Nephroquiz for the Beginner
(Section Editor: T. J. Rabelink)

A swollen face in a girl on haemodialysis

MB, a 14-year-old Arab, Moslem girl suffered from end-stage renal insufficiency (ESRI) due to reflux nephropathy. She had been on haemodialysis since the age of 9 years. She had mostly been followed in our paediatric renal unit but over the previous 2 years she was treated elsewhere, where she was clearly underdialysed. During those last 2 years she slowly developed a gross deformation of the right side of her face (Figure 1). Her general condition was rather precarious. She was depressed and listless with a bad appetite, she had difficulty in walking and was anaemic (Hb: 7.2 g/dl; HTC: 24.6%).

The treatment with sc. erythropoietin, oral calcium salts and 1-α hydro vitamin D3 was very erratic indeed. The major blood chemistries at this time showed (predialysis): urea 32.3 mmol/l, creatinine 854 μmol/l, total protein 72 g/l of which 2.9 albumen, claim 3.9, phosphorus 4.0 mol/l, alkaline phosphatase 437 IU and PTH 769 pg/ml.

Question

What is the most likely diagnosis? How would you proceed to prove the diagnosis and what do you suggest as therapy?

Fig. 1. Photograph of the face of MB, showing severe deformation with an enormous swelling of the entire right side.
Changes in divalent ions with a high alkaline phosphatase and PTH level suggest some form of renal osteodystrophy (ROD), due to secondary hyperparathyroidism (SHPT) [1]. Bone tumours seen within this framework are known as ‘brown tumours’ (BT). The diagnosis of ROD was indeed conferred by an X-ray survey of the skeleton, including the findings of ‘pseudotumours’ of the maxilla and mandible with severe deformation of the sinuses (Figure 2). Two additional, milder cases were seen in Arab children with ESRI treated at our institution.

The types of uraemic bone disease seen in children are either due to a high or a low turnover of bone. The latter form is nowadays often iatrogenic (adynamic bone status), due to overcorrection of the abnormalities of divalent ion metabolism [2,3]. On imaging ROD can present as osteopenia, osteosclerosis, osteitis fibrosa cystica, and soft tissue calcifications. Brown tumours are far less common [4,5]. Histologically these ‘tumours’ consists of giant cell lesions, which have a brown colour, due to the presence of haemosiderin-laden macrophages. Brown tumours are typically found in the skull and neck and in particular in the jawbones, especially in children and adolescents. Brown tumours occasionally also occur in adults and can be found in other locations such as in the orbit or the spine.

The presence of fibro-osseous conditions in the craniofacial bones often poses a difficult diagnostic dilemma from a radiological point of view. Radiological evaluation, either by conventional X-ray pictures, CT or magnetic resonance imaging (MRI), are used to assess the type and severity of these bone lesions. An accurate diagnosis can be made on the basis of the clinical and laboratory data combined with the skeletal findings on imaging. A bone biopsy can help to establish the diagnosis but is often inconclusive, since there are similarities in the histological appearance of brown tumours, fibrous dysplasia, giant cell tumour, aneurysmal bone cyst and ossifying or non-ossifying fibromas.

Non-compliance with medical therapy was the major cause of the development of these preventable lesions in our patients. Only total surgical removal of brown tumours in the skull and jaw can give long lasting success. Two of our patients underwent subtotal parathyroidectomy. Following surgery we not only observed growth arrest of the brown tumours, but even a subtle regression in size. Total excision of the large tumour in the index case was not feasible.

Suggested reading


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