Original Article

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Correction of the axial and appendicular deformities in a patient with Silver-Russel syndrome

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ABSTRACT

Background: Scoliosis and limb length discrepancy are the major orthopaedic abnormalities in patients with Silver-Russel syndrome (SRS). In this paper, we describe a series of orthopaedic interventions in an attempt to overcome the progressive pathologic mechanism in a 7-year-old girl who manifested the full phenotypic features of SRS. **Materials and Methods:** Unilateral hip dislocation, progressive scoliosis and limb length discrepancy have been dealt with through Pemberton osteotomy, spinal fusion and Taylor-Spatial-Frame respectively. **Results:** In order to correct the axial and the appendicular deformities a sum of seven operations were performed (between the age of 7 years and 13 years). Pemberton osteotomy was performed to treat dislocation of her right hip because of developmental dysplasia of the hip. Spinal fusion (spondylodesis) of segments Th3-L5 was done to correct her scoliosis. And, to overcome the limb length discrepancy of 15-cm we used Taylor-Spatial-Frame with percutaneous distal corticotomy of the femur, and the proximal tibia, as well as the foot, were performed. We were able to minimize the limb length discrepancy to 5 cm. The girl became able to walk with the aid of a below knee orthosis and through lifting the left limb with 5-cm height shoe. **Conclusion:** Limb lengthening surgery in patients with multiple malformation complex as in SRS is associated with high recurrence risk because of; muscular hypotonia, overtubulation of the long bones, and the poor bone regenerative quality. Our interventions were principally directed towards improving the cosmetic outlook, functions and the biomechanics.

Key words: Limb length discrepancy, Silver-Russel syndrome, Tyalor Spatial Frame

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INTRODUCTION

Silver-Russel syndrome (SRS) is characterized by intrauterine dwarfism accompanied by postnatal growth deficiency, small triangular face, thin lips with downturned corners and micrognathia. Silver *et al.*[1] described two unrelated children with congenital hemihypertrophy, low birth weight, short stature and elevated urinary gonadotropins. Russell[2] described five unrelated children with intrauterine growth retardation and characteristic facial features, and two children manifested bodily asymmetry. The thorax is slender associated with café-au-lait spots over the trunk and thighs. Hyperhidrosis and a muscular hypotrophy with body asymmetry are characteristic. There is delayed closure of the anterior fontanelle, hypoplasia of the mandible and hip dislocation. The hands usually manifesting brachyphalangia of the 5th finger associated with pseudoepiphysis of the second metacarpal. In 20% of cases, there is a history of abnormal pregnancy (threatened abortion, eclampsia, viral disease and drug ingestion).[3,4]

Netchine *et al.*[5] and Bartholdi *et al.*[6] accounted primarily on the phenotypic characterisation as a baseline tool of diagnosis. We describe a girl with a constellation of pre and postnatal growth deficiency, thoraco-lumbar scoliosis of 40° Cobbs angle, bilateral hip dysplasia with a unilateral hip dislocation, congenital hemihypertrophy and she manifested elevated urinary gonadotropins. The overall phenotypic features were compatible with SRS. Parents showed normal heights with no peculiar facial features suggestive of the syndrome. The latter raises the possibility of autosomal recessive pattern of inheritance.[7]

Clinical report

This child was referred to our department for the 1st time at the age of 7 months because of congenital hip dislocation of the right hip. She was a product of full term pregnancy, and she manifested pre and

postnatal growth deficiency. Family history was non-contributory.

At the age of 7 years, her growth was − 3SD and she manifested prominent forehead, triangular face, which is abnormally small compared to the normally developed cranium (pseudohydrocephalus). Strabismus, wide frontal area, depressed nasal bridge and small snob nose, thin lips with downturned corners, her mouth gave the shape of inverted V. Arched palate, crowded teeth and micrognathia were present. The thorax is slender with poorly developed musculature overwhelmed by apparent hemihypertrophy and body asymmetry with subsequent development of limb length discrepancy of 15-cm [Figure 1]. Her subsequent course of development was of marked retardation in acquiring the skills of gross motor development. Her neurological development had been within normal limits, though she manifested poor schooling achievement and poor communicative skills, and her vision showed anisometropia. Thoraco-lumbar scoliosis of 80° Cobbs angle, bilateral hip dysplasia with a unilateral hip dislocation was detected.

