Fibromatosis Arising in Association with Neuromuscular Hamartoma of the Mandible

Leena Yaseen Taher · Muhammad Saleem ·
Suresh Velagapudi · Anas Dababo

Received: 21 September 2012 / Accepted: 28 December 2012 / Published online: 12 January 2013
© Springer Science+Business Media New York 2013

Abstract Aggressive fibromatosis is a benign but locally-aggressive tumor, which most often affects the muscles of the shoulder, the pelvic girdle, and the thigh. It usually affects adolescents and young adults. Desmoplastic fibroma, considered the bone counterpart of soft tissue fibromatosis, is a rare tumor that usually affects the metaphyseal or diaphyseal portions of long bones or, less commonly, the jaw. Neuromuscular hamartoma, a rare developmental lesion composed of mature elements of both striated muscle and nerve, is usually diagnosed in infants and children and affects large nerve trunks. Rarely, it can affect the head and neck region. Occasional cases showing an association between aggressive fibromatosis and neuromuscular hamartoma have been reported in the literature. Here we present a unique case of an adult patient with desmoplastic fibroma of the mandible in association with neuromuscular hamartoma.

Keywords Neuromuscular hamartoma · Desmoplastic fibroma · Aggressive fibromatosis · Mandible

Introduction

Deep extra-abdominal fibromatosis (also known as aggressive fibromatosis or desmoid tumor) is a benign but locally aggressive tumor. It manifests during the first three decades of life and shows no gender predilection. Despite the bland microscopic appearance, it frequently carries a high rate of recurrence. Treatment is usually carried out by a wide local excision, if possible. Postoperative radiation should be considered in incompletely excised lesions. Desmoplastic fibroma (DF) is the bone counterpart of soft tissue aggressive fibromatosis. It affects the metaphyseal or the diaphyseal parts of long bones and less commonly the jaw (about one-fifth of DF cases). The radiologic features of desmoplastic fibroma are not specific and usually show an osteolytic expansile intramedullary lesion, which can break through the thinned cortex and involves the adjacent soft tissue. Neuromuscular hamartoma is a rare developmental nodular lesion comprising mature skeletal myofibers of variable sizes in an intimate relationship with nerve fibers. There have been few reported cases of fibromatosis in association with and/or complicating a neuromuscular hamartoma, which cannot be explained by just a coincidence.

Case Report

A 28-year-old female presented with a 6 month history of trismus and right-sided painless facial swelling accompanied by numbness. There was no history of fever, night sweats, or weight loss. Her past medical/surgical history was unremarkable. Clinical examination revealed diffuse right-sided

L. Y. Taher
Department of Pathology, Taibah University, Al-Madina,
Saudi Arabia

L. Y. Taher (✉)
Department of Pathology (32), College of Medicine and King
Khaled University Hospital, P.O. Box 2925, Riyadh 11461,
Saudi Arabia
e-mail: leena.taher@gmail.com

M. Saleem · S. Velagapudi
Department of Otorhinolaryngology Head and Neck Surgery,
King Faisal Specialist Hospital and Research Centre, Riyadh,
Saudi Arabia

A. Dababo
Department of Pathology, King Faisal Specialist Hospital and
Research Centre, Riyadh, Saudi Arabia

Fig. 1 An axial CT image demonstrating the destruction of the right mandibular ramus by an infiltrative mass

facial swelling. There were no other masses or lymphadenopathy. Computerized tomography (CT scan) demonstrated an expansile mass involving the right ramus of the mandible, measuring $4 \times 3 \times 3$ cm with remarkable bone thinning and craniocaudal extension (Fig. 1). The radiologic diagnosis was that of a “benign bony cyst or a dentigerous cyst, although malignancy cannot be excluded”. The patient had a previous biopsy at a local hospital, which was interpreted as “reactive periosteal fasciitis”. Upon receiving the outside material, our diagnosis was consistent with desmoplastic fibroma (fibromatosis) of the mandible. Subsequently, the patient underwent partial right hemimandibulectomy with reconstruction. Grossly, there was a $7 \times 4 \times 3$ cm tan-gray rubbery intramandibular lesion with focal extension into the adjacent soft tissue (Fig. 2). Histologically, the lesion was composed of a bland fibroblastic proliferation with variable cellularity and dense collagen deposition (Figs. 3, 4). Beta catenin demonstrated nuclear positivity (Fig. 5). Interestingly, there were multiple foci of neuromuscular hamartoma, characterized by intimately related mature skeletal muscle myofibers (Fig. 4) and nerve fibers. The largest focus was noted within the mandible (Figs. 6, 7) and several other smaller neuromuscular hamartomas were also observed within the adjacent soft tissue (Fig. 8). Although the morphologic appearance of the lesion is very characteristic, immunohistochemistry was performed to highlight the small skeletal myofibers and nerve axons, e.g. positive staining with desmin (Fig. 9) and neurofilament protein (NFP) (Fig. 10), respectively.

