Clinical Presentation of Malignant Transformation from Fibrous Dysplasia to Osteosarcoma: A Case Report

Saahiti Denduluri, BA[1]; Alexander Straughan, MD[1]; Neal Godse, MD[1]
[1]Department of Otolaryngology, University of Minnesota, Minneapolis, MN

Introduction

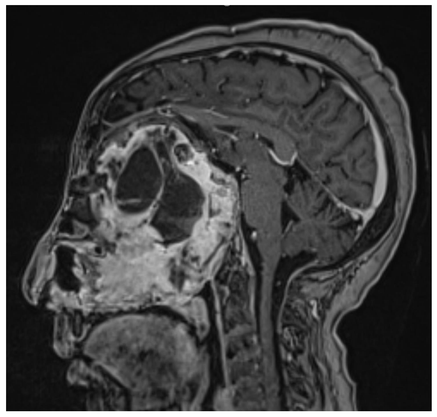
Fibrous dysplasia (FD) is a benign bone disorder characterized by the replacement of normal bone with abnormal fibrous tissue.[1] While generally considered a benign condition, FD has been associated with rare instances of malignant transformation, most commonly to osteosarcoma.[2] The infrequency of this transformation makes it a diagnostic challenge, highlighting the importance of vigilant monitoring.

Case Presentation

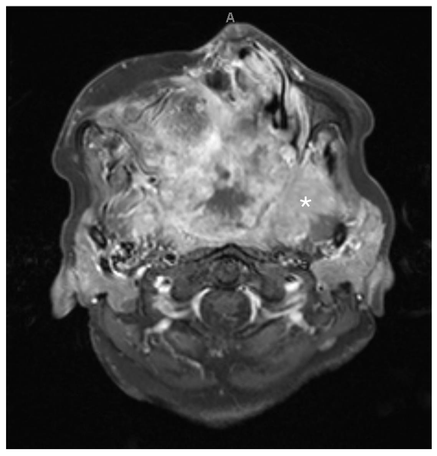
A 37-year-old female with a long-standing history of right craniofacial FD presented to our clinic with facial pain and hearing loss. At 20-years-old, she had undergone her first partial tumor excision followed by several craniofacial reconstructive surgeries, with goals of tumor reduction and cosmesis. After being lost to follow up for nearly 10 years, the patient re-presented with worsening left facial pain. Interval imaging demonstrated progressive, contrast enhancing T1 and T2 intermediate tumor involving the left pterygopalatine fossa as well as multiple pulmonary nodules, suggesting potential malignant degeneration and metastatic disease. Histopathology from endoscopic endonasal debulking biopsy demonstrated dedifferentiated (high-grade) osteosarcoma with PET scan demonstrating lung and bone metastases.

Images

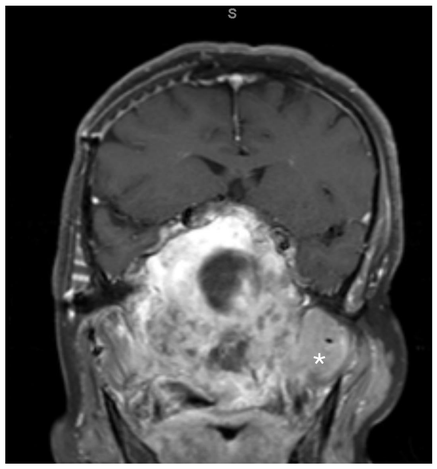
A.



B.



C.



Sagittal (A), Axial (B), and Coronal (C) MRI imaging demonstrated known fibrous dysplasia with involvement of the face, facial bones, left pterygoid muscles, and extension into the anterior skull base fossa and prepontine cistern. * indicates the region of tumor progression concerning for malignant degeneration.

Discussion

Malignant transformation of FD to osteosarcoma is a rare but clinically significant event, with reported frequencies ranging from 1-3% depending on the anatomical location and specific features of disease.[3,4] Craniofacial sites are frequently involved in reported cases.

Malignant transformation can often present with signs of new or worsening pain, swelling, or neurological symptoms, which should prompt further investigation.[5] These symptoms may mimic nonmalignant and locally aggressive FD, thus histology is an important diagnostic factor.[5] Radiographs suggesting malignant osteosarcoma may feature poorly marginated osteolytic lesions, cortical destruction, soft tissue extension into the bony cortex, or bone with a radiolucent "ground-glass" pattern.[5] Molecular analysis, including GNAS mutation testing, can also help distinguish malignant transformation from benign aggressive variants.[2]

Treatment of malignant transformation of FD often requires surgical resection for improved outcomes, whereas chemotherapy poses no improved benefit for the prognosis of patients.[6]

Conclusion

We aim to present this rare case to spread awareness about the notable potential for malignant transformation from FD to osteosarcoma. Lifelong follow-up with attention to specific changes in symptoms like new onset pain, numbness, tinnitus, vision changes, and radiologic findings is critical.

References

1. Kim DY. Current concepts of craniofacial fibrous dysplasia: pathophysiology and treatment. *Arch Craniofac Surg.* 2023 Apr;24(2):41-51. doi: 10.7181/acfs.2023.00101. Epub 2023 Apr 20. PMID: 37150524
2. Sugiura Y, Kanda H, Motoi N, et al. Osteosarcoma arising in fibrous dysplasia, confirmed by mutational analysis of GNAS gene. *Pathol Res Pract*. 2018 Feb;214(2):318-324. doi: 10.1016/j.prp.2017.10.018. Epub 2017 Nov 12. PMID: 29268950.
3. Furmah P, Aminoshariae A, Mupparapu M, et al. Frequency of spontaneous osteosarcomatous transformation of craniofacial fibrous dysplasia: a systematic review. *Oral Surg Oral Med Oral Pathol Oral Radiol*. 2023 May;135(5):678-685. doi: 10.1016/j.oooo.2022.12.007. Epub 2022 Dec 24. PMID: 36828758.
4. Wagner VP, Carlos R, Romañach MJ, et al. Malignant transformation of craniomaxillofacial fibro-osseous lesions: A systematic review. *J Oral Pathol Med*. 2019 Jul;48(6):441-450. doi: 10.1111/jop.12867. Epub 2019 May 15. PMID: 31062892.
5. Hussaini AS, Swanson DD, Nguy PL, et al. Malignant Sarcomatous Degeneration of Craniofacial Fibrous Dysplasia. *J Craniofac Surg*. 2022 Sep 1;33(6):1787-1790. doi: 10.1097/SCS.0000000000008437. Epub 2021 Dec 16. PMID: 36054890
6. Weber V, Stigler R, Lutz R, et al. Systematic review of craniofacial osteosarcoma regarding different clinical, therapeutic and prognostic parameters. *Front Oncol*. 2023 Mar 24;13:1006622. doi: 10.3389/fonc.2023.1006622. PMID: 37035145