Dysmorphic facies and diffuse posterior spine ankylosis in a patient with unusual form of spondyloenchondrodysplasia (Spranger type IV)

Ali Al Kaissi · Farid Ben Chehida ·
Maher Ben Ghachem · Klaus Klaushofer ·
Franz Grill

Received: 13 April 2012 / Revised: 9 August 2012 / Accepted: 16 September 2012 / Published online: 30 September 2012
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Abstract We describe a male patient, who was seen for the first time at the age of 8 years because of short trunk dwarfism. Spine radiographs showed platyspondyly with irregular areas of increased and decreased mineralization (irregular spotted appearance within lytic lesions located along the posterior vertebral bodies of the entire spine). Skeletal survey showed no enchondromatous lesions of the short/long tubular bones. At the age of 17, progressive spine stiffness associated with stooping posture developed. 3DCT scanning showed pathological transformation of the spinal enchondromas into generalized ossification and thickening of the posterior vertebral elements (vertebral laminae, supraspinal, and interspinal ligaments, respectively) causing effectively the development of a diffuse posterior spinal ankylosis. We report what might be a unique subtype of spondyloenchondrodysplasia (Spranger type IV).

Keywords Spondyloenchondrodysplasia ·
Diffuse posterior spine ankylosis · CT scan

Introduction

Enchondromas are common, benign, and usually asymptomatic hyaline cartilage forming neoplasms in the metaphyses and diaphyses of the short and long tubular bones of the limbs, especially the hands and feet. They have been considered as a developmental disorder caused by the failure of normal endochondral bone formation. On conventional radiographs, enchondromas present as multiple, oval-shaped, linear, and/or pyramidal osteolytic lesions with well-defined margins in the metaphysis and/or diaphysis of the long tubular and in the flat bones [1, 2].

Enchondromatosis is a group of common skeletal dysplasias with extremely variable patterns of bony involvement. There are seven well-differentiated forms of enchondromatosis: Ollier disease, Maffucci syndrome, metachondromatosis, spondyloenchondrodysplasia, spondyloenchondromatosis with basal ganglia calcification, dyspondyloenchondromatosis, and genochondromatosis types I and II [3–5].

Spondyloenchondrodysplasia is a rare autosomal recessive skeletal dysplasia with vertebral dysplasia and enchondroma-like lesions in the pelvis and long bones. The vertebral bodies show dorsally accentuated platyspondyly with disturbance of ossification [6].

The pathogenesis of enchondromatosis is still not clearly understood. A genetic basis for some cases of enchondromatosis has been suggested, although a Mendelian inheritance pattern has not been identified.

Clinical report

The boy was seen for the first time at the age of 8 years because of short trunk dwarfism. Spine radiographs at the age of 13 years showed irregular spotted areas within lytic

A. A. Kaissi (✉) · K. Klaushofer
First Medical Department, Ludwig Boltzmann Institute
of Osteology, Hanusch Hospital of WGKK and AUVA Trauma
Center Meidling, Heinrich Collin Str. 30, 1140 Vienna, Austria
e-mail: ali.alkaissi@osteologie.at

A. A. Kaissi · F. Grill
Pediatric Department, Orthopedic Hospital of Speising,
Speisinger Str. 109, 1130 Vienna, Austria

F. B. Chehida
Department of Imaging Studies, Center of Radiology,
Ibn Zohr Institute, Tunis, Khadra 1003, Tunisia

M. B. Ghachem
Department of Pediatric Orthopedic Surgery,
Children Hospital of Tunis, Jabari 1007, Tunisia

Fig. 1 Spine radiographs at the age of 13 years showed irregular spotted areas within lytic lesions along the entire spine and were located mostly through the posterior end plates (Fig. 1).

lesions along the entire spine and were mostly located through the posterior end plates (Fig. 1).

Physical examination in our department at the age of 17 years revealed a narrow frontal area, depressed nasal bridge and coarse facies, short trunk dwarfism, normal limbs length, and normal hands and feet (Fig. 2). Movements of the spine showed moderate limitations in all planes associated with mild pain. He manifested stooped position resembling ankylosing spondylitis (Fig. 3). Hands were normal and showed no associated abnormalities (Fig. 4).

There were normal movements of the weight-bearing joints as well as the shoulder joints.

Neurological examination was normal. Hearing, vision, and intelligence were normal. Testing the muscle strength, sensation, and reflexes (Hoffman's, Babinski's, and Chaddock's reflexes) of the upper and lower limbs were negative. Bowel and the bladder sphincter control were normal. His erythrocyte sedimentation rate (ESR) was unremarkable.