Discussion

Fibromatosis is a locally aggressive tumor that can damage nearby structures causing organ dysfunction and tends to

Fig. 2 The gross specimen of the posterior compartment of the right mandible showing a tan-gray, rubbery mass

Fig. 3 Desmoplastic fibroma: spindle cell proliferation of fibroblastic origin in a collagenous stroma. (H&E; $\times 10$)

recur even after wide resection. It is common in the abdomen, shoulder girdle, thigh, and gluteal regions of adults. Desmoplastic fibroma of bone is identical to the fibromatosis of soft tissue. It usually affects the long bones but is very rare in the mandible. In 1965, the first report about a desmoplastic fibroma of the jaw was presented by Griffith and Irby [1]. Neuromuscular hamartomas are rare lesions of the peripheral nerve characterized by the presence of mature neural tissue and well-differentiated striated muscle exhibiting an intimate relationship between the two components. Most cases of neuromuscular hamartomas occur in infants and children affecting large nerves, like the sciatic and the brachial nerve [2, 3], or intracranial nerves such as the trigeminal nerve [4, 5]. These lesions are considered benign, although the pathogenesis is unknown and many theories regarding their etiology have been

Fig. 4 Desmoplastic fibroma infiltrating through the muscle bundles of the surrounding soft tissue. (H&E; ×20)

Fig. 7 Muscle fibers intimately associated with a nerve trunk. (H&E; ×10)

Fig. 5 Positive nuclear staining for β-catenin. (β-catenin; ×40)

Fig. 8 Neuromuscular hamartoma in the soft tissue. (H&E; ×20)

Fig. 6 A neuromuscular hamartoma within the mandibular bone. (H&E; ×10)

Fig. 9 Immunohistochemical staining for desmin highlighting the skeletal muscle differentiation. (Desmin; ×10)

Fig. 10 Immunohistochemical staining for NFP demonstrating the neural component of neuromuscular hamartoma. (NFP; $\times 10$)

proposed. Karyotypic analysis in a case reported by Kim et al. [6] was normal, which favors the opinion that this lesion is a malformation, not a true neoplasm. Development of aggressive fibromatosis in association with neuromuscular hamartoma has been reported in the literature. Boman et al. [2] reported a case of neuromuscular hamartoma involving the left sciatic nerve in a 4-year-old girl with equinism (“clubfoot”) and spasticity. A first surgical procedure, with muscle biopsy, did not yield a diagnosis of hamartoma. This was followed by the occurrence of a firm mass at the same location. Partial excision was subsequently performed, and the mass was revealed to be comprised largely of fibrous tissue. Bonneau et al. [7]

discussed a case of a 14-year old girl with multiple small neuromuscular hamartomas surrounding a recurrent massive fibromatosis of the right axilla. Mitchell et al. [8] reported a classic case of neuromuscular hamartoma in a female patient where the lesion appeared to grow after incomplete initial resection. Re-excision revealed fibromatosis. Despite its wide excision, recurrence of the fibromatosis required an amputation. Blouin et al. [9] conducted the most recent literary study wherein 10 cases were reviewed (Table 1), and a diagnosis of neuromuscular hamartoma was established. Four of these cases, including 2 males and 2 females presenting with lesions involving the sciatic nerve (3 cases) and brachial plexus (1 case), developed aggressive fibromatosis. The diagnosis of neuromuscular hamartoma was made on the basis of either open or CT-guided biopsy. All patients presented with a new enlarging mass between 4 months and 8 years post-operatively. Biopsy revealed aggressive fibromatosis in 3 of the 4 patients; in the other patient, although the imaging study was consistent with aggressive fibromatosis, the limited biopsy didn’t confirm the diagnosis. Blouin et al. [9] remarked that the occurrence of aggressive fibromatosis in patients with neuromuscular hamartoma is more than a coincidence, since both lesions are rare. Physical factors such as trauma or surgery may contribute to this process. No endocrine abnormality has been reported, but it is difficult to eliminate this possibility. Genetic alteration is unlikely to be the dominant predisposing factor for the development of aggressive fibromatosis in neuromuscular hamartoma patients. He also concluded that there is a direct relationship between the occurrence of aggressive

