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Review

Desmoid tumors in neurosurgery: A review of the literature☆

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ABSTRACT

Desmoid tumors (DTs) are rare myofibroblastic neoplasms, which are mostly sporadic, but sometimes associated with familial adenomatous polyposis syndrome. Neurosurgical cases of DT have been very scarce. We review the literature concerning neurosurgical DTs and describe the first case of a cicatricial DT after the resection of vestibular schwannoma, presenting as a painful swelling in the retrosigmoid scar.

Contrary to other localizations in the body, standard-of-care wide margin resection cannot be performed in intracranial and spinal DTs. Therefore, maximally safe resection followed by radiotherapy when tumor margins are not free can be proposed as a treatment strategy in neurosurgical DTs.

Contents

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1. Introduction

Desmoid tumors (DTs), also called aggressive fibromatosis or desmoid-type fibromatosis, are histologically benign myofibroblastic neoplasms that exhibit slowly infiltrative growth. DTs do not metastasize but can be locally invasive and often recur after surgical excision [8,11,55]. It was recently shown that DTs arise from mesenchymal stem cells [68]. They are very rare, accounting for approximately 0.03% of all neoplasms and have an incidence of 2–4 per million per year [11,52]. The first DT description was made by MacFarlane in 1832 [31].

The majority of the cases are sporadic. Sporadic DTs appear to be three times more frequent in women than in men with a peak in the third and fourth decade [39,52]. The most frequent locations of sporadic DTs are the extremities, trunk musculature, head and neck and the abdominal cavity [1,52]. They mostly present as painless progressive swellings [39]. Besides these sporadic cases, there is group of DT with a clear association with familial adenomatous polyposis (FAP) or Gardner's syndrome and other adenomatosis polyposis coli (APC) gene mutations [66]. Up to 29 percent of the FAP patients harbor DTs, a frequency 850 times greater than the general population [31]. Most FAP-associated DTs arise in the small bowel or the mesentery and cause more mortality, morbidity and recurrence when compared to sporadic DTs [38].

Based on the female predominance, an association with sex hormones is presumed. The direct relationship of the growth rate to the level of endogenous estrogen in the female patients and the demonstration of significant amounts of estradiol but not progesterone receptors in the tumor cytosol suggest that the growth rate of DT is regulated by steroid sex hormones [21]. Anti-estrogenic treatment was effective in about half of the cases in which it was applied [67]. Some cases, particularly with abdominal wall localization, are thought to be associated with local myofibroblastic trauma such as during pregnancy [2,21] or after traffic accidents [4,41]. Exceptionally, cases of DT have been found in surgical scars, especially after mammoplasty [35].

The current standard of care for initial management is surgical removal with wide margins [55]. If for functional or cosmetic reasons these tumor-free margins cannot be obtained, adjuvant radiotherapy decreases recurrence rates according to most studies [1,23,42]. It has been suggested that the outcome of radiotherapy alone is equal to that of combined surgery and radiosurgery, though this remains unclear [15,25]. Pharmaceutical therapy including chemotherapy is controversial [31].

In the neurosurgical literature, DTs are only anecdotally described. Firstly, there are a few, mainly pediatric, case reports of primary intracranial DTs [64]. Next, sometimes a neurosurgical approach is needed when primary head and neck DTs affect the skull base and secondarily grow intracranially [64]. Furthermore, DTs can compromise the brachial or lumbosacral plexus [13], peripheral nerves [59], or the spine and/or spinal cord [55]. Finally, a few authors reported on DTs arising in neurosurgical scars [26].

We describe a case of cicatricial DT encountered in the retrosigmoid scar after vestibular schwannoma resection and review the literature on neurosurgical DTs.

2. Methods

The described patient has given written consent for submission of the case report to the journal. On November 1st 2014 we searched the MEDLINE database for "desmoid tumor" (total of 2112 articles), "desmoid-type fibromatosis" (total of 1555 articles) and for "aggressive fibromatosis" (total of 1502 articles). We further selected cases within the scope of neurosurgery.

2.1. Case

In January 2006, brain Magnetic Resonance Imaging [MRI] revealed a left-sided vestibular schwannoma in a 56-years old female with tinnitus and vertigo. Due to tumor growth on serial imaging, radiosurgical (Gamma Knife, Elekta, Stockholm, Sweden) treatment was performed in March 2007, complicated by a transient left facial nerve palsy. Following further tumor volume increase (Fig. 1a), a surgical resection via retrosigmoid approach was performed in October 2009. More than 2 years later, in January 2012, because of a painful progressive retro-auricular swelling, revision surgery was performed demonstrating a dense layer of yellowish tissue overlying the dura. No pathological specimen was retrieved.

Since the retro-auricular swelling recurred soon, she was referred to our hospital in August 2012. On clinical examination there was a firm swelling behind the left ear, about 4 cm in diameter, with a normal appearance of the overlying skin and the scar (Fig. 1d), painful at palpation. No neurological deficits were found on clinical examination.

Ultrasound imaging showed a hyporeflective and inhomogeneously vascularized lesion with no clear margins. Fine needle puncture biopsy was inconclusive. On computed tomography there was no bony infiltration nor erosion. MRI demonstrated an ovular and lobulated structure between the sternocleidomastoid and paraspinal muscles and the craniotomy cavity, T2-hypointense and slightly T1-hyperintense, with important but inhomogeneous contrast enhancement and measuring 40 mm × 50 mm × 50 mm (Fig. 1b and c).

In November 2012, the retrosigmoid scar was reopened and a well-encapsulated tumor was found. Peritumoral dissection along the skin and the cervical muscles unto the craniotomy margins was relatively easy. Upon further dissection at the craniotomy site, we noted an important infiltration of the dura, which could be readily dissected from the underlying and indurated arachnoid. Macroscopically, we performed an apparently complete resection.

Pathological examination demonstrated bundles of buckled fusiform cells, moderately densely embedded in a collagenous matrix and surrounded by multiple vessels. No clear cellular atypia, increase of mitosis or necrosis was noted (Fig. 1e). Tumor cells expressed alpha-smooth muscle actin and beta-catenin, mainly cytoplasmatic, but also nuclear (Fig. 1f). The tumor margins were not tumor free. The pathological diagnosis of a DT was made.

Because of the positive tumor margins, adjuvant radiotherapy was performed (28 × 2 Gy) starting from December 2012. At last follow-up 2 years after tumor removal, there were no clinical or radiological signs of tumor recurrence.

3. Discussion

Neurosurgeons can grossly encounter DTs in six occasions.

3.1. Primary intracranial

Literature research showed very few intracranial cases of DT, all cases being pediatric (Table 1). Except for the peculiar case of Chung [3] with an APC mutation and a medulloblastoma followed by two intracranial and one spinal metachronous DTs, no patients were older than 3 years, suggesting a congenital origin. All were resected without recurrence, taking into account the limited length of follow-up.

Due to its rarity, intracranial DT is poorly recognized. Histopathological differentiation with other spindle cell lesions such as fibrosarcoma, reactive fibrosis, nodular fasciitis, fibrous