Paget's disease diagnosis

Because early diagnosis and treatment of Paget's disease is important, after age 40, siblings and children of someone with Paget's disease should have an alkaline phosphatase blood test every two or three years. If the alkaline phosphatase level is above normal, other tests such as a bone-specific alkaline phosphatase test, bone scan, or X-ray can be performed.

Hyperostosis frontalis interna (HFI) is a benign irregular nodular thickening of the inner table of the frontal bone that is almost always bilateral. The midline is spared at the insertion of the falx.

Unilateral cases have been reported, and in these cases one must R/O other etiologies such as meningioma, calcified epidural hematoma, osteoma, fibrous dysplasia, an epidural fibrous tumor, or Paget’s disease.

Conditions with Basilar invagination associated with softening of bone include Paget’s disease.

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1. lab work (serum markers may be normal in monostotic involvement):

a) serum alkaline phosphatase: usually elevated (this enzyme is involved in bone synthesis and so may not be elevated in purely lytic Paget’s disease [1] ; mean 380 ± 318 IU/L(normal range: 9–44) [2].

Bone-specific alkaline phosphatase may be more sensitive and may be useful in monostotic involvement

b) calcium: usually normal (if elevated, one should R/O hyperparathyroidism)

c) urinary hydroxyproline: hydroxyproline is found almost exclusively in cartilage. Due to the high turnover of bone, urinary hydroxyproline is often increased in PD with a mean of 280 ± 262 mg/24 hrs (normal range 18–38) [3].

2. bone scan: lights up in areas of involvement in most, but not all [4]. cases

3. plain x-rays:

a) localized enlargement of bone: a finding unique to PD (not seen in other osteoclastic diseases, such as prostatic bone mets)

b) cortical thickening

c) sclerotic changes

d) osteolytic areas (in skull →osteoporosis circumscripta; in long bones →“V” shaped lesions).

Osteoporosis circumscripta is sometimes observed at the skull vault and corresponds to the initial stage of Paget's disease diagnosis of the bone. Differentiating osteoporosis circumscripta from other reasons for osteolytic images of the vault may be difficult.

Bou Antoun et al., report a case of osteoporosis circumscripta of the frontal bone. A lucent rim seen on CT scan, which was enhanced on gadolinium-enhanced MRI, delineated the abnormal bone. The

patient was a 50-year-old woman who had CT scans of the skull for chronic sinusitis. Pathology examination showed typical bone changes of Paget's disease. The lucent and enhancing rim sign may help in differentiating Paget's disease from other conditions [5].

e) spinal Paget's disease often involves several contiguous levels. Pedicles and lamina are thickened, vertebral bodies are usually dense and compressed with increased width. Intervening discs are replaced by bone

Consider Paget's with a dense vertebra on x-ray in an older patient, commonly involving several contiguous vertebrae.

4. CT: hypertrophic changes at the facet joints with coarse trabeculations.

[1]
Rothman RH, Simeone FA. The Spine. Philadelphia 1992
[2], [3], [4]
Altman RD, Brown M, Gargano F. Low Back Pain in Paget's Disease of Bone. Clin Orthop. 1987; 217:152–161
[5]
Bou Antoun M, Kemel S, Polivka M, Bresson D, Laredo JD. The lucent (CT) and enhancing (MR) rim, a sign of Paget's disease of the skull: case report. Skeletal Radiol. 2018 Nov;47(11):1567-1570. doi: 10.1007/s00256-018-2957-x. Epub 2018 May 5. PubMed PMID: 29730701.