Perthes disease in a child with Silver-Russell syndrome

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Citation

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Abstract

Silver-Russell is a rare, congenital syndrome characterized by growth failure, facial dysmorphism and asymmetry. We report a case of a child diagnosed with SRS aged three who later developed Perthes disease of the left hip. The diagnosis, Management and possible association between the conditions are discussed

INTRODUCTION

Silver et al[1] in 1953 and Russell[2] in 1954 described two series of children with low birth weight, asymmetry, growth retardation and characteristic facies. The term Silver- Russell syndrome (SRS) has been adopted since the 1970's and over 400 cases have been reported worldwide, although strict diagnostic criteria do not exist probably reflecting the heterogeneous etiology of the condition[3]. Growth failure is the main abnormality and management is aimed at addressing this. We report the case of a young girl with SRS who also developed Perthes'disease of the left hip five years later.

CASE REPORT

A three year old Caucasian girl was referred to the growth clinic in her local hospital because of post natal growth below the 0.4th centile. Her birth weight was 2.1kg at 37 weeks. There were no problems in pregnancy or the neonatal period. On examination, obvious findings included a triangular face and small chin, overcrowding of the teeth in her lower jaw, clinodactyly of both little fingers and 2.5cm shortening of the right lower limb and a small right foot. There were no feeding difficulties in infancy or excessive sweating. Head circumference was normal, no café au lait spots were present and no learning difficulties were apparent. There were no other systemic abnormalities. A clinical diagnosis of SRS was made and chromosome analysis undertaken, which was normal (46XX). She was managed with a right shoe raise and nutritional support and continued to grow parallel and below the 0.4th centile. On a routine review, now aged eight, she was noted to be limping on her left leg which had developed over several months and the limb length discrepancy was now only 2cm. Orthopaedic examination revealed 10° fixed flexion deformity of the left hip and moderate restriction of range of motion. Trendelenberg's test was positive bilaterally. Thigh foot angle and trans-malleolar axis were equal. Plain radiographs of the pelvis showed Perthes' disease suggesting involvement of the left femoral head and collapsed lateral pillar with slight extrusion of the epiphysis (figures 1,2). This was entirely new in comparison to a previous pelvic radiograph 3 years previously (which was normal). A hip Arthrogram was now performed confirming severe total head involvement (figures 3 a,b,c). However as the femoral head was still well contained within the acetabulum, a trial of conservative management was agreed at an Orthopaedic case conference at Great Ormond Street Children's hospital in London. This approach resulted in a progressive improvement in the range of motion and symptomatic improvement in the left hip over the next few months. Periodic follow up has demonstrated continued clinical and radiological improvement without operative intervention.

Figure 1

Figures 1 and 2; Anteroposterior and ‘frogs legs' views showing obvious Perthes disease affecting the left hip

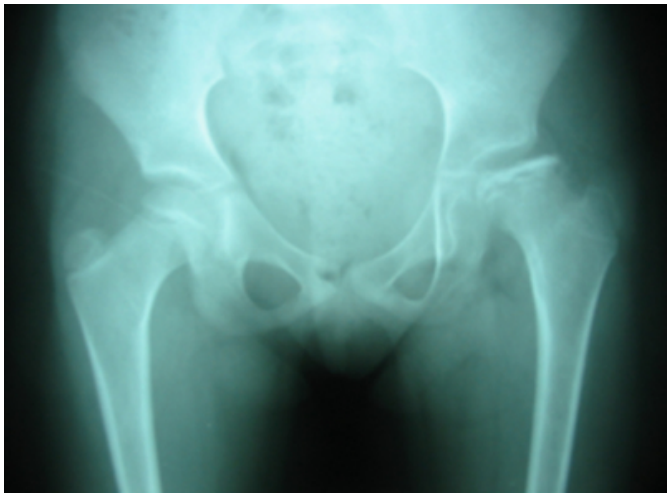


Figure 2

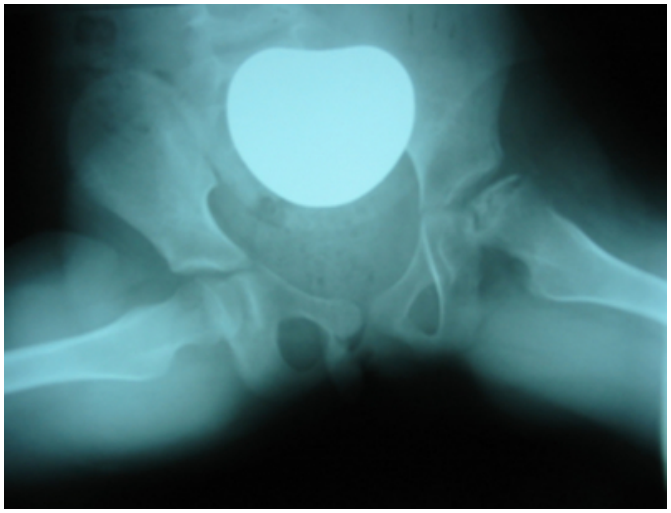


Figure 3

Figures 3 (a,b,c) Left hip arthrograms in abduction and external rotation, abduction and adduction showing signs of Perthes

