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Original article

Desmoid tumours of the head and neck in children: Review of management

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ABSTRACT

Objective: Desmoid tumours of the head and neck in children are rare, local invasive and potentially fatal tumours. The purpose of this review is to discuss the management of these tumours in the light of a case series and a review of the literature.
Material and methods: This retrospective study summarised the medical data of children treated for desmoid tumours of the head and neck between 1976 and 2014.
Results: Five of the 6 children were treated by radical surgical resection, with positive surgical margins (R1) in 2 cases, followed by recurrence requiring further resection. One child with a very advanced lesion was treated by weekly methotrexate and vinorelbine chemotherapy for 18 months, allowing 93% reduction of tumour volume without recurrence.
Conclusions: Desmoid tumours of the head and neck in children are more aggressive than their adult counterparts and are associated with high morbidity and mortality and a high recurrence rate. CT and MRI imaging assessment should preferably be performed before biopsy. External beam radiotherapy must be avoided in children as it is less effective than in adults, and is responsible for long-term cosmetic and functional sequelae and even a risk of second tumours. Treatment is surgical whenever radical resection is possible. In patients presenting an excessive risk of morbidity and mortality, chemotherapy devoid of long-term adverse effects (such as methotrexate in combination with a Vinca alkaloid) can be proposed. Long-term follow-up must be ensured due to the risk of recurrence.

1. Introduction

Desmoid tumours are fibroblastic and myofibroblastic tumours with intermediate malignancy (locally invasive with no metastatic potential) that, according to the World Health Organisation (WHO), arise from muscle and fascia of the deep soft tissues [1]. It is a rare disease with an annual incidence of 2.4 to 4.3 new cases per million inhabitants [2].

Desmoid tumours can occur at any age. They are ubiquitous, but can be divided into three groups of tumour sites: intra-abdominal and mesenteric (8%), abdominal wall (49%) and extra-abdominal (43%) [2,3].

Head and neck desmoid tumours represent 12% to 15% of all extra-abdominal desmoid tumours in adults [4,5] and are more commonly observed in children with an estimated incidence of between 22% and 35% of all extra-abdominal desmoid tumours [6–8].

Desmoid tumours are histologically benign tumours, with no metastatic potential, but with a high propensity to local invasion and recurrence [4,9], resulting in significant morbidity and occasionally death, particularly in the case of head and neck tumours. These tumours have also been demonstrated to be more aggressive in children [9–12].

No management guidelines are available for desmoid tumours of the head and neck in children, in whom the long-term adverse effects of surgical and medical treatment must be taken into account.

The objective of this study is to discuss the management of desmoid tumours of the head and neck in children based on a case series and a review of the literature.

2. Material and methods

This was a two-centre, retrospective, observational study including all children treated for desmoid tumour of the head and

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neck between 1976 and 2014 in the Paediatric Otorhinolaryngology department of Lille Regional University Hospital and the Paediatric Oncology department of Centre Oscar Lambret (cancer centre) in Lille.

Epidemiological, clinical, imaging and treatment data were collected from the patients' medical records.

First-line treatment was surgery whenever complete radical resection was possible. The status of surgical margins was described according to the International Union Against Cancer (UICC) R classification.

When radical surgery was not possible, systemic chemotherapy was administered in line with the European Paediatric Soft Tissue Sarcoma (EpSSG) 2005 guidelines for Non-Rhabdomyosarcoma Soft Tissue Sarcoma (NRSTR), combining methotrexate 30 mg/m^2 IV once weekly + vinorelbine 20 mg/m^2 IV once weekly [13].

Response evaluation after chemotherapy was based on 2D measurements according to the Response Evaluation Criteria in Solid Tumors (RECIST) criteria [14].

Follow-up was based on regular clinical and radiological evaluation. Recurrence was defined by reappearance of a tumour at least one month after stopping treatment.

3. Results

The study population comprised 6 children under the age of 15 years, with a male predominance (83%; 5 boys for 1 girl). The mean age at diagnosis was 5.5 years and the median age was 2.8 years (range: 6 days to 15 years).

The presenting symptom was isolated painless swelling in 4 of the 6 patients (66.7%). Associated symptoms were present in 2 cases (33.33%): dyspnoea or neuropathic pain with upper extremity paresis.

The lesion was located in the left medial canthus in one case, the submandibular region in two cases, the parapharyngeal space in one case, the neck and left shoulder in one case (Fig. 1), and the right external ear in one case.

All patients presented an isolated lesion with no multifocal lesions at the time of diagnosis.

Data for each patient are summarised in Table 1.

