Aggressive Infantile (Desmoid-type) Fibromatosis of the Maxilla

A Case Report and New Classification

C Ogunsalu, S Barclay

ABSTRACT

This paper describes the clinical, radiographic and histologic findings of an aggressive infantile (desmoid-type) fibromatosis of the face in a seven-year-old black Jamaican male. This condition is rare in the head and neck region and its occurrence in the maxilla is exceptional. The differential diagnosis, management and long term follow-up of this case are also mentioned. The need for a less aggressive surgical management in this child and long-term follow–up is stressed. Also, its occurrence in someone of African descent has not been reported previously. The absence of recurrence, eight years after surgery is significant. This paper discusses the differential diagnosis and treatment of aggressive infantile fibromatosis and suggests a classification of the condition.

INTRODUCTION

Aggressive fibromatosis is a non-metastasizing tumour-like fibroblastic growth of unknown pathogenesis involving voluntary muscle as well as aponeurotic and facial structures. Histologically, it is indistinguishable from an abdominal fibromatosis. The lesion has a strong tendency for local recurrence and aggressive infiltrating growth. It is most common in the shoulder girdle, the thigh and gluteal region of growing adults (1).

Aggressive fibromatosis of the oral or para-oral structures is a very uncommon finding. Melrose and Abram (2) reported three cases involving the jaws of children. They discussed the protean nature of this lesion. Our case presented as a rapidly enlarging lesion post-trauma, although some cases may be quite slow in growth; pain may or may not be a finding. Most cases of aggressive infantile fibromatosis were clinically misdiagnosed as fibroma, fibrous histocytoma, granuloma, cyst, ameloblastoma, fibrous dysplasia or sarcoma. Sarcoma was the initial diagnosis in this case; however, the histopathological confirmation as aggressive fibromatosis was prompt and management was appropriate.

It is said that some childhood lesions proliferate very rapidly and histologically can be very cellular and active so that a diagnosis of sarcoma is seriously entertained and radical treatment carried out. Occasionally, the histomorphology may fail to reflect the biological behaviour of the lesions which can occasionally be localized, such as myofibromatosis, and as such be amenable to conservative surgery. On the other hand, some childhood fibrous lesions such as infantile fibromatosis may grow in a persistent and infiltrative manner. These lesions continue to present a difficult diagnostic problem, especially to pathologists (3–5). The two lesions commonly confused with infantile fibromatosis are myofibromatosis and fibrosarcoma. It is very important to distinguish between these conditions as they may display quite different clinical behaviour. The distinction between infantile fibromatosis and fibrosarcoma is a problem for pathologists as well as being of academic interest. Fortunately, the infantile fibrosarcoma may display attenuated behaviour compared to its adult counterpart. They have reduced metastatic potential but may recur locally much like the fibromatosis (5). It is claimed that the treatment for both should be complete excision.

Case Report

A seven-year-old black Jamaican boy presented to us on February 11, 1995, with a painless firm swelling of the left cheek and infra-orbital region. The swelling was not attached to the skin but clinically appeared to be bony hard. It measured about 2 cm in the longest diameter. The mother associated the occurrence of the lesion to a previous trauma to the face following an alleged assault. She claims that the lesion has increased rapidly in size over a short period.

Radiographic Findings

Radiologically, the occipitomental view (standard) showed an extensive radiopaque lesion of the left zygomatic bone, extending up to the region of the zygomatico-frontal suture line (Fig. 1). No bony destruction of the orbital or antral wall was noted. On initial examination, a diagnosis of facial osteoma was made; however, malignant tumours such as osteosarcoma and fibrosarcoma were also considered based on the history of recent onset of swelling and rapid growth thereafter.

Treatment – excisional biopsy

The treatment was that of a total excision of the lesion – with nibbling of bone at the base in all areas. At surgery, the lesion was found to consist of soft (fibrous) tissue essentially

From: School of Dentistry, Faculty of Medical Sciences, The University of the West Indies, St Augustine, Trinidad and Tobago, West Indies.