Table 1 Cases of neuromuscular hamartoma that are associated with definite or suspected aggressive fibromatosis

Authors	Number of cases	Location of neuromuscular hamartoma	Onset of fibromatosis after diagnosis of neuromuscular hamartoma
Bonneau and Brochu [7]	1	Right axilla	Initial operation
Chen [11]	1	Right median nerve (axilla)	Short period post-operation
Awasthi et al. [3]	1	Left brachial plexus	After 35 years
Boman et al. [2]	1	Left sciatic nerve	After 1 year
Mitchell et al. [8]	1	Right axilla and chest wall	5 months post-operation
Tiffée and Barnes [10]	1	Left face and orbit	1 month post-operation
Blouin et al. [9]	4	(1) Right sciatic nerve (2) Left sciatic nerve (3) Right sciatic nerve (4) Right brachial plexus	8 years post-biopsy 3 years post-biopsy 4 months post-biopsy
Present case	1	Right mandible	Enlarging mass by MRI (no diagnostic biopsy) Initial biopsy

fibromatosis and neuromuscular hamartoma. It has been suggested that the development of fibromatosis may be related to the degeneration of neural and skeletal muscle components due to the loss of trophic stimuli. This process could lead to either spontaneous regression of neuromuscular hamartoma, or to the development of aggressive fibromatosis. Some studies opine that neuromuscular hamartoma is the result of the entrapment of mesenchymal cells in the developing nerves [10] where neuroectoderm is capable of mesenchymal differentiation, specifically into skeletal muscle [6]. Our case is unique in that the fibromatosis/desmoplastic fibroma arose in association with neuromuscular hamartoma within the mandibular bone in an adult patient.

Conclusion

To our knowledge, this is the first case report of desmoplastic fibroma/aggressive fibromatosis and neuromuscular hamartoma within the mandibular bone. We believe that the fibromatosis arose in association with a preexisting neuromuscular hamartoma.

References

1. Griffith JG, Irby WB. Desmoplastic fibroma. Report of a rare tumor of the oral structures. *Oral Surg Oral Med Oral Pathol.* 1965.
2. Boman F, Palau C, Floquet A, Floquet J, Lascombes P. Neuromuscular hamartoma. *Ann Pathol.* 1991;11:36–41.
3. Awasthi D, Kline DG, Beckman EN. Neuromuscular hamartoma (benign “triton” tumor) of the brachial plexus. Case report. *J Neurosurg.* 1991;75:795–7.
4. Castro DE, Raghuram K, Phillips CD. Benign triton tumor of the trigeminal nerve. Case report. *AJNR Am J Neuroradiol.* 2005;26:967–9.
5. Vajramani G, Devi I, Santosh V, Hegde T, Das BS, Das S, et al. Benign triton tumor of the trigeminal nerve. *Childs Nerv Syst.* 1999;15:140–4.
6. Kim DH, Hong EK, Lee JD. Subcutaneous neuromuscular hamartoma. A case report. *Korean J Pathol.* 1999;33:62–4.
7. Bonneau R, Brochu P. Neuromuscular choristoma. A clinicopathologic study of two cases. *Am J Surg Pathol.* 1983;7:521–8.
8. Mitchell A, Scheithauer BW, Ostertag H, Sepehrnia A, Sav A. Neuromuscular choristoma. *Am J Clin Pathol.* 1995;103:460–5.
9. Blouin MNH, Scheithauer BW, Amrami KK, Durham SR, Spinner RJ. Fibromatosis: a potential sequela of neuromuscular choristoma. *J Neurosurg.* 2012;116:399–408.
10. Tiffie JC, Barnes EL. Neuromuscular hamartomas of the head and neck. *Arch Otolaryngol Head Neck Surg.* 1998;124:212–6.
11. Chen KT. Neuromuscular hamartoma. *J Surg Oncol.* 1984;26:158–60.