Esetismertetés

Osteogenesis imperfecta and spondylo-costal dysplasia in a male child

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INTRODUCTION – Co-existence of osteogenesis imperfecta with another bone dysplasia is rare. That of osteogenesis imperfecta and spondylo-costal dysplasia has not yet been reported.
CASE REPORT – The authors present a 13 year-old boy. He was disproportionate with shortening of the upper segment, and the upper and lower extremities, and he had dysmorphic face. His parents were healthy.
Radiographs revealed osteogenesis imperfecta and spondylo-costal dysplasia.
CONCLUSION – Patients with two bone dysplasias have a much worse prognosis. Superimposition of any skeletal dysplasia on abnormal osteogenesis enhances the disposition to deformities and fractures.

osteogenesis imperfecta, spondylo-costal dysplasia

Osteogenesis imperfecta és spondylocostalis dysplasia egy fiúgyermek esetében

BEVEZETÉS – Az osteogenesis imperfecta és más típusú csontdysplasia együttes előfordulása ritka. Osteogenesis imperfecta és spondylocostalis dysplasia együttes megjelenését még nem közölték.
ESETISMERTETÉS – A szerzők egy tizenhárom éves fiúgyermek esetét mutatják be, akinél szembetűnő volt a törzs és a végtagok rövidsége, valamint a torz arc. A fiú szülei egészségesek voltak.
A röntgenképek alapján megállapítható volt a rendellenes csontképzés és a spondylocostalis dysplasia.
KÖVETKEZTETÉS – A kettős csontdysplasiában szenvedő betegek prognózisa rosszabb. A kóros osteogenesishez társuló egyéb csontdysplasia növeli a deformitások és a törések veszélyét.

osteogenesis imperfecta, spondylocostalis dysplasia

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Association of two skeletal dysplasias or a skeletal dysplasia with dysostosis in a child of healthy parents is uncommon. We report a boy with a negative family history who showed combined features of osteogenesis imperfecta and spondylo-costal dysplasia.

CASE REPORT

The propositus was an 11 year-old Czech boy referred to the ambulant centre for diagnosis *(Fig. 1.)*. The patient is the only child of young non-consanguinous, healthy parents whose family history

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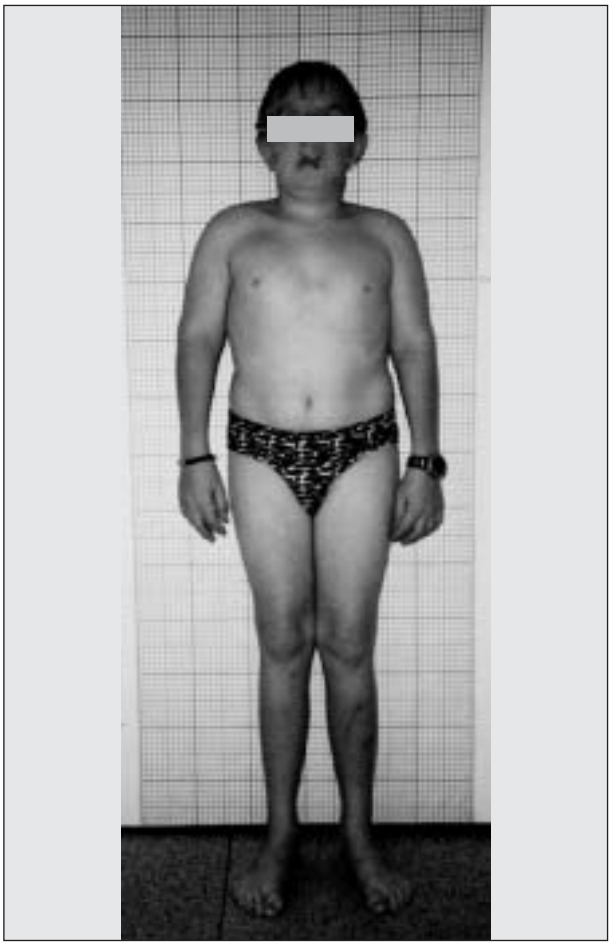