Correspondence: Dr C Ogunsalu, School of Dentistry, Faculty of Medical Sciences , The University of the West Indies, St Augustine, Trinidad and Tobago, West Indies. Fax: (868) 645-3823, e-mail: chrisogun@yahoo.com

and not osseous tissue as was suggested by the clinical and radiological examination. All the tissues obtained at surgery were submitted for histopathology.

Sections of tumour, or histology, showed a diffuse growth of mature appearing fibroblasts arranged in distinct bundles and fascicles associated with variable amount of collagen. It appeared that the growth began in relation to fascia or periostium and small foci of ossification were noted at the periphery (Fig. 2 a & b). Also vascular spaces are seen. The overall picture is that of infantile (desmoid-type) fibromatosis.

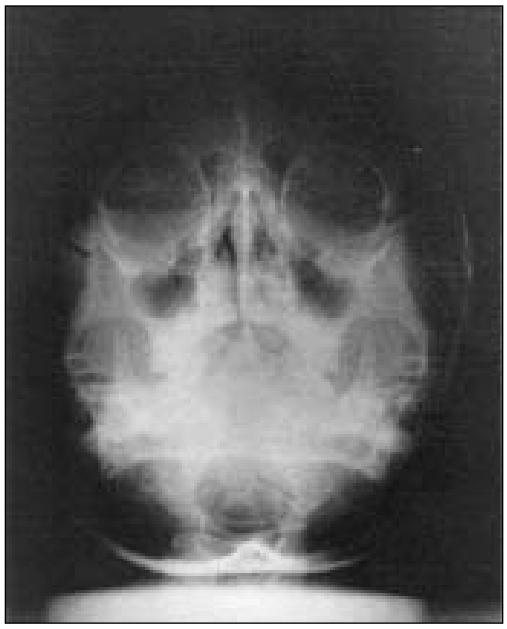

Fig. 1: Occipitomental radiograph showing radiopaque lesion of the left zygomatic bone (see arrow)

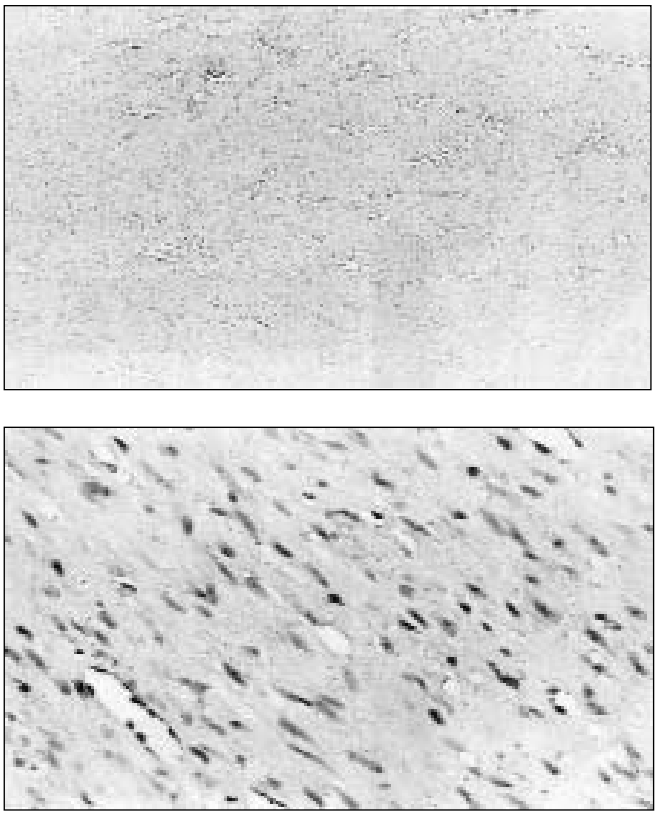

Fig. 2a & b: Low power x 85 and high power x 200, respectively, showing mature fibroblast arranged in distinct bundles and fascicles associated with variable amount of collagen

DISCUSSION

Aggressive fibromatosis belongs to a sizeable group of the so-called "miscellaneous locally aggressive fibrous lesions". They are non-metastasizing and must always be differentiated from fibrosarcoma, particularly the well-differentiated type. In the past, many of these benign but locally aggressive lesions have been confused with sarcoma and it is only in recent years that the pathologists have been able to separate these lesions with any assurance (1).

