

## Commentary

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We were asked recently by our orthopaedic colleagues for advice on a young patient with Gorham's syndrome. He had the characteristic osteolytic lesions, predominantly affecting the vertebral column, and bilateral pleural effusions. Unable to recall this syndrome on the list of potential causes for a pleural effusion, we turned to the text books. With each edition the lists get longer – for example, *Respiratory Medicine* by Brewis *et al*<sup>1</sup> cites 38 possibilities but Gorham's syndrome is conspicuous by its absence. So, off to MedLine; there at last was Gorham's. This syndrome is the common thread in two case reports in this issue of *Thorax*. McNeil *et al* report a case complicated by a pleural effusion successfully treated with radiotherapy while Riantawan *et al* describe a case of fatal chylothorax.

Gorham's syndrome is a rare condition of unknown aetiology first reported by Jackson in 1838 and described in more detail, along with the characteristic histological features, by Gorham in 1954. It consists of proliferation of thin walled vascular or lymphatic channels in association with massive osteolysis and the consequent disappearance of bones. The lymphohaemangiomas may extend into adjacent bones, viscera, or soft tissues and are accompanied by local fibrosis. Spinal disease may produce vertebral collapse and consequent neurological disability or lead to chest deformity and respiratory compromise. As illustrated by the case reported by McNeil, spinal involvement seems to be associated with a worse outcome while disease at more peripheral sites frequently arrests spontaneously. Osteolysis of the thoracic cage can lead to pleural effusions, chylous or non-chylous. The former are said to occur if the thoracic duct is invaded but the mechanism underlying the latter is unclear. Extension of the vascular proliferation to involve the pleura is perhaps the most plausible explanation.

Approximately 150 cases of Gorham's syndrome are to be found in the literature, with no sexual bias and more commonly affecting younger people. The true prevalence is difficult to ascertain since not every case will have been written up. Moreover, cases will be missed if a haemorrhagic pleural effusion with lytic lesions in the bones, such as that reported by McNeil *et al*, is simply ascribed to malignancy without histological proof. If you've never heard of Gorham's syndrome, you're unlikely to diagnose it.

In these days of "evidence based medicine" how do we decide on treatment of such a rare condition? Few centres have accumulated enough cases to publish a series. The Massachusetts General Hospital has only seen four cases since 1965.<sup>2</sup> Hence, guidance on management must come from case reports such as those in this issue, bearing in mind possible publication bias. Are more difficult cases preferentially reported or are editors more willing

to accept papers showing beneficial treatments than those with less favourable outcomes?

Chylothorax seems to occur in around 17% of patients with Gorham's syndrome. The available evidence suggests that early surgical intervention to ligate the thoracic duct before malnutrition and sepsis occur reduces mortality. Would the outcome in the case described by Riantawan *et al* have been different had the diagnosis been made earlier? Non-chylous effusions have been described in a number of patients. They are associated with a better outcome than chylothorax and in several cases effusions have resolved spontaneously. Others have been usefully treated by drainage, with or without pleurodesis. Radiotherapy applied to the lytic lesions has also resulted in resolution of associated pleural effusions.

What about treating the lesions themselves? Radiotherapy has been used in a number of cases with varying degrees of success, and debate about its usefulness exists. What does seem to be important is the dose used. Treatment failure is associated with smaller doses – that is, a combined dose of under 30 Gy. The patient described by McNeil *et al*, who showed arrest of the disease process, received a total dose of 40 Gy, supporting this view. Surgery may be useful in cases of limited disease where local resection, with or without reconstructive bone grafting, has achieved some favourable results. However, occasionally the grafted bone may become affected too. Amputation has been used for disabling disease of distal limbs with good success. In some circumstances palliative measures are indicated – for instance, spinal supports to prevent cord compression. These will obviously not alter the disease process itself. Finally, it must be remembered that spontaneous regression does occur in a small number of cases.

In summary, Gorham's syndrome is rare but probably underdiagnosed. It should feature on a differential diagnosis for any pleural effusion, especially if osteolysis is present, highlighting the importance of "not forgetting the bones" when looking at a chest radiograph. Often the diagnosis is made by a radiologist and not a physician which should be borne in mind when it comes to acknowledgements and authorship.<sup>3,4</sup> Many cases will arrest spontaneously, but chylothorax should be treated promptly by surgical ligation of the thoracic duct. Radiotherapy is probably useful if given in a high enough dose.

If you've seen a case of Gorham's syndrome then you can probably cross it off your differential diagnosis of pleural effusion since you will have retired before you come across another!

- 1 Brewis RAL, Corin B, Geddes DM, Gibson GJ. In: *Respiratory medicine*. London: WB Saunders, 1995.
- 2 Dunbar SF, Rosenberg A, Henry MD, *et al*. Gorham's massive osteolysis. The role of radiation therapy and a review of the literature. *Int J Radiat Oncol Biol Phys* 1993;26:491–7.
- 3 Rose DSC. Signing up for authorship. *Lancet* 1996;347:1412.
- 4 Horton R, Smith R. Signing up for authorship. *Lancet* 1996; 347:780.

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