

Case Report

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Congenital growth disorder: Silver Russell syndrome - A case report

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Abstract

Silver Russell syndrome is a congenital growth disorder with features of craniofacial dysmorphism and asymmetrical limbs. This case highlights failure to thrive (height and weight are less than -2SD), hemihypertrophy of the left side of the body and dysmorphic facial profile in a 13 year old boy. The parents have been counselled regarding etiology of the disease and the child is being treated with growth hormone therapy.

Keywords: Silver russell syndrome; Growth disorder; Limb asymmetry.

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Introduction

Silver Russell syndrome (SRS) is a rare genetic disorder that combines intrauterine growth retardation, craniofacial dysmorphism and limb asymmetry [1]. It may combine various disorders with clinically related phenotypes or may result from disruption of a single biochemical or endocrinological pathway [1-3].

Estimated prevalence of the disorder is 1/100,000 [1,4,5].

It is associated with an increased risk of delayed developmental milestones, speech and language problems, and learning disabilities [6].

Case report

A 13 year old boy from a non consanguineous marriage presented to the department of Paediatrics with complaints of fail-

ure to thrive and hemihypertrophy of the left side of the body since birth. He was delivered by LSCS at 37 weeks of gestation and birth weight was 1.8 kilograms. There is a history of admission to the Neonatal Intensive Care Unit (NICU) for 7 days.

Developmental milestones were achieved at appropriate ages. No history of other family members suffering from similar complaints. Younger sibling does not show similar complaints and is taller than the patient. Surgery for cryptorchidism on the left side was done at 10 years of age.

On examination, the boy was conscious and cooperative. His vitals were within normal limits. Intelligence and psychomotor development was appropriate for his age.

His height was 112 cm: less than -2SD and weight was 16 kg: Less than -2SD (according to growth references for boys by

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World Health Organization). Triangular face with small chin, small mouth, low set ears were noted. Hypoplasia of 5th finger (Figure 1) and clinodactyly of 4th and 5th toes (Figure 2) were noted in this boy.

Laboratory investigations including blood counts and urine were normal. IGF-1 levels were found to be low.

Radiological images show delayed bone age.



Figure 1:



Figure 2:

Considering history, clinical examination and radiological evidence, this patient was diagnosed as a case of Silver Russell Syndrome (SRS) and was planned for Growth Hormone (GH) Therapy.

Discussion

Silver Russell Syndrome (SRS) was first described in 1953 [1,3]. The disorder is clinically and genetically diverse and various modes of inheritance have been estimated involving chromosomes 7, 8, 15, 17 and 18, however, only chromosomes 7 and 17 have been consistently implicated in clinical diagnosis of SRS and SRS like disorders [3].

7-10% of patients with this SRS have a defect in a gene called the maternal Uniparental Disomy (UPD) for chromosome seven and in 35%-50% have hypomethylation of the Imprinted Control Region 1 (ICR1) at 11p15.5 [3,7,8]. It combines severe intrauterine restriction, postnatal failure to thrive, craniofacial dysmorphism, and limb asymmetry [1]. The musculoskeletal manifestations in Russell silver syndrome were studied in 25 cases. The most common manifestations were short stature (25 cases),

limb length discrepancy (23 cases), clinodactyly (19 cases), metacarpal bone and phalangeal abnormalities (13 cases), scoliosis (nine cases), foot syndactylism (five cases) and developmental dysplasia of the hips (three cases) [2,9,10].

Clinical diagnosis of Silver Russell Syndrome (SRS) is considered if a patient scores at least four of six criteria according to the Netchine-Harbisson clinical scoring system [3]. Testing for genetic causes confirms diagnosis in 60% patients [11,12].

Our patient had low birth weight and poor postnatal growth. The boy also had hemihypertrophy of the left side of body, dysmorphic facies, clinodactyly of toes and delayed bone age. The patient also had a history of cryptorchidism. The diagnosis was made on the basis of clinical evidence and radiological findings. For lack of facilities, genetic testing could not be done at our hospital.

Management of Silver Russell syndrome is based on growth hormone therapy [13] and a multidisciplinary approach of management is done in the form of speech therapy, physiotherapy, bone lengthening surgeries and psychological counselling of patient and family.

Conclusion

Silver Russell syndrome is a rare genetic disease. Its diagnosis is primarily by clinical evidence. Genetic counselling for identification of the underlying molecular subtype helps in treatment with regard to specific risk factors. Management is through a multidisciplinary approach and close parental guidance [13].

Declarations

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Informed consent: The patient and his family provided verbal consent for submission of the case report provided personal identifiers are not included. All details that might disclose the identity of the patient have thus been omitted. Conflict of interest: The authors have no conflict of interest to declare.

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