Article is available online at http://www.webio.hu/por/2004/10/4/0237

CASE REPORT

Desmoplastic Fibroma-Like Tumor of Maxillofacial Region Associated with Tuberous Sclerosis

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Desmoplastic fibroma is a rare primary tumor of bone that histologically and biologically mimics the extra-abdominal desmoid tumor of soft tissue. It usually presents in patients during the first three decades of life and often involves the mandible or long bones of the skeleton. Its clinical behavior is characterized by a locally aggressive, infiltrating, and destructing course, often with invasion of surrounding tissues but without metastasis. We present herein the clinicopathological features of a desmoplastic fibroma-like tumor involving the left maxillofacial region in a 14-year-old Hispanic boy with tuberous sclerosis. (Pathology Oncology Research Vol 10, No 4, 237–239)

Keywords: desmoplastic fibroma, desmoid tumor, bone tumor, tuberous sclerosis

Introduction

Desmoplastic fibroma of bone is a rare, locally aggressive lesion that histologically resembles a desmoid tumor of the soft tissues. Although considered a benign lesion, it has a high rate of local recurrences after incomplete surgical excision. Review of the literature revealed scattered previous reports of desmoplastic fibroma-like lesions within the jaws of patients with tuberous sclerosis.[1-2] In fact, these desmoplastic fibroma-like tumors have been suggested to be part of the tuberous sclerosis complex.[2]

Case Report

A 14-year-old Hispanic boy with tuberous sclerosis presented for evaluation of a slowly enlarging, non-painful osseous mass involving the left maxillofacial region, present for 3 years. Clinical examination revealed facial angiofibromas and a left hemi-facial mass, approximately 8x6 cm in size (*Figure 1*). Computed tomographic scans displayed an aggressive bone tumor with trabeculated, "soap bubble" appearance (*Figure 2*). The mass showed invasion of the orbit and soft tissues of the floor of the mouth.

After taking incisional biopsies of the intrabony mass and its soft tissue extension, microscopic examination of the specimen revealed fibroblastic-like spindle cells proliferating within a delicate mesenchymal stroma, containing intertwining collagen fascicles (*Figure 3a*). No cellular pleomorphism was observed and scant mitotic figures were identified (*Figure 3b*). Immunohistochemical labeling was performed, and the tumor cells proved positive for vimentin (*Figure 3c*), while actin and S-100 were negative.

The lesion was locally infiltrative and uncapsulated; the surgical procedure consisted of resection of the tumor and facial reconstruction. The pathological analysis of the specimen confirmed the diagnosis of desmoplastic fibroma-like tumor. Four months later, the patient was diagnosed with a 6-cm renal angiomyolipoma using echography and CT scan performed for hematuria. A nephrectomy was done. No follow-up information is available for this treated patient.

Received: Aug 27, 2004; *accepted:* Sept 18, 2004

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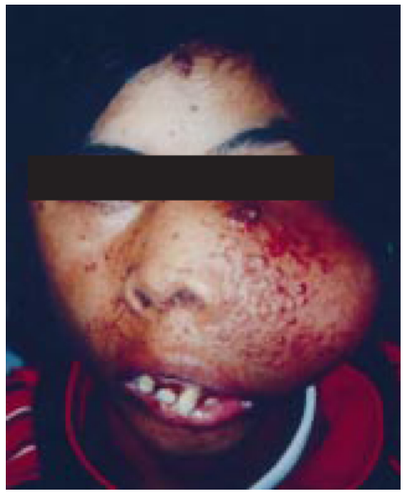

Figure 1. *Clinical appearance of facial angiofibromas and giant left maxillofacial tumor.*

