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CASE REPORT

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Spondylocarpotarsal synostosis syndrome (not rare but infrequently recognised syndrome)

Zespół zrostowy kręgosłupa, nadgarstka i śródstopia (rzadko rozpoznawany, nierzadko występujący)

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Summary

Spondylocarpotarsal synostosis syndrome – a rarely recognised entity – is characterised by malsegmentation of the spine and carpal/tarsal fusions (1,2,5–9). The main reason for the rarity of its diagnosis is that radiographs of the hands and feet are not routinely performed in children with scoliosis and/or kyphoscoliosis, and fusion of the carpal/tarsal bones may not be evident in preschool children. We report four patients with spondylocarpotarsal synostosis syndrome; the diagnosis in three of these was not made until radiographs of the hands were performed. The amount of scoliosis was much less evident in two of these patients (sibs) than in the others.

Key words: Scoliosis • Malsegmentation • Unsegmented bar • Carpal fusion • Spondylocarpotarsal synostosis syndrome

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Background

Scoliosis due to malsegmentation is a common spinal abnormality. Its syndromic association with carpal/tarsal fusion is often missed. We report four patients with scoliosis due to vertebral malsegmentation and spinal bars, associated with carpal and/or tarsal fusions; the scoliosis was very severely progressive in two patients. The diagnosis of spondylocarpotarsal synostosis syndrome (SSS) was not made in three patients until the radiographs of the hands and feet were performed.

Case reports

Patient I

This 16 year-old girl presented for orthopaedic consultation for evaluation of spinal deformity. She was born to a 29 year-old primigravid mother after a normal pregnancy. The 35 year old father was a second degree relative. Birth weight, length and head circumference were around the tenth centile. Her motor development was delayed with walking commenced at two years. Mental development

was normal. She was seen at six years for evaluation of back deformity and a diagnosis of congenital scoliosis was made.At 16 year follow up her height was 138cm (-3SD), weight 49kg (-1SD), head circumference 52 cm (normal). Her face, vision and hearing were normal. There was a kyphosis with scoliosis convex to the right, and generalised ligamentous laxity. The abdominal and renal ultrasound were normal. Biochemical investigations including routine blood and urine examinations, serum Ca and P and karyotype were normal, Radiographic examination documented scoliosis convex to the right with T4-T5 and T6-T10 block vertebrae and a unilateral unsegmented bar on the left. (Fig 1A & B) There were also bilateral carpal and tarsal fusions (Fig 1C & D).

Patient II

This 4 year-old girl with minor dysmorphic features and development delay presented for evaluation of spinal deformity.She was the result of the 5th pregnancy (third child) of a 33year-old mother and 35 year-old father. Delivery was normal. The parents were not related but from the same geographical area. Two older sibs are normal. Birth weight was 2900 g, length 47 cm. Because of short stature and dysmorphic features a syndrome was suspected but no diagnosis was made. At 4 years 9 months development of motor skills and language were delayed. Height was 86 cm (-4.9SD = 11cm below 3rd centile), weight 11.5 kg (just above 25th centile). Upper segment 50.3cm (-4.9SD), lower segment 36cm (-4.7SD). Pertinent physical findings included proportional shortening of all the segments of the body, left sided thoraco-lumbar scoliosis, prominent forehead, wide nasal bridge, epicanthal folds, long philtrum and low set ears. There was premature loss of the deciduous teeth. The joints were hypermobile with the exception of the forearms where restriction of supination was present. There was minor valgus deformity of the knees, and flat feet with abduction of the forefeet. There was simian crease in the palm of the left hand. Vision and hearing were normal. Renal and abdominal ultrasound were all normal. Routine blood and urine examinations, thyroxin and parathormone levels and markers of bone turnover were all normal. The karyotype (Fish) was 45X (2.5%) 46 XX (97.5%). Radiographic examination documented thoraco-lumbar scoliosis convex to the left. There were block vertebrae T7-11 with bilateral unsegmental bar. There was narrowing of the disc spaces T2-3, L2-3, L5-S1. 11 ribs were counted (Fig 2A,B). There were bilateral carpal and tarsal fusions (Fig 2C,D).