Figure 1. A photograph of the patient, a 13 years nine months old boy. Note short trunk in comparison with the lower extremities

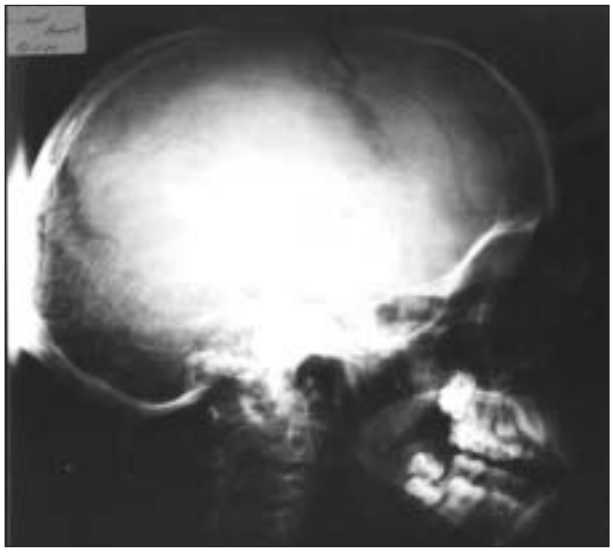

Figure 2. "Helmet skull" with multiple Wormian bones. Hypoplastic-dysplastic teeth

does not include any bony disorders. He was born at 38 weeks gestation with a birth weight 2450 g, length 47 cm. Wide open sutures and large fontanelles were noted at birth, and closure of the large fontanelle was delayed until the age of 10 years. Ultrasound examination of the brain after birth and CT of the brain at the age of five years were normal. He sustained fractures, left femur at six years and right clavicle at 10 years. At the age of 13 years nine months his height was 139.7 cm (<3rd percentile), weight 45 kg (>97th percentile). He was disproportionate with shortening of the upper segment (–2.9 SD) and predominantly rhizomelic shortening of the extremities: arms –3 SD, forearms –1.7 SD, hands –0.4 SD. There was also some shortening of the lower extremities –1.7 SD. He was brachycephalic with a dysmorphic face, slight exophthalmus and long philtrum. The palate was high and the teeth were hypoplastic/dysplastic. The sclerae were white. Radiographs revealed changes consistent with osteogenesis imperfecta and spondylo-costal dysplasia *(Figs. 2–4.)*. The former consisted of generalised osteoporosis, acetabular protrusion, helmet type of cranium with multiple Wormian bones, triangular facial bones and dentinogenesis imperfecta. The latter presented as butterfly vertebra T5, malsegmentation of T6–11 and scoliosis. There were 13 ribs on the left and the 5th and 4th left ribs were fused. The routine blood and urine examination were normal.

Discussion

The clinical history, phenotype of the patient and radiographic examination are characteristic of osteoporosis imperfecta type IV, and a dysostosis manifested by malsegmentation of the spine and rib abnormalities. There are two major subtypes of the latter: the spondylo-thoracic dysostosis (Jarcho-Levin syndrome)[1] usually with early fatal outcome, and the benign autosomal dominant or autosomal recessive spondylo-costal dysostosis (SCD)[2]. Spondylot-horacic dysostosis is characterised by ribs which flare in a fanlike pattern. Spondylo-costal dysostosis may present with minor or dramatic rib malformations but do not have a fanlike thoracic

