial dysmorphism will disappear. Growth hormone therapy may improve growth in some patients and they should have good nutritional evaluation, physical therapy, occupational therapy and some corrective surgery or disability support.

References

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