Short Case Report

Facial fibromatosis: benign and aggressive, yet treatable!

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Abstract – Fibromatosis or desmoid tumors are locally aggressive neoplasms that have a propensity for local invasion and recurrence. The mainstay of treatment is excision with negative margins and the role of radiotherapy is controversial. Desmoids arising in the head and neck area are rare and pose a dilemma to the surgeon due to large number of vital structures that preclude resection with wide margins. This leads to a high incidence of recurrence. We present a case of a 24-year-old male who presented with an asymptomatic left sided facial swelling causing cosmetic deformity. A subcutaneous lipoma/fibroma/neuroma was suspected and a fine needle aspiration done was inconclusive. Computed tomography revealed a subcutaneous swelling that was excised. Histopathology revealed it to be a desmoid tumor. At the end of 4-year-follow-up, he has no local recurrence.

Introduction

Fibromatosis refers to a locally aggressive, slow growing neoplasm arising due to uncontrolled proliferation of musculoaponeurotic fibrous tissue. As opposed to its malignant counterpart fibro sarcoma, fibromatosis or desmoid tumors have few mitotic cells and absent necrosis on microscopy and do not metastasize. Despite its benign nature, the local aggressiveness of this tumour combined with its propensity for recurrence makes it a frustrating problem [1]. It may be abdominal, intra-abdominal or extra-abdominal. Fibromatosis affecting the head and neck comprises 10–25% of extra-abdominal disease [2]. The large number of vital structures coursing through the face combined with the invasive nature of this disease is what leads to the significant morbidity associated with facial fibromatosis, as complete resection may not always be possible [3].

Case report

A 24-year-old male with no comorbidities presented with complaints of an asymptomatic, gradually growing left sided facial swelling of 6 months duration. There was no other

positive history. General examination was normal. Local examination revealed a hard, non-tender, nodular, partially fixed 4 × 3 cm swelling around the zygomatic arch (Fig. 1). There were no other swellings or palpable cervical lymph nodes. Intraoral examination was unremarkable. Examination of facial nerve, mouth opening and jaw movements were normal. A differential diagnosis of subcutaneous lipoma/fibroma/neuroma was considered and a fine needle aspiration cytology (FNAC) performed, but was inconclusive. A contrast enhanced computed tomography (CECT) of the area done revealed a well-defined, round-to-oval peripherally enhancing lesion superficial to left masseter muscle measuring 17.1 mm (transverse) × 19.1 mm (anteroposterior) × 23.5 mm (vertical) dimension with no infiltration and normal adjacent fat planes. Left parotid gland was also normal (Fig. 2). An incisional biopsy was planned but the patient did not consent for the same, hence an excision was carried out under general anesthesia. Intra-operatively, the tumor was hard, but not infiltrating the surrounding structures (masseter, Stenson's duct, and branches of facial nerve) and was excised *in toto* (Fig. 3) and was sent for frozen section (FS) that was reported to be a benign looking fibromatosis. The margins were re-excised and also were found to be free of tumor cells at FS. Final histopathology revealed a spindle cell hypocellular tumor with a dense collagen background, no cell atypia, pleomorphism or mitotic figures

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Fig. 1. Lump round the left zygomatic arch.

Fig. 3. Excised specimen.

Fig. 4. Photomicrograph showing sparse spindle shaped cells, whorly pattern, no abnormal cell lines (H and E, $\times 200$).

Fig. 2. CECT showing a well-circumscribed, non-invasive, tumor (arrows).

(Fig. 4). Immunohistochemistry was beta catenin positive (Fig. 5). Re-excised margins were clear of tumor cells. Hence a final diagnosis of fibromatosis of left infratemporal region was made. He made a good postoperative recovery and has been kept under close follow-up: the patient has undergone MRIs periodically (yearly) and after 4 years there has been no recurrence.

