

Paget's disease 1: epidemiology, causes and clinical features

This article outlines the epidemiology, causes and clinical features of this long-term bone disorder

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ABSTRACT Sutcliffe, A. (2009) Paget's disease 1: epidemiology, causes and clinical features. *Nursing Times*; 105: 6, 14–15.

The first in this two-part unit on Paget's disease outlines the epidemiology, pathophysiology, causes and clinical features of this long-term bone condition.

INTRODUCTION

In 1877 Sir James Paget, an eminent British surgeon, described 'osteitis deformans' in an elderly man with progressive skeletal deformities (Paget, 1877). This condition, which became known as Paget's disease of bone, is characterised by rapid bone remodelling and the formation of bone that is structurally abnormal.

Although many people are symptom free, Paget's disease can result in pain, deformity, increased risk of fracture and other complications including osteoarthritis and nerve-compression syndromes. The condition cannot be cured but treatments can control its progression and minimise complications.

While Paget's disease is common, initial diagnosis is often missed and many healthcare professionals, including nurses, fail to recognise early symptoms and are uncertain about management. Patients commonly present in both primary and acute care. In the latter, they are managed in various disciplines including care of the elderly, rheumatology and orthopaedics.

EPIDEMIOLOGY

Paget's disease occurs in 1–2% of white adults over 50 and is more common in men. Prevalence increases substantially with age and by the eighth decade of life it may be present in approximately 8% of men and 5% of women (van Staa et al, 2002).

There is marked ethnic and geographical clustering, with the disease being common in some parts of the world but relatively rare in others. Clinical observations indicate that it is

LEARNING OBJECTIVES

1. Know the clinical features of Paget's disease.
2. Understand why the long-term condition occurs.

found more often in Europe, the US, Australia and New Zealand but is rare in Scandinavia, the Indian subcontinent, China, Japan and other countries in South East Asia.

A large European radiographic study showed that rates in the UK were higher than in most other western European countries (Detheridge et al, 1982). A further survey showed a marked focus of Paget's disease in a cluster of six Lancashire towns (Barker et al, 1980). Over the past 25 years the prevalence and severity of disease has reduced substantially in the UK and New Zealand and remained fairly stable in the US.

PATHOPHYSIOLOGY

Bone is a living tissue that is constantly renewed through a remodelling process that provides a mechanism for self-repair and adaptation to stress. This takes place at different sites on the bone surface and is largely regulated by two types of bone cells, osteoclasts and osteoblasts.

Osteoclasts degrade bone by attaching to bone surface and secreting acids and enzymes into the bone cavity. The osteoblasts then synthesise and mineralise new bone matrix within the cavity created.

In Paget's disease, the osteoclasts are increased in size and number and contain many more nuclei than normal and, consequently, they resorb bone more quickly. In response, the osteoblasts deposit new bone at an increasingly rapid rate. This rapid rate of bone turnover leads to the production of bone that may be larger in size than normal but is characterised by disorganised

architecture and reduced mechanical strength, leaving people at increased risk of developing fractures and deformities.

CAUSES

For many years, the causes of Paget's disease were poorly understood but this has changed considerably over the past decade.

Genetic factors are now recognised to play an important role, with approximately 15–40% of people having a positive family history. Siris et al (1991) estimated that the risk of developing Paget's disease is 7–10 times greater in first-degree relatives of patients than in the general population.

In most families, the disease is inherited as a simple autosomal dominant trait with high penetrance by the sixth decade.

Anomalies have been identified in four genes that cause Paget's disease or related syndromes. The most common is an anomaly of the sequestosome 1 (SQSTM1) gene, and people with this anomaly tend to have more extensive disease than those without.

It is also possible that exposure to certain paramyxoviruses, particularly measles and canine distemper viruses, may influence the development of Paget's disease but research in this area remains inconclusive.