The group consists chiefly of the following: nodular fasciitis (psuedosarcomatous fibromatosis); aggressive fibromatosis (extra-abdominal desmoid); proliferative myositis; fibrous histocytoma (fibroxanthoma); atypical fibroxanthoma (and malignant variant) and desmoplastic fibroma of the bone.

It is unclear whether or not trauma played a role in the aetiology of this case, or whether it was co-incidental. All these lesions are quite uncommon in the oral cavity. Aggressive infantile fibromatosis may cause destruction of the bone when in apposition with such. Fortunately, this lesion did not destroy the underlying bone.

The microscopic appearance of these lesions is quite uniform, however, consisting of cellular interlacing bundles of elongated fibroblasts showing no pleomorphism, little or no mitotic activity and no giant cells (1). Typically, it shows numerous slit-like vascular spaces not associated with inflammation.

The treatment of this patient was that of complete surgical excision with generous margin, without any residual facial disfiguration even though the diagnosis had not been confirmed at the time of surgery. There has been no evidence of recurrence, after eight years but we recommended long-term follow-up.

Clinically and radiologically, it is almost impossible to make a diagnosis of infantile fibromatosis, a condition which may or may not be aggressive. When aggressive, a diagnosis of malignant neoplasm is usually suggested until the result of histopathology is available. Interesting but quite disturbing is the widely accepted fact that it can be difficult to differentiate it from low-grade fibrosarcoma and as such eventuate in mismanagement of the cases.

The consideration of the clinical course, radiological finding and histopathology finding can assist in making a

Table 1: Summary of literature review for cases of aggressive infantile fibromatosis – (1974 – 2000)

Case No.	Author	Year	Age	Gender	Anatomic area central = c, peripheral = p, no precise detail =?	Preliminary diagnosis	Time of diagnosis	Radiographic findings
1	Pindborg and Hjrting-Hanson	1974	3 years	Male	Left inferior border of the orbit	-	-	Radiolucency
2	Wilkins *et al*	1975	3 years		Anterior palate?	-	-	Radiolucency
3	Melrose and Abram	1980	5 years	Female	Palate, right maxillary ridge (p)	-	1 month	Loss of interdental bone
4	Melrose and Abram	1980	3 years	Female	Mandible and floor of mouth	-	-	-
5	Bertrand *et al*	1981	9 ½ years	Female	Right maxilla (?)	-	6 months	-
6	Goopfert *et al*	1982	2 years and 9 months	Male	Right upper gingival (p)	-	-	-
7	Tagwa *et al*	1989	3 years	Female	Submandibular region	-	-	-
8	Carr *et al*	1992	2 years	Male	Left mandible (c)	-	2 years	Radiolucency
9	Carr *et al*	1992	2 years	Male	Left mandible (c)	-	1 year	Radiolucency
10	Ramanathan and Thomas	1997	7 years	Female	Parotid gland	-	-	-
11	Sato *et al*	2000	3 years	Male	Mandible (c)	Malignancy	-	
12	Donohue *et al*	1990	14 years		Maxilla (left palate) (c)	Fibroma	-	
13	Ogunsalu and Barclay	2003	7 years	Male	Left cheek and infraorbital region	Fibrosarcoma and osteosarcoma	-	Radiopacification

definitive diagnosis. Unlike fibrosarcoma, the aggressive infantile fibromatoses never metastasize (3), although they have a potential to produce fatal outcome from extension into vital organs.

In the early 1950s, Stout (4) reported on the state of confusion around the term fibromatosis and noted that there was nothing to gain by retaining the older names such as non-metastasizing fibrosarcoma.

A review of 241 cases of juvenile fibromatosis by Stout (5) distinguished it from fibrosarcoma by identifying features that would be indicative of potentially metastatic behaviour. The description 'aggressive' is based on the invasive characteristic of the lesion and the rate of growth. It is our opinion that, to avoid further confusion, other synonyms such as extra-abdominal desmoid, juvenile fibromatosis, congenital fibrosarcoma should be discontinued and the term aggressive or non-aggressive infantile fibromatosis retained based on the rate of growth and clinical course with or without treatment, as it is documented that this tumour has been known to regress without any form of treatment.