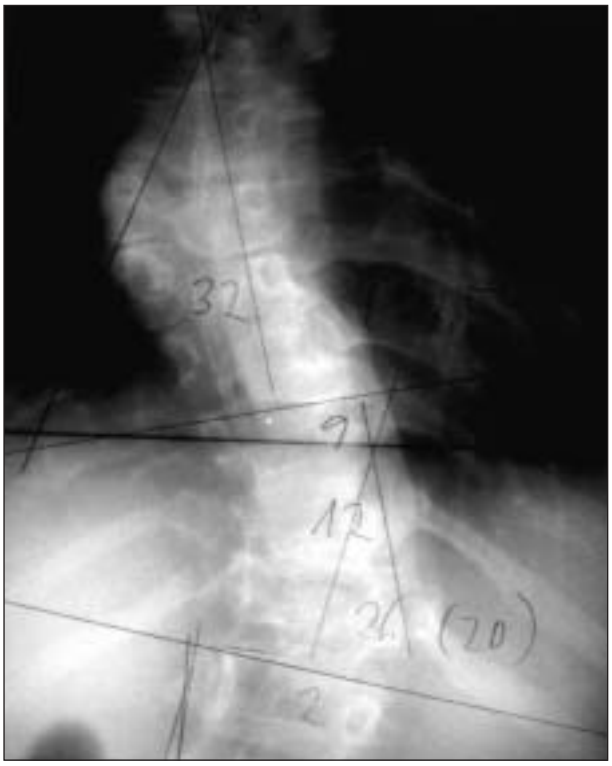

Figure 3. Malsegmentation of the thoracic spine. Butterfly vertebra T5. Scoliosis

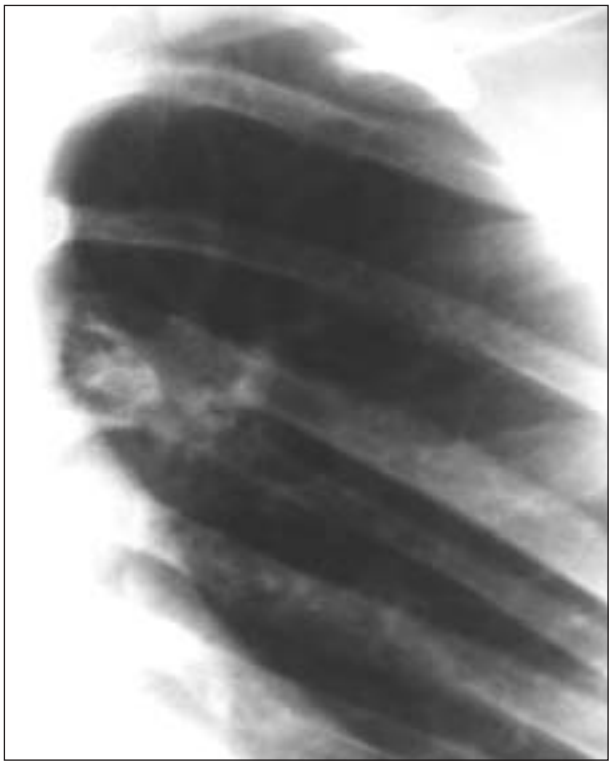

Figure 4. Posterior fusion of the left 4th and 5th ribs

configuration[2]. The related Cuvesdem syndrome is characterised by costo-vertebral segmentation defect with mesomelia and peculiar facies[3]. In our patient there was predominantly rhizomelic shortening of the extremities. We do not know if our patient represents a dominant (new mutation) or recessive type of spondylo-costal dysostosis. Recently two patients of healthy parents with two bone dysplasias were reported – a boy with Ollier disease and Trevor's dysplasia epiphysealis hemimelica[4] and a girl with achondroplasia and enchondromatosis[5]. *Al-Ismail* et al reported association of Ollier disease with fibromatosis in a women[6]. Our patient is at risk from complications of each of these disorders. The clinical course of his mild form of osteogenesis imperfecta, will be handicapped by the dextro-convex scoliosis of the thoracic spine and sinistro-convex scoliosis of the thoraco-lumbar spine. The patient should be watched for progress of scoliosis and appearance of neurologic symptoms.

References

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