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# Spondylometaphyseal dysplasia sutcliffe type – case report

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## Summary

**Background:** The first case of spondylometaphyseal dysplasia Sutcliffe type (SMDST) from Africa is reported.

**Case report:** A boy with waddling gait was diagnosed at the age of two years as bilateral idiopathic coxa vara. Skeletal survey performed 15 months later documented beside bilateral coxa vara, metaphyseal infractions and spinal changes. The hallmark of this spondylometaphyseal dysplasia are bilateral coxa vara, metaphyseal infractions and minor spinal changes.

**Conclusions:** In terms of differential diagnosis, it is relevant that the manifestations of SMDST resemble those of non-accidental injury and idiopathic coxa vara.

**Key words:** coxa vara • non-accidental injury • spondylometaphyseal dysplasia

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## Background

Spondylometaphyseal dysplasia Sutcliffe type (SMDST) was delineated as a separated entity in 1965 [6]. Thereafter, in 1974, a father and his daughter with SMDST were reported as having a variant of spondylometaphyseal dysplasia [1]. The fifth patient with SMDST, and the third reported under the accepted designation was not described until 1989 [2]. A year later Langer et al [5] reported 7 affected persons and coined the term "Spondylometaphyseal dysplasia corner fracture type". Two further cases with SMDST, one with severe genu valgus deformity, were reported in 1992 [3]. In 1993 Kozlowski et al [4] reported three children with SMDST. A fourth patient with bilateral coxa vara had rectangular platyspondyly with smooth vertebral plates, which were different from the oval shaped vertebral bodies of SMDST. This individual probably had a variant form of SMDST. Those authors stressed that corner fractures are not a unique feature of SMDST but also occur in other complex generalized bone disorders – spondyloepimetaphyseal dysplasias.

The clinical features of SMDST are short stature and waddling gait. As there are no other dysmorphic features beside moderate shortening of stature (adult height <150 cm) bone dysplasia is often not diagnosed.

This rare disorder enters into the *differential diagnosis* of any individual with coxa vara and an underlying skeletal dysplasia. Equally, the metaphyseal infarctions could be confused with non-accidental injuries and the diagnosis of SEDST therefore warrants consideration in the assessment of children with apparent non-accidental injury (child abuse, battered child).

## Case report

The boy was referred to the Department of Human Genetics in Cape Town at the age of 2 years with bilateral coxa vara which had been detected during investigation of abnormal gait. A cleft lip and cleft palate had been repaired during infancy, but he was otherwise well. Clinically, he was normal, apart from proportionate small stature. Radiographs revealed gross coxa vara with irregularity of the metaphyses of the femoral necks. Those of the skull, spine and hands were reported as normal. The parents were not related and there were no affected kin. The child was seen again at the age of 3 years and 3 months. He still had a disturbed gait,