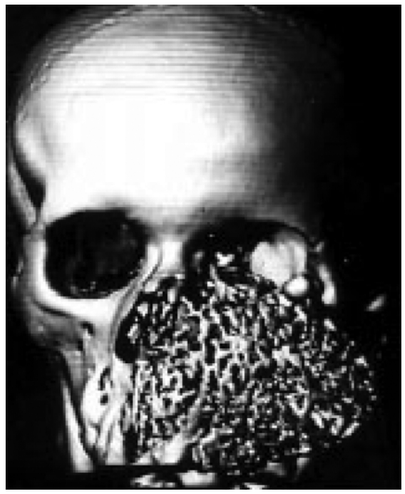

Figure 2. *Three-dimensional computed tomography shows an expansile, trabeculated bone tumor.*

Discussion

In 1958, Jaffe[3] first distinguished desmoplastic fibroma from other intraosseous fibrous tumors. It is one of the rarest bone tumors, representing the intraosseous counterpart of the soft tissue desmoid tumor or fibromatosis. In a review by the Mayo Clinic, a series of approximately 30,000 bone tumor cases included only 27 cases of desmoplastic fibroma.[4] There is only 1 case among the 776 benign bone tumors in the Dutch bone tumor registry.[5] Dahlin and Unni[6] presented 9 cases of DF in a series of 8542 primary bone tumors (0.11%). Böhm et al.[7] reviewed 191 cases of DF reported in 80 publications. In their review, the age of patients ranged from 15 months to 75 years, with a reported mean age of 23 years. Approximately 16% of the patients were in their first decade, 35% were in their second decade, and 23% were in their third decade. Seventy-four percent of the patients were younger than 31 years, and only 6% were older than 50 years. According to published data on tumor location in 184 patients, DF most often involved the following bones: mandible (22%), femur (15%), pelvic bones (13%), radius (12%), and tibia (9%).

In the maxillofacial location desmoplastic fibroma usually presents as a painless, slow-growing, firm mass, and less commonly reported symptoms include loose teeth, recurrent sinusitis and exophthalmos.

Radiographically, desmoplastic fibromas are expansile, lytic lesions, often with internal trabeculation and "soap bubble" appearance. Cortical thinning and disruption are common findings, often associated with a soft tissue mass.[8] Computed tomography is superior to standard radiography in assessment of the cortical continuity.[9]

Histologically, desmoplastic fibroma consists of a mildly to moderately cellular matrix of fibrocollagenous stromal tissue, lacking cellular pleomorphism, nuclear hyperchromatism, or mitosis. Both the lack of a capsule and the infiltrative nature of this lesion are hallmarks of desmoplastic fibroma.

The differential diagnosis includes low-grade fibrosarcoma, fibrous dysplasia, congenital fibromatosis, nonossifying fibroma and odontogenic fibroma. Several histologic findings, including increased number and size of cells, greater nuclear pleomorphism, and increased mitotic activity, suggest a well-differentiated fibrosarcoma. The hallmark of fibrous dysplasia is formation of immature woven bone and a bony matrix that characteristically lacks the osteoblastic rim. The solitary congenital fibromatosis (infantile myofibromatosis) is most commonly seen in the craniofacial bones of patients less than 2 years old. It may be distinguished from desmoplastic fibroma by its cellular morphology, nodular growth pattern, and immunohistochemical profile to include positive reactivity for smooth muscle actin and S-100. The fibrogenic stroma on nonossifying fibroma contains plumper cells in a more cellular storiform pattern, with scattered giant cells and clusters of lipophages and hemosiderin-laden macrophages. The odontogenic fibroma is composed of stellate fibroblasts, often arranged in a whorled pattern with fine collagen fibrils and considerable ground substance. Small foci of odontogenic epithelial rests may or may not be present.

