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Localized Infantile Myofibromatosis

Report of a Case Originating in the Mandible

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Summary

A case of congenital solitary myofibromatosis occurring in the mandible is presented. Recognition of this entity is of clinical importance as the lesion can be easily confused with juvenile fibromatosis. In contrast to this latter disease which exhibits a locally aggressive growth pattern requiring radical surgery, myofibromatosis consists of well-circumscribed nodules amenable to simple excision. Moreover, myofibromatosis may show spontaneous regression. Histological features permitting the differentiation of myofibromatosis from juvenile fibromatosis are discussed.

Key-Words

Myofibromatosis – Fibrous jaw tumour – Fibromatosis – Infancy – Childhood

Introduction

Fibromatoses in children show a variable picture and can differ considerably from each other in behaviour. Multiple as well as solitary forms do occur (*Allen*, 1977). One of these childhood forms of fibromatosis is an entity known as congenital generalized fibromatosis (*Stout*, 1954). This is a disseminated disease process consisting of multiple nodular lesions composed of collagen-forming spindle cells which are widely distributed throughout the whole body. *Kauffman* and *Stout* (1965) divided congenital generalized fibromatosis into a variant in which the lesions were confined to the subcutis, muscle and skeleton for which they employed the designation multiple as opposed to the generalized form in which there is also visceral involvement. Occasionally, solitary forms of the disease have been reported (*Enzinger*, 1965; *Kindblom* et al., 1977) and this form turned out to be the most frequently occurring manifestation (*Chung* and *Enzinger*, 1981). As the involved cells display staining characteristics of fibroblasts as well as smooth muscle cells it has been proposed that the term infantile myofibromatosis should be preferred to congenital generalized fibromatosis (*Chung* and *Enzinger*, 1981).
Recognition of infantile myofibromatosis is of clinical importance as the lesion consists of well-circumscribed nodules that are amenable to conservative surgery, thus contrasting with other types of childhood fibromatoses that may behave in an aggressive manner that calls for more extensive surgical intervention.
We report a case with a solitary manifestation of infantile myofibromatosis in the lower jaw and will discuss the aspects of this disease that permit differentiation from other fibrous proliferations of childhood that occur in the oral cavity.

Case History

A newborn male infant was seen because of a congenital swelling of the anterior part of the left mandible. The patient was the first son of healthy parents. Pregnancy and delivery had been uneventful. At examination, a 2 cm. sized lump was seen originating from the anterior of the left mandible and bulging into the overlying buccal soft tissues. A corresponding extra-oral swelling was also evident. Radiographs showed that the lesion had caused loss of bone and displacement of tooth germs in the involved area of the jaw (Fig. 1).

After an incisional biopsy the lesion was removed with a margin of adjacent normal tissue. The patient has remained free of disease for 10 years.
Histologically, the biopsy as well as the surgical specimen showed spindle cells which were arranged in short curving fascicles (Fig. 2). The cells themselves were slightly elongated and contained blunt-ended nuclei with a wrinkled outline. There were also cells with tapering processes which, when stained with Masson's trichrome and PTAH staining methods, revealed the presence of intracytoplasmic longitudinal fibrils indicating a smooth muscle character (Fig. 3). The central part of the lesion was occupied by multiple slit-like vascular spaces and showed some resemblance to an haemangiopericytoma (Fig. 4). These lesional vessels were surrounded by round to polyhedral cells that merged with the adjacent spindle cells. Coagulation necrosis was also present in this central area (Fig. 5). Collagen fibres separated individual cells and cell clusters and focally, in relatively less cellular areas, a slightly hyaline ground substance was present. Some smaller nodules of

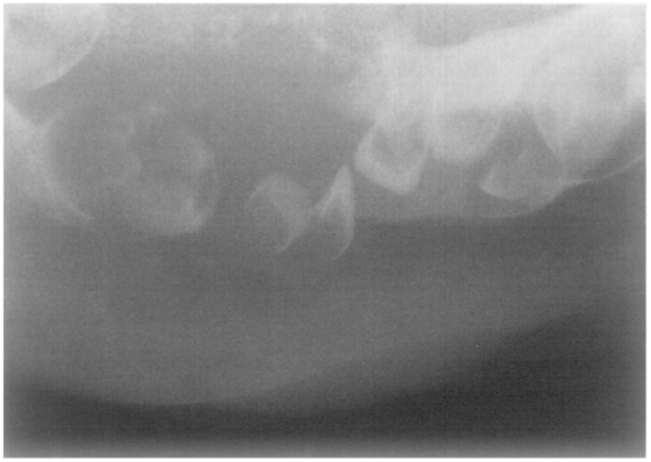

Fig. 1 Radiograph of anterior mandible. Although the X-ray is poor in contrast, loss of bone and displacement of tooth germs can be discerned.