The diagnosis was confirmed by histological examination of surgical biopsies. Local extension was evaluated by contrast-enhanced computed tomography (CT) and/or magnetic resonance imaging (MRI).

No aetiological factor was identified with the exception of patient 3, who presented a neonatal desmoid tumour in whom the forehead presentation requiring delivery by caesarean section may have constituted a traumatic factor responsible for the tumour.

Five patients were treated surgically. Nerve and/or vascular invasion was observed in 5 patients, requiring sacrifice of the hypoglossal nerve (XII) and external carotid artery in 2 cases, and the marginal mandibular branch of the facial nerve (VII) in 1 case.

Histological examination of the operative specimen revealed microscopic invasion of the surgical margins (R1) in 2 patients.

With a mean follow-up of 80 months (range: 12 to 194 months), recurrence was observed in 2 patients after a mean interval of 19.5 months. In the first case (patient 1), the recurrence was situated in the right parapharyngeal space 35 months after surgery and was associated with a distinct second tumour situated in the base of the skull. The second patient (patient 4) presented 2 recurrences involving the external ear, 4 months and 24 months after the initial surgery. These two patients presented positive surgical margins (R1) on histological examination of the initial specimen. All cases of recurrence were treated by surgical resection.

The postoperative functional sequelae of nerve lesions in 2 patients were responsible for minimal symptoms. The main complaint concerned the scar or facial asymmetry. Secondary reconstruction of the external ear was performed after resection of most of the ear in patient 4.

One patient presented an extensive lesion not accessible to surgical resection at the time of diagnosis (patient 6). This lesion involved the mediastinum and retrotracheal space, and had invaded the common carotid artery, brachial plexus and recurrent laryngeal nerves (Fig. 1A and B). Chemotherapy (methotrexate + vinorelbine) was administered for 18 months. This patient experienced hepatic toxicity of methotrexate that resolved after stopping treatment. Chemotherapy achieved a partial response with 93% reduction of the tumour volume over 18 months (Fig. 1C and D). No recurrence was observed with a stable radiological appearance of the residual tumour volume 64 months after stopping treatment. A parasympathetic syndrome (sweating) of the left upper extremity was still present after more than 3 years of follow-up.

4. Discussion

Although the present series is one of the largest series of desmoid tumours of the head and neck in children yet to be reported, it presents a number of limitations: the small study population and the selection bias inherent to the retrospective nature of the study. However, it would be very difficult to conduct a prospective study in view of the very low incidence of this tumour. This retrospective study confirms the difficult management of desmoid tumours of the head and neck in children that can be life-threatening.

This series illustrates the young age at diagnosis (median age: 2.8 years), as also reported in the literature [4,6,7]. The head and neck is a predominant site of desmoid tumour in the paediatric population, as the study by Kruse based on 179 cases of desmoid tumour of the head and neck in patients of all ages revealed a mean age of 16.87 years, with 57.32% of patients under the age of 11 years [4]. Several radiological and clinical studies have suggested that desmoid tumours are more aggressive in children, with a more invasive appearance, more rapid growth, a higher recurrence rate, and a less favourable response to radiotherapy [9–11,15].

The leading clinical symptom is a painless swelling, sometimes associated with signs of local compression. The diagnosis can be suspected on MRI: low-intensity signal or intermediate signal (isointense to muscle) on T1-weighted images with marked gadolinium enhancement [15]. On T2-weighted images, desmoid tumours present an intermediate to high-intensity signal. Non-contrast-enhanced bands of low-intensity signal are fairly characteristic and correspond to the dense acellular collagen stroma. The tumour infiltrates adjacent tissues. These signs were observed in the patient with the largest tumour (Fig. 1).

Nevertheless, a biopsy must be performed to confirm the diagnosis. The histology of desmoid tumour has been extensively described [1,3]. Macroscopically, desmoid tumours present features of a non-encapsulated and poorly circumscribed scarred fibrous mass with invasive margins and adherent to adjacent tissues. Section of these tumours reveals a shiny greyish-white appearance. Histologically, they are characterized by a uniform and monoclonal proliferation of differentiated fibroblasts and myofibroblasts with low mitotic activity and a benign appearance with no nuclear atypias. These cells are surrounded and separated by a dense collagen stroma, forming intertwined bands in various directions. The tumour infiltrates adjacent tissues, inducing atrophy of adjacent striated muscle cell and the formation of multinuclear giant cells. Micro-haemorrhages are frequent, but invasion of nerves and blood vessels is never observed [4]. Although nerve and vessel invasion is not observed on histological examination,

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