Surgical management ranging from simple curettage to segmented resection has been recommended for treatment of this tumor. Segmental resection is preferred when a lesion displays signs of aggressive behavior and extension into surrounding soft tissues. Significant rates of recurrence have been associated with desmoplastic fibroma, depending on the completeness of surgical removal.[7]

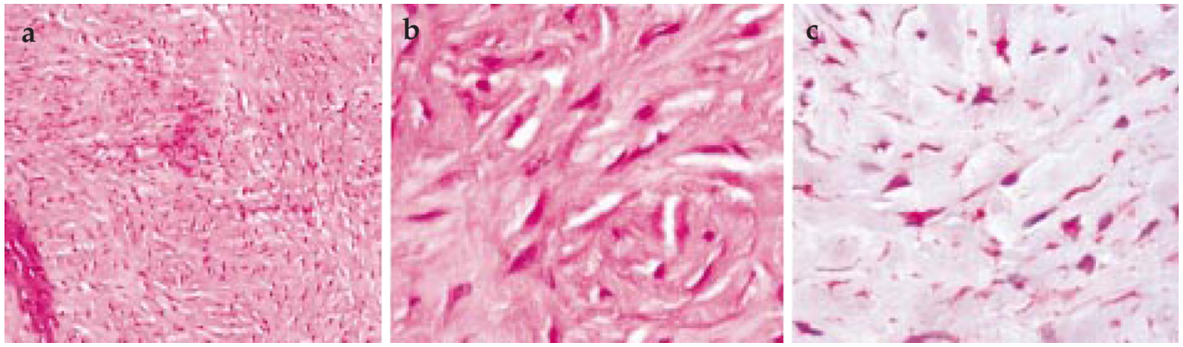

Figure 3. *(a) A moderately cellular neoplasm composed of bland spindle cells in a background of numerous wavy collagen fibers (HE, ×10). (b) Fibroblastic cells without atypia and pleomorphism (×40). (c) Positive staining for vimentin was seen in the neoplastic cells (×40).*

Tuberous sclerosis is a hamartoneoplastic syndrome exhibiting multiple hamartomatous proliferations that may involve a variety of organ systems. The process is autosomal dominant, but two thirds of the cases are sporadic and appear to represent new mutations. These mutations involve either one of two recently described genes: TSC1 or, more commonly, TSC2. Both of these gene products are believed to contribute to the same intracellular biochemical pathway that seems to have a tumor suppressor function.[10] The multiple hamartomatous growths that are seen in this disorder are thought to arise from disruption of the normal tumor suppressor function of these genes. The prevalence of tuberous sclerosis is between 1 in 10,000 and 1 in 23,000 in the general population. The extraneural lesions involve the skin (facial adenofibroma, hypomelanotic macule, shagreen patch, fibroma, café au lait spot), heart (rhabdomyoma), kidney (angiomyolipoma), lung (lymphangiomatosis), spleen (hemangiomatosis), liver (hemangiomatosis) and bone (fibrous dysplasia).

Oral manifestations of tuberous sclerosis include developmental enamel pitting on the facial aspect of the anterior permanent dentition in 50% to 100% of patients.[11] Multiple fibrous papules affect 11% to 56% of patients. The fibrous papules are seen predominantly on the anterior gingival mucosa, although the lips, buccal mucosa, palate, and tongue may be involved. Less common oral manifestations include hemangiomas, facial asymmetry, high arched palate, bifid uvula, cleft lip/palate, delayed eruption, and diastemas.[12]

In the literature we found some reports of intraosseous fibrous lesions of the jaws indistinguishable from desmoplastic fibroma in patients with tuberous sclerosis. Miyamoto et al.[1] in 1995 reported an example with significant clinical, radiographic, and histopathologic features. The patient was a 29-year-old Japanese woman who manifested a well-defined radiolucency of the anterior mandible, approximately 7×5×3.5 cm. The tumor was diagnosed as desmoplastic fibroma. Miyamoto et al. suggested the possibility that the tumor depicted in their report might represent an intraoral manifestation of tuberous sclerosis. Damm et al.[2] in 1999 reported four additional cases and made an extensive review of the literature, concluding that these intraosseous fibrous lesions of the jaws or "desmoplastic fibroma-like tumors" are thought to represent a manifestation of tuberous sclerosis rather than a coincidental finding. We suggest a clinicopathological correlation in any case of desmoplastic fibroma-like tumor occurring in the jaw of patients with tuberous sclerosis.

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