Fibrous Dysplasia: Osteosarcoma

16

Skull deformity, predominantly frontal, has been something this 62-year-old lady has lived with since childhood. A diagnosis of craniofacial fibrous dysplasia was established by the previous X-ray, CT and MRI exams, as well as by bone biopsy, and the appearances were stable for years. The patient's personal medical history also included surgery and chemotherapy for breast cancer 12 years ago (Fig. 16.1).

Six months before admission to the hospital, the patient noticed a moderate enlargement of the deformity in the left frontal region: at that time, FNA confirmed fibrous dysplasia. Further growth warranted a follow-up CT exam (Fig. 16.2).

A localised resection of the expanded bone in the left frontal region was done. Intraoperative biopsy confirmed fibrous dysplasia. However, postoperative extended histopathology analysis revealed osteosarcoma on grounds of previous fibrous dysplasia. The resection borders could not be determined in the available tissue specimen. Another surgery was done, this time larger in extension (Fig. 16.3).

16.1 Craniofacial Fibrous Dysplasia

Fibrous dysplasia (FD) is a tumour-like, non-neoplastic congenital disease, probably caused by a somatic mutation early in embryonic life, featuring defective osteoblastic differentiation and maturation. Immature bone is intermixed with excessively proliferated fibrous tissue.

It may affect a single bone (monostotic, 70% of cases, most common in ribs) or multiple bones (polyostotic, usually unilateral limb lesions). Any bone may be affected. Craniofacial fibrous dysplasia (CFD) occurring in multiple adjacent craniofacial bones is regarded as monostotic, and it accounts for up to 25% of monostotic form. It may be one of the features in McCune-Albright syndrome [1]. CFD behaves as a chronic, slowly progressive mass lesion, usually self-limiting, rarely progressing after the third decade of life. Complications are usually caused by compression of skull foramina, nerves and vessels—such as visual loss, proptosis, hearing loss and headache.

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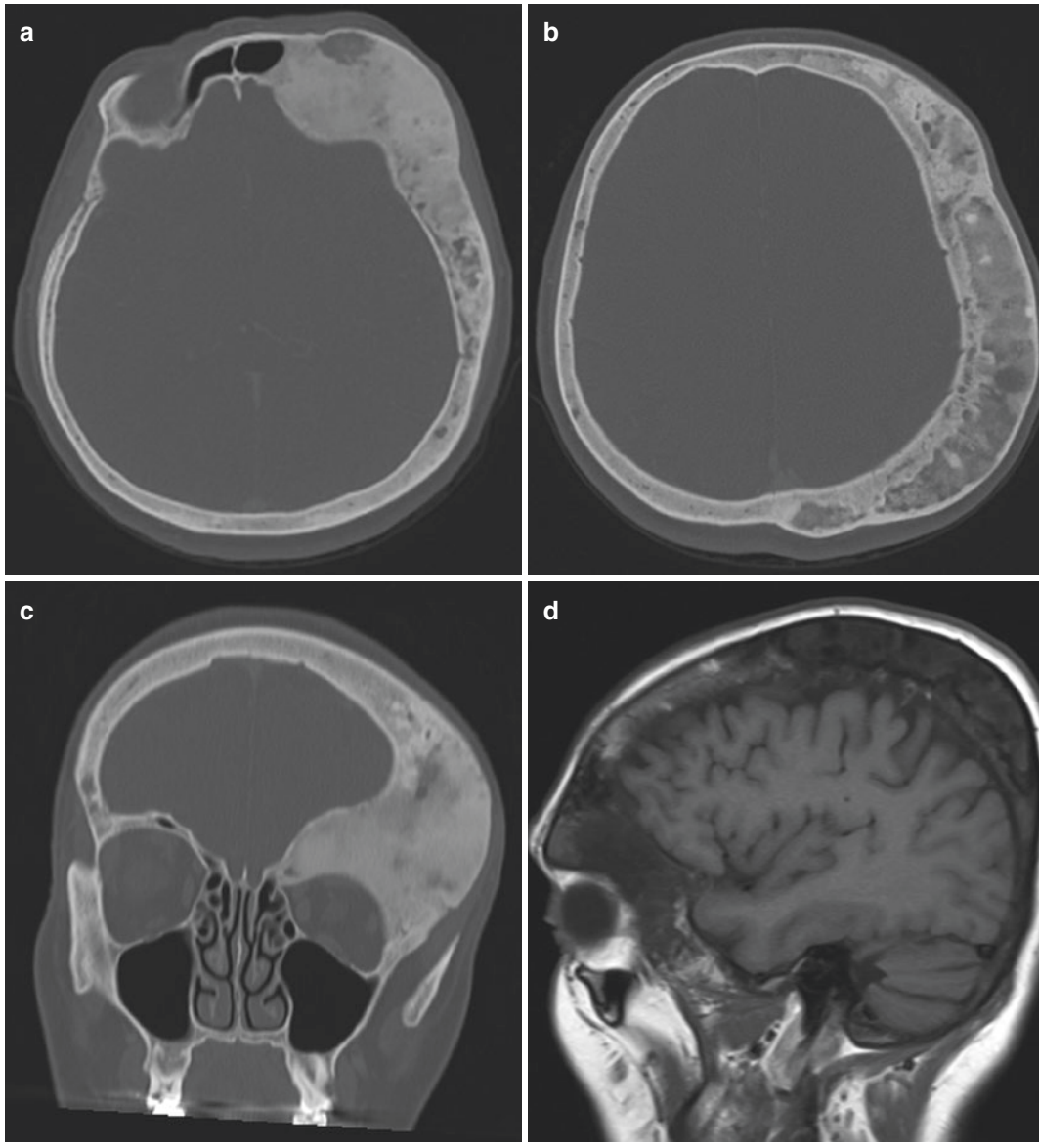

