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Case Report

A rare presentation of Russell–Silver syndrome accompanied by torticollis: a case report

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ABSTRACT

Russell–Silver syndrome (RSS) is a clinically and genetically heterogeneous condition characterized mainly by intrauterine and postnatal growth retardation. It is also associated with several dysmorphic features, such as a triangular face, prominent forehead, body asymmetry, and feeding difficulties. The syndrome is usually sporadic. Because the clinical spectrum is broad, both the frequency and the severity of findings vary among individuals. Here, we present a child who had fifth-finger clinodactyly, growth retardation, body asymmetry, relative macrocephaly, and a triangular facial appearance consistent with RSS, and who also had torticollis a rare accompanying finding.

Keywords: Russell–Silver syndrome, Torticollis, Genetic

INTRODUCTION

Russell–Silver syndrome (RSS) was first described by Silver in 1953 and by Russell in 1954. RSS occurs worldwide and affects males and females equally.[1] The true incidence is not known, but it is estimated to be between 1 in 35,000 and 1 in 100,000 live births.[2] The diagnosis is based on the criteria defined in the Netchine–Harbison Clinical Scoring System (NH-CSS).[1] The clinical picture may change with age, and the most obvious findings are usually seen during childhood. The typical facial appearance includes a small triangular face, narrow chin, and prominent forehead, which can give the impression of macrocephaly.[3] Most individuals with RSS have normal intelligence, but motor and speech delays are common.[1]

Although many patients show the classic phenotype, others may have milder or atypical presentations.[4] In this report, we present a case who met the NH-CSS criteria and presented with torticollis a complaint rarely described in the literature and not included in the diagnostic criteria.

CASE REPORT

A 7-year-old girl was presented to our clinic because her family noticed a tilt of the neck. On examination, she had marked short stature for her age. Detailed evaluation revealed a triangular facial appearance, prominent forehead, hypertelorism, micrognathia, and relative macrocephaly (Figures 1–3). Body asymmetry was present, including right-sided hemihypertrophy and kyphoscoliosis (Figure 4). Both hands showed clinodactyly of the fifth fingers (Figures 5a and b). Her height was 110 cm. The patient demonstrated poor communication and limited cooperation during the examination. Her family admitted her school performance was poor. She had been born weighing around 3000 g and had no known additional medical conditions. There was no consanguinity between the parents. She had never received a diagnosis before. Routine complete blood count and biochemical tests were normal. Based on the clinical findings, the patient was diagnosed with RSS. Her family was informed, and she was referred to the genetics department, pediatric endocrinology for evaluation of growth retardation, and pediatric surgery for assessment of

torticollis. However, because the family lived outside the city, they declined further diagnostic evaluation and treatment at our hospital.

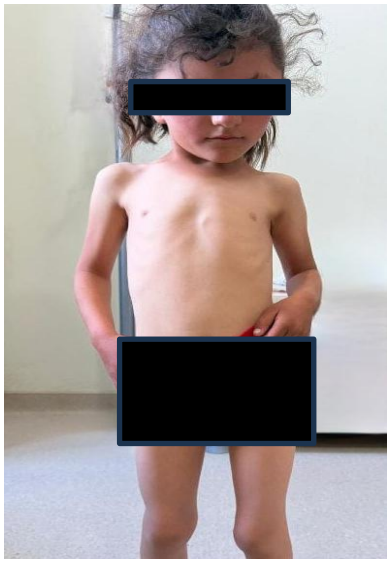

Figure 1: Relative macrocephaly, triangular face, torticollis, micrognathia, prominent forehead, and body asymmetry.

Figure 2: Hypertelorism, macrocephaly, prominent forehead, micrognathia, and triangular facial appearance.

Figure 3: Torticollis.

