Helen F Ashdown, Ling-Pei Ho and Jayne E Haynes

Lumps, bumps and diagnostic stumps

HF Ashdown, MRCP, DCH, academic clinical fellow in general practice, University of Oxford, Oxford. **L-P Ho**, FRCP, MD, DPhil, consultant in respiratory medicine, Oxford Sarcoidosis Service, Churchill Hospital, Oxford. **JE Haynes**, MRCGP, DRCOG, DFFP, GP, 19 Beaumont Street Surgery, Oxford.

Address for correspondence

Helen Ashdown, University of Oxford, Department of Primary Care Health Sciences, Radcliffe Observatory Quarter, Woodstock Road, Oxford, OX2 6GG, UK.

E-mail: helen.ashdown@phc.ox.ac.uk

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INTRODUCTION

Numquam ponenda est pluralitas sine necessitate (plurality ought never be posed without necessity), proposed William of Ockham (Occam) in the early 14th century. Commonly known as 'Occam's razor', this principle asserts that a single unifying diagnosis is more likely to explain multiple simultaneous presenting symptoms than several different diagnoses.[1] In primary care, patients frequently present with multiple symptoms. This case report follows a patient who presented to different GPs within our practice with various symptoms, that were subsequently explained by one unifying diagnosis.

CASE REPORT

A previously well 52-year-old university academic, of Asian descent, presented initially with blurred vision. An ophthalmologist diagnosed bilateral anterior uveitis, which responded well to dexamethasone eye drops.

Around the same time, he consulted a different GP for a right groin lump. In addition to a reducible hernia there was also bulky inguinal lymphadenopathy which was felt clinically to be lymphoma. A full blood count was normal. A fine needle aspiration showed no evidence of malignancy, but a probable reactive lymphadenopathy. Following excision biopsy, histology of the lymph node was reported as numerous epithelioid granulomas, with Ziehl-Nielson staining showing aggregates of acid-fast bacilli, consistent with mycobacterial infection.

Meanwhile, he had presented to a third GP with an enlarging painless breast lump that was considered to be potentially malignant, and he was referred via the 2-week-wait bureau to a breast surgeon.

A fine needle aspiration of the breast lump showed granulomatous inflammation rather than malignancy. At this time, a chest X-ray, which had been arranged by the ophthalmologist, showed bilateral hilar lymphadenopathy. His serum angiotensin-converting enzyme (ACE) was raised, and he was referred to a respiratory physician who undertook high resolution CT and diagnostic bronchoscopy, which confirmed a diagnosis of sarcoidosis. A further review of his original histology from his inguinal lymph node biopsy was requested and the previously reported acid-fast bacilli were shown to be contaminants.

The patient presented with several features that caused concern: the breast lump could be rarely observed male breast cancer, making this a sarcoid-like reaction to solid tumour; groin lymph nodes are an unusual presentation for lymphadenopathy associated with sarcoidosis, therefore lymphoproliferative disease, HIV infection and tuberculosis were differential diagnoses. He subsequently avoided treatment apart from inhaled corticosteroids and has increased his 10-mile cycle routine which contributes to both wellbeing and control of his disease.

Our patient describes that for him the diagnosis came as somewhat of a relief that he only had one disease and not a combination of lymphoma, tuberculosis, and breast cancer.

DISCUSSION

Sarcoidosis is a multisystem granulomatous disease which can affect any organ.[2,3] In a general practice with 10 000 patients, one to two patients will develop sarcoidosis each year, although this will vary with practice population as the disease is more common in certain racial groups, such as African–Americans. It predominantly affects young and middle-aged people. The cause is not known, but it may occur in genetically susceptible people exposed to unknown environmental triggers.[2]

Histologically the disease is characterised by the formation of granulomas which may resolve spontaneously, but in 20–25% of patients will result in pulmonary fibrosis.[3] Granulomas are a non-specific finding found

in other conditions such as lymphoma, mycobacterial and fungal infections, and malignancy.[2] Granulomatous deposits show up on 18FDG-PET scans and this can also lead to them being confused with malignancy.[4]

Over 90% of patients have respiratory symptoms at presentation, including shortness of breath, cough, or wheeze, so in this respect our case is unusual, although similar cases have previously been reported.[5] Sarcoidosis is also frequently found as an incidental finding on chest X-ray as bilateral hilar lymphadenopathy or pulmonary infiltrates.[3] Respiratory examination is usually normal, although in more advanced disease there may be fine inspiratory crackles due to pulmonary fibrosis. A common acute presentation is Löfgren's syndrome, which presents with arthralgia, erythema nodosum, and bilateral hilar lymphadenopathy;[2] this carries a good prognosis with most patients resolving within 2 years.[6]

Other clinical features include skin involvement (24%) (maculopapular lesions, lupus pernio, or infiltration of scars or tattoos by granulomas, as well as erythema nodosum).[5] There is ocular involvement in 12% patients, usually anterior uveitis, but ocular involvement may be asymptomatic so all patients should undergo slit lamp examination.[2] Extrathoracic lymphadenopathy is also common (15%)[2] but involvement outside of lymph-node sites such as in breast tissue is rare: we found one reported case of sarcoidosis of the breast.[4] Sarcoidosis may also affect the liver (18%), kidney (5%) (hypercalcaemia, interstitial nephritis and renal calculi), nervous system (5%) (aseptic meningitis and nerve palsies), and heart (2%) (cardiomyopathy or rhythm disturbance, and, rarely, sudden death).[2]

Diagnosis is predominantly based on clinical and radiological findings with supportive histology. The disease should usually be confirmed and managed in secondary care, usually by respiratory physicians. Useful investigations include full blood count, calcium studies, renal and hepatic function, chest X-ray, baseline spirometry, and baseline ECG. Serum ACE tests are commonly done but have a relatively poor sensitivity and specificity. However it is useful for following burden of disease and response to treatment. Further investigations usually involve biopsy of the most easily accessible site, including staining for acid-fast bacilli to exclude mycobacterial infection, high resolution CT, and bronchoscopy.[3]

Prognosis is variable. Two-thirds of patients remit within a decade of diagnosis with no long-term consequences, but up to one-third of patients have progressive disease leading to significant organ impairment. It is fatal in fewer than 5% of patients and this is usually due to pulmonary fibrosis.[3]

For most patients, treatment is not necessary as the disease is asymptomatic and resolves spontaneously. Regular analgesia can be useful for painful erythema nodosum.[2] Oral corticosteroids are used if there is evidence of progressive organ dysfunction (for example, impaired lung function) aiming to wean these over approximately 6–12 months. Second-line and corticosteroid-sparing treatment such as methotrexate, azathrioprine, hydroxychloroquine, and tumour-necrosis factor inhibitors have been used but with limited evidence for success, and treatment options are sparse.[6]

Sarcoidosis is a classic example of a disease which can present with apparently disconnected symptoms, usually initially in primary care, and so GPs are well-placed to make the diagnosis early, both to avoid unnecessary investigations for alternative diagnoses and enable initiation of early treatment if needed. However, this may be challenging particularly when a patient chooses to see different GPs about different problems. This case shows the importance of looking back through the records and considering whether problems may be linked: remember Occam's razor.

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The patient provided written consent for this article to be published.

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