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Desmoplastic fibroma of the mandible in young children—a case series

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Abstract. Desmoplastic fibromas are rare, benign, but aggressive lesions, affecting predominantly young people, with an affinity for the mandible. Four patients with desmoplastic fibromas of the mandible, seen between 1995 and 2015 with long-term follow-up, were identified. Three were treated by wide mandibular resection and immediate reconstruction with rib grafts, and one was treated with chemotherapy. In the three resected cases, there has been no recurrence and all rib grafts were successfully incorporated. The case treated by chemotherapy has persistence of the tumour, but it is not progressing. Desmoplastic fibromas in young children respond well to wide mandibular resection and immediate reconstruction with rib grafts. Chemotherapy may halt progression.

Key words: desmoplastic fibroma; benign tumour of jaws; jaw tumours in children.

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Desmoplastic fibromas are rare, benign, yet aggressive fibrous lesions of bone, with high recurrence rates. The lesion was first described in bone by Jaffe in 1958[1] and was later reported in the jaws by Griffith and Irby.[2] They account for 0.06% of all tumours of bone.[3] The mandible is the bone most frequently affected by desmoplastic fibromas (22–27%), followed by the femur (15%), pelvis (13%), radius (12%), and the tibia (9%).[4–6] Fibromatosis is a term first used by Stout in 1948[7] and represents the soft tissue counterpart of desmoplastic fibroma. There have been many confusing attempts to classify these lesions, including desmoplastic fibromas, extra-abdominal desmoids, juvenile fibromatosis, aggressive juvenile fibromatosis, infantile fibromatosis, and infantile myofibromatosis.[8–11] Gold, recognizing that biological activity in fibromatosis was not predictable from their histological morphology, suggested anatomical classification of intraosseous (endosteal), paragnathic , and combined.[12] This endosteal classification is what Jaffe described as desmoplastic fibroma.

The reported age range for presentation of desmoplastic fibromas of the jaws is from 12 months to 46 years, with an average age at diagnosis of 14 years. There is no predilection by sex, and pathognomonic symptoms do not exist. Patients may note a painless swelling as a presenting sign in some instances, difficulty with mouth opening, or more rarely, a pathological fracture.[6]

There is typically no associated lymphadenopathy or mobility of teeth, but displacement of teeth can occur. Radiographically, these lesions can be ill-defined or well-circumscribed unilocular or multilocular radiolucencies with the potential to perforate through buccal and lingual cortices. A periosteal reaction is uncommon, but the periosteum itself can be the origin of the desmoplastic fibroma.[4]

Histopathologically, fibromatosis and desmoplastic fibromas are identical. They are described as showing small uniform elongated fibroblasts within a stroma of abundant collagen fibres. A key feature

separating these lesions from fibrosarcomas is a lack of mitoses, cellular atypia, and pleomorphism.[13] Flucke et al. identified the first case providing a genetic relationship between desmoid tumours and desmoplastic fibromas when assessing seven head and neck desmoids in children.[14] All patients were β-catenin-positive, but they noted that one of two desmoplastic fibromas showed a pT41A mutation, a CTNNB1 gene mutation found in approximately 85% of desmoids. The head–neck desmoids/desmoplastic fibromas show heterogeneous mutations associated with the CTNNB1 gene in contrast to more homogeneous changes with tumours at other sites.[14]

Histologically it is difficult to differentiate from other benign entities including nodular fasciitis, fibrous dysplasia, myofibromas, non-ossifying fibromas, and odontogenic fibroma.[13] Even more concerning is histological similarity to malignant lesions such as low-grade fibrosarcomas and osteosarcoma. Unlike ossifying fibromas and sarcomas, bone is never an integral part of the lesion. Fibrous dysplasia can mimic desmoplastic fibromas in areas devoid of osteoid and areas that are predominantly fibrous tissue. Fibrous dysplasia is further characterized by bone deposition and shorter compact nuclei rather than the fibroblasts, collagen, and elongated slender nuclei found in desmoplastic fibromas.[4]

Key histological characteristics of fibrosarcomas and osteosarcomas are spindle cells with cellular atypia, mitotic figures, and cellular pleomorphism. Fibrosarcomas have a low to moderate cellular proliferation of spindled cells, often uniform and without the classic 'herring-bone' pattern of a high grade fibrosarcoma. In contrast desmoplastic fibromas are hypocellular and have spindle cells organized in a more unidirectional pattern whose nuclei are without atypia, pleomorphism, or mitotic figures. Further, Woods et al. have stated that the lesional cells in desmoplastic fibromas display indistinct cell borders, with stroma that often encases thin spicules of trabecular bone exhibiting reactive changes.[13] Osteosarcomas also have irregular scattered bony trabeculae. Knowing this, desmoplastic fibromas should be a diagnosis of exclusion.