Fig. 16.1 Non-contrast-enhanced axial (**a**, **b**) and coronal (**c**) CT and sagittal T1WI (**d**), axial T2WI (**e**) and coronal T2WI (**f**) MRI images of the (monostotic; see text) fibrous dysplasia involving the left frontal, parietal, sphenoid and temporal bone. Note the facial asymmetry with left orbital deformity (**c**, **f**). CT images demonstrate loss of normal corticomedullary differentiation in the expanded bones, replaced by a ground-glass pattern with focal lucencies and scleroses. MRI images show heterogenous bone signal

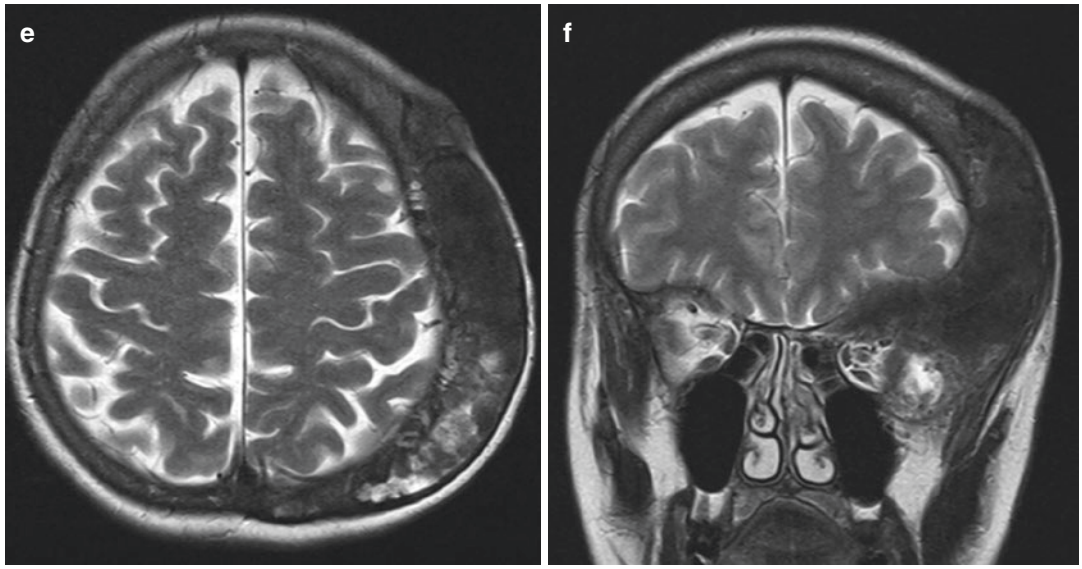

Fig. 16.1 (continued)

CT imaging features ground-glass expansile lesion centred in the medullary bone layer, with inner cortical scalloping and heterogenous sclerosis. There is no periosteal reaction.

MR imaging features consist of heterogenous signal, mostly intermediate in T1WI and low in T2WI and heterogenous contrast enhancement.

The transition to normal bone is often indistinct.

Differential diagnosis includes Paget disease which usually spares facial bones and is more sclerotic; intraosseous meningioma which tends to be sclerotic, does not spare the cortical bone and often abuts the intracranial compartment; sclerotic metastases which are usually smaller in size and focal in distribution; and cemento-ossifying fibroma which is usually distinct from the adjacent normal bone.