Skeletal survey showed; anteroposterior (AP) hand radiographs showed clinodactyly of the 3rd-5th fingers respectively associated with noticeable unilateral hypoplasia of the middle phalange of the left hand of the second and fifth fingers, pseudoepiphysis of the first proximal metacarpophalnageal joint [Figure 2]. AP pelvis radiograph showed bilateral dysplasia of the capital femoral epiphyses (more marked on the left side with subsequent development of right hip dislocation) [Figure 3]. Standing preoperative AP hips to-ankles radiograph of the child showed apparent limb length discrepancy of 15-cm associated with overtubulation/slender long bones of the long bones [Figure 4]. Brain magnetic resonance imaging (MRI) and spinal MRI were normal. Serum CK and plasma lactate were normal. Electomyography showed minimal myopathic changes, though past muscle MRI imaging showed minimal, non-specific and not diagnostic changes. Hypothyroidism, hyperthyroidism, hyperparathyroidism and Vitamin D deficiency were excluded. She manifested high serum and urine levels of gonadotropin in her prepubertal years.

Between the age of 7 years and 13 years, seven operations were performed. At the age of 7 years and 6 months, the patient underwent arthrotomy, hip reduction, derotation varising osteotomy of the right femur (with shortening of 15 mm) and acetabuloplasty (Pemberton osteotomy) to treat dislocation of her right hip due to developmental dysplasia of the hip.

Although radiographs after 6 weeks of hip spica cast showed a good result, there was persisting instability of the right hip. Open reduction, soft-tissue release, partial purse-string suturing and resection of the capsule were performed 5 months later. Osteosynthesis material was surgically removed 21 months after the first operation.

At the age of 11, spinal fusion (spondylodesis) of segments Th3-L5 was performed in another hospital because of progressive scoliosis (80° Cobb angle) [Figure 5].

Due to the hemihypotrophy of the patient's left body half, she developed limb length discrepancy

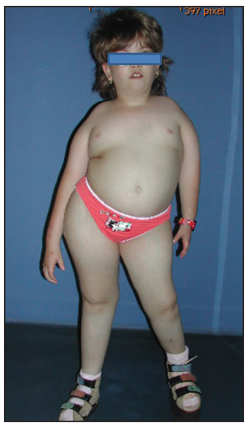

Figure 1: Photo of the patient at the age of 7 years showed, growth was -3SD and she manifested prominent forehead, triangular face, which is abnormally small compared to the normally developed cranium (pseudohydrocephalus). Strabismus, wide frontal area, depressed nasal bridge and small snob nose, thin lips with downturned corners, her mouth gave the shape of inverted V. Arched palate, crowded teeth and micrognathia were present. The thorax is slender with poorly developed musculature overwhelmed by apparent hemihypertrophy and body asymmetry with subsequent development of limb length discrepancy of 15 cm

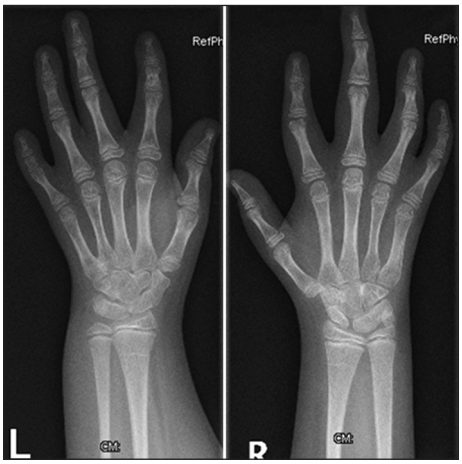

Figure 2: Anteroposterior hand radiographs showed clinodactyly of the 3rd-5th fingers associated with noticeable unilateral hypoplasia of the middle phalange of the left hand of the second and fifth fingers, pseudoepiphysis of the first proximal metacarpophalnageal joint

