CLINICAL IMAGE PEER REVIEWED | OPEN ACCESS

Adult presentation of diffuse osteopetrosis masquerading as right hip osteoarthritis

Carolyn Ward, Steven Verga, Kimberly Lynch, Payal Parikh

CASE REPORT

A 41-year-old female with no contributory personal or family medical history came to our hospital due to right hip pain for one month, previously diagnosed at outside hospital as osteoarthritis. Patient had no history of fracture, bone pain, or trauma-related injuries. X-rays, magnetic resonance imaging and bone scans were taken of the right femur, hip, and lumbar spine which demonstrated diffuse abnormal bone sclerosis with cortical thickening. Figure 1 is a complete spine X-ray which showed the classic rugger-jersey appearance consistent with osteopetrosis in the setting of normal parathyroid hormone ruling out hyperparathyroidism. Figure 2 is a whole body bone scan showing the diffuse process of her disease with abnormally increased uptake in almost all her bones especially at the ends of long bones.

Laboratory examination of the patient showed alkaline phosphatase 90 IU/L, vitamin D_{25} 18.7 ng/ml, calcium 8.5 mg/dl, ionized calcium 4.5 mg/dl, PTH 42 pg/ml, protein 7.6 g/dl, albumin 4.2 g/dl, CA 15-3 (<8 U/ml), CA27-29 (<12 U/ml), CEA (1.3 ng/ml). Given that her parathyroid harmone (PTH) was not elevated and she had low-normal calcium, hyperparathyroidism was unlikely. Her normal alkaline phosphatase and mildly low vitamin D_{25} also made osteomalacia unlikely. Serum markers of Acid Phosphatase Prostatic, CA 27-29, CA 15-3, and CEA were negative ruling out malignancy. Patient had no other signs of symptoms of multiple myeloma as well as no protein-albumin gap. The diagnosis of osteopetrosis was made.

Although this patient presented with localized pain in her right hip, imaging found that her disease was diffuse and extensive. The patient was seen in hospital by

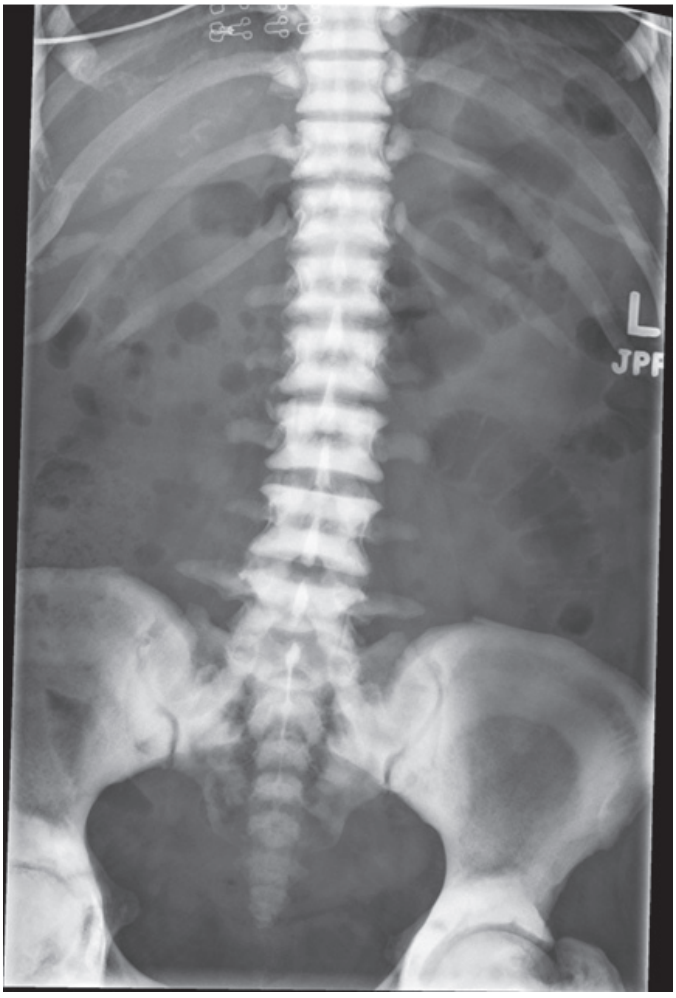

Figure 1: X-ray of thoracic and lumbar spine showing classic rugger-jersey appearance.

Carolyn Ward[1,2], Steven Verga[1,2], Kimberly Lynch[2], Payal Parikh[1,2]

Affiliations: [1]MD, Department of Internal Medicine, Robert Wood Johnson University Hospital, New Brunswick, NJ, USA; [2]MD, Rutgers-Robert Wood Johnson Medical School, Piscataway, NJ, USA.