Epidemiological studies and observational data have suggested that potential environmental triggers may include low

BOX 1. CLINICAL FEATURES

● Pain	Bone pain Joint pain
● Deformity	Bowing of long bones Skull deformities
● Fracture	Fissure fracture Complete fracture
● Neurological	Deafness Other cranial nerve palsies Spinal cord compressions
● Malignant change	Tumour formation



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dietary calcium intake or vitamin D deficiency in childhood, repetitive mechanical loading and occupational exposure to toxins.

CLINICAL FEATURES

In many cases, there may be few symptoms and the condition is found if an X-ray or blood test is performed for another reason.

Any bone can be affected but Paget's disease is most commonly found in the spine, skull, pelvis, femur or tibia. The condition may be monostotic, affecting a single bone or part of a bone, or polyostotic, involving two or more bones.

While Paget's disease may progress within a bone, its appearance at new sites after initial diagnosis is uncommon. This information can be reassuring to patients who often worry about its progression to new areas. For clinical features, see Box 1.

Pain is the most common symptom, with studies showing it is experienced by 80% of patients (Wermers et al, 2008). Arising from the affected bone, it is often described as 'persistent and nagging'. The pain usually occurs at rest, particularly at night, and is not relieved by exercise. Pagetic bone has an increased vascularity and pain may be accompanied by an unpleasant sensation of increased heat or pressure around the affected site. The direct cause of the pain may be difficult to characterise and

distinguishing bone pain from the pain associated with coexisting musculoskeletal disorders can be problematic.

Deformity in the long bones is more likely to occur in untreated long-standing disease and may be associated with pain. A bowed limb is typically shortened, resulting in specific gait abnormalities that can lead to abnormal mechanical stresses. Fissure fractures may occur on the convex aspect of bones of a deformed limb and it has been estimated that approximately 50% of those with fissure fractures will develop a complete fracture. Clinical fractures may be traumatic or spontaneous and usually occur in long bones with active areas of advanced disease.

Paget's disease in the skull may be asymptomatic but common complaints in up to one-third of patients include an increase in head size with or without frontal deformity, and headache, sometimes described as a band-like tightening around the head. Extensive skull involvement can cause softening and flattening at the base of the skull so that the odontoid process extends upwards as the skull sinks downward upon it.

The rate of hearing loss in Paget's disease is fairly high and it is thought that this is caused by pagetic bone invading the cochlear capsule and changing its electrical properties. Cranial nerve deficits can arise but these are rare and vary in presentation.

In the lumbar spine, Paget's disease can lead to vertebral fractures and possible spinal stenosis, occasionally leading to nerve impairment. This probably occurs because the hypervascular pagetic bone 'steals' blood from the spinal cord, causing ischaemia.

Malignant change is rare, occurring in fewer than 1% of cases. Although it may be possible to resect the tumour and give chemotherapy or radiotherapy, prognosis is poor, with death usually within 1–3 years.

Associated non-specific conditions are:

- Osteoarthritis – this is common and is related to bone placing additional pressure on joints, exacerbating degenerative changes;
- Cardiac complications – rare cardiac failure related to increased output required to respond to vascular pagetic bone;
- Hypercalcaemia – occasionally associated with long-term immobility;
- Gout – slightly increased incidence;
- Kidney stones – slightly higher incidence.

Studies from the UK (van Staa et al, 2002) and US (Wermers et al, 2008) have suggested that the clinical features and complications

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associated with Paget's disease lead to increased morbidity. Van Staa et al showed that people with Paget's disease had a lower survival rate compared with age-matched controls. However, findings from the recent US study did not suggest that Paget's disease was associated with lower overall survival and no clinical factors were identified that were associated with an increased risk of death.

CONCLUSION

In recent years, advances have been made in defining the pathogenesis of Paget's disease, with an increased understanding of bone cell biology and genetic causation.

Although prevalence is declining in this country, patients with more severe disease continue to present with pain, deformity, fractures and other associated conditions.

It is important that nurses are aware of this condition and are able to recognise its features and complications. ■

● Part 2 of this unit, to be published in next week's issue, discusses the clinical management of Paget's disease.

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