of 15-cm with shortening of the left femur, tibia and foot. Limb lengthening surgery of the femur and tibia was performed when she was 12 years old, using Taylor-Spatial-Frame with percutaneous distal corticotomy of the femur and for the proximal tibia including the foot into the external fixator [Figure 6a and b]. The foot frame was removed 3 months later. Follow-up X-rays after 9 months showed a very thin bone and weak bony regenerate at the callotasis site with a consecutive risk of regenerate fracture. To prevent fracture, intramedullary stabilization was performed when the frames were disassembled, a Rush rod was inserted in the femur, which was 4 mm in thickness. For the tibia, we used elastic stable intramedullary nail, 2 mm in thickness, also note the diminution of the leg length discrepancy from 15 to 5 cm and the girl became able to walk with the aid of a below knee orthosis and through lifting the left limb with 5-cm height shoe [Figure 7].

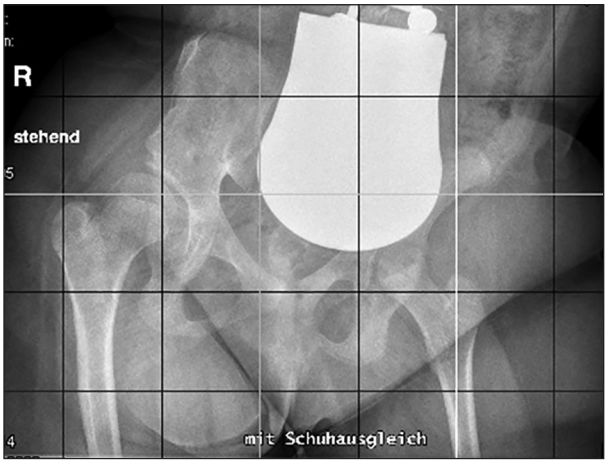

Figure 3: Anteroposterior pelvis radiograph showed bilateral dysplasia of the capital femoral epiphyses (more marked on the left side with subsequent development of right hip dislocation)

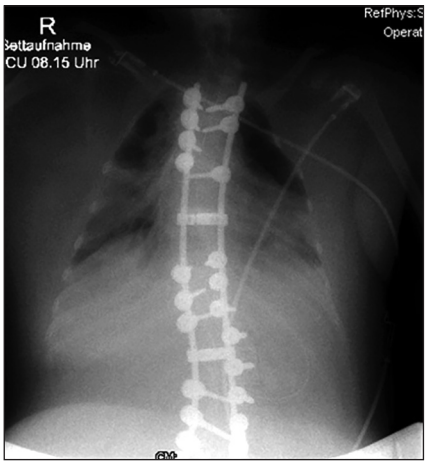

Figure 5: Anteroposterior spine radiograph at the age of eleven, showed spinal fusion (spondylodesis) of segments Th3-L5 was performed in another hospital because of progressive scoliosis (80° Cobb angle)

DISCUSSION

The syndrome of short stature of prenatal onset, triangular facies, body asymmetry, variation in the pattern of sexual development and other abnormalities including cafe au lait pigmentation and clinodactyly of the fifth fingers was independently described by Silver *et al*. 1953 and by Russell in 1954.[1,2] Birth weight is usually <2200 g at full term, and birth length is about 44 cm, the placenta is small. Short stature is maintained throughout childhood, height usually being below the third percentile; adult height averages 149-cm in males and 138 in females; females seen to gain some subcutaneous fat after puberty the facies is characterized by pseudohydrocephaly due to the relative smallness of the face and the calvaria, while appearing large is really somewhat smaller than normal