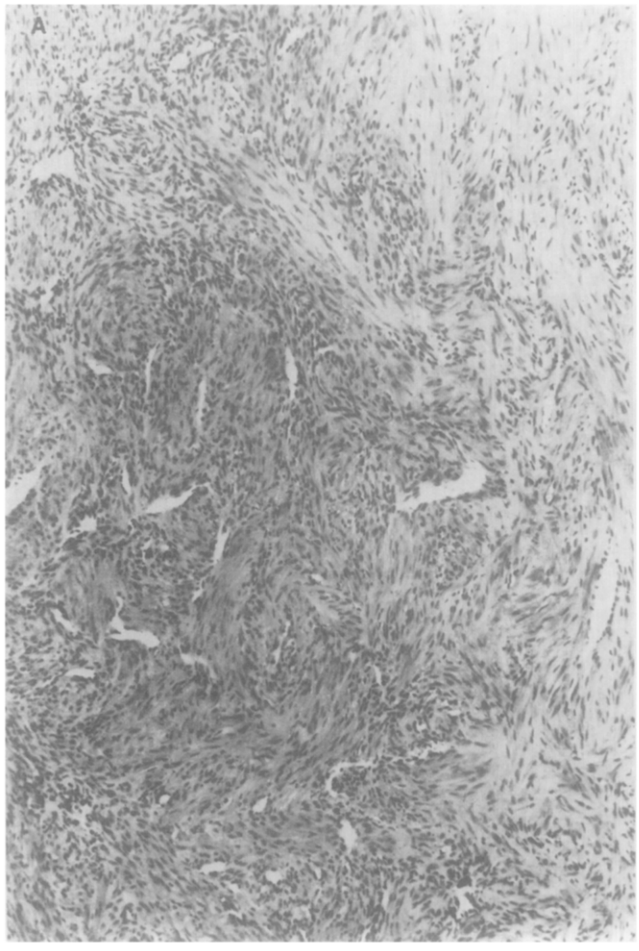

Fig. 2a Photomicrograph shows lesion composed of short, sharply bent fibrous fascicles with intervening slit-like vascular spaces (A).

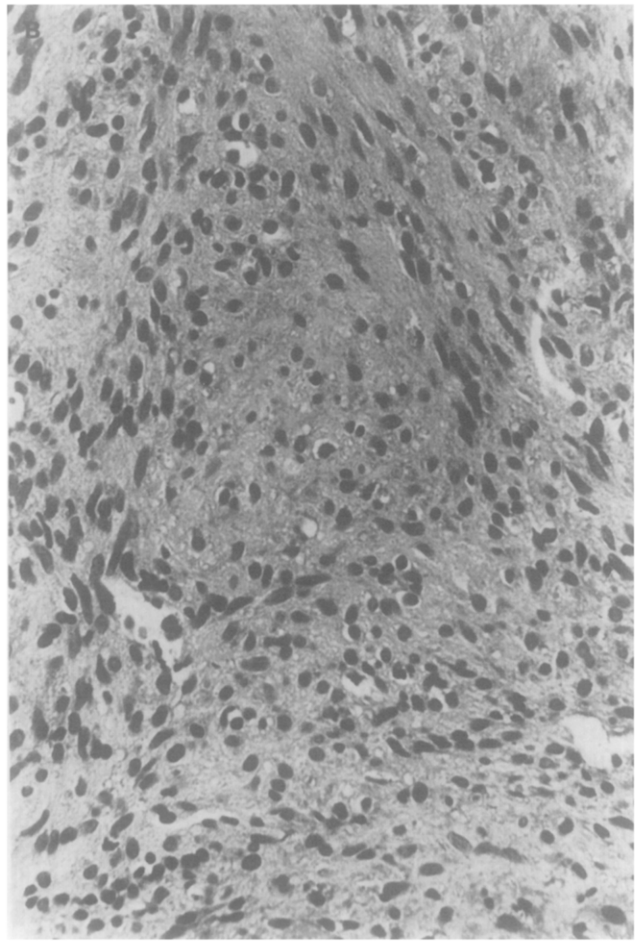

Fig. 2b Higher magnification shows the blunt-ended nuclei with wrinkled outline that are typical of smooth muscle cells (B). HE, A × 36, B × 150.

