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Orbital Desmoid Tumor Complicated with Corneal Abscess: A Challenging Case

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Abstract **Case Report**

Desmoid tumor is a benign fibrous neoplasia arising from the musculoaponeurotic structures of the muscles. They are characterized by their infiltrative and locally aggressive behavior with tendency for recurrence. Orbital location is extremely rare, only a few separate cases have been reported in the literature and are mainly pediatric. In this article, we report the case of an adult patient affected with a locally aggressive desmoid tumor involving the orbit that was unsuccessfully treated with hormonal therapy then by chemotherapy which induced a hematological toxicity before undergoing the ultimate surgical removal followed by adjuvant radiotherapy. The aim of the article is to describe this unusual location for desmoid tumors and outline the challenges in the decision-making process and the therapeutic alternatives. The management of desmoid tumors is mainly based on the function preservation, but in case of symptomatic patients or aggressive course of the disease, the surgical approach must be considered.

Keywords: Desmoid tumor, orbit, corneal abscess, chemotherapy, exenteration, radiotherapy.

INTRODUCTION

Desmoid tumors, also known as aggressive fibromatosis, extra-abdominal desmoid, or well-differentiated non-metastasizing fibrosarcoma [1] is a rare, locally invasive, yet non metastasizing histologically benign fibrous neoplasm. It arises from connective tissue, fascial planes, and musculoaponeurotic structures of the muscles [2–5]. Although considered benign, desmoid tumors, DTs are often locally aggressive, they may infiltrate adjacent organs, muscles, blood vessels, nerves and bones, causing severe pain, significant impairments, disfigurement, and disease or treatment-related morbidity and mortality [3, 6, 7]. Their locally aggressive behavior and high recurrence rate makes them challenging to treat. Head and neck DTs represent 11-15% of extra-abdominal desmoids [8]; nevertheless, orbital involvement is rare and can be secondary to contiguous invasion of lesions originating from adjacent structures. Orbital DTs have been described in children, but only rare adult cases have been reported.

CASE REPORT

A 52 years old woman previously treated for type 2 diabetes and high blood pressure presented to the ophthalmology department with an 8 months history of right ocular pain and exophthalmos followed by progressive loss of visual acuity. The initial examination found a healthy patient with a mildly proptosed right eye with restricted eye movements in all positions and diplopia. A craniofacial MRI showed a neoplasm located in the medial portion of the orbit, entrapping the medial rectus and extending to the anterior ethmoidal cells (Figure 1). A surgical biopsy through internal paracanthal approach revealed the presence of fusiform cellular elements arranged in bundles, immersed in abundant, dense connective tissue with areas of fibrosis and low cell density. No atypical nuclear or mitotic figures were seen. On immunohistochemistry the cells stained with b-catenin and smooth muscle actin antibodies and were negative with anti CD34, desmin and S.100 protein antibodies. This profile was consistent with the diagnosis of a desmoid tumour. The patient underwent a medical treatment with tamoxifen for five years. After an initial regression the mass rapidly evolved causing a complete loss of vision and an important exophthalmos. The latest MRI revealed an increase of the mass volume which extended to the maxillary sinus and the optic nerve. The therapeutic strategy was switched to intravenous Vinorelbine in association with methotrexate. After the fourth dose the patient developed a bone marrow aplasia and a corneal

abscess which indicated the cessation of medical treatment and the patient was readdressed to our facility for surgical excision. Clinical examination found an altered patient with negative luminous perception in the right eye, exophthalmos, chemosis, corneal abscess and hypopyon (Figure 2). The patient underwent an exenteration of the orbital content under general anaesthesia (Figure 3). The initial diagnosis of orbital desmoid tumour was confirmed with both the lower and medial orbital walls being infiltrated. Negative tumoral margins could not be achieved, thus, the patient then received 56 Gy in radiation distributed over 28 sessions. Follow-up of the patient has been performed for 30 months with no signs of recurrence.