Patient III

This boy was investigated at 5 months of age because spine deformity was noticed. Radiographs at that time showed regions of fusions of the neural arches of contiguous vertebrae (at C4-6, T12-L1, L4-S2) with abnormal proximity of the related vertebral bodies. Deficient development of neural arches was seen at T11-12, with related wide separation of the vertebral bodies. Segmental lordosis and kyphosis have occurred secondary to these abnormalites (Fig 3A). There were no other abnormalities on physical examination and mental development was normal. At five years studies showed the changes of growth, but little change in the abnormalities, except for extension of fusion at L1-2 and abnormally wide separation, with neural arch maldevelopment, at L2-4 (Fig 3B). Radiographs of the hands documented early fusion between capitate and hamate (Fig 3C). Clinical examination at 13 years and 10 months showed thoraco-lumbar scoliosis with multiple curves which are fairly well balanced. Thoracic lordosis was prominent, resulting in major protuberance of the scapulae, and the whole thoraco-lumbar spine showed very restricted movement. Height 129cm (3rd centile=147 cm) Upper segment 61 cm, lower segment 68 cm). The patient has no problems with daily activities. He plays sport and rides a horse well, but he has been warned to avoid contact sports. He shows no weakness or restricted mobility related to the hands, wrists or forearms. His school achievement is average and he relates well to classmates.

Patient IV

This girl is the younger sister of Patient III; there is another normal sib and there is no relevant family history. The patient was investigated at three months because of spinal deformity of the affected brother. Radiographs at three months showed vertebral fusions in four areas with segments of scoliosis at the cervico-thoracic and lumbo-sacral junctions, both convex to the right side. There was also lower thoracic lordosis (Fig 4A,B). Because of affected brother – CASE 3 – SSS syndrome was diagnosed. Follow up at 8 years and 6 months; height 113cm (3rd centile=117 cm); upper segment 56 cm, lower segment 57 cm. The patient had no problems with daily activities and played sport normally. There was no clinical hand or wrist abnormality. Scoliosis with multiple curves was present; the curves were pertly balanced, but elevation of the left shoulder was evident. There was some thoracic lordosis. There was moderate restriction of thoraco-lumbar spine movement. Mental development and physical examination otherwise were normal.

Radiograph of the left hand at 8 years 7 months showed elongation of the triquetrum in the transverse plane and fusion of this bone to the lunate. Bone age was 6years 10 months (Fig 1C).

Discussion

Congenital scoliosis – a common abnormality – is usually due to malsegmentation or malformation of the spine. It occurs as an isolated anomaly or as a major component of a syndromic association [11]. The latter includes spondylocostal dysplasia (SCD), spondylothoracic dysplasia (STD), ischiovertebral dysplasia (IVD), cerebrofaciothoracic dysplasia (CFTD), Robinow syndrome (RS), and other rare private syndromes [3,4]. All of them are characterised by variable vertebral abnormalities (block vertebrae, hemivertebrae, butterfly vertebrae), severe rib changes (missing, bifid, fused) and **absence of carpal/tarsal fusion**. Scoliosis develops usually later in life and is less severe than in SSS.

In SSS scoliosis is malignantly progressive early in life and steadily during the growth. SSS should be suspected in children of short stature with congenital progressive scoliosis. Uncharacteristic slightly dysmorphic face, cleft palate and hearing defects may be present. The mental