Discussion

Fibromatosis also called desmoid tumors have historically been called by many names including fibrosarcoma grade 1, desmoplastic fibroma and desmoma. However as this tumour has virtually no malignant potential the term fibrosarcoma no

Fig. 5. IHC showing positivity for beta catenin.

longer includes desmoid tumors [3]. Although desmoids may occur anywhere in the body, majority of them arise from the abdomen with only 10–25% arising in the head and neck, with an incidence of 2 to 4 cases per million patients each year.

Even in this rare subset of patients, tumors of the face are rare, with the cervical and supraclavicular area accounting for 40–71% of cases [4].

Desmoid tumors usually present as painless, indolent masses that are clinically fixed to underlying bone or muscle. Symptoms relate to the site of its presence. Local invasion and compression can lead to pain, dysphagia, and hoarseness, proptosis, drooling of saliva or nasal bleeds [3–5]. In a few patients impingement of the trachea or infiltration of the carotids can lead to life threatening emergencies [2]. There is no accepted etiology for desmoid tumors. However an increased risk associated with Gardner's syndrome, possibly implying a genetic component has been documented [6]. There are numerous reports of antecedent trauma to the area, such as from thermal, blunt injury or prior surgery, giving credence to the theory that immature fibroblasts stimulated by injury develop an uncontrolled proliferative drive, though the majority of head and neck desmoids have no reports of antecedent trauma [2,4,7]. There is also an evidence of hormonal influence, supported by the increased incidence in women during puberty and pregnancy and regression of tumors following menopause and castration. de Bree *et al.* have reviewed and summarized cases of head and neck facial fibromatosis reported in the literature [4].

The tumors are firm to hard, rubbery and demonstrate a whorl-like pattern on cut section. Histologically they comprise of spindle shaped cells in a collagen matrix with no atypical figures and almost no mitotic figures with occasional lymphocytic invasion at the periphery. The most common differential diagnoses are reactive fibrosis, myxoma, nodular fasciitis, keloids, rhabdomyosarcoma or well-differentiated fibrosarcomas. The diagnostic workup of a suspected desmoid should begin with a MRI or CT to assess local invasion followed by an incisional biopsy to confirm the diagnosis [4].

The mainstay of treatment of desmoids is surgical excision with negative margins. Given the difficulty of attaining this in the head and neck, resulting in a high recurrence rate of between 46–62%, numerous adjuvant strategies have been proposed [2]. In most of the cases majority of recurrences of head and neck desmoid tumors occur in the first 2 years, but duration of recurrence varies from months to more than 10 years, and therefore close follow-up for at least 2, but preferably more years should be considered [8–10]. Adjuvant or even definitive radiation has been tried with varied results. There is no consensus regarding the role of radiation with some studies demonstrating only a transient control in size and other advocating radiation for residual disease [7,11]. Other treatment modalities such as Selective Estrogen Receptor Modulators (SERMs), Non Steroidal Anti Inflammatory Drugs (NSAIDs), steroids and castration have been tried with transient benefit [4,7].

Even though local control is seldom achieved by initial surgery, the prognosis of this disease remains controversial. Microscopic positive margins may be followed with observation

alone and re-excision reserved for grossly residual disease or tumors progressing on follow-up. However, there are no factors which will predict how the residual tumor will behave, making close observation of grossly residual disease an option. The lack of clarity in the management of grossly residual disease stems from the lack of high level evidence and limited series available to reach a solid conclusion on post-operative radiation [12]. We recommend a multidisciplinary approach towards this benign, locally aggressive, yet curable disease comprising of a radiologist (to mark out the tumor extent), a pathologist (for a definitive diagnosis on a FNAC/incisional biopsy, tumor margins), surgeon (for a tumor free margin resection), a plastic surgeon (for reconstruction, if there is a large area of tissue loss) and a radiotherapist (for postoperative radiation), if indicated.

Conflicts of interest: The authors declare that they have no conflicts of interest in relation to this article.

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