Corresponding Author: Carolyn Ward, 1 Spring St Unit 2205 New Brunswick, NJ 08901; Email: carolynleeward@gmail.com

Received: 05 August 2017
Accepted: 21 August 2017
Published: 07 September 2017

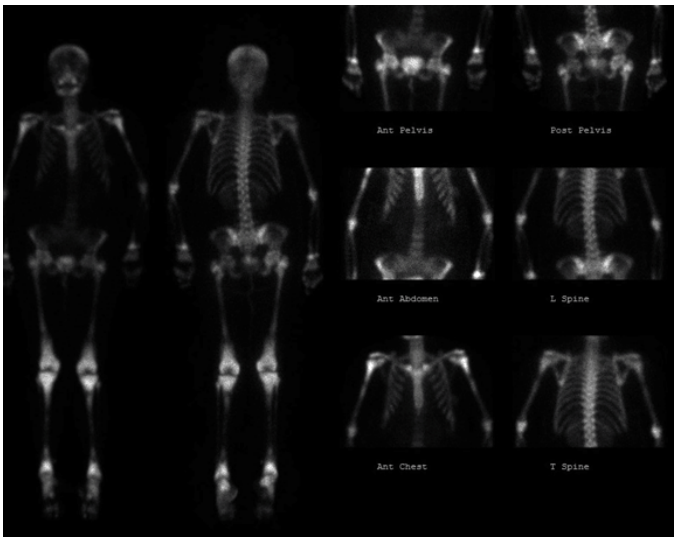

Figure 2: Whole body bone scan showing diffusely increased activity especially at ends of long bones.

orthopedic surgery who recommended outpatient total right hip replacement given the severity of bone changes and uncontrolled pain. Patient was discharged on an oxycontin taper with percocet as needed as well with osteopetrosis require special considerations and close outpatient follow-up.

DISCUSSION

Osteopetrosis, defined by a defect in osteoclast bone resorption, is a group of heritable conditions that was first described radiographically by Albers–Schonberg. Abnormally dense bones and increased bone density is characteristic of the disease which usually presents in childhood. Presentation in adulthood without family history is very rare. The most common form in adulthood is autosomal dominant osteopetrosis type II. This type is caused by a defect in the chloride channel CLCN7 which is coupled to osteoclast H+-ATPase and has radiographic findings of rugger-jersey appearance of the spine and pelvis along with the typical bone in bone appearance of all types of osteopetrosis [1]. Patients with this condition have an increased rate of fractures, and based on a case series reported in an academic journal 78% of studied patients had fractures and 27% hip osteoarthritis. 52.8% of their patients underwent orthopedic surgery with an abnormally high number of complications, including intraoperative fractures and nonunion [2]. Given the increased rates of orthopedic injuries, surgical interventions, and postoperative complications, differentiating the diagnosis of osteopetrosis, a diffuse disease, from uncomplicated osteoarthritis is imperative.

We describe a case of a female presenting with localized right hip pain that was found to have extensive bony disease including classic imaging of rugger-jersey spine. The rugger-jersey spine is highly specific for osteopetrosis but also commonly seen in hyperparathyroidism as well as seen in Paget's disease, osteomalacia, and metastatic disease. The striped appearance of sclerotic bands in between thoracic and lumbar vertebral bodies gives the appearance of a striped rugby jersey. Excess osteoid volume accumulates giving the opacified appearance of these stripes [3]. The other possible causes of these bony changes were ruled out in our patient, and she was diagnosed with osteopetrosis.

CONCLUSION

Adult onset osteopetrosis is rare, but given its diffuse nature, high risk of future fractures with need of surgical intervention, and higher percentage of post-operative complications, it should be considered in patients presenting with diffuse bone disease. Classic imaging findings include 'bone in bone' sclerosis and the rugger-jersey spine. Patients are typically followed closely to prevent and minimize any complications from osteoarthritis, fractures, and trauma.

****

Keywords: Diffuse osteopetrosis, Hip pain, Osteoarthritis, Rugger-jersey spine

How to cite this article

Ward C, Verga S, Lynch K, Parikh P. Adult presentation of diffuse osteopetrosis masquerading as right hip osteoarthritis. J Case Rep Images Orthop Rheum 2017;2:20–22.

Article ID: 100007Z14CW2017

****

doi:10.5348/Z14-2017-7-CL-4

****

Author Contributions

Carolyn Ward – Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Steven Verga – Analysis and interpretation of data, Revising it critically for important intellectual content, Final approval of the version to be published
Kimberly Lynch – Acquisition of data, Drafting the article, Final approval of the version to be published

Payal Parikh – Substantial contributions to conception and design, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

Guarantor
The corresponding author is the guarantor of submission.

Conflict of Interest
Authors declare no conflict of interest.

Copyright

REFERENCES

1. Tolar J, Teitelbaum SL, Orchard PJ. Osteopetrosis. N Engl J Med 2004 Dec 30;351(27):2839–49.
2. Bénichou OD, Laredo JD, de Vernejoul MC. Type II autosomal dominant osteopetrosis (Albers–Schönberg disease): Clinical and radiological manifestations in 42 patients. Bone 2000 Jan;26(1):87–93.
3. Wittenberg A. The rugger jersey spine sign. Radiology 2004 Feb;230(2):491–2.