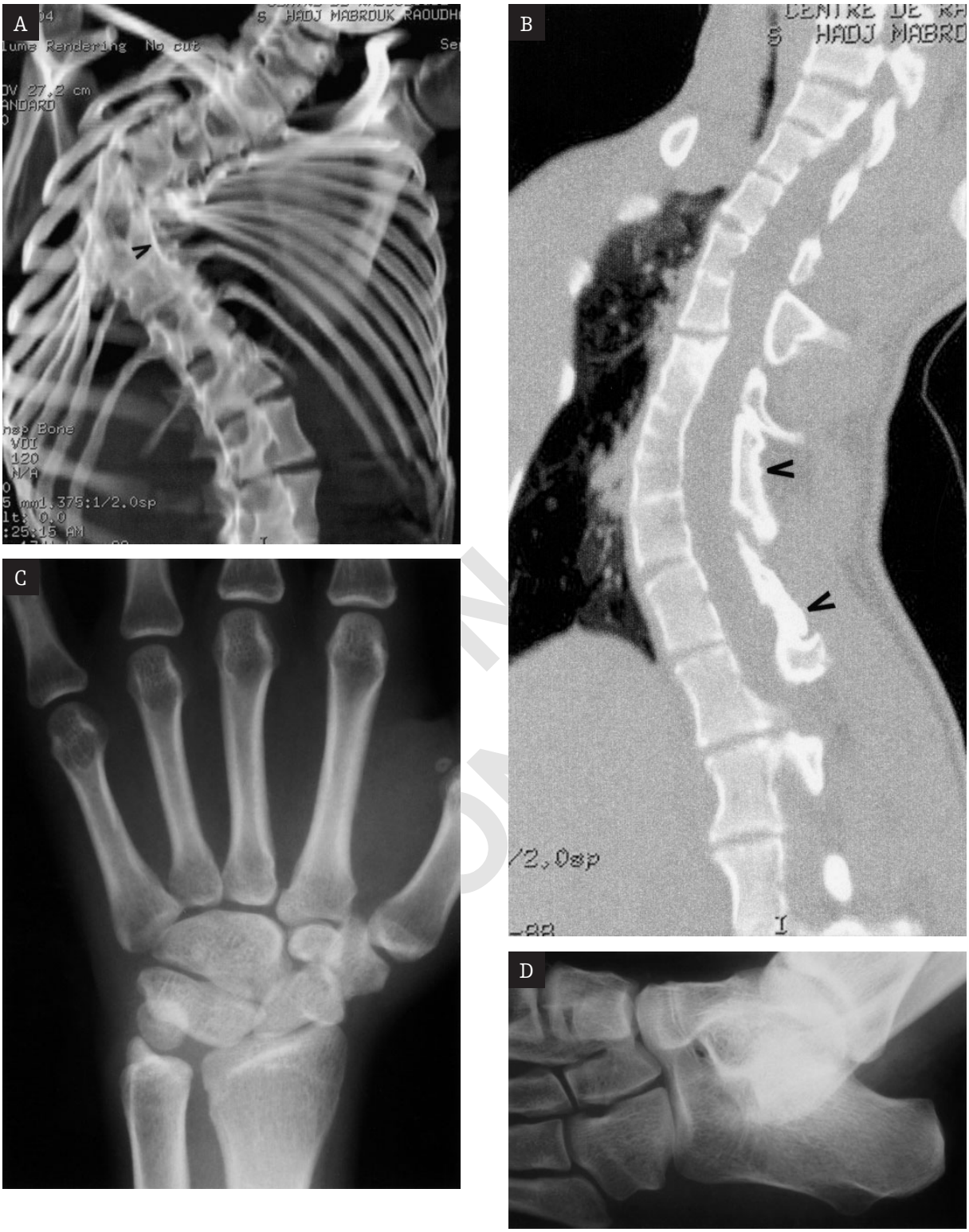


Figure 1 A-D. **A**. & **B**. Scoliosis convex to the right. Note unilateral unsegmented spinal bar on the left -T7-T11 (arrowed), slightly hypoplastic/ dysplastic ribs, fracture at the distal end of 10 and 11 left ribs, block vertebrae T4/T5 and T6-T10. **C**. Left Hand. Synostosis between capitate-hamate and lunate-triquetrum **D**. Left foot. Multiple fusions: talo-calcanear, talo-navicular, naviculo-calcanear, cuneiform 2 – cuneiform 3.

Rycina 1 A-D. **A**. & **B**. Boczne, prawostronne skrzywienie kręgosłupa. Lewostronny zrost łuków kręgowych T7-T11 (strzałki). Cienkie, nieznacznie zniekształcone żebra, złamania w dalszej części 10 i 11 lewego żebra, zrost kręgów T4/T5 I T6-T10. **C**. Lewa ręka. Zrost kości główkowatej i haczykowatej, księżycowato-trójgraniastej. **D**. Lewa stopa. Liczne zrosty kości śródstopia: skokowo-piętowej, skokowo-łódkowatej, łódkowato-piętowej, klinowatej 2,3.