Histological similarities have prompted further means of analysis with immunohistochemical markers. In the literature review by Woods et al., including 152 patients and three of their own, immunohistochemical markers including S100, SMA, vimentin, MSA, Ki-67, and since 2002 β-catenin, have been utilized to delineate gnathic desmoplastic fibromas from other similar entities.[13]

Although these lesions do not metastasize, they do infiltrate and are locally aggressive, resulting in a high recurrence rate if not completely removed. In 1996, Böhm et al. reviewed 196 cases of desmoplastic fibroma of bone (194 in the literature and two of their own) with regards to clinical and histological data, with a focus on treatment and recurrence. DNA analysis of desmoplastic fibromas showed a higher incidence of proliferation in the intrabony form when compared to fibromatosis (24% vs. 8%, respectively).[5] Due to the aggressive nature of this benign lesion, treatment recommendations vary from aggressive surgical resection to conservative enucleation. Enzinger and Weiss reported that fibromatosis in infancy and childhood appeared more aggressive then in older children and could easily be confused with fibrosarcomas. They further classified fibromatosis under the age of 5 years as aggressive infantile fibromatosis.[15] Carr et al. argue that one should not use the term aggressive for infantile fibromatosis, as all infantile fibromatosis shows unpredictable behaviour and all should be treated aggressively.[16]

Four cases of desmoplastic fibroma of the jaw are described herein. The four patients were treated in the department of oral and maxillofacial surgery of the authors' institution and were followed up for an average of 14 years. No recurrence was noted.

Materials and methods

Four young children with desmoplastic fibromas of the mandible, who were treated and followed up at the study institution in San Francisco, USA between 1995 and 2015 , were identified. Three patients (cases 1 to 3) were under regular follow-up and charts were available. One patient (case 4) was lost to follow-up after 6 years. Institutional review board approval was given to contact this patient.

Case 1

An 8-year-old Caucasian female was noted to have had an asymptomatic, firm, 1-cm expansile lesion of the left mandible for 1.5 years. A panoramic radiograph showed an ill-defined, loose, trabecular bone structure in the left mandible, extending from the angle forward to the bicuspid region (Fig. 1). The lower left first molar was displaced anteriorly and the second molar was displaced posteriorly. Computed tomography (CT) reported a well-circumscribed, expansile, lytic lesion of the mandibular body measuring 2.6 cm × 1.6 cm × 2.4 cm (Fig. 2). An incisional biopsy reported a benign connective tissue spindle cell proliferation with a differential diagnosis of odontogenic fibroma, desmoplastic fibroma, or chondromyxoid fibroma. The patient subsequently underwent a supraperiosteal dissection of the left mandible from the canine region to the angle and the lesion was resected with 1-cm margins (Fig. 3). The defect was immediately reconstructed with right ribs 5 and 6. Final pathology noted a desmoplastic fibroma of 1.8 cm at its greatest dimension, with negative margins (Fig. 4). The patient had an uneventful postoperative course, with adequate graft consolidation. Nine years later, the patient elected to proceed with secondary reconstruction using a bicortical iliac crest bone graft (Fig. 5) and later was

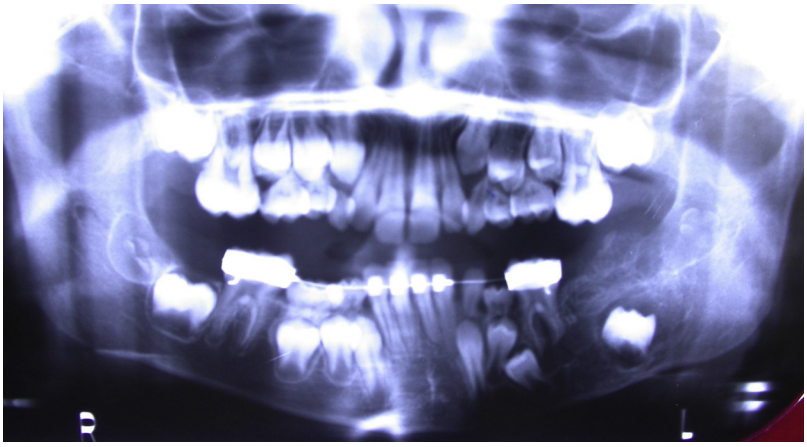

Fig. 1. Initial panoramic radiograph showing abnormal bone formation and displaced teeth in the left posterior mandible.