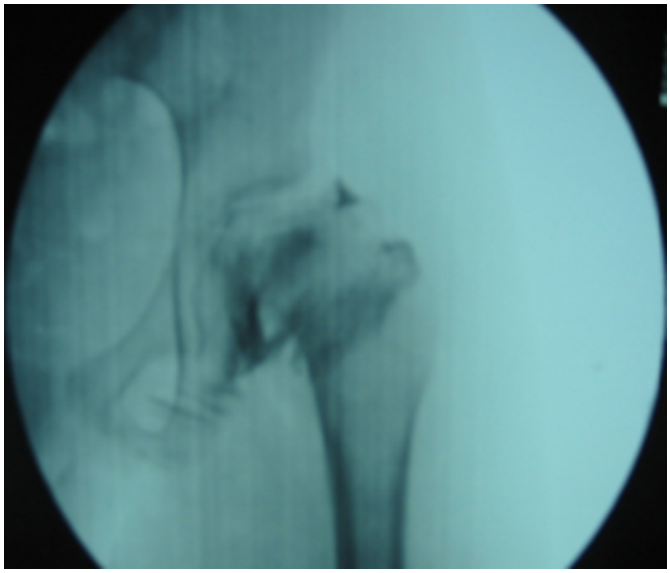


Figure 4

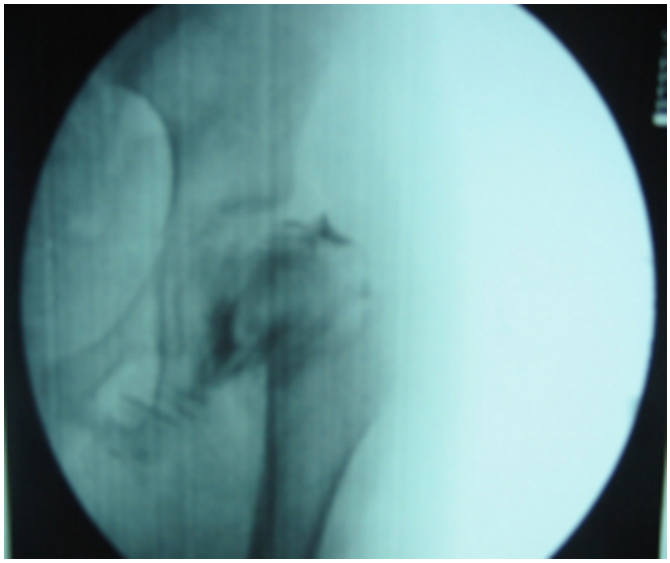
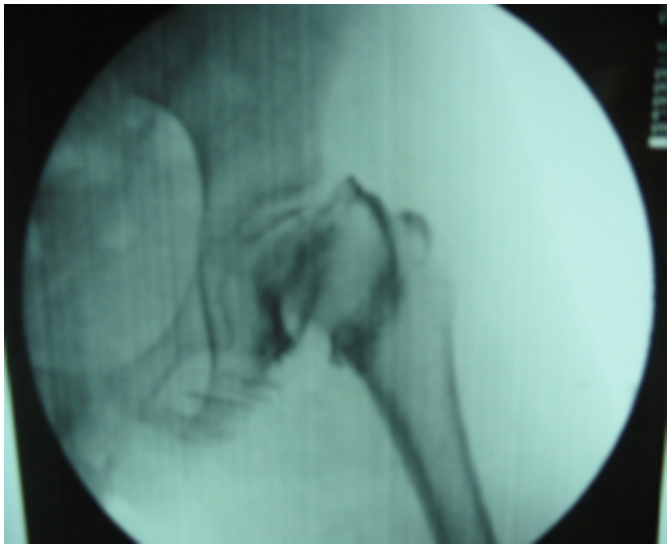


Figure 5



DISCUSSION

Silver Russell syndrome is a congenital syndrome of unknown aetiology, usually sporadic and has equal sex incidence. Various patterns of inheritance and chromosomal abnormalities have been suggested, most commonly involving chromosomes 7 and 17[3]. Approximately 10% of patients have uniparental disomy (UPD) of chromosome 7, which was not present in this child. It has been suggested that diagnosis should include at least 3 of the following 'major' criteria[4]: Low birth weight (? 2 standard deviations), poor postnatal growth, preservation of Occipitofrontal circumference, classic facies and general asymmetry. The child in our case has all 5 of these. Other criteria include feeding difficulties and fasting hypoglycaemia in infancy, clinodactyly of the fifth finger, genital anomalies and learning difficulties in one third[5]. Management comprises a multidisciplinary approach aimed at optimizing growth with appropriate nutritional support and use of growth hormone in some. Appropriate use of occupational therapy and physical therapy also needs to be made. Overall prognosis is good, especially in this case as no learning difficulties are present. The objective in this case was to prevent severe degenerative osteoarthritis in the future and followed traditional lines for Perthes disease with success.