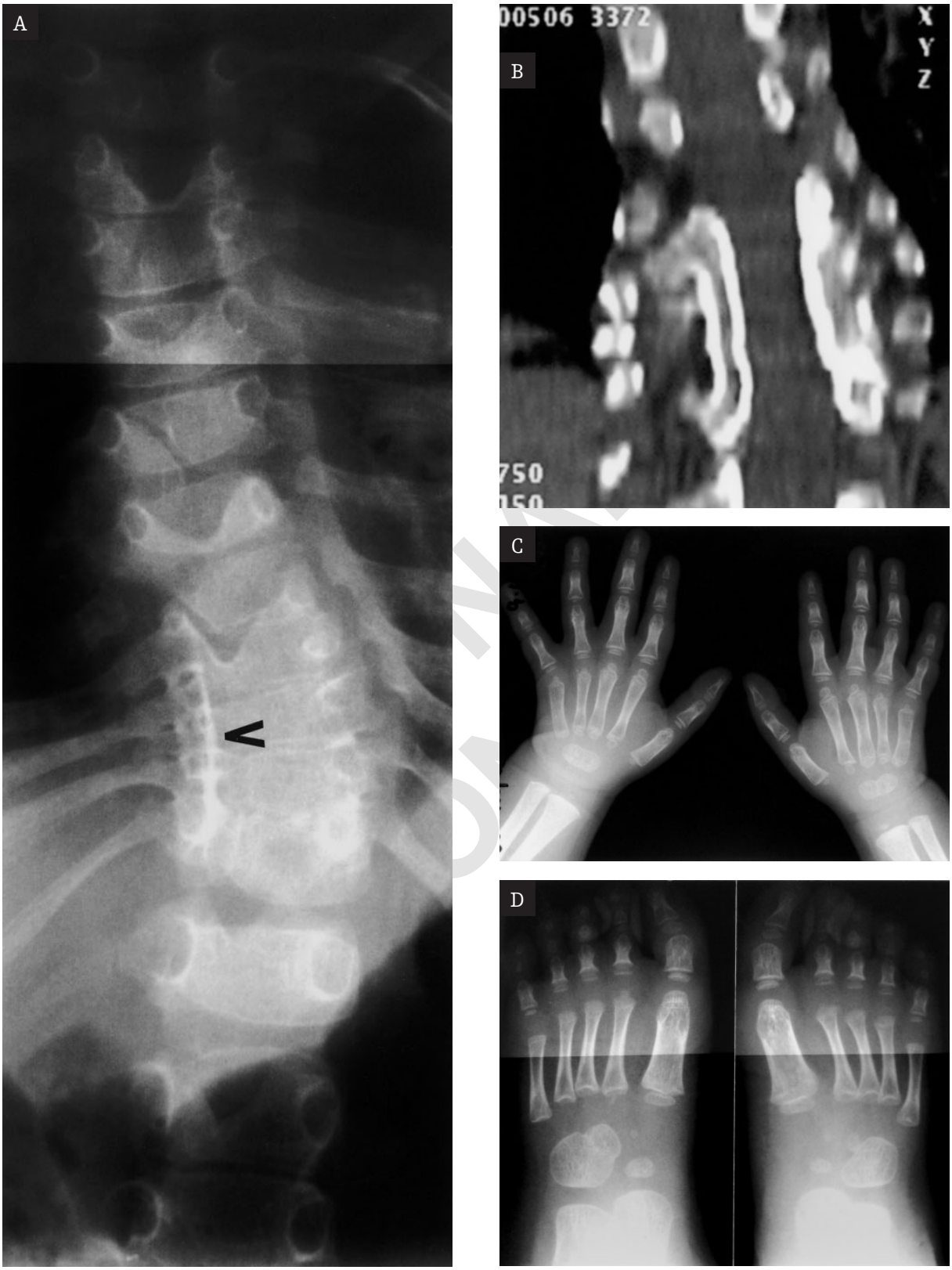
