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Case Report Open Access

Osteopathia Striata with Concomitant Fibroblastic Osteosarcoma of the Femur. A Potential Malignant Risk?

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Abstract

We report a case of a 33-year old female with an unusual presentation of high-grade osteosarcoma superimposed on osteopathia striata. Few cases of malignancy arising in sclerosing bone dysplasia have been reported in literature. Of the few reported ones, osteosarcoma was associated with melorheostosis in two cases, with osteopoikilosis in the one case, and with combined melorheostosis and osteopathia striata in one case. To the best of our knowledge the current case is the first one of a high-grade osteosarcoma arising from isolated osteopathia striata, not combined with other forms of sclerosing bone dysplasia.

Introduction

Osteopathia striata is an uncommon sclerosing bone dysplasia that remains mostly asymptomatic. First described by Voorhoeve in 1924 [1], it is characterized by linear streaks of altered bone density in the metaphyses and diaphyses of long bones, pelvis, and scapulae, thought to arise from disturbances in the endochondral bone formation. Initially thought to be an autosomal dominant disorder, recent molecular analysis confirmed an X-linked dominant inheritance, with germline mutations of the *WTX* gene, causing osteopathia striata congenita with cranial sclerosis [2]. The radiologic findings of osteopathia striata are characteristic, suggesting the diagnosis; however, the molecular analysis can confirm it .

Here we report an unusual case of high-grade osteosarcoma likely arising from osteopathia striata. We review the literature and attempt to clarify questions of potential malignant risk associated with sclerosing bone dysplasia. To our knowledge, only one case of osteosarcoma arising in a background of osteopathia striata combined with melorheostosis has been reported in literature, but none of osteosarcoma arising from isolated osteopathia striata.

Case Report

A 33-year-old woman presented with pain in the left thigh. Imaging revealed a diaphyseal soft tissue mass superimposed on a previously undetected benign sclerotic condition of the left femur, in the background of sclerosis (Figure 1). Endosteal scalloping was present in the femoral diaphysis, suggesting a slowly growing process, possibly a malignant transformation taking place in an otherwise benign bone condition. The distal femoral metaphysis and the proximal tibia showed radiographic changes consistent with osteopathia striata (Figures 2 and 3). Radiographic characteristics consistent with osteopathia striata were also appreciated in the patient's contralateral lower extremity (right femur, tibia, and fibula - Figure 4). Abnormality in the left ileo-sacral region was also noted.

A needle biopsy of the diaphyseal soft tissue mass indicated malignancy; an 8-cm mass was removed during a subsequent extra-articular surgery with diaphyseal allograft placement (Figure 5). Decalcified sections of the tumor with bone revealed abundant malignant osteoid formation in a high-grade osteosarcoma of the femoral diaphysis, with prominent fibroblastic features, in a background of extensive sclerosing bone dysplasia, prominent cement lines, and focal marrow fibrosis, consistent with osteopathia striata (Figures 6-8). Areas of woven bone were also present on polarized light examination (Figure 9). Many areas of osteosarcoma displayed features morphologically indistinguishable from leiomyosarcoma. Immunohistochemical markers were negative for desmin, actin, CD-34, S-100, and wide spectrum keratins. The soft tissue portion of the tumor showed foci of malignant osteoid formation.

Postoperative imaging showed characteristics of osteopathia striata in the residual distal femoral and proximal tibial metaphyses. Radiographs in four other family members (father, mother, brother and daughter) showed changes consistent with osteopathia striata in father and brother (Figures 10 and 11).

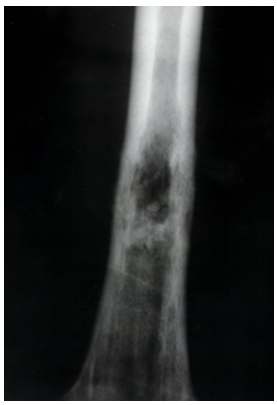

Figure 1: X-ray - Osteosarcoma in the background of osteopathia striata, left femur diaphysis.

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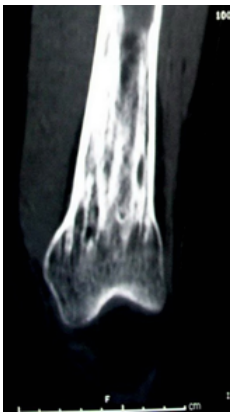

Figure 2: CT - Osteosarcoma in background of osteopathia striata, left femur metaphysis.

