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Case Report

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Low-grade fibromyxoid sarcoma of the oral cavity in an infant: The youngest patient in the world: A case report

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Abstract

Background: Low-grade fibromyxoid sarcoma (LGFMS) is a rare tumour in the head and neck. The literature reports less than 50 cases with different locations and variable ages from 22 months to 84 years. It can be considered a benign tumour, but potentially metastasizing and surgical excision is the primary treatment.

Methods: A 11-month-old girl presented with a history of neoformation of the buccal rim on the left. A bio exercises was performed, and a histological examination of the excised tumour revealed an LGFMS.

Results: The surgical excision revealed clear margins; no adjuvant treatments were necessary. A long-term follow-up was planned, and there was no recurrence or metastasis at five years.

Conclusion: This report describes the histopathological evaluation of an LGFMS of the oral cavity in the youngest patient in the world. A small lesion must be excised and biopsied in infants since it could reveal a rare tumour.

Keywords: c.

Introduction

Low-grade fibromyxoid sarcoma (LGFMS) is a rare tumour; it is a malignant variant of fibroblastic and myofibroblastic tumours according to the 2020 WHO classification of soft tissue tumours [1]. It usually occurs in the deep soft tissues of the trunk and extremities in young or middle-aged adults; the median age of patients ranges from 29 to 40 years [2], and less than 400 cases were reported in the literature. The disease was first described by Evans et al. in 1987 as a soft tissue tumour characterized by bland histologic features and a paradoxically aggressive clinical course [3]. Subsequent reports confirmed the existence of this entity of new sarcoma. However, they contrasted the innocuous histologic features and confirmed an aggressive clinical behaviour with recurrences noted in 15 of 22 (68%), metastases in 9 of 22 (41%), and death from disease in 4 of 22 (18%) patients with follow up [4-6]. According to Evans, metastasis can occur several years after the diagnosis; the median time was five years in a range from 0 to 42 years [2].

Due to the rarity of this tumour, a careful anatomopathological analysis is always necessary. The strongest cytoplasmic marker of LGFMS is MUC 4 [7], and immunohistochemically, the majority of cells are strongly positive for vimentin and generally negative for actin, desmin, S-100, protein, cytokeratin, CD34 and CD56 [8]. A recent study from Gjorgova Gjeorgjievski et al.

[9] can help in differential diagnosis, and this case report would also like to make an effort to the importance of pathologist to gain the correct diagnosis. Additionally, this paper increases the number of LGFMS in the head and neck region; nowadays, around 50 cases, less or more, were described with different locations and variable ages from 22 months to 84 years [10]. The treatment of choice is a wide surgical resection of the primary lesion within clear margins and eventually chemotherapy and radiotherapy if required [11,12].

We present one case of LGFMS of the oral cavity that was diagnosed and treated at our department in an infant of eleven months. This report aims to confirm histological and immunohistochemical aspects of LGFMS needful for diagnosis and differential diagnosis. Another purpose is to provide an example of clinical and radiological follow-up since no guidelines exist. Lastly, to the best of our knowledge, this is the youngest patient in the world affected by LGFMS.

Case report

Patient description

In February 2017 an 11 months-old female with a very small lesion (about 4 mm) of the left upper internal lip mucosa presented to the ENT outpatient clinic at Meyer Children's Hospital-IRCCS of Florence. The aspect was not suspicious of malignity because of its consistency and mobility. The neoformation was soft and jelly, translucent, free from the superficial and deep tissues miming a mucocele of a minor salivary gland. The superficial mucosa was normal and smooth. No pain was detected during palpation. The parents noticed the lesion, and it came to our attention. Even if the little child did not suffer from it, no feeding problems or pain episodes were reported. An excisional biopsy under general anaesthesia was proposed and performed.

Biopsy performance and results

The biopsy consisted of a complete removal of the lesion, the surgical specimen measuring about 1x1x0.4 cm. However, as the surgeon expected, the intraoperative aspect and consistency were not the usual ones of the mucocele. The lesion was hard in consistency and adherent to surrounding tissue, but intraoperatively no other lesions were detected or suspicious tissue. Despite the initial suspicion of benignity, the macroscopic aspect was already deposed for something malignant, and the final histological analysis revealed the correct diagnosis. The specimen was analyzed twice, firstly at the pathology Unit of Meyer's Children's Hospital and secondly at the pathology unit of the University of Padova, which was an Italian referral department for sarcoma and its varieties.

The specimen was routinely fixed in neutral buffered formol and paraffin-embedded. Five-micrometer sections were stained with hematoxylin and eosin. In contrast, additional sections were mounted on electrostatic slides and used for immunohistochemical studies using the standard streptavidin-biotin technique and commercially available antibodies.

The final histology described a lesion composed of bland spindle cells, showing a biphasic pattern with fibrous hypocellular heavily collagenized areas and more cellular myxoid nodules, focal fascicular growth pattern and occasional arcades of small blood vessels were seen. The immunohistochemical testing showed positive immuno-staining for MUC-4 and vimentin, negativity for S-100, keratins, SMA and desmin and weakly positivity for CD34. The proliferative index, determined to estimate the percentage of the Ki-67 positive neoplastic cells in the total of the tumour cells, was about 15-20%.

Morphologic features and immunohistochemical results were consistent with the diagnosis of LGFMS.

The margins were reported free from the tumour with a distance of 5 mm along the circumference of the entire specimen.

According to surgeon experience, histological examination, and literature reported about LGFMS, the case was also discussed with the pediatric oncologist, and the surgical excision was considered enough as primary treatment. Based on this decision, a clinical and radiological follow-up was planned until February 2022.

Follow-up

According to a pediatric oncologist and radiologist of Meyer Children's Hospital IRCCS head and neck magnetic resonance imaging (MRI) and chest and abdomen computed tomography (CT) were performed to exclude locoregional and distant metastasis. The only lesion detected was an hepatic cist, unrelated to LGFMS, followed by abdomen ultrasonography (US) every twelve months for the first two years. During the third year of follow-up, new chest and abdomen CT and head and neck MRI were performed, and at the successive annual control, an abdomen US was taken. In the fifth year, the head, neck, chest and abdomen MRIs were still negative. The clinical evaluations were repeated every six months for the first two years and annually in the last three years; no lesions were detected in the head and neck through the physical examination completed with fibre optics.

Discussion

LGFMS is a rare tumour in the head and neck region. The most recent literature review takes into account 45 cases of localization in the head and neck; among them, the oral cavity represents only 5 cases (2 cheek, 1 hard palate, 1 tongue and 1 buccal mucosa) and a case series of 15 patients in which only one has an LGFMS of the oral cavity (1 lower lip) [9,10]. Apart from the patient with the localization in the hard palate, the interesting thing is that all other patients were younger than eighteen. However, nobody was younger than one year old, so our patient was still the youngest patient reported.

The pediatric population can be affected by this variant of sarcoma, and an increasing number of studies reported LGFMS in children [5,6,11,13,14]. The sites of origin differ from the most known LGFMS, which arise in the deep tissues of the trunk and extremities; in children, the superficial tissues like the subcutis and dermis are preferred locations, and a less aggressive behaviour characterizes it. Specifically, Billings et al. revealed that superficial LGFMS was more common than previously recognized, may affect children at a higher rate (37%) than that deep LGFMS, and that metastasis occurred less frequently within the superficial tumours; this last consideration was also reported by Folpe et al. [11,12]. The most common sites of metastasis were the lung, pleura and chest, followed by bone and liver [2]. Since this tumour could recur up to 15 years after the

end of therapy, a long-term follow-up was recommended. Our patient had no recurrences or metastasis in the first five years of follow-up. However, she will be followed until she is 18 years old in our department; this behaviour agrees with that reported by Evans et al. [2].

Regarding the follow-up, it represented an essential issue in managing these patients. Radiological exposure and radiation-induced tumours constitute a problem, especially for children. However, radiological examinations are the only way to detect a recurrence or distant metastasis. For this purpose, a paper by Sargar et al. debates CT and MRI imaging in LGFMS used for diagnosis [15], but most of the case report and review data about follow-up is not available. In this report from a Children's Oncology Group published in 2015, the MRI characteristics of LGFMS were defined: a complex solid-cystic tumour with fibrotic tissue that appeared hypointense to muscle on T1 and T2-weighted images and exhibited a variable degree of enhancement but also these authors did not delineate the regular radiological follow-up [15]. Unlikely, we did not have any pre-operative imaging. However, all MRIs performed after the surgery were negative for recurrences or new lesions.

Concerning the LGFMS clinical features, they were very specific. They mainly showed slow-growing, painless single masses with a hard texture, easily misdiagnosed as benign tumours, like in our case. The correct diagnosis was anatomopathological; however, at the histological examination, several lesions had to be considered in differential diagnosis, including spindle cell lesions such as fibromatosis, nodular fasciitis, collagenous fibroma, benign nerve sheath tumours, collagenized neurofibroma, myxofibrosarcoma [9]. The immunohistochemical helped diagnose correctly since marker MUC4 was highly sensitive and specific [16]. Some cases of LGFMS with negativity for MUC4 were reported by Linos et al. A genetic study was performed in this case, and some typical chromosomal translocations confirmed the diagnosis [17]. In our case, the histological aspect of the lesion with the immunohistochemical analysis performed twice was needed for the correct diagnosis as reported above. Considering all these aspects, an expert pathologist should analyze the specimen, or it should be sent to a referred centre, in which histological, immunohistochemical and genetic analysis could be performed for a definite diagnosis.

This patient had let us not to discourage the resection of a lesion even if the aspect is similar to benign ones and the child is very young. The unluckily malignant lesions could also affect infants, and a biopsy of neoformation had to be done for the correct diagnosis. According to the size of the lesion, it could be performed pre-operative imaging, since some pathologies have radiological pathognomonic findings or in order to plan the best curative treatment.

Conclusion

In conclusion, LGFMS affect pediatric patients also in the head and neck; the oral cavity represents a possible localization, and for this reason, the lesions observed in oral mucosa, even if a benign origin was suspected, need to be analyzed. Although this tumour has the possibility of distant metastasis, this patient may have a favourable prognosis with long-term close follow-up because the tumour was solitary, well capsulated, confined to the localized area and showed a low rate of mitosis at histology. Our case shows that early surgical resection with negative margins will produce a fair outcome in terms of survival.

Declarations

Data availability statement: The co-author will provide data if requested.

Ethical approval: All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Informed consent: Informed consent was obtained from the parents.

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Conflicts of interest: The authors state that they have no conflicts of interest.

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