Knee Discomfort in a 38-year-old Man*

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A 38-year-old, previously healthy, nonsmoking man has been referred to you for evaluation of digital clubbing (Fig 1, 2), which was noted during a routine pre-employment physical examination. He also has an abnormal knee radiograph (Fig 3). His only complaint is aching pain and the feeling of swelling just above his knees for about 6 months. He also volunteers the information that his fingers look no differently now than they did when he was a teenager. Physical examination is normal, except that he appears older than his stated age and has skin changes and clubbing of 10 digits, as shown in the accompanying figures. Which of the following tests will be needed to establish his diagnosis?

A. Chest CT scan
B. Serum liver function tests
C. Right-to-left shunt measurement on 100% oxygen
D. Colonoscopy
E. No further testing is necessary

*From the ACCP-SEEK program, reprinted with permission.

Items are selected by Department Editors Richard S. Irwin, MD, FCCP, and John G. Weg, MD, FCCP. For additional information about the ACCP-SEEK program, phone 1-847-498-1400.

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Answer: E. No further testing is necessary.

The patient under discussion has primary hypertrophic osteoarthropathy (HOA), also referred to as idiopathic or hereditary HOA, or pachydermoperiostosis. Clues to this are multiple and include the following: 1) the history of clubbing has been present since earlier in life; 2) the patient has coarse, hypertrophied skin on the hands and face, making him appear older than his stated age (ie, pachyderma); 3) clubbing of the fingers appearing to be at an advanced age (eg, “drumstick” configuration); and 4) the knee radiograph (Fig 4) showing typical changes of HOA at the distal, lateral ends of the right femur. The thin opaque lines (see arrows) of new bone are separated from the underlying denser cortex by a narrow, radiolucent periosteal band. Because the diagnosis of this condition can be made clinically, no further testing is necessary. However, when routine bone radiographs are occasionally negative in either primary or secondary causes of HOA, whole-body bone scanning with Tc 99m methylene diphosphonate would be necessary to establish the presence of HOA.

The characteristic bone scan finding of HOA is

Table 1—Secondary Causes of Hypertrophic Osteoarthropathy*

<table>
<thead>
<tr>
<th>Generalized</th>
<th>Pulmonary</th>
<th>Cystic fibrosis</th>
<th>Pulmonary fibrosis</th>
<th>Chronic infections</th>
<th>Cancer (primary or metastatic)</th>
<th>Arteriovenous fistulae</th>
<th>Mesothelioma</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cardiac</td>
<td>Congenital cyanotic diseases</td>
<td>Infective endocarditis</td>
<td></td>
<td></td>
<td></td>
<td></td>
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</tr>
<tr>
<td>Hepatic</td>
<td>Cirrhosis</td>
<td>Carcinoma</td>
<td></td>
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</tr>
<tr>
<td>Intestinal</td>
<td>Crohn disease</td>
<td>Ulcerative colitis</td>
<td>Chronic infections</td>
<td>Laxative abuse</td>
<td>Polyposis</td>
<td>Malignant tumors</td>
<td></td>
</tr>
<tr>
<td>Mediastinal</td>
<td>Esophageal carcinoma</td>
<td>Thymoma</td>
<td>Achalasia</td>
<td>Miscellaneous</td>
<td>Graves disease</td>
<td>Thalassemia</td>
<td>Diverse malignancies</td>
</tr>
</tbody>
</table>

*Adapted from Martinez-Lavin.
†POEMS = polyneuropathy, organomegaly, endocrinopathy, M protein, and skin changes.
‡This category includes isolated case reports of diverse entities that are continually being published.
increased uptake of radionuclide in a linear pattern along the periosteal surfaces of the involved bones (Fig 5). The radius, ulna, tibia, and femur are most commonly affected; the phalanges, clavicles, ribs, scapulae, and mandible are less commonly affected.

Primary HOA (ie, clubbing and periosteal proliferation of the tubular bones not associated with an internal illness) is inherited in an autosomal dominant pattern and affects men more frequently than women (approximately 9 to 1). It is characterized by osseous and soft-tissue proliferation, digital clubbing, and glandular and hypertrophic skin changes. The skin involvement can consist of acne, hyperhidrosis, and seborrhea, and/or coarse facial features that may progress to deep furrowed, cerebroid-appearing skin of the forehead (ie, cutis verticis gyrata) and hypertrophy of the eyelids (ie, blepharoptosis). Patients with this disorder can display a wide spectrum of skeletal and skin abnormalities. As in our patient, the osteoarthropathy can be expressed clinically at a later time than the skin changes and clubbing. With respect to onset of manifestations, there appears to be a bimodal distribution, with one peak being during the first year of life and the other at age 15. The disease eventually runs a self-limited course, ending in adulthood, with survival not being affected.

When confronted with a patient with clinical features of HOA, it is important to consider the differential diagnoses of secondary causes before diagnosing primary HOA. These are listed in the accompanying Table 1. Colonoscopy is listed as a possible test to order if the clinician is going to investigate the multiple possible intestinal causes of secondary HOA.

**Selected Readings**