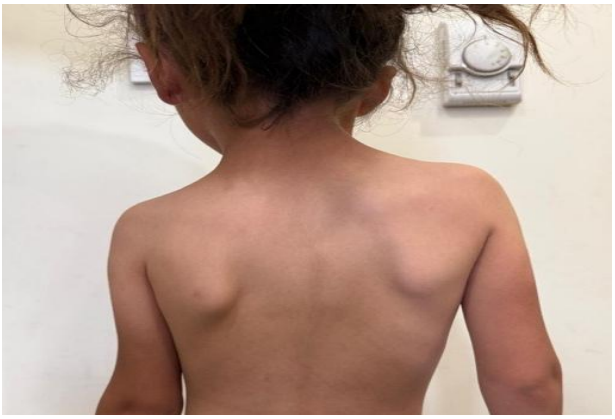

Figure 4: Scapular asymmetry and kyphoscoliosis.

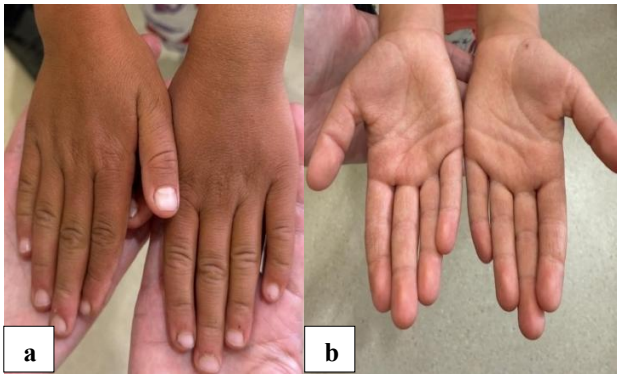

Figure 5 (a and b): Clinodactyly of the fifth fingers.

DISCUSSION

The earliest sign of RSS is abnormal growth. Most affected children are born small for gestational age, with a birth weight and/or length more than two standard deviations below the mean.[5] Our patient, however, was born with a normal birth weight. The NH-CSS criteria include small size for gestational age, postnatal growth retardation, relative macrocephaly at birth, prominent forehead, body asymmetry, and feeding difficulties. A clinical diagnosis is made when at least four of these criteria are met.[3] If at least three criteria are present and clinical suspicion is high, molecular testing is recommended to confirm the diagnosis.[6] In our case, the patient had a prominent forehead, relative macrocephaly, growth retardation, and body asymmetry, fulfilling the clinical diagnostic criteria. Additional supportive features although not specific to RSS include triangular facies, fifth-finger clinodactyly, micrognathia, reduced muscle mass, low-set or posteriorly rotated ears, downturned mouth, delayed closure of fontanelles, genitourinary anomalies (cryptorchidism, hypospadias), speech and motor delay, dental crowding, toe syndactyly, café-au-lait spots or pigmentary changes, scoliosis/kyphosis, gastroesophageal reflux, feeding difficulties, growth hormone deficiency, and early puberty.[3,6] Our patient exhibited triangular facies, fifth-finger clinodactyly, micrognathia, speech delay, and kyphoscoliosis.

Epigenetic mechanisms play a key role in the pathogenesis of the syndrome.[3] The most common epigenetic causes are loss of methylation at chromosome 11p15 (50%) and maternal uniparental disomy of chromosome 7 (10%).[5,7] Genetic testing confirms the clinical diagnosis in approximately 60% of cases.[6] However, 30–40% of patients who meet the NH-CSS clinical criteria have negative molecular or cytogenetic tests.[5] A negative test result does not exclude the diagnosis.[2] Despite advances in genetics, diagnosis of RSS still relies primarily on clinical evaluation.[4] Genetic testing could not be performed in our patient because the family lived outside the city. Growth hormone (GH) therapy is recommended in RSS regardless of whether GH deficiency is present.[5,8] Children with short stature or body asymmetry may be sensitive about body image, which can affect self-esteem, peer relationships, and social interaction.[5] Our patient was referred to pediatric endocrinology for appropriate follow-up.

Torticollis results from contracture or fibrosis of one sternocleidomastoid muscle, leading to ipsilateral head tilt and contralateral rotation.[9] The coexistence of torticollis with RSS has been reported only in two cases in the literature.[8,9] Our patient had also torticollis.

CONCLUSION

Patient met the major diagnostic criteria for RSS and also presented with torticollis. Since torticollis has been reported only rarely in association with RSS, this case is noteworthy and may represent one of the very few documented examples in the literature. This coexistence suggests that torticollis may be an uncommon clinical manifestation of RSS.

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