Short reports

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 Multifocal osteolysis with nephropathy

 Apart from the well-known specific osteodystrophy associated with chronic renal failure, skeletal changes are rarely found with nephropathy. We describe a patient with an osteolysis localized to the carpal and tarsal bones, associated with a glomerulonephritis, atypical in its age of onset and in its rapid progression to death.

 Case report

 An only child of unrelated English parents presented at the age of 5 years with uraemia, hypertension, and heart failure after a short history of anorexia and lethargy. She had attended another hospital several times from the age of 18 months because of a limp, but had no bone or joint pain. Both ankles, particularly the right, became progressively stiff and corrective shoes were worn for a short time because of a slight valgus deformity and in-toeing. There was no family history of arthritis or renal disease. On examination she was short (10th centile) and had pronounced epicanthic folds with mild hypertelorism. Her palate was high. Her last recorded height at the age of 3 years was on the 25th centile. Both wrists were puffy and telescoped and there was marked limitation of active and passive extension, but no signs of inflammation. She had marked pes cavus and nodular thickening of the plantar fascia.

 Blood pressure was 170/100 mmHg and there were signs of heart failure. Plasma creatinine was 5.2 mg/100 ml (459 μmol/l), calcium 3.8 mg/100 ml (0.95 mmol/l), phosphorus 10 mg/100 ml (3.23 mmol/l), albumin 2.7 g/dl (27 g/l), and Hb 7.8 g/dl, alkaline phosphatase 16 KA units/100 ml, and serum parathormone 380 pg/ml. Immunoglobulins and C3 component of complement were normal. There was moderate, poorly selective proteinuria (IgG/albumin clearance 88%) but no haematuria. Intravenous urography showed poor concentration of contrast and no obstruction; cystography was normal. Renal biopsy showed an advanced sclerosing glomerulonephritis with widespread interstitial fibrosis and tubular atrophy. Less severely affected glomeruli had some increase in mesangial matrix and segmental epithelial proliferation. On immunofluorescence there was some IgM in unsclerosed segments of glomeruli.

 X-rays of hands (Fig. 1b) showed the same appearance in both, grossly narrowed carpal spaces and absence of most of the carpal bones. The remainder were irregular in outline and density and the proximal ends of the metacarpals were abnormally pointed. X-rays of hands 3½ years previously (Fig. 1a) were almost normal, but the capitates and bases of the 2nd metacarpals had irregular margins. X-rays of both feet showed the tarsal bones to be irregular in outline particularly on the right (Fig. 2b). 3½ years previously there was slight irregularity of the cuneiforms on the right (Fig. 2a). X-rays of long bones, spine, and chest were normal.

 Treatment resulted in short-lived clinical and biochemical improvement and 2 months after original presentation she died. At necropsy her kidneys were small, with granular surfaces, and thin cortices. Bone histology showed severe osteoporosis and no renal osteodystrophy. Parathyroid glands were normal.

 Discussion

 Disappearing carpal and tarsal bones have been described both with nephropathy, and as an isolated entity. Thieffry and Sorre’l-Déjerine (1958) have described it in 3 generations of a family with autosomal dominant transmission and Torg et al. (1969) reported a recessively inherited form. None has had renal disease. Marie et al. (1956) described a girl with progressive erosion of the carpal and tarsal bones, and adjacent long bones, with distal muscular wasting and considerable incapacitation. She died at the age of 22 years of renal failure with hypertension, which had become evident some 2 years previously (Marie et al., 1963). Subsequent descriptions (Lagier and Rutishauser, 1965; Torg and Steel, 1968; MacPherson, Walker, and Kowall, 1973) have been similar, with difficulty in walking, the commonest presenting feature in the
first 5 years of life. Deformity of the hands and feet, with muscle weakness, was progressive and renal disease became evident at a later stage, with haematuria and proteinuria, leading to death in late teens or early twenties generally complicated by hypertension. None of these patients, though radiologically indistinguishable from the inherited familial forms, has had affected parents or sibs. Arthritic symptoms were more common in the inherited forms, but the osteolysis was less severe and after some years no longer progressed. Normand, Dent, and Smellie (1962) described a child with apparently noninherited multifocal osteolysis without a nephropathy. However, glomerulonephritis became evident later and she died of renal failure at the age of 14 years (E. G. L. Bywaters, personal communication, 1975). A unilateral form was reported by Dérot et al. (1961), but the renal disease in this patient seems to us to have been acquired, being of sudden onset, nonprogressive and followed a sore throat with raised antistreptolysin-O titre.

Other histological studies have also shown severe osteolysis in remaining carpal bones, or fibrous tissue in their area and no evidence of renal osteodystrophy there or elsewhere in the skeleton. The glomerulonephritis has been too far advanced in this and other patients for morphological classification; however, in the present patient it was much more rapidly progressive and far outstripped the advance of the osteolysis. Since renal osteodystrophy nearly always occurs in patients dying with chronic renal failure, its absence in all the described cases of osteolysis seems remarkable.

Summary

A patient with progressive osteolysis of the carpal and tarsal bones with glomerulonephritis of unusual severity is described. There was a notable

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A patient with progressive osteolysis of the carpal and tarsal bones with glomerulonephritis of unusual severity is described. There was a notable
absence of osteodystrophy in this and other reported cases who had chronic renal failure.

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REFERENCES


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‘Juvenile’ myasthenia gravis in early infancy

An infant aged 6 months is described with clinical features resembling the ‘juvenile’ form of myasthenia gravis. As far as we are aware this has not previously been described at this young age.

Case report

A female, aged 6 months was admitted to the Royal Children’s Hospital on 8 October 1974, with a short history of lethargy and hypotonia. She had been a normal and active infant until 2 weeks before admission when she became drowsy and lethargic, and appeared to have difficulty in clearing secretions from her pharynx. Her local doctor treated her with antibiotics and she apparently improved.

One week later she again became floppy, lethargic, and drowsy, and again developed noisy gurgling respirations. She was unable to feed properly, and her eyelids were drooping. She was admitted to a local hospital where pneumonia was diagnosed, and was treated with parenteral antibiotics. She required frequent aspiration of secretions and was fed through a nasogastric tube. However, her condition did not improve and after a brief respiratory arrest due to retained secretions she was transferred to this hospital.

Examination showed a floppy but alert infant with bilateral ptosis (Fig. 1). There was little spontaneous movement and she was hypotonic. However, muscle bulk and deep tendon reflexes were normal; there was no fasciculation of her tongue; pain sensation was normal; cough reflex was diminished; and auscultation of her chest showed evidence of retained secretions.

The combination of weakness, ptosis, and hypotonia, together with normal muscle bulk and deep tendon reflexes, suggested the diagnosis of myasthenia gravis and this was subsequently confirmed by her response to a neostigmine test. She was given 0.1 mg atropine, and 0.25 mg neostigmine 30 minutes later intramuscularly.