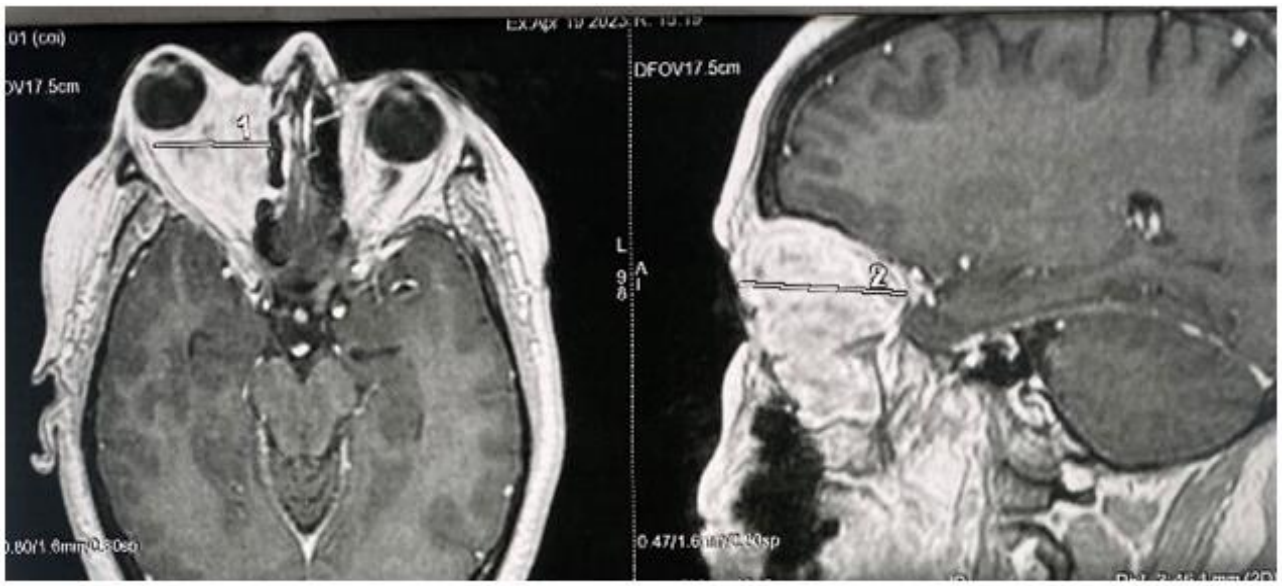

Figure 1: Facial MRI showing a neoplasm located in the medial portion of the orbit, entrapping the medial rectus and extending to the maxillary sinus

Figure 2: Photograph of the patient showing a proptosed right eye with chemosis and corneal abscess

Figure 3: Intraoperative photograph of the patient showing during the exenteration

DISCUSSION

Desmoid tumour is a rare neoplasm of unidentified aetiology that was first described by McFarlane in 1832 [5, 7]. They account for only 0.03% of all tumours and nearly 3% of all soft tissue neoplasms [9, 10]. The worldwide annual incidence is estimated at 3-6 cases per million [3, 6, 11, 12]. The majority of DTs are sporadic, with approximatively 50% arising in the abdominal region [13]. Head and neck DTs represent 8-15% of extra-abdominal DTs [13] and orbital involvement is even more exceptional. According to the World Health Organization (WHO), DT is a clonal fibroblastic proliferation that arises in the deep soft tissues and is characterized by infiltrative growth and a tendency toward local recurrence but an inability to metastasize [2]. It is considered as an intermediate locally aggressive fibroblastic tumor [14].

The exact physio pathological mechanism is unclear, however mutations in CTNNB1, the gene encoding for ß catenin are reported to being associated with 85% of sporadic DT [15]. These mutations lead to an excessive nuclear ß catenin accumulation which stimulates cell proliferation and differentiation [4, 15].

Clinical presentation of DT varies depending of the location, the volume and the degree of invasion, it ranges from asymptomatic to impairment or loss of function of a vital organ. The orbital location is exceptional, nineteen cases have been reported [5, 7, 8, 16–27] and only eleven were primary orbital tumors [5, 7, 16, 17, 19, 20, 22, 25–27], four of which are reported in adult patients [7, 19, 22, 26]. The clinical representation included a painless swelling [7, 16, 21, 26], painful orbital mass [5, 8], loss of visual acuity [5, 16–19, 23, 26], proptosis [8, 16, 18–20, 23, 25, 26], restricted ocular movements and diplopia [8, 18, 19, 21, 23], ptosis [7, 16, 17, 19], pale optic nerve [23, 25], hyperoptic shift [25], corneal abrasion [26], nasal bleeding [18] and nasal obstruction [23].

Reported clinical differential diagnosis of orbital DT included epidermoid cyst [7, 26], cellulitis [16], idiopathic inflammatory pseudotumor [19], rhabdomyosarcoma [23].

DTs are locally aggressive neoplasms that tend to infiltrate neighboring structures. MRI is the imaging modality of choice for diagnosis, local extent evaluation and for follow-up. CT can be more valuable in the analysis of bony erosion. Reported infiltrated structures include the maxillary sinus [5, 8, 21], ethmoid [18, 21], frontal sinus [18], sphenoid [24], temporal and infratemporal fossa [5, 8, 24], nasal cavity [23], anterior cranial fossa [23] and middle fossa [24].

Histologically the tumor is composed of homogeneous proliferation of mature fibroblasts arranged in spindle form of long fascicles and embedded within a variably dense collagenous stroma. The features suggestive of malignancy such as hyperchromatic nuclei, nuclear atypia, necrosis, mitotic activity and anaplasia are not seen. DT immunohistochemistry is characterized by nuclear ß -catenin positivity along with positivity for smooth muscle actin, vimentin, ß-cyclooxygenase-2 (COX-2), and frequently estrogen receptors beta, and by negativity for Desmin, S100, CD34, S-100 protein or cytokeratin [3, 4, 28, 29]. The tumor in our case tested positive for with ß catenin and smooth muscle actin antibodies and were negative for anti CD34, Desmine and S.100 protein antibodies. Up to 40% of DTs are still misdiagnosed in spite of these histological markers [12, 30].