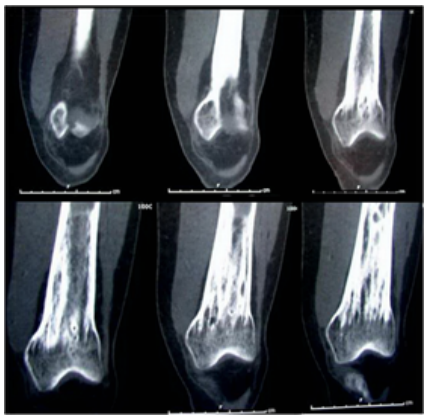

Figure 3: CT – Osteosarcoma in the background of osteopathia striata, multiple views, left femur.

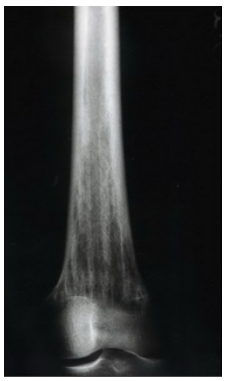

Figure 4: X-ray - Osteopathia striata in the right femur.

Patient underwent post-operative chemotherapy; however, six months after the treatment initiation a 6-cm mass was found in the left upper lobe of the lung with pleural seeding. Biopsies of the lung nodule, left chest wall, pericardium, and pleura showed metastatic high-grade spindle/pleomorphic sarcoma with focal malignant osteoid formation, all histomorphologically similar to the original femoral osteosarcoma (Figures 12 and 13).

The patient succumbed to disease nine months after the initial diagnosis.

Discussion

Osteopathia striata is a rare sclerosing bone dysplasia with characteristic radiographic features including longitudinal sclerotic striations of long bones. Sclerosing bone dysplasia comprises of a unique set of skeletal abnormalities with a wide range of radiologic, clinical, and genetic characteristics. The hereditary dysplasia includes osteopetrosis, pyknodysostosis (Maroteaux-Lamy disease),

Figure 5: X-ray - Left femur, post-op.

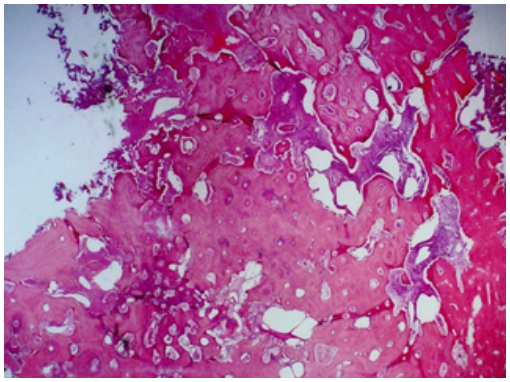

Figure 6: H&E - Infiltrating osteosarcoma in the background of osteopathia striata, left femur.

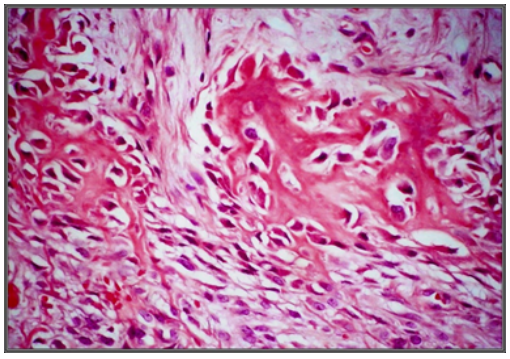

Figure 7: H&E - Osteosarcoma with malignant osteoid, left femur.

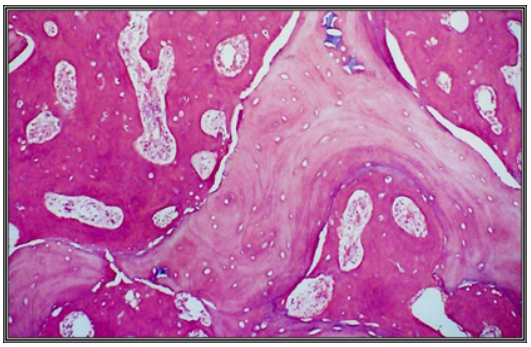

Figure 8: H&E - Osteopathia striata, mature and woven bone, left femur.

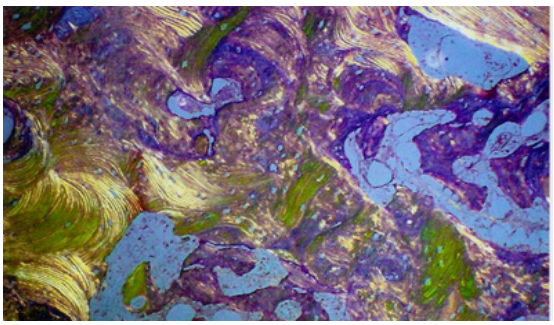

Figure 9: Osteosarcoma in the background of osteopathia striata (polarized light).