Investigations of mucopolysaccharides, oligosaccharides, and chromosomes were normal. Lateral spine

Fig. 2 A narrow frontal area, depressed nasal bridge and coarse facies, short trunk dwarfism, normal limbs length, and normal hands and feet

radiograph at the age of 17 years showed two different pathologies; at the level of the thoracic vertebrae, platyspondyly associated with irregular spotted appearance within a lytic areas containing rings and arcs of chondroid calcification associated with scalloping of the posterior end plates and areas of irregular mineralisation. At the level of the lumbar vertebrae, it showed platyspondyly and irregular spotted lesions and the rings and arcs have been replaced with diffuse hyperostosis of the posterior vertebral elements (Fig. 5).

Coronal reformatted spine CT scan showed extensive ossification of the supra and interspinous ligaments of the entire spine, and no enchondromatous involvement of the long bones, ribs, and clavicles and dysplasia of the shoulder joints (Fig. 6). Sagittal reformatted thoracic spine CT scan confirmed the ossification of the supra and the intra-spinal ligaments causing the development of diffuse posterior bony-ankylosis associated with bands of dense calcification with the areas of demineralization within the vertebral bodies. Also, the progressive narrowing of the

Fig. 3 Stooped position resembling ankylosing spondylitis was evident

Fig. 4 Hands were normal and showed no associated abnormalities

intervertebral foraminae (Fig. 7). Coronal reconstruction CT scan confirmed the diffuse thickening and ossification of the posterior vertebral elements along the lumbar and the sacral regions giving a melting wax appearance (Fig. 8).

Fig. 5 Lateral spine radiograph at age of 17 years showed two different pathologies; at the level of the thoracic vertebrae, platyspondyly associated with irregular spotted appearance within a lytic areas containing rings and arcs of chondroid calcification associated with scalloping of the posterior end plates and areas of irregular mineralisation. At the level of the lumbar vertebrae it showed platyspondyly, and the irregular spotted lesions and the rings and arcs have been replaced with diffuse hyperostosis of the posterior vertebral elements

Anteroposterior hand radiograph showed no enchondromatous lesions (Fig. 9). Anteroposterior pelvis radiograph showed hypoplasia of the ileae, shortened femoral necks, and mild sclerosis of the acetabulae associated with small areas containing rings and arcs of chondroid calcification within the metaphyses, narrowing of the joint spaces because of hip dysplasia (Fig. 10).

Discussion

Enchondromatosis encompasses several different subtypes of which Ollier disease and Maffucci syndrome are the most common, while other subtypes (metachondromatosis, spondyloenchondrodysplasia, dyspondyloenchondrodysplasia and cheirospondyloenchondromatosis) are rare clinical entities. The genes causing the different enchondromatosis dysplasia are largely unknown [1–7].

Fig. 6 Coronal reformatted spine CT scan showed extensive ossification of the supra and interspinous ligaments of the entire spine, no associated enchondromatous involvement of the long bones, ribs, and clavicles and notable dysplasia of the shoulder joints

Fig. 8 Coronal reconstruction CT scan confirmed the diffuse thickening and ossification of the posterior vertebral elements giving a melting wax appearance

Fig. 7 Sagittal reformatted Lumbo-Sacral spine CT scan confirmed the ossification of the supra and the intraspinal ligaments causing effectively the development of diffuse posterior bony ankylosis associated with bands of dense calcification with areas of demineralization within the vertebral bodies. Note the progressive narrowing of the intervertebral foraminae

Fig. 9 Anteroposterior hand radiograph showed no enchondromatous lesions

Spondyloenchondrodysplasia is a rare autosomal recessive skeletal dysplasia with vertebral dysplasia and enchondroma-like lesions in the pelvis and long bones. The vertebral bodies show dorsally accentuated platyspondyly with disturbance of ossification. Clinical abnormalities such as short stature, rhizomelic micromelia, increased lumbar lordosis, barrel chest, facial anomalies, and clumsy movements are the main features [6].

Fig. 10 Anteroposterior pelvis radiograph showed hypoplasia of the iliae, shortened femoral necks, and mild sclerosis of the acetabulae associated with small areas containing rings and arcs of chondroid calcification within the metaphyses, narrowing of the joint spaces because of hip dysplasia

Spranger et al. [7] called Ollier disease and Maffucci syndrome types I and II, enchondromatosis; metachondromatosis, type III; and spondyloenchondrodysplasia, type IV.

Halal and Azouz [8] added three more subtypes to the Spranger classification [7].

Subtypes of enchondromatosis associated with spine involvement are well documented in the literature (Table 1) [6–16].