Histological differential diagnosis for a cytologically bland spindle cell mass includes Gardner fibroma, desmoplastic fibroma, superficial fibromatosis, nodular fasciitis, myofibroma, collagenous fibroma, gastrointestinal stromal tumor, leiomyoma, hypertrophic scars and keloids and even fibrosarcoma, low grade fibromyxoid sarcoma and low-grade leiomyosarcoma [12, 31-33].

Up till this day the treatment of desmoid tumors hasn't been standardized. The choice of treatment modality may depend on extent/location of disease and institutional capabilities. Based on the clinical situation and patient preference, any of these treatment options may potentially be first- or second-line [34]. Surgery has been considered for years as the mainstay of DTs treatment [5]. However, radical surgery may result in significant function loss and mutilation. According to the NCCN guidelines surgery is only considered a first line treatment in restricted situations if agreed upon by a multidisciplinary tumor board [5, 12, 34]. Cryoablation, High-Intensity Focused Ultrasound and radiofrequency ablation have been accepted as less morbid than surgery and can be performed as primary or salvage therapy for desmoid tumors [4]. Other primary treatment modalities comprise radiotherapy, as primary treatment, it may be considered as a valid alternative to surgery and be used if medical therapies are not available or not active [11], it can also be used for lesions where recurrence would be technically challenging to resect and would lead to significant morbidity [34]. As adjuvant treatment, radiotherapy has been used in cases of positive surgical margins to decrease the local recurrence rate [5, 34] even though the level of evidence for adjuvant radiotherapy is low [11]. The recommended dose for radiotherapy is 50–56 Gy in daily fractions of 2 Gy [6]. Anti-hormonal agents such as tamoxifen or toremifene have been used alone or in association with NSAIDs as a first line therapy, and has showed considerable clinical benefits with low toxicity [10], it is even considered by some authors as the most effective noncytotoxic drug treatment in adults [33] although they are not mentioned in the NCCN recommendations [12]. Chemotherapy options include a "low-dose" regimen with methotrexate plus vinblastine or vinorelbine, or conventional dose chemotherapy using anthracycline based regimens as similarly instituted for the treatment of soft tissue sarcomas should be reserved for patients with rapidly growing and symptomatic unresectable or advanced tumors, for residual and progressive disease after initial local treatment, and, in rare cases, as induction treatment to facilitate adequate surgical resection [10, 11, 33]. Toxicity is primarily hematologic, which was the case with our patient. Other systemic therapy options include pegylated liposomal doxorubicin tyrosine kinase inhibitors (sorafenib, imatinib, pazopanib) [3, 11] with very promising results. More investigational therapies are under trial such as gamma-secretas inhibitors (Nirogacestat, AL101 and AL102), beta-catenin inhibitor Tegavivint, monoclonal antibodies (Nivolumab and Ipilimumab) and mTOR inhibitor Sirolimus [3, 4, 12]. As for the watch and wait policy, it is nowadays the most recommended as a first line approach by the NCCN and the Desmoid Tumor Working Group (DTWG) for managing tumors that are asymptomatic or mildly symptomatic and not progressing or in an anatomical location where progression would not be morbid [11, 34], however since the orbital location is associated with high morbidity and functional impairment risk this watchful approach is rarely recommended.

Treatment selection depends mainly on the aggressivity of the neoplasm and the need for a faster response to treatment. In the presented case, a watch-and-wait approach could not be adopted because the tumor was having a rapidly aggressive growth infiltrating the adjacent anatomical structures and causing loss of visual acuity. Tamoxifen was chosen as first line treatment but with no favorable results which indicated the switch for a more aggressive systemic approach using Vinblastine and Methotrexate which resulted in a medullar aplasia complicated by a corneal abscess. Surgical exenteration was then the last available resort associated with adjuvant radiotherapy since complete tumor-free margins could not be achieved.

Head and neck DTs are associated with a higher recurrence rate, they are estimated to recure in 25 to 80% of cases [20, 22], which indicates a long term surveillance. The NCCN recommends imaging with CT or MRI every 3–6 months for 2–3 y, then every 6–12 months thereafter (34). On the other hand, death is extremely exceptional, long-term survival is nearly 100%. Abdelkader and al reported a case of a patient with orbital DT who died from the extensive local destructive recurrence and bleeding [8].

CONCLUSION

Desmoid tumors are benign yet locally aggressive neoplasms. The orbital location is extremely exceptional and challenging to manage. Since this is a nonmalignant tumor with no risk of distant metastasis nor malignant transformation, function preservation and quality of life are considered fundamental goals to be reached in their management.

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Author Contributions

Conceptualization: Hamza Sobhi
Investigation: Badreddine Belmadani, Hamza Sobhi
Writing – original draft: Hamza Sobhi, Badreddine Belmadani

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Ethics approval and consent to participate

Our study is exempted from ethical approval. Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Consent for Publication

Written informed consent was obtained from the patient for publication of this case report and accompanying images.

Availability of data: Data can be obtained from the corresponding author upon reasonable request.

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