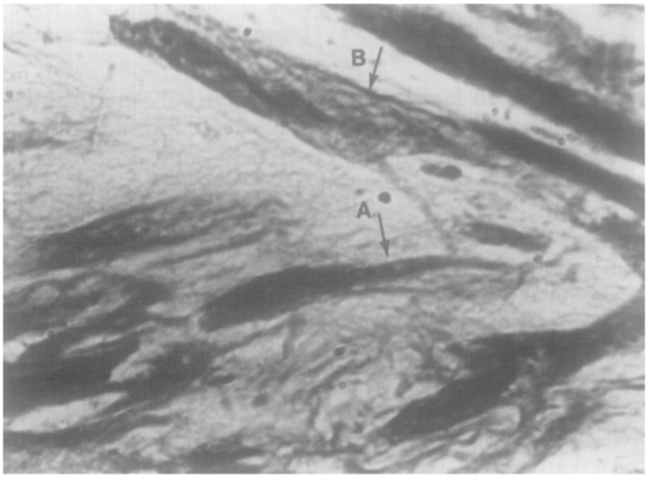

Fig. 3 High power view reveals tapering cytoplasmic extensions filled with PTAH-positive material (arrow A). In another cell, individual fibrils are visualized by this staining method (arrow B). PTAH, × 600.

identical histology were seen lying apart from the large nodule that made up the bulk of the lesion.

At that time a diagnosis of angioleiomyomatous hamartoma was made based on the presence of smooth muscle cells and vessels. When reviewing the sections later, the histological appearances were considered to be similar to the lesions that were described by *Chung* and *Enzinger* (1981) as typical of infantile myofibromatosis. As a result the original diagnosis was changed to congenital solitary myofibromatosis.

Discussion

Within the juvenile fibromatoses, various types are discerned that differ considerably with respect to behaviour and prognosis (*Enzinger*, 1965). *Chung* and *Enzinger* (1981) summarized the fibrous tumours of infancy and childhood (Table 1). Their classification conforms largely to that compiled by *Allen* (1977). Most of these entities are readily recognizable by their characteristic location and microscopic appearance. These include fibrous hamartoma of infancy, digital fibromatosis, fibromatosis colli, gingival fibromatosis, hyaline fibromatosis and calcifying aponeurotic fibroma.

In the head and neck region, diagnostic problems concentrate upon the differentiation of solitary infantile myofibromatosis from infantile or juvenile fibromatosis. Infantile or juvenile fibromatosis is a lesion that behaves aggressively as is demonstrated by an infiltrative growth pattern and