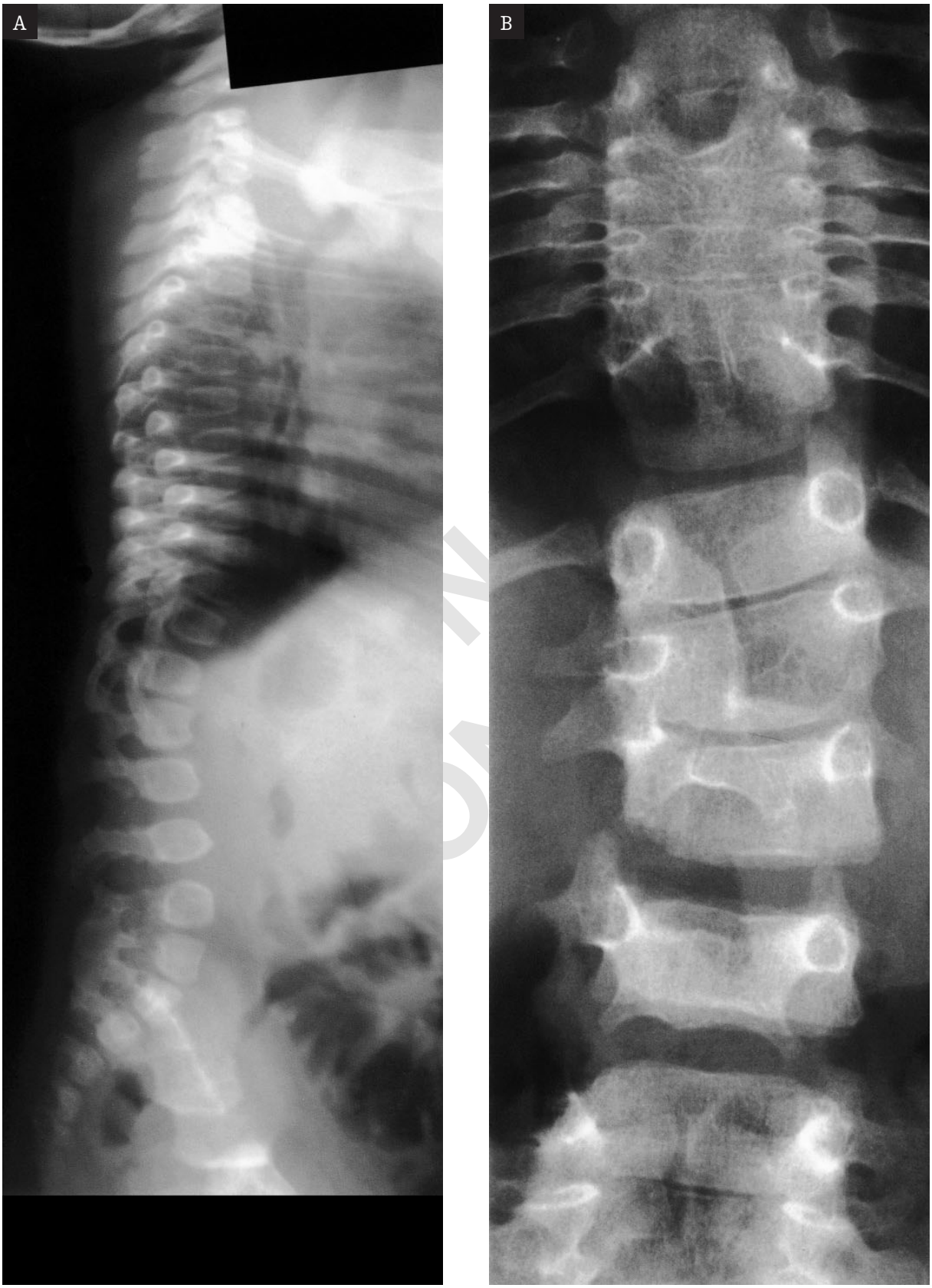