Al Kaissi et al. [17] described progressive vertebral fusion in a girl and her father in connection with unusual form of spinal enchondromatosis. Both individuals showed normal facies but had rigidity over the back associated with bouts of pain referred to the lower extremities.

None of the above-mentioned entities manifests dysmorphic facies, stooping posture, and diffuse spinal ankylosis as seen in our patient.

It is empirical to differentiate our patient from other forms of severe platyspondyly and joint disorders as seen in spondyloepimetaphyseal dysplasia (SEMD). SEMD is a heterogeneous group of skeletal dysplasias in which

Table 1 Comparison between our patient and patients with other subtypes of multiple enchondromas and vertebral involvement

Condition	Clinical features	Hand/long tubular bones involvement	Vertebral involvement	CNS involvement	Inheritance
Our patient	Dysmorphic facies, and stooping like posture with no rhizomelic micromelia	–	Vertebral enchondromatosis with progressive spinal ankylosis	–	Suggested AR
Enchondromatosis with irregular vertebral lesions (Spranger type I)	Multiple enchondromas	Involvement of long tubular and flat bones	Irregular dysplasia of vertebral bodies	–	Unknown
Enchondromatosis with mild platyspondyly (Spranger type II)	Generalized enchondromatosis	Severe involvement of hands and feet	Mild platyspondyly	Skull deformity	Unknown
Spondyloenchondrodysplasia (Spranger type IV)	Multiple enchondromas with severe platyspondyly	Minimal	Platyspondyly	Spasticity, calcification of basal ganglia	AR/AD
Enchondromatosis (Spranger type VI)	Generalized enchondromatosis	Brachymetacarpia and brachyphalangia	Moderate flattening (hazel nut deformity) in lumbar region	–	Unknown
Enchondromatosis type VII of Halal and Azouz	Generalized enchondromatosis	Moderate involvement of hands and feet	Irregular vertebral bodies	–	Unknown
Enchondromatosis type IX of Halal and Azouz	Generalized enchondromatosis	Minimal	Concave vertebral bodies	–	Unknown
Dyssspondyloenchondromatosis	Multiple enchondromas	Unequal limb length and multiple enchondromas present in the long tubular bones and flat bones	Platyspondyly (anisospondyly) which mimics segmentation defects of the spine	–	Unknown

Table 1 continued

Condition	Clinical features	Hand/long tubular bones involvement	Vertebral involvement	CNS involvement	Inheritance
Cheirospndyloenchondromatosis	Multiple enchondromas	Severe hand and foot involvement	Platyspondyly and erosions of the iliac crest	Mental retardation	Unknown
Pseudoachondroplastic-like spondyloenchondrodysplasia (Beighton)	Multiple enchondromas	Acromesomelia	Mild platyspondyly	–	Unknown
Enchondromatosis (Wallis)	Multiple enchondromas	–	Mild platyspondyly	Sensi-neural hearing loss	Unknown

simultaneous involvement of the epiphyses, metaphyses, and vertebrae produce severe dwarfism with deformity and shortening variously prevalent in the trunk and limbs. The main forms are Kniest syndrome (facial dysmorphism, myopia, spinal deformity, bowing and torsion of the limbs, and short stature), parastremmatic dwarfism (similar to Kniest dysplasia, but more severe). Other forms are SEMD (with joint laxity) (kyphoscoliosis and joint laxity), SEMD (Hall type) (joint dislocation is a prominent feature), SEMD (Irapa) type described among the Indians of Venezuela, and spondylo-megepiphysio-metaphyseal dysplasia (short stature mainly due to a short trunk with defective vertebral ossification and excessive epiphyseal development) [7, 16, 18].

None of the above-mentioned syndromic entities fit our patient.

On the other hand, severe platyspondyly and joint dysplasia can be a manifestation of mucopolysaccharoidoses (MPSs) [7, 16]. We initially proceeded by ruling out MPSs through vigorous laboratory investigations. MPSs were totally eliminated.

Conclusion

The radiographic features in our patient were consistent, but not completely diagnostic with spondyloenchondrodysplasia (SED). SED is characterized by multiple enchondromata of the long bones, and severe platyspondyly. Our case appears to be a unique subtype of SED. Ossification of the posterior elements and absence of enchondromatous lesions of the long bones and hip and shoulder joints dysplasia were additional features. We suggest that the development of heterotopic bone formation along the spinal ligaments had occurred through an abnormal ossified enchondral bone formation. The pathological composites of the spine have been clarified using scanning technology.

Acknowledgments We wish to thank Dr. Gen Nishimura, Department of Radiology, Nasu-Chuo Hospital (Tokyo) Japan, for confirming the diagnosis.

Conflict of interest None whether financial or non financial.

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