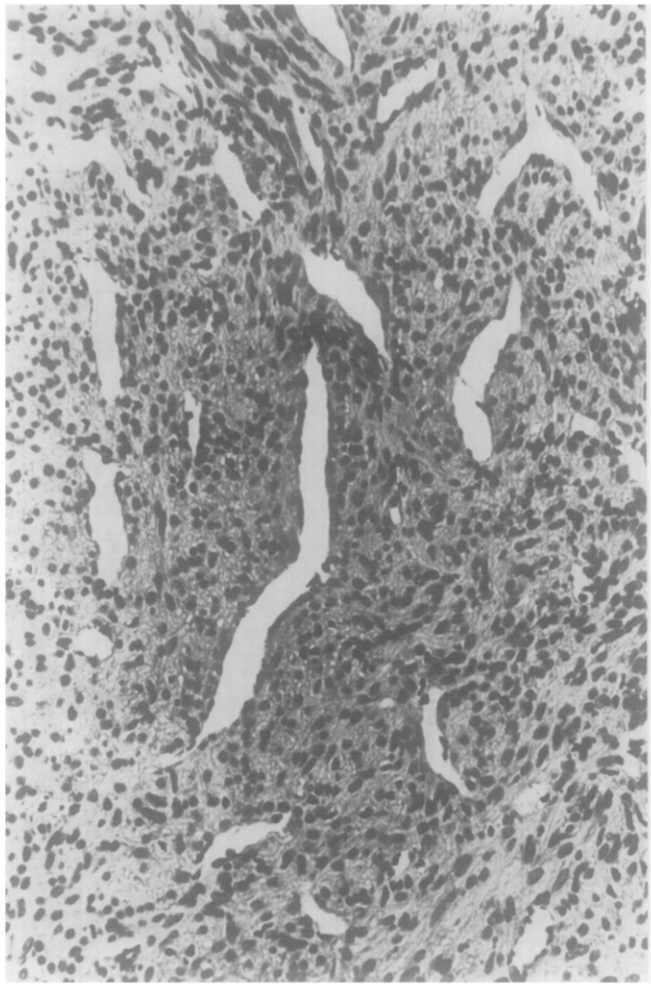

Fig. 4 Central area of the lesion mimicks an haemangiopericytoma. Vascular spaces are seen that are surrounded by cuboidal cells with dense hyperchromatic nuclei. HE, × 100.

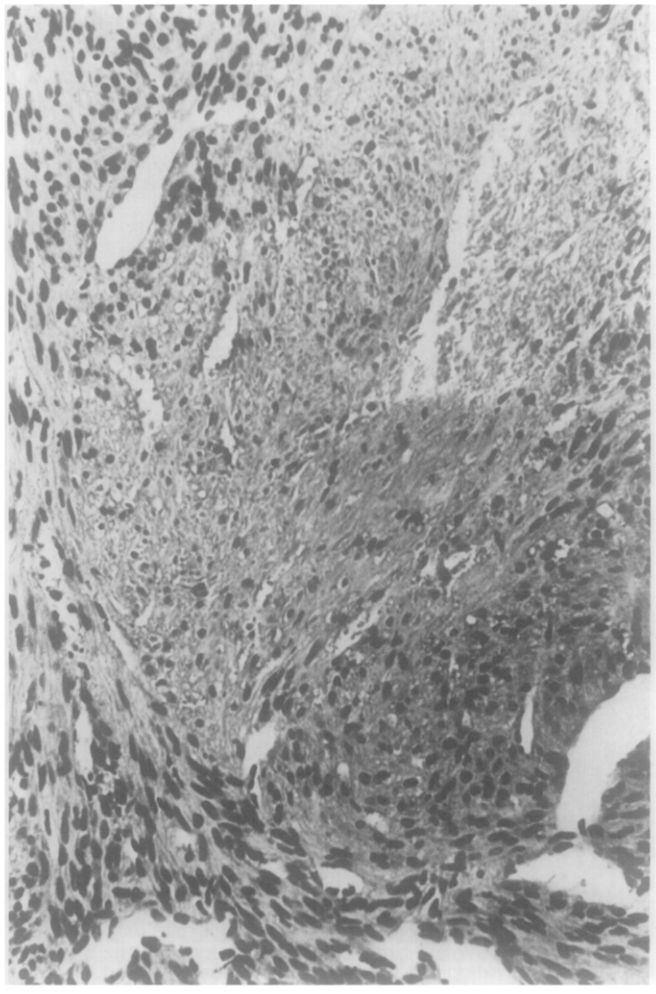

Fig. 5 Photomicrographs shows an area of coagulation necrosis that is rather sharply demarcated from the adjacent tumour tissue. HE, × 100.

recurrence after incomplete removal. Its occurrence in and around the oral cavity has been documented by several case studies which all stressed the necessity of adequate surgical intervention (*Takagi* and *Ishikawa*, 1973; *Larsson* and *Björlin*, 1976; *Henefer* et al., 1978; *Melrose* and *Abrams*, 1980; *Reihl-Ehlers*, 1980; *Reich*, 1981; *Rodu* et al., 1981). The differentiation of this type of fibromatosis from infantile fibrosarcoma may be a matter of concern for the pathologist but bears not much clinical significance as it is a well established fact that in infants fibrosarcomas display an attenuated biological potential with clinical problems arising through multiple recurrences and not through metastatic disease (*Stout*, 1962; *Dehner* and *Askin*, 1976; *Soule* and *Pritchard*, 1977). Thus it is obvious that treatment of juvenile fibromatosis as well as infantile fibrosarcoma should be directed entirely toward complete removal of the local lesion.