The risk for malignant transformation in FD is approximately less than 1% in the monostotic form and up to 4% in the polyostotic form, being the most frequent in McCune-Albright syndrome patients [2]. Prior radiation exposure is also recognized as a risk factor for malignant transformation. The most common sites of malignant transformation in monostotic form of fibrous dysplasia are facial and skull bones. Osteosarcoma accounts for approximately 70% of malignant transformation cases, followed by fibrosarcoma (20%) and chondrosarcoma (10%). The appearance of the benign fibrous dysplasia makes malignant transformation difficult to identify. Sarcomatous transformation may appear in form of cystic osteolytic areas, cortical destruction and heterogeneously enhancing soft tissue mass, such as in this case. The patient should be instructed to bring any change in symptoms to physician's attention. Rapid growth, especially in adults, pain without history of trauma and significant change in radiologic appearance are some of the signs of possible malignant transformation. Yearly X-rays are advocated for screening [3]. The cure for FD or ways to prevent malignant transformation still do not exist.

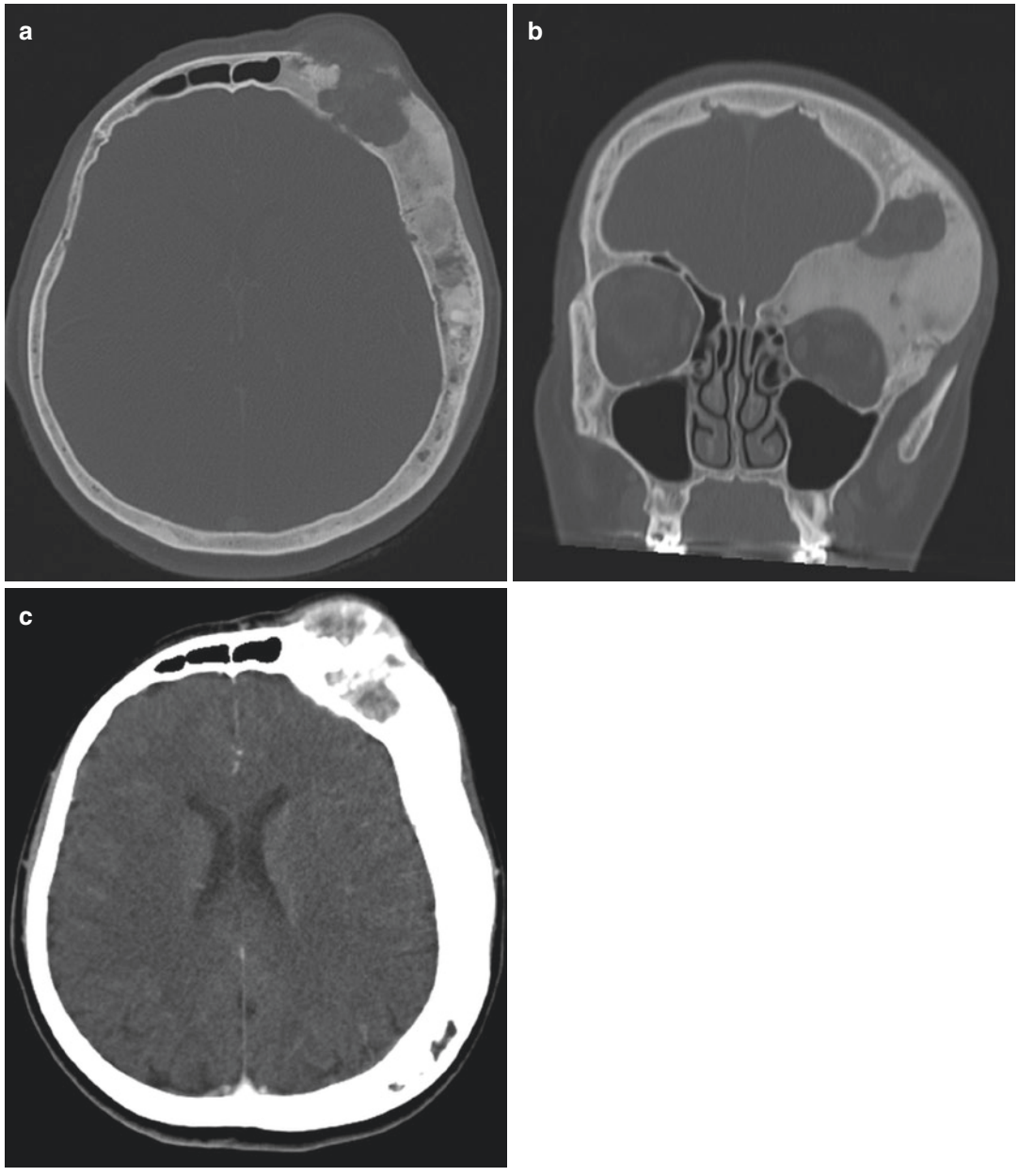

Fig. 16.2 Contrast-enhanced follow-up CT images of the head—note the left frontal bone defect (**a**, **b**) caused by an irregularly enhancing (**c**) osteolytic expansile lesion, not evident in Fig. 16.1