Figure 2 A-D. **A**. Block vertebrae T7-T11. Note unsegmented spinal bar T6-T11 (arrowed). **B**. 3D reconstruction lower thoracic spine shows bilateral unsegmented bars at the level T6-T11. **C**. Hands. Capitate-hamate fusion. **D**. Feet. Cuboid-cuneiform 3 fusion.

Rycina 2 A-D. **A**. Zrost kręgów T7-T11. Prawostronny zrost luków kręgowych T6-T11 (strzałka). **B**. Rekonstrukcja 3D dolnego odcinka kręgosłupa piersiowego uwidacznia obustronne boczne zrosty łuków kręgowych T6-T11. **C**. Zrost kości główkowatej-haczykowatej. **D**. Zrost kości sześciennej i klinowatej 3.



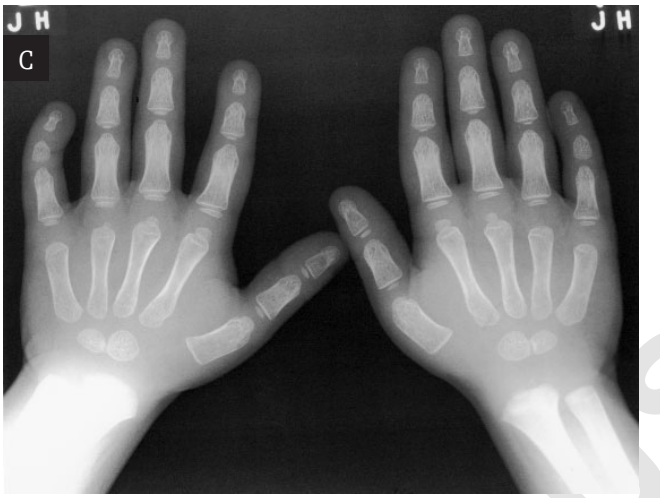


Figure 3 A-C. **A**. Lateral spine at 5 months. Fusion between neural arches and narrowing of disc spaces, especially posteriorly at C4-6. Abnormally wide separation of vertebrae at T11-12 with maldevelopment of neural arches. Neural arch fusion at T12-L1 with partial fusion of vertebral bodies posteriorly: similar fusion of neural arches at L4-S2. Pointed shape of vertebral bodies at C6 and C7 anteriorly and T8 posteriorly. Lordosis at T6-11, kyphosis at T12-L4. **B**. Lateral lower thoracic and lumbar spine at 5 years. Additional features to above, except for growth changes are: abnormal separation, with deficient development of neural arches at L2-4, abnormal proximity T12-L1 and neural arch fusion T12-L2 and slight scoliosis, convex to the left at T12-L3. **C**. Hands at 5 years. There is early fusion between capitate and hamate. Bone age 4 years.

Rycina 3 A-C. **A**. Boczny kręgosłup w wieku 5 miesięcy. Zrost łuków kręgowych i zwężenie przestrzeni międzykręgowych, zwłaszcza w części tylnej na poziomie C4-6. Znaczne poszerzenie przestrzeni międzykręgowej T11-12 ze zniekształceniem rozwojowym łuków kręgowych. Zrost łuków kręgowych T12-L1 z częściowym zrostem tych kręgów w odcinku tylnym. Podobny zrost łuków kręgowych L4-S2. Zaostrzony kształt kręgów C6 I C7 w odcinku przednim oraz T8 w odcinku tylnym. Lordoza T6-11 i kyphoza T12-L4. **B**. Kręgosłup lędźwiowy I dolny piersiowy w wieku 5 lat. W porównaniu z Ryc. 3A oprócz wzrostu stwierdza się: zwiększenie odległości I niedorozwój łuków kręgowych L2-4, zbliżenie T12-L1, zrost łuków kręgowych T12-L2 oraz niewielkie lewostronne skrzywienie na poziomie T12-L3. **C**. Ręce w wieku 5 lat. Wczesny zrost kości główkowatej i haczykowatej. Wiek kostny 4 lata.

development of the patients is normal. Radiographic documentation of severe scoliosis subsequent to failure of normal spine segmentation, resulting in block vertebrae, and fusion of posterior elements makes SSS highly probable. This probability is enhanced if a unilateral, bilateral or rarely posterior unsegmented bar is present. Rib abnormalities in SSS are not primary but secondary to scoliosis. Therefore the ribs are less affected in SSS than in SCD, STD, IVD, CFTD and RS. Fusion of the ribs is not a feature of SSS. There are usually 12 slightly deformed and dysplastic ribs. **Radiographs of the hands and feet establish the diagnosis**.