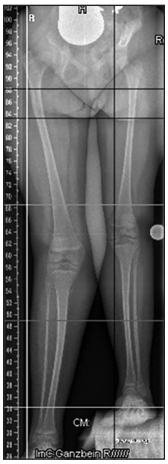

Figure 4: Standing pre-operative anteroposterior hips to-ankles radiograph of the child showed apparent limb length discrepancy of 15 cm associated with overtubulation/slender long bones of the long bones

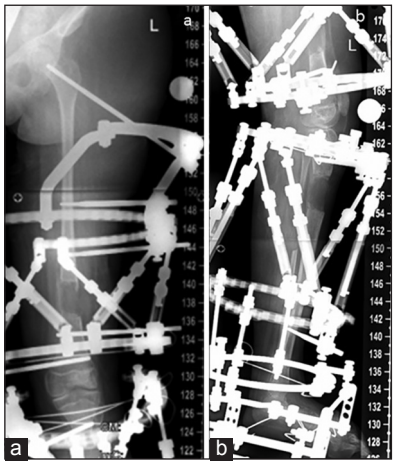

Figure 6: (a and b) Anteroposterior radiograph of the left femur and lateral radiograph of the left tibia and fibula showed limb lengthening surgery of the femur and tibia was performed when she was 12 years old, using Taylor-Spatial-Frame with percutaneous distal corticotomy of the femur and for the proximal tibia including the foot into the external fixator

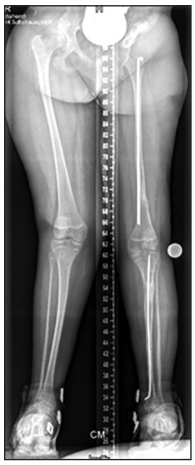

Figure 7: Standing anteroposterior radiograph of the lower limbs showed a Rush rod was inserted in the femur, which was 4 mm in thickness. For the tibia we used elastic stable intramedullary nail, 2 mm in thickness, also note the diminution of the leg length discrepancy from 15 to 5 cm and the girl became able to walk with the aid of a below knee orthosis and through lifting the left limb with 5-cm height shoe

the forehead is prominent and bossed and the face is triangular with the chin is small and pointed in most of the cases the sclerae are bluish in infancy, the eyes seem large, the mouth appears wide and the corners are often turned downward, the upper lip vermilion is thin, the pinnae may protrude and the appearance becomes markedly less striking with age congenital asymmetry has been noted in 80% of cases. Recent studies on developmental delay in these children have shown that most require special education. It has been considered that 20% of cases of threatened abortion, eclampsia, viral diseases and drug ingestion are in fact victims of SRS. Price *et al*.[7] proposed diagnostic criteria for SRS of:

1. Birth weight below or equal to -2SD from the mean;
2. Poor postnatal growth below or equal to -2SD from the mean at diagnosis;
3. Preservation of occipitofrontal head circumference;
4. Classic facial phenotype; and
5. Asymmetry.

Silver-Russel syndrome is one of a large group of conditions categorized as intrauterine growth retardation or low birth weight dwarfism differential diagnosis includes a plethora of conditions with short stature and precocious sexual development mullibery nanism to be excluded which is an autosomal recessive disorder associated with pericardial constriction and characterized by eye findings, and 3M syndrome cafe au lait pigmentation and or body asymmetry may be seen with NF I, hemihyperplasia, McCune Albright, Proteus syndrome, and Klippel-Trenaunay-Weber and del 18p and trisomy 18 mosaicism and Beckwith-Wiedmann hemihypertrophy.[8]

