Paraspinal soft tissue masses in a patient with a metabolic bone disease

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The 49-year-old female patient presented to the emergency department with cough and shortness of breath for a few days. Her chest radiograph demonstrated a right upper lobe consolidation representing pneumonia. Incidentally, she was noted to have a lobulated paraspinal mass in her left hemithorax (Fig. 1). The patient was noted to have a history of pyknodysostosis, multiple related fractures including C1 vertebral body and hearing loss. Prior imaging workup performed included plain film radiography and CT of the face and neck. Plain films of the face and cervical spine revealed an obtuse gonial angle (Fig. 2), a hallmark feature of this entity. Other characteristic changes noted on CT face included calvarial thickening, non-closure of cranial sutures, underpneumatization of sinuses and mastoid air cells, and diffuse sclerosis of the visualized bones. CT of the neck demonstrated bilateral lobulated paraspinal soft tissue masses left larger than right (Fig. 1 *). These findings were consistent with extramedullary hematopoiesis. On further review of her charts, the patient had a history of long-standing fatigue, for which she was being followed by a hematologist. Her most recent hemoglobin (Hgb) was low – 7.1 and so was her platelet count – 95,000. Prior records confirmed that these counts had been low for several years. Her blood smear showed nucleated RBC, metamyelocytes, and myelocytes. Her TSH was elevated at 6.87. Her iron saturation was 22% and ferritin 98. Her Vitamin B12 and folate levels were normal. Her ESR and reticulocyte count were elevated at 46 and 3.1% respectively. There was no hemolysis on her blood smear. Her HIV, HBV and HCV test were normal. On clinical examination, she had hepatosplenomegaly, which was felt to represent additional sites of extramedullary hematopoiesis. In the background of

pyknodysostosis, she was suspected to have myelophthisic anemia. Her WBC count had been reported as normal on several occasions and hence was not pancytopenic, and the pneumonia was likely an incidental finding.

Pyknodysostosis is a rare autosomal recessive disorder estimated to affect about 1 in 1.7 million people worldwide. A mutation in the 1q21 gene leads to miscoding of Cathepsin K, an osteoclast lysosomal enzyme, causing accumulation of matrix proteins and type I collagen, leading to generalized osteosclerosis and brittle bones [1].

Characteristic features include short stature, craniofacial abnormalities, frequent fractures, and higher risk of osteomyelitis [1]. Patients suffer from dolichocephaly, delayed closure of sutures or persistently open fontanelles (Fig. 2), multiple wormian bones, hypoplastic paranasal sinuses, beaked appearance of the nose, and frontoparietal bossing [2, 3]. There are also peculiar mandibular and dental manifestations (Fig. 2). A highly obtuse gonial angle of the mandible is pathognomonic of pyknodysostosis, often with prognathism, with higher risk of mandibular fracture and osteomyelitis [3] (Fig. 2). Patients often have a high arched palate with delayed eruption of both deciduous and permanent teeth and persistence of deciduous teeth leading to crowding and increasing vulnerability to dental caries. Hypodontia or supernumerary teeth, hypercementosis, and hypoplasia of the mandible and maxilla may also be seen [2] (Fig. 2). Dental caries that are often severe and necessitate procedures also predispose these patients to mandibular osteomyelitis. Patients may also have blue sclera, pectus excavatum, scoliosis, distal clavicular or acromial dysplasia, broad hands and feet, short terminal phalanges, nail deformities, and hearing loss [3, 4]. Other manifestations include obstructive sleep apnea and snoring [5]. On imaging, bones demonstrate generalized osteosclerosis with relative sparing of the medulla [4]. However the bones are brittle and are vulnerable to easy fracturing, most commonly seen in the lower limbs. The spine demonstrates spondylolisthesis, possibly with defects in segmentation and spoon-shaped vertebral bodies [4].

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Myelophthitic anemia is a type of anemia that occurs in patients with diseases affecting the bone marrow, where normal marrow cells are replaced by fibrosis, granulomas, or neoplastic cells. Hence, it can be seen in the setting of leukemia, lymphoma, myeloproliferative disorders, and cancers. These processes can eventually lead to “extramedullary hematopoiesis” referring to masses producing hematopoietic precursors outside of the medullary cavity of the bone (Fig. 1). Myelophthitis is rarely associated with pyknodysostosis and usually found with osteopetrosis. We present this case to illustrate that though extramedullary hematopoiesis is rarely seen in pyknodysostosis, however, literature review has previously reported a few case reports of anemia associated with disorder [5, 6].

Acro-osteolysis of the distal phalanges is characteristic and helps to differentiate it from cleidocranial dysostosis. Clavicular involvement is less frequent in pyknodysostosis. Other differentials of sclerotic bone disease include osteopetrosis, where there is more significant medullary involvement, and short stature, open fontanelles, clavicular involvement, and distal phalangeal involvement are not seen. Osteogenesis imperfecta can also have a similar radiographic appearance, but patients suffer from more severe and frequent fractures and choanal atresia [7, 8].

Though imaging and dental findings of pyknodysostosis are characteristics, other clinical features and imaging findings must all be used to arrive to a conclusive diagnosis. Gene mutation analysis confirms the diagnosis. Treatment is supportive, aiming to prevent fractures and resulting deformities.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

Informed consent Informed consent and IRB are not mandated for single patient case report as per our institution and hence were not obtained.

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