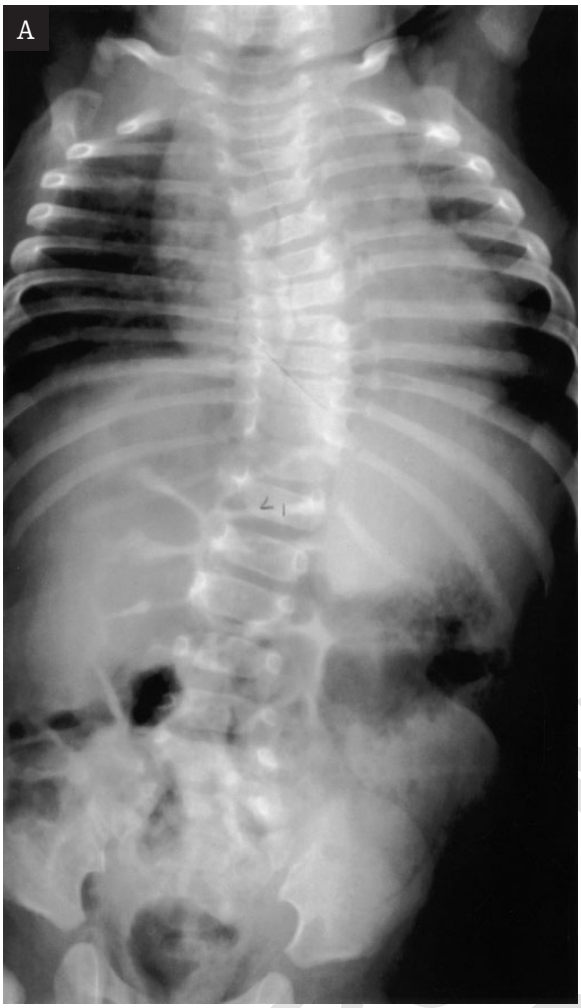
Carpal and tarsal fusions do occur in tarsal/carpal coalition syndrome characterised by fusion of the carpals, tarsals, and phalanges. Short first metacarpals may cause brachydactyly, and humeroradial fusion may occur [12]. Vertebral anomalies are not a feature of this syndrome.

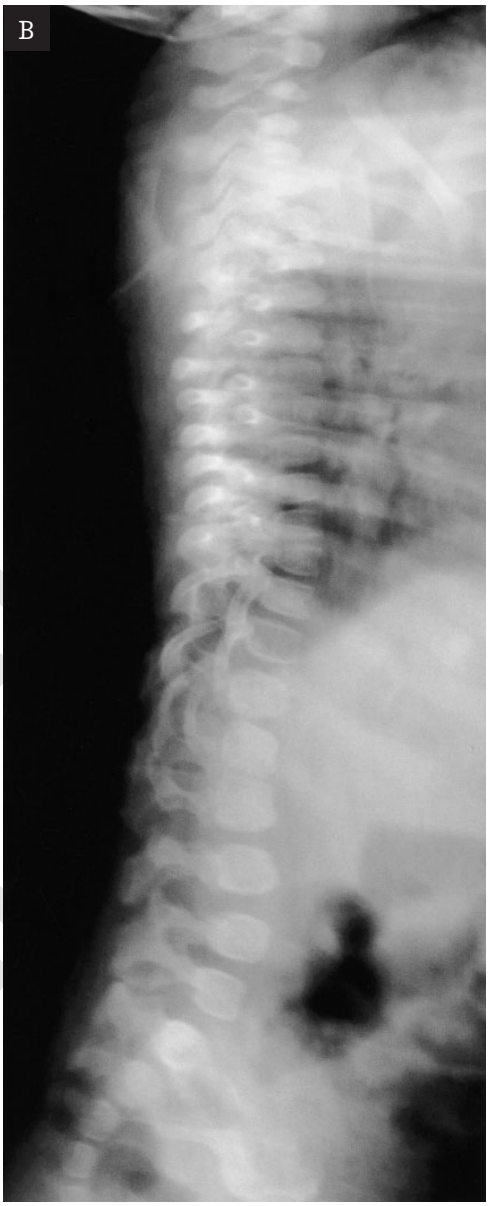
The most common isolated carpal fusions are capitate/hamate and lunate/triquetrum. Fusions going across rows are always suggestive of a syndromic association. In the feet the most common fusions are talo-calcaneal and calcaneo-navicular. They may be syndromic or non-syndromic. Tarsal fusions involving cuneiforms, cuboids and metatarsals are nearly all syndromic [10].