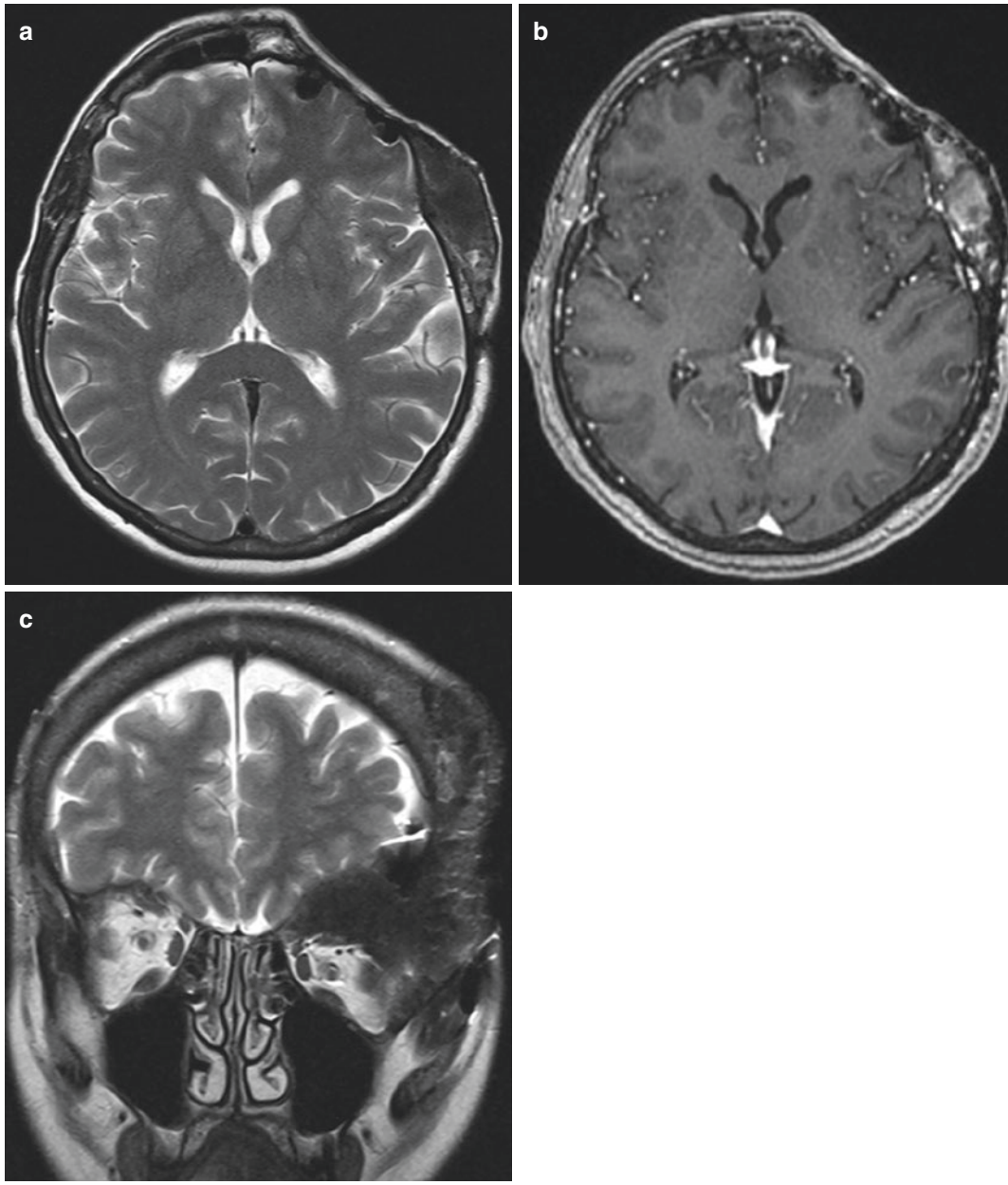

Fig. 16.3 Postoperative MRI of the head—axial T2WI (**a**), axial post-contrast T1WI (**b**), coronal T2WI (**c**). There are postoperative bony defects and characteristic post gadolinium enhancement of the remaining dysplastic bones (**b**)

References

1. Larheim TA, Westesson P-LA (2006) Maxillofacial imaging, vol 81. Springer, Berlin
2. Riddle ND, Bui MM (2013) Fibrous dysplasia. Arch Pathol Lab Med 137(1):134–138
3. Mardekian SK, Tuluc M (2015) Malignant sarcomatous transformation of fibrous dysplasia. Head Neck Pathol 9(1):100–103