Interesting Case

Erdheim–Chester disease: a rare cause of renal failure

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Keywords: Erdheim–Chester disease; histiocytosis; renal failure; urinary tract obstruction

A 52-year-old woman was admitted to our department because of renal failure. She had initially presented at the age of 50, with a thyroid mass requiring hemithyroidectomy. The histological diagnosis of the thyroid mass was that of a pseudo-inflammatory histiocytic tumour. For the next 2 years, the patient had no other problems.

One week before admission, the patient noted vomiting and decreased urine volume. On admission, she had elevated blood pressure (180/80 mmHg) and serum creatinine was 1060 mmol/l (1 year previously, serum creatinine levels were normal). The patient had lower haemoglobin levels (9 g/dl) and hyperkalaemia (serum potassium of 7 mmol/l). The rest of the serum biochemical analysis was in the normal range.

In spite of a bladder catheter being placed, urine volume was minimal and a haemodialysis session was performed via a femoral catheter.

A computed tomography scan (with intravenous medium contrast) was performed and showed bilateral compression of renal parenchyma by retroperitoneal mass (Figure 1).

Left mass measured 8 × 9 × 13 cm and affected the right kidney, suprarenal gland and spleen. Right kidney showed diffuse infiltration and light hydronephrosis. A biopsy of retroperitoneal mass was performed.

The left renal parenchyma was thinned and showed chronic hydronephrosis. Therefore, we decided to perform decompression of the right kidney. Left pyelography showed upper hydronephrosis and a nephrostomy was placed.

The patient was free of symptoms and did not require dialysis after the procedure and her serum creatinine level stabilized at around 265 mmol/l.

Histological examination of the biopsy revealed an accumulation of foamy histiocytes in a background of fibrosis admixed with chronic inflammation (Figure 2).

The histiocytic cell population was not immunoreactive for S100, CD21, CD23, CD34, ALK1 and LMP1. These histological findings were similar to the thyroid mass. On the basis of these findings, a diagnosis of Erdheim–Chester disease was made. No other lesions in eyes, long bones, lungs and heart were found in this patient.

Discussion

We report the 61st case of Erdheim–Chester disease since the first two descriptions by Chester in 1930. It is an extremely rare, non-familiar, histiocytic disorder of unknown origin, usually occurring in middle-aged to older patients. Manifestations range from a focal asymptomatic process to a fatal systemic disease that...
may involve virtually all organ systems. Focal manifestations include leg pain with typical radiological findings: symmetrical cortical thickening a metaphyseal sclerosis are characteristic in bone involvement [1]. Half of these patients will have multifocal disease: fever, orbital tumour, lung fibrosis, pericardial infiltration, hypothalamic/pituitary involvement with diabetes insipidus, urinary tract obstruction and renal failure.

Diagnosis of Erdheim–Chester disease derived from the classic radiographical findings in the long bones and the biopsy with histological confirmation of an extra-osseous site.

The histological diagnosis is suggested in the presence of a pleomorphic inflammatory reaction with foamy histiocytes in sclerotic nodes.

In Erdheim–Chester disease, electron microscopy does not reveal Birbeck granules and immunohistochemical study does not demonstrate staining for S-100 protein and leukocyte antigen CD1, in contrast with histiocytosis X [2].

Retroperitoneal involvement may occur in isolation or as a component of disseminated disease. Kidneys and retroperitoneum are frequently involved (18 of 60 patients) but this affectionation is rarely symptomatic, abdominal CT disclosed hydronephrosis in seven cases, enlarged kidneys in six, retroperitoneal infiltration in five and pelvic infiltration in one [3]. Renal involvement may be caused by direct invasion of the renal sinus and parenchyma, as seen in our patient, or by more distal ureteral obstruction [4].

Treatment usually includes oral steroids or, in advanced cases, chemotherapy, radiotherapy or immunotherapy, but these treatments have little overall affect on progressive disease. Patients with renal involvement are often treated using palliative percutaneous nephrostomy to preserve renal function, as in our patient.

Recently, a case treated by surgery has been published with good results [5].

Conflict of interest statement. None declared.

References


Received for publication: 2.12.06
Accepted in revised form: 11.2.07