Discrepancy in limb lengths is a common orthopaedic problem, arising from either shortening or overgrowth of one or more bones in the limb. It may be caused by a multitude of conditions. Any correction of limb length disparity must be performed after thorough analysis and assessment of its aetiology, pathophysiology, and clinical consequences. Any change in limb length interferes with the dynamics of the locomotor system and its compensatory mechanism. Several studies examined the morbidity of limb length discrepancy in children and its possible association with gait abnormality, joint pain, and premature osteoarthritis. A gait analysis study of 35 children with limb length discrepancy ranging from 0.8% to 15.8% of the length of the long extremity (0.6-11.1 cm) found that when the discrepancy 5.5% or more, more mechanical work was performed by the long extremity and there was a greater vertical displacement of the centre of body mass.[9] It can be inferred from this finding that a limb with a discrepancy of 5.5% or more is less energy efficient. A force plate study concluded that in adults with limb length discrepancy of 1-3 cm, the shorter limb sustains a greater proportion of the load and loading rate.[10]

There are several methods of limb lengthening such as distraction osteogenesis, which has rapidly become the accepted method of limb lengthening.[9] The key elements in successful distraction osteogenesis are a low-energy, minimally invasive corticotomy and gradual rhytmitic distraction at the corticotomy site, beginning 5-7 days after the cortoicotomy. The Ilizarov technique for limb lengthening with external fixation using circular frames and tensioned fine wires has been successfully modified by using segmental half-pins rather than wires for fixation.[11] Optimal regenerate bone formation is associated with daily fixator lengthening of up to 1 mm, done at three or more widely spaced intervals throughout the day. Predictable lengthening can be achieved with monolateral external fixators, circular Ilizarov fixators and variants, hybrid fixation systems (external fixators with lengthening over an intramedullary nail, and fully implantable intramedullary lenghthening devices). A circular external fixation system, as originally developed by Ilizarov, is extremely versatile, adaptable with the use of 90° and 120° arches, and usable across joints, especially in situations such as lengthening for congenital short femur, in which knee subluxation and dislocation is a predictable complication.[12]

Callotasis or callus distraction is a technique of limb lengthening developed by De Bastiani *et al*.[13] in which the callus formed in response to a proximal submetaphyseal

corticotomy is slowly distracted by the use of dynamic axial fixator (Orthofix). The distraction begins 10-14 days post corticotomy. When the required lengthening is obtained, the fixator is held rigid until radiographic evidence of good consolidation of the callus is seen. At this time, the locking screw of the fixator is released, and dynamic axial loading is started and continued until corticalisation is demonstrated radiographically (then the device and screws are removed).

The Taylor-Spatial-Frame (Smith and Nephwe, Memphis, TN) is a computer-assisted system for limb lengthening and deformity correction. The power of the spatial frame lies in its precise control over the final limb length and alignment and its ability to correct a residual deformity. The stability of this multiplanar circular fixator permits earlier weight-bearing and provides an ideal environment for both new-bone formation and soft tissue healing.[14]

Limb length, bony translation, and bone segment rotation can be simultaneously controlled. Any deformity occurring during limb lengthening can be easily treated as a secondary residual correction without modifying the frame. As greater length is achieved, the struts are simply and easily exchanged in the office with no need for anaesthesia or sedation. Controversy exists regarding the advantages and disadvantages of the Taylor-Spatial-Frame in children. Iobst and Dahl[15] studied fifteen paediatric patients who underwent lengthening with the Taylor system concluded that the time required for lengthening was significantly greater than for six patients who were treated using the traditional Ilizarov frame with clickers. This result is believed to be related to increased bone travel with concomitant deformity correction as well as the greater stiffness of the Taylor-Spatial-Frame. Kristiansen *et al*.[16] studied 20 tibia lengthening using the Taylor-Spatial-Frame found no difference in the time required for lengthening compared with 27 tibia lengthening using Ilizarov frame. The advantages of the Taylor-Spatial-Frame for treating limb length discrepancy with deformity include the ability to simultaneous correct length, angulation, rotation and translation without the use of hinge constructs as well as the ability to fine-tune the deformity correction with residual adjustments. Even though, a high recurrence rate of 90% after the first, and 60% after the second corrective procedure, respectively, were observed in a retrospective follow-up, but nevertheless limb lengthening was found to lead to significant improvement in the quantifiable stance phase parameters of gait and limp.[17]

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