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Incidentally Found Fingers and Toes Swelling
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A 58-year-old man presented to the clinic with a simple gastroenteritis. He was found to have a striking bilateral fingers and toes clubbing (Figure 1). The patient reported having this deformity since childhood. He is a non-smoker and denied any concurrent cardiopulmonary disease, arthralgia, or bone deformity. Physical examination was otherwise unremarkable including normal heart and lung findings. EKG, Chest X-ray, blood gasses, chemistry panel, and blood count were unremarkable. X-ray failed to show any bony overgrowth or periosteal reaction.

What is the most likely diagnosis? What would be your management, and what would you tell the patient?

DIAGNOSIS
Isolated congenital clubbing.

Isolated congenital clubbing is an incomplete form of primary hypertrophic osteoarthropathy (PHO), also known as pachydermoperiostosis (1). Isolated congenital clubbing is a rare disorder characterized by thickened nails, and swelling of the terminal segments of the fingers and toes. This type of genodermatosis results from connective tissue overgrowth between the nail and the phalanx. The complete form of PHO is characterized with periostosis (bone overgrowth), hyperhidrosis, seborrhea, in addition to pachyderma (skin thickening). The condition can be sporadic or inherited. The latter is secondary to a mutation in the HPGD gene located on chromosome 4q33-q34, resulting in increased prostaglandin E2 (PGE2) production. PGE2 stimulates osteoblasts and osteoclasts, and results in a chronic localized vasodilation in the distal phalanx (2).

DIFFERENTIAL DIAGNOSIS
The differential diagnosis of isolated congenital clubbing includes any disease entity that can result in digit swelling:
Thyroid acropachy:
Thyroid acropachy is a severe manifestation of autoimmune thyroiditis. It causes bilateral swelling of fingers but comes with stiffness and pain of the affected areas. It can also be associated with pretibial myxedema, periosteal reaction, and exophthalmos (3).

Heberden’s node:
This is a classic manifestation of osteoarthritis, usually associated with pain and other joint involvement. The nodes affect the distal interphalangeal joint and not the nail (4).

Acromegaly:
Acromegaly causes blunting of the fingers and toes due to bone and cartilage overgrowth and deformity. It does not cause swelling or clubbing. Acromegaly is secondary to growth hormone overproduction, classically by a pituitary tumor (5).

Pseudoclubbing (PC):
Asymmetric digital swelling characterizes PC. It is associated with acro-osteolysis identified on X-ray by apical tufts resorption. This results in soft-tissue collapse and subsequent deformity mimicking clubbing. The exact pathophysiology remains unclear; however, PC has been seen in conditions associated with bone remodeling such as renal osteodystrophy, acrometastasis, and subungual hematoma among others (2).

MANAGEMENT
A good history and physical exam are sufficient to make the correct diagnosis and to spare the patient unnecessary, and usually invasive, testing: clubbing is present since childhood with the absence of signs or symptoms associated with secondary clubbing, such as cardiopulmonary disease, inflammatory bowel disease, and malignancy. Reassurance is sufficient, and no treatment is needed. However, with any clinical suspicion, it may be necessary to rule out secondary causes of clubbing.
Figure Legend:
Figure 1: Fingers and toes clubbing with nail thickening.

References: