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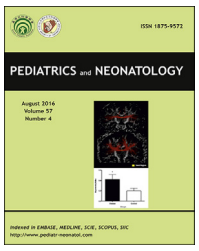

Images

# Spondylocarpotarsal synostosis syndrome: "Bat wings" spinal fusions and "ladybug" carpal coalitions

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A pediatrician noticed a hump in a 1-year-old girl during a chest examination for cough and fever. The girl had suffered from intrauterine growth retardation and had been born preterm at 34 weeks, through a caesarean section, in the context of preeclampsia. The child's weight was 1.88 kg at birth. Growth rate was slower than expected for age (at 2 years and 6 months, height was on the 2.9th percentile and weight was on the 1.8th percentile; at 3 years and 6 months, there was slowing in growth with height on the 0.4th percentile and weight on the 1.8th percentile; and at 4 years and 6 months, there was further slowing in growth with height on the 0.1th percentile and weight on the 0.4th percentile). Radiographs of the spine were taken, and in view of the findings, computed tomography (CT) was performed and surveillance was extended to the remainder of the skeleton. Radiographs of the spine demonstrated left convex thoracolumbar scoliosis, with a left rib hump (Fig. 1). CT demonstrated multiple vertebral fusions (the fusion of posterior elements resembling bats with their wings expanded on 3D reconstruction, Fig. 2A). Surveillance of the hands demonstrated bilateral symmetric double osseous carpal coalitions (CC) of the hamate-capitate and lunate-triquetral bones, resembling a "ladybug" (Fig. 2B).

The association of these findings suggests the clinical–radiological diagnosis of spondylocarpotarsal synostosis syndrome (SCT), an extremely rare skeletal disorder characterized by progressive vertebral, carpal, and tarsal fusions. Individuals with SCT are short, have frontal bossing, scoliosis and/or kyphosis, anteverted nares, and potentially a cleft palate and in some cases hearing loss, problems with tooth enamel, or hyperlaxity.[1] The majority of affected individuals show mutations in the FLNB gene, which encodes a cytoskeletal protein known as filamin B. The result of the mutation is an abnormally short filamin B protein that is unstable and breaks down, acting as a stimulus for

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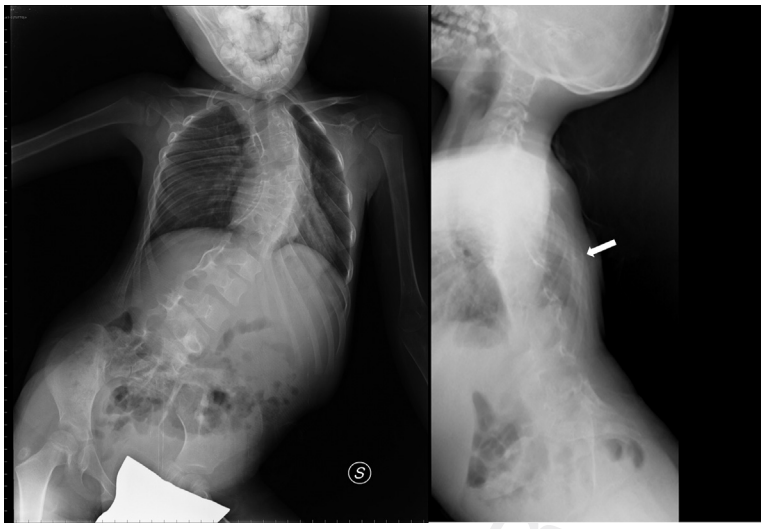

**Figure 1** Conventional radiograph of the spine (posterior-anterior and lateral view) showing scoliosis with rib hump (white arrow).

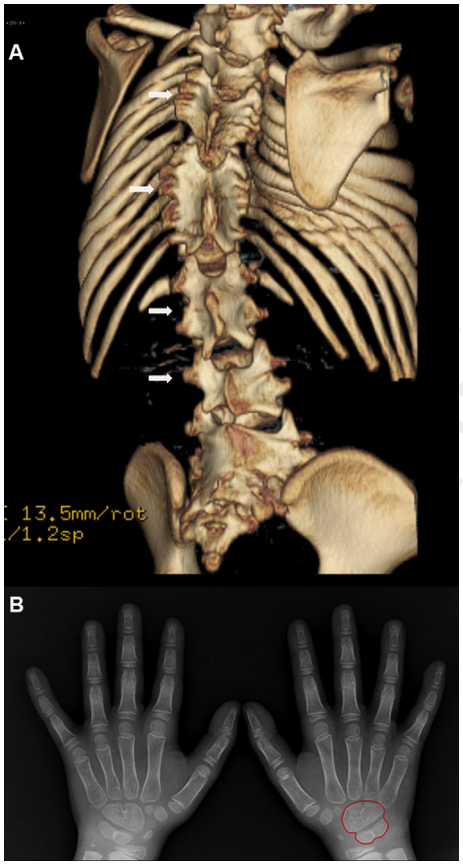


**Figure 2** **A)** 3D reconstruction of a CT scan showing multiple vertebral body fusions. The fusion of posterior elements resembles bats with their wings expanded (white arrows); **B)** conventional radiograph of the hands (posterior-anterior projection) showing a bilateral identical carpal coalition of the hamate-capitate and lunar-triquetral bones that resembles ladybug shape.

the ossification of cartilage. SCT due to FLNB mutations exhibits an autosomal recessive inheritance pattern. In a few individuals with no FLNB mutations, the syndrome exhibited an autosomal dominant inheritance pattern.[2] Mutations in MYH3, which encodes embryonic myosin heavy chain 3, have been detected in some families with autosomal dominant SCT.[3] However, in some cases, the specific genetic mutation remains unknown.[2] Our patient underwent genetic study, with testing for mutation of the FLNB gene, but no specific genetic mutation could be demonstrated.

Routine skeletal surveillance should be considered in children with congenital scoliosis to avoid missing a number of bony dysplasias and complex syndromic causes. Thus, SCT may potentially represent an underdiagnosed syndrome.

## Conflicts of interest

All authors have contributed to the manuscript and approved the final submission. All authors have no funding or conflict of interest to declare.

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