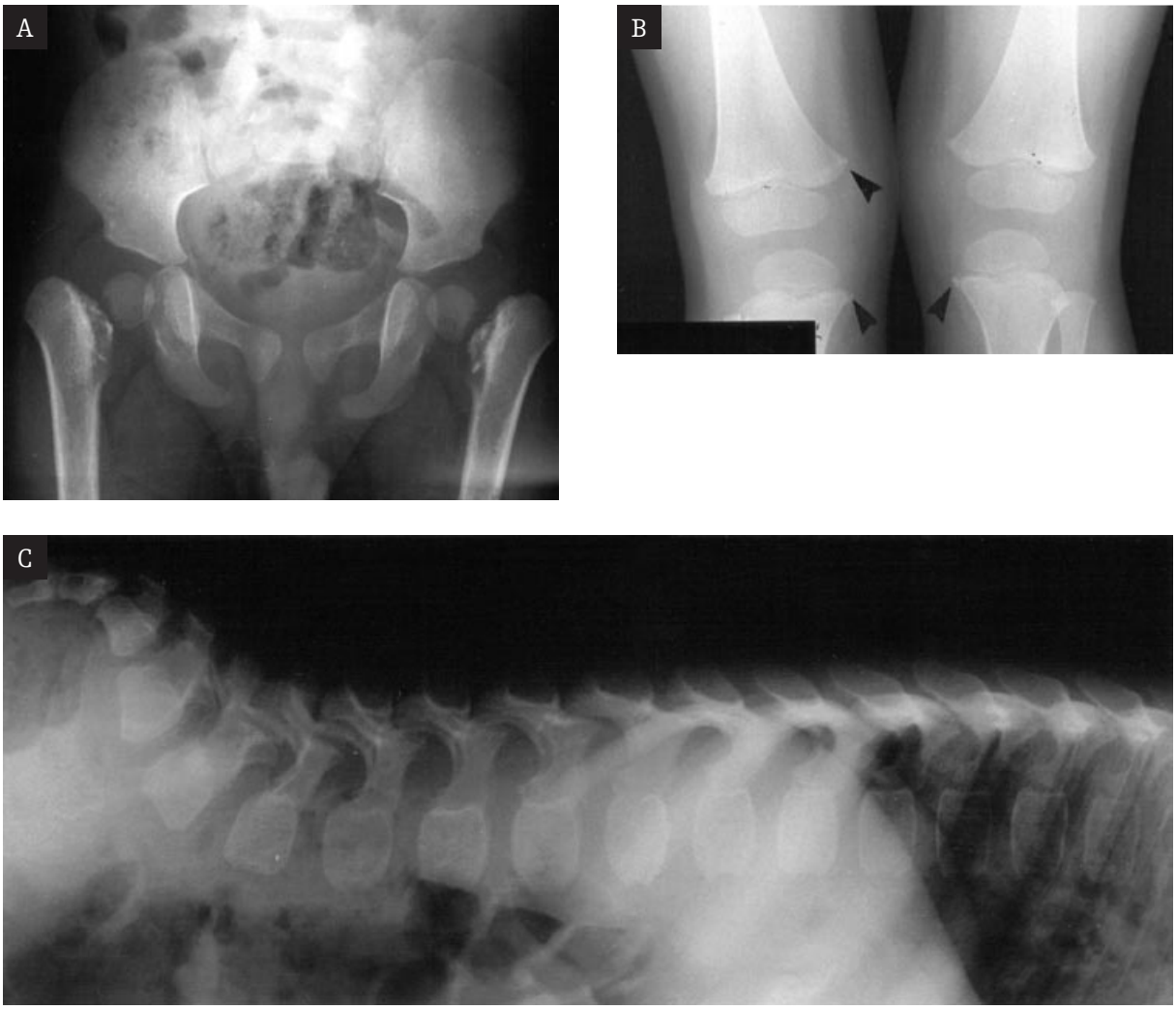


**Figure 1.** Boy 3 year 3 month-old. **A**. Bilateral coxa vara. Severe metaphyseal changes. Vertical epiphyseal plates. Short femoral necks. Club shaped proximal femora. **B**. Minimal metaphyseal changes. Small chip fractures (arrowed). **C**. Oval shaped vertebral bodies.

but was otherwise well. His height was 84.5 cm (below the 3rd centile). The upper segment was 48.5 cm, and the lower segment 46 cm. Radiographs showed severe coxa vara with vertical epiphyseal plates, extremely short femoral necks and club-shaped proximal femurs. The dysplastic changes in the spine were characterized by persisting ovoid vertebral bodies. A striking finding was "corner fractures" of the lower medial femoral metaphyses and upper tibial metaphyses (Fig 1A–C).

## Discussion

SMDST is characterized clinically by short stature and a waddling gait. The diagnostic radiographic findings consist of *bilateral coxa vara, minor to moderate spinal changes, progressive metaphyseal dysplasia* and *corner fractures* [1–3, 5, 6]. The most important radiographic feature is bilateral severe coxa vara with grossly normal femoral heads which are separated from the femoral shaft by a vertical growth plate. In two reported cases hip radiographs performed in the first year of life were described as normal [5]. The spinal changes are characterized by preservation of the infantile oval shape of the vertebral bodies during the first few years of life. Later, some flattening and anterior wedging of the vertebral bodies is apparent. Metaphyseal involvement is characterized by corner fractures and metaphyseal irregularities. Corner fractures present as small triangular bone fragments at the periphery of the metaphyses adjacent to the growth plate and are usually already evident when coxa vara is detected. Metaphyseal irregularity appears later. Both metaphyseal abnormalities progress with age.

What are the minimum criteria for diagnosis of SMDST? Metaphyseal changes of the proximal femoral metaphyses with coxa vara are the most important radiographic sign although in one of the cases documented by Langer et al [5] and in one of Kozlowski et al [4] only one hip was affected. Corner fractures are the most frequent and evident metaphyseal abnormalities. Irregularity of the metaphyses is very variable and in the first few years of life the changes may be minimal. Similarly, spinal involvement may be easily overlooked if not carefully sought. Persistence of infantile (oval) shape of the vertebral bodies in the first few years of life may precede the irregularity of the vertebral plates and flattening of the vertebral bodies. As few of the patients with congenital coxa vara have long term skeletal survey, the

relationship between isolated idiopathic or congenital coxa vara and SMDST is uncertain. They may represent the spectrum of the same disorder, and it is possible that molecular investigations will eventually answer this question.

**The differential diagnosis** of SMDST is with other bone dysplasias presenting with coxa vara and/or corner fractures [4]. In all these disorders the pattern of bony changes including the spine and metaphyseal lesions is different and confusion with other conditions is unlikely. Corner fractures are important radiographic sign in the battered child syndrome, Menkes kinky hair syndrome and scurvy. However the clinical history, phenotype of the patient, and different pattern of other bony changes is unlikely to cause confusion.

## Conclusions

We have reported the case of a child with waddling gait diagnosed at the age of 2 years as idiopathic coxa vara. Skeletal survey performed 15 months later documented SMDST. The case reported is an example that in a child with a waddling gait and bilateral coxa vara bone dysplasia and non-accidental injury should be excluded, before the diagnosis of idiopathic coxa vara is accepted.

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