How Is Paget's Disease of Bone Diagnosed?

Symptoms of Paget's disease

Many people with Paget's disease do not know they have it. There may be no symptoms, or the symptoms may be very mild. Sometimes the symptoms are mistaken for something else, such as arthritis. In other cases, complications, which vary depending on which bones are affected, have developed before Paget's disease is diagnosed.

Methods used to diagnose Paget's disease

Three tests – an x-ray, blood test, and bone scan – are commonly used to discover Paget's disease. Sometimes the disease is found by accident when one of these tests is done for another reason. X-rays are used to make the final diagnosis.

X-rays. In x-ray images, bones affected by Paget's disease have a particular appearance that is different from other bones. Accordingly, a doctor will use x-rays, alone or with a blood test or bone scan, to confirm whether or not a person has Paget's disease of bone.

Blood test (measurement of serum alkaline phosphatase). Sometimes blood test results are what first alert doctors to the possibility that a person has Paget's disease. When blood contains a higher-than-usual level of a chemical substance called serum alkaline phosphatase (SAP), it is a sign that the disease may be present. SAP is a type of enzyme made by bone cells that is overproduced by pagetic bone.

A mild increase in SAP, up to twice the usual level, may indicate Paget's disease or another condition, such as liver disease or a bone fracture that is in the process of healing. However, a SAP level greater than twice the usual level strongly suggests Paget's disease, especially if the person's serum calcium level, phosphorus level, and kidney function are normal.

Bone scan. A bone scan is a test that helps identify which bones have been affected by Paget's disease. The procedure is also a useful way to determine the extent and activity of the disease. In a bone scan, a safe amount of a radioactive substance is injected into the person's vein. The substance circulates through the bloodstream and "highlights" places in the skeleton where Paget's disease may be present. If the scan suggests that the person has the disease, the affected areas usually are x-rayed to confirm the diagnosis.
Bone biopsy. In rare cases, a biopsy (a small sample of tissue removed for analysis) is taken from bone that is suspected of having Paget's disease. Bone biopsies are done when x-ray images do not confirm the presence or absence of the disease.

Urine test. Although urine tests are not used to detect or diagnose Paget's disease, they may be used to monitor a person's response to treatment for the disease. In this test, a sample of the person's urine is analyzed for the presence of substances called bone markers. These substances provide information about bone resorption – the process of breaking down and taking up of bone by the body. One such substance is \( N \)-telopeptide.

After the diagnosis

Several effective treatment options are available for people who are diagnosed with Paget's disease. Information on these treatments is available from the NIH Osteoporosis and Related Bone Diseases ~ National Resource Center.

If you need more information about available resources in your language or another language, please visit our website or contact the NIH Osteoporosis and Related Bone Diseases ~ National Resource Center.