Table 1 Classification of fibrous tumours of infancy and childhood (*Chung* and *Enzinger*, 1981)

1. Fibrous hamartoma of infancy
2. Infantile digital fibromatosis
3. Fibromatosis colli
4. Infantile myofibromatosis
   a. solitary
   b. multicentric
5. Infantile (desmoid-type) fibromatosis
6. Hyaline fibromatosis
7. Gingival fibromatosis
8. Calcifying aponeurotic fibroma

The differentiation of infantile myofibromatosis from juvenile fibromatosis is, on the contrary, not an academic question. In contrast to the infiltrative behaviour of juvenile fibromatosis, infantile myofibromatosis manifests itself as well-circumscribed nodules that can be managed in a more conservative way. Death from infantile myofibromatosis is only seen through complications from visceral locations in the multicentric form. Moreover, spontaneous regression, particularly of the osseous lesions may occur as is reported by several authors (*Teng* et al., 1963; *Heiple* et al., 1972). When encountering infantile myofibromatosis in its multiple form, distinguishing this lesion from juvenile fibromatosis does not cause many problems as the latter lesion rarely occurs in a multicentric form. The solitary form of infantile myofibromatosis can be distinguished from

juvenile fibromatosis on histological grounds. The spindle cells in juvenile fibromatosis lack the smooth muscle features that are exhibited by the spindle cells in infantile myofibromatosis. These smooth muscle features consist of blunt-ended nuclei with wrinkled outlines, fusiform cellular contour and an eosinophilic cytoplasm in which PTAH or Masson's trichrome staining methods reveal the presence of myofilaments. Moreover, a vascular pattern mimicking an haemangiopericytoma may be present in some areas.

With respect to the intra-osseous location of infantile myofibromatosis, another differential diagnostic problem arises. In this site the lesion has to be differentiated from desmoplastic or odontogenic fibroma. Both lesions, however, also lack smooth muscle characteristics and with the possibility of infantile myofibromatosis in mind, establishing the exact nature of the lesion should not give rise to too many difficulties.

When returning to the present case, the histology was fully compatible with infantile myofibromatosis as delineated by *Chung* and *Enzinger* (1981). Lesional cells exhibiting fibroblastic as well as smooth muscle characteristics, vascular areas mimicking haemangiopericytoma and areas of coagulation necrosis are all features consistent with that diagnosis. As there were no other lesions, the disease manifested itself in its solitary form. Our unawareness about this entity when we encountered this patient resulted in treatment as advocated for aggressive fibromatosis, consisting of removal of the lesion with a margin of adjacent normal tissue. Our current understanding of it would have resulted in more limited treatment. Therefore, we thought it appropriate to bring this entity to the attention of the profession.

References

Allen, P. W.: The fibromatoses: A clinicopathologic classification based on 140 cases. Am. J. Surg. Path. 1 (1977) 305

Chung, E. B., F. M. Enzinger: Infantile myofibromatosis. Cancer 48 (1981) 1807

Dehner, L. P., F. B. Askin: Tumours of fibrous tissue origin in childhood. A clinicopathologic study of cutaneous and soft tissue neoplasms in 66 children. Cancer 38 (1976) 888

Enzinger, F. M.: Fibrous tumours of infancy. In: Tumours of Bone and Soft Tissue. Chicago: Year Book Medical Publishers, Inc. (1965) 375

Heiple, K. G., E. Perrin, M. Aikawa: Congenital generalized fibromatosis. A case limited to osseous lesions. J. Bone Jt. Surg. 54-A (1972) 663

Henefer, E. P., H. C. Bishop, A. Brown: Juvenile fibromatosis with invasion of the mandible: report of two cases. J. Oral. Surg. 36 (1978) 965

Kauffman, S. L., A. P. Stout: Congenital mesenchymal tumours. Cancer 18 (1965) 460

Kindblom, L. G., G. Termén, J. Säve-Söderbergh, L. Angervall: Congenital solitary fibromatosis of soft tissues, a variant of congenital generalized fibromatosis. Acta Path. Microbiol. Scand. 85-A (1977) 640

Larsson, A., G. Björlin: Aggressive