Bridget *et al* (6) observed chromosomal abnormalities in their analysis of 26 cases of desmoid tumour (6). We suggest that trauma, such as birth trauma and childhood trauma, is probably necessary to initiate the condition in those with genetic predisposition. It is for this reason that we suggest that a complete paediatric history be an important aspect in the investigation of these patients. None of the published cases indicated that paediatric history including perinatal and postnatal history was taken. We reviewed 12 published cases of aggressive infantile fibromatosis (7–15) of the jaws as shown in Table 1. The male: female ratio was 1:1, with an average age of 4.94. The age range was 2 – 9.5 years. The maxilla was more involved (nine cases) than the mandible (three cases). The radiographic finding was that of radiolucencies of the affected jaw mainly. However, the index case herein reported presented with radiopacification of the maxilla, a reason for favouring the diagnosis of osteogenic sarcoma initially.

In their classical paper of 1992, Carr *et al* (12) described the clinicopathological finding of two unusual cases of infantile fibromatosis of the mandible of two-year-old children. These tumours, though highly aggressive initially, underwent spontaneous regression in the absence of definitive treatment. These authors avoided the term aggressive in the title of their classical article. In the light of our current clarification of terms, they should have called these two cases atypical aggressive infantile (desmoid-type) fibromatosis. Further, we suggest that if the term atypical or non-aggressive is not found desirable, then aggressive infantile fibromatosis should be retained and classified as

grades A, B, C and D: grade A – clinically aggressive, histologically confirmed tumour and clinically recurrent after aggressive surgical treatment; grade B – clinically aggressive, histologically confirmed tumour and clinically non-recurrent after aggressive surgical treatment; grade C – clinically aggressive, histologically confirmed tumour and clinically non-recurrent after non-aggressive surgical treatment; grade D – clinically aggressive, histologically confirmed tumour and clinically regressive after no definitive surgical treatment.

This classification is as such retrospective and best done after at least two years post surgical follow-up. Table 2 shows the type of treatment and our new retrospective grading for aggressive infantile fibromatosis. From a review of the English-speaking literature, it is obvious that despite the variability in the clinico-pathologic behaviour and progression of all pathologies ascribed "aggressive infantile fibromatosis", no classification has been developed. It is for this reason that we favour our new classification.

Table 2: Showing type of treatment and new retrospective grading of aggressive infantile fibromatosis

Case	Type of Treatment	Recurrence	Grade
1	Aggressive surgical resection	none	**B**
2	Aggressive surgical resection	yes – after one month	**A**
3	Aggressive surgical resection	no	**B**
4	Not sure	none	**?**
5	Aggressive surgical resection	lost to follow-up	**?**
6	Not sure	none	**?**
7	Resection	none	**B**
8	No definite treatment	regression	**D**
9	No definite treatment	regression	**D**
10	Complete surgical excess of the patoid gland	none	**B**
11	Marginal mandibulectomy +reconstruction with iliac crest bone.	none	**B**
12	Excision of lesion	recurrence	**B**
13	Excision of lesion (conservative but total)	none	**C**

CONCLUSION

Aggressive infantile (desmoid-type) fibromatosis of the face may appear to be malignant in nature based on the clinical presentation and radiographic findings, particularly when it has caused destruction of bone. Pathologists are now able to separate this lesion from sarcomas with much greater certainty and assurance. Local excision is the treatment of choice with much emphasis on long-term follow-up. The clinical features and radiographic findings should at least point to the diagnosis of aggressive fibromatosis or low grade fibrosarcoma Subsequently, the absence of features that would be indicative of potentially metastatic behaviour should allow a definitive diagnosis of aggressive infantile fibromatosis to be made without the use of the previously utilized multitude of diagnostic terms. Finally, we suggest that the aetiology is likely trauma (perinatal, postnatal, or early childhood trauma) in a rare group of genetically predisposed patients.

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