Legg, Calve and Perthes independently described the condition which now bears their names in 1910[6]. It most commonly presents in boys aged 4-9 with unilateral limp and reduced range of hip motion and is more common in caucasians. Aetiology is unknown but predisposing factors include slipped upper femoral epiphysis, trauma, steroids, sickle crisis and developmental dysplasia of the hip (DDH)[7].

The pathophysiology is widely accepted as avascular necrosis of the femoral head due to compromise of the blood supply to the proximal femoral epiphysis and plain radiology confirms the diagnosis. Treatment goals are to reduce load bearing on the femoral head and maintain it within the acetabulum. There is little consensus on the optimum treatment strategy, the options being conservative or surgical. However, it is recognized that age at presentation over 8 is associated with a worse prognosis making surgery more likely and immediate prognosis depends on the extent of deformity before treatment[8,9,10]. The long term risk is of secondary osteoarthritis.

In this case Perthes disease developed several years after Silver-Russell syndrome was diagnosed raising the issue of a potentially rare association or pure coincidence. The genetic basis to Perthes disease is multifactorial and does not follow classical mendelian inheritance. Delayed skeletal maturity is a well recognized feature in Silver-Russell syndrome and is strongly associated with Perthes. Biomechanically, limb length discrepancy can result in abnormal weight bearing on one leg, potentiating stress on the femoral head. However, neither Perthes nor delayed skeletal maturity appears more common in cases of sporadic hemihypertrophy[11,12]. Indeed, isolated hemihypertrophy has been linked to neoplasia[13]. Therefore development of Perthes for this child may have been due to a combination of excessive weight bearing on the affected leg and delayed skeletal maturity, with or without a genetic predisposition.

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References

1. Silver HK, Kiyasu W, George J, Deamer WC. Syndrome of congenital hemihypertrophy, shortness of stature, and elevated urinary gonadotrophins. Paediatrics 1953;12:368-375
2. Russell A. A syndrome of intra-uterine dwarfism recognizable at birth with cranio-facial dysostosis, disproportionately short arms and other anomalies. Proc R Soc Med 1954;47:1040-1044
3. Megan PH, Stainer P, Preece M, Moore GE. Silver-Russell syndrome: a dissection of the genetic aetiology and candidate chromosomal regions. J Med Genet 2001;38:810-819
4. Price SM, Stanhope R, Garrett C, Preece MA, Trembath RC. The spectrum of Silver-Russell syndrome: a clinical and molecular genetic study and new diagnostic criteria. J Med Genet 1999;36:837-842
5. Lai KYC, Skuse D, Stanhope R, Hindmarsh P. Cognitive

abilities associated with the Silver-Russell syndrome. Arch Dis Child 1994;71:490-496
6. Maxwell L, Kealey D. Surgical management of Perthes disease. (Protocol) Cochrane Database of Systematic Reviews 2002, Issue 2. Art. No.: CD003829.
7. Harris GD. Legg-Calve-Perthes disease. e-medicine.com/ Orthopedic surgery. July 14, 2004.
8. Noonan KJ, Price CT, Kupiszewski SJ, Pyevich M. Results of femoral varus osteotomy in children older than 9 years of age with Perthes disease. J Pediatr Orthop. 2001 Mar-Apr;21(2):198-204.
9. Yasuda T, Tamura K. Prognostication of proximal femoral growth disturbance after Perthes disease. Clinical Orthopaedics 1996;329:244-254
10. Shigeno Y, Evans GA. Quantitative correlation between the initial and final femoral head deformity in Perthes' disease. J Pediatr Orthop B. 1996 Winter;5(1):48-54
11. Viljoen, D.; Pearn, J.; Beighton, P. Manifestations and natural history of idiopathic hemihypertrophy: a review of eleven cases. Clin. Genet 1984. 26: 81-86.
12. Stoll C, Alembik Y, Steib JP, De Saint-Martin A. Twelve cases with hemihypertrophy: etiology and follow up. Genet Couns. 1993;4(2):119-26
13. Hoyme HE, Seaver LH, Jones KL, Procopio F, Crooks W, Feingold M. Isolated hemihyperplasia (hemihypertrophy): report of a prospective multicenter study of the incidence of neoplasia and review. Am J Med Genet. 1998 Oct 2;79(4):274-8.

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