About 20 cases of SSS were reported up to 2004. SSS is an autosomal recessive disorder with location of the gene in chromosome 3p14 [8].

Conclusion

In patients with congenital scoliosis, radiographs of the hands and feet should be performed routinely. They are important not only in establishing SSS, but also help to confirm or exclude other bone dysplasias such as Robinow syndrome, dyschondrosteosis or some forms of enchondromatosis, all of which may be associated by scoliosis.





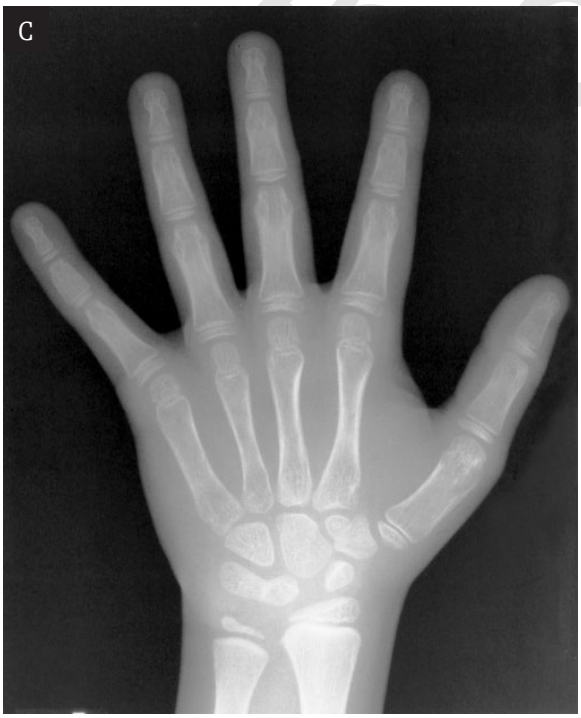


Figure 4 A-C. **A**&**B**. Spine at 3 months. Major scoliosis T2-L1, convex to left. Fusion neural arches C4-5. Pedicles T6-8 right side clustered and probably fused, perhaps fusion on the left side too. Lordosis T7-10, narrow disc spaces T8-9. Narrow disc T12-L1 right side. Abnormal shape and position T12-L1 pedicles both sides. Neural arch fusion L4-5. **C**. Left hand 8 years and 7 months. Elongated triquetrum transversely and fusion with lunate. Bone age 6 years 10 months.

Rycina 4 A-C. **A**&**B**. Kręgosłup w wieku 3 miesięcy. Znacznego stopnia lewostronne skrzywienie na poziomie T2-L1. Zrost łuków kręgowych C4-5. Nasady łuków T6-8 po stronie prawej znacznie zbliżone, prawdopodobnie zrośnięte. Prawdopodobny zrost po stronie lewej. Lordoza T7-10, zwężenie szpary międzykręgowej T8-9 oraz T12-L1 po stronie prawej. Nieprawidłowe położenie I zniekształcenie nasady łuków T12-L1. Zrost łuków kręgowych L4-5. C. Lewa ręka w wieku 8 lat 7 miesięcy. Zrost poprzeczne wydłużonej kości trójgraniastej z kością księżycowatą. Wiek kostny 6 lat 10 miesięcy.

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