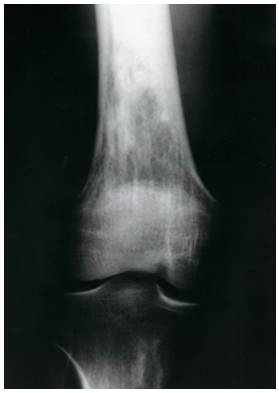

Figure 10: X-ray - Patient's father – osteopathia striata, left femur.

osteopoikilosis, osteopathia striata (Voorhoeve disease), progressive diaphyseal dysplasia (Camurati-Engelmann disease), hereditary multiple diaphyseal sclerosis (Ribbing disease), and hyperostosis corticalis generalisata (endosteal hyperostoses). Each disorder represents disruption in the bone ossification pathway. Some of these entities have similar genetic pathways, possibly representing phenotypic variants of the same disease. The nonhereditary dysplasia includes intramedullary osteosclerosis, melorheostosis (Leri disease) and overlapping syndromes [3].

Osteopathia striata is viewed as presence of disturbances in development associated with the process of enchondral bone formation. Although referred to as an autosomal dominant, some studies suggested an X-linked inheritance. Most cases were reported in association with cranial sclerosis, which was lacking in our patient. Osteopathia striata is often a marker of other disorders, particularly associated with focal dermal hypoplasia (Goltz-Gorlin syndrome or Goltz syndrome) [4].

More recently, an association with cranial sclerosis with and without cranial nerve dysfunction has been described [5]. Males with osteopathia striata with cranial sclerosis show mutations of the *AMER1* gene located on the long arm of the X chromosome; they don't usually survive and the survival is associated with various internal organs malformations [6]. Other clinical associations include skin

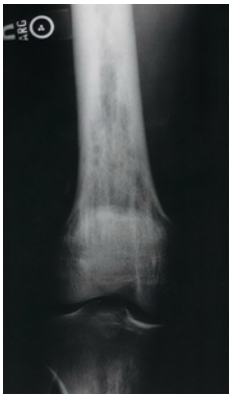

Figure 11: X-ray - Patient's brother – osteopathia striata, left femur.

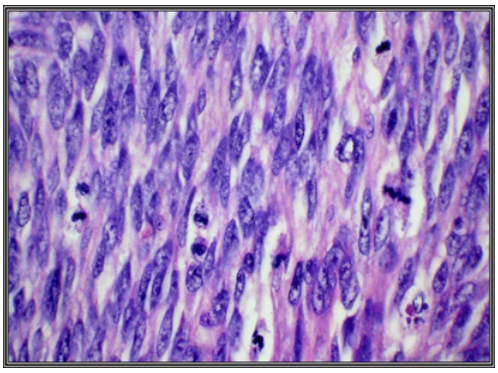

Figure 12: H&E - Pulmonary metastasis, mostly fibroblastic.

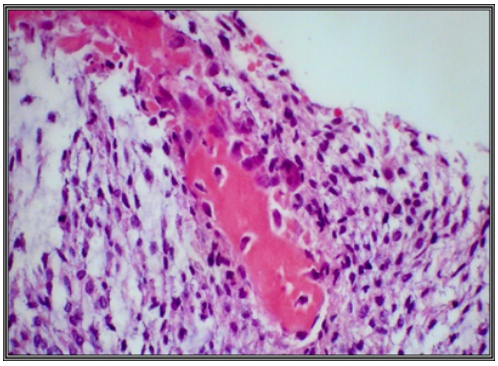

Figure 13: H&E - Pulmonary metastasis, malignant osteoid.

abnormalities, mucosal papillomas, eye, renal, and dental anomalies. Therapy is mainly symptomatic, directed at relieving pain.

To the best of our knowledge the current case is the first one of a high-grade osteosarcoma arising from isolated osteopathia striata, not combined with other forms of sclerosing bone dysplasia. Few cases of malignancy arising in sclerosing bone dysplasia have been reported in literature. Of the few reported ones, osteosarcoma was associated with melorheostosis in two cases [7,8], with osteopoikilosis in the third case [9], and with combined melorheostosis and osteopathia striata in the fourth case [10].

Our case illustrates an unusual concurrent osteopathia striata and osteosarcoma in a female, emphasizing the various clinical associations of this disorder. When osteopathia striata is diagnosed, other abnormalities such as cranial osteosclerosis and internal anomalies should be looked for. In addition, family members should be screened for the presence of this skeletal abnormality, as demonstrated in our case. A close monitoring might be beneficial for patients since osteosarcoma might develop. With more cases of simultaneous osteopathia striata and osteosarcoma to be reported in the future, a better understanding of the optimal follow-up and management in these patients will emerge.

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