Case 1494

Eurorad

Hajdu-Cheney Syndrome

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Patient: 40 years, female

Clinical History:

The clinical history and the imaging findings suggest the diagnosis of acro-osteolysis syndrome of Hajdu-Cheney.
Imaging Findings:

A patient presented with slowly progressive deformity of the distal ends of the fingers.
For a few months there have been recurrent periods of painful swelling at both hands. Now she also complains of pain in the neck and at both shoulders. She had to stop her work as an ironer. No other systemic complaints were mentioned. Laboratory findings (urinary amino-acids, calcium, phosphorus, alkaline phosphatase, rheumatoid factor, anti-nuclear factor and serum complement) were normal.
On radiography of the skull, lateral view, there are a mild dolichocephaly, wormian bones (arrow), a prominent squama of the occipital bone and an enlarged sella turcica.
A-P view of the skull (not shown) reveals a small hypoplastic maxilla, hypoplastic sinuses and total loss of teeth.
Radiography of both hands and right wrist A-P view, figure 3 shows acro-osteolysis of the middle and distal phalanges and of the proximal interphalangeal joint of the left fourth finger with soft tissue swelling and telescoping. The resorption occurs in a band-like fashion across the waist of the middle and distal phalanges. There are erosive lesions in the carpal bones and right distal ulna. The pisiform bone is hypertrophic.
Radiography of both feet, A-P view shows acro-osteolysis at the middle and distal phalanges of the second to fifth toe and at the interphalangeal joint of the great toes. Erosions of several proximal interphalangeal joints is noted.
The clinical history and the imaging findings suggest the diagnosis of acro-osteolysis syndrome of Hajdu-Cheney.
Discussion:

Hajdu-Cheney syndrome (HCS) is a rare autosomal dominant disorder with variable expressivity. The onset of the syndrome is in early childhood but clinical manifestations are usually delayed, until the second or third decade. The phenotype of affected patients with HCS is characteristic. They are short, proportionated with coarse face and a short neck. Facial anomalies include, proptosis, full cheeks, a long philtrum, small mouth with malocclusion, premature loss of teeth and low-set ears. Bone malformations of the skull are wormian bones, delayed closure of cranial sutures, progressive mandibular ramus osteolysis, hypo- or aplastic sinuses and bathrocephaly. Bathrocephaly is a deep occipital fossa with protuberance of the squamous portion of the occipital bone. This can lead to an extreme basilar impression, with gradual neurological deterioration, with bulbar, pyramidal and cerebellar signs and symptoms, sometimes leading to death.
In 90% of the cases there is acro-osteolysis of the distal phalanges. Acro-osteolysis in HCS consists of resorption of tufts and band-like areas of lucency across the waist of the middle and terminal phalanges, leading to transverse gaps in the phalanges. This process, with shortening of the bones, leads to pseudoclubbing and telescoping of the fingers. The radiological appearance is identical to that in occupational acro-osteolysis.
Erosive lesions in the wrist and hypertrophy of the pisiform bone visible in our patient, have not been previously

described in HCS. Demineralisation of the vertebral bodies, with transverse striation can be observed. Laboratory analysis is usually unremarkable and renal function, which is altered in other osteolysis syndromes, is normal in this condition. Sometimes internal organ anomalies involving kidneys (polycystic) and the intestine (malrotation) are seen. Intelligence is usually (borderline) normal. The differential diagnosis of acro-osteolysis of the phalanges is extensive and includes: s cleroderma, psoriatic arthropathy, neuropathic disease, sarcoidosis, occupational acro-osteolysis, thermal injuries, epidermolysis bullosa, progeria and acro-osteolysis of Shinz.
Differential Diagnosis List: Acro-osteolysis syndrome of Hajdu-Cheney

Final Diagnosis: Acro-osteolysis syndrome of Hajdu-Cheney

References:

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