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# GROWTH FAILURE WITH HEMIHYPERTROPHY

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A 4-year-old male child born of non-consanguineous marriage, first by birth order presented with growth failure. He was born of cesarean section at full term gestation with birth weight of 1.5 kg. He also had delayed motor development with delayed speech development. There was history of excessive sweating but no documented hypoglycemia. On examination, height was 89 cms [<3rd standard deviation (SD)], weight was 10 kgs (<3rd SD) and body mass index (BMI) was 12.65 (<3 SD) as per World Health Organization (WHO) growth charts. Right side of his body was less developed as compared to left, the difference ranging from 0.5 cm to 1.5 cms. He had a limb length discrepancy of 1 cm with right being smaller. Other clinical findings were frontal bossing, arthrogryposis, high arched palate, syndactyly, triangular face, micrognathia, downward slanting of angle of mouth, clinodactyly of little fingers and wide gap between great toe and 2nd toe (Figure 1 and 2). Complete blood count picture of patient showed hemoglobin 7.2 gm/dl, mean corpuscular volume (MCV) 64fl, mean corpuscular hemoglobin concentration (MCHC) 24 g/

**Figure 1.** Wide gap between great toe and 2nd toe.

**Figure 2.** Triangular face, downward slanting of angle of mouth, hemihypertrophy of left side.

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dl, with peripheral blood smear showing microcytic, hypochromic anemia, suggestive of iron deficiency anemia. Bone age was 1.2 years on radiographs. There was no cardiac lesion on echocardiography.

*What is the diagnosis?*

Silver Russell Syndrome (SRS) is also known as Silver Russell dwarfism. It is a growth disorder approximately affecting 1:50,000 to 1:1,00,000.[1] SRS is characterized by congenital hemihypertrophy and growth retardation.[2] SRS occurs in all populations and affects males and females in equal numbers.[3] About 30-60% of children with SRS have changes (loss of methylation/hypomethylation) affecting the ICR1 region on chromosome 11 (11p15 LOM).[3] This, in turn, affects the activity of two genes (maternally expressed H19 and paternally expressed IGF2) which are believed to play a role in the development of SRS.[3] SRS is genetically heterogeneous, meaning that different genetic abnormalities are known to cause the disorder. About 5-10% of individuals with SRS have been found to have both copies of chromosome 7 from their mother, rather than one from each parent. This is called maternal uniparental disomy of chromosome 7.[4] Silver Russell syndrome is a growth disorder characterized by slow growth before and after birth. Netchine - Harbinson Clinical Scoring System is a sensitive diagnostic scoring system. Four out of six criteria are required to suspect the clinical diagnosis.[5] They include a) small for gestational age (birth weight ≤2 SD for gestational age), b) postnatal growth failure, c) relative macrocephaly at birth, d) frontal bossing, e) body asymmetry (limb length discrepancy ≥0.5 cm and BMI ≤2 SD, f) feeding difficulty. Our patient had a birth weight of less than -2 SD, poor postnatal growth and development delay. The boy also had hemihypertrophy of the left side, dysmorphic facies, skeletal deformities and delayed bone age. Diagnosis of silver Russell Syndrome was done on basis of clinical examination. Management of SRS requires a multidisciplinary approach. They also require physical, occupational, speech and language therapy.

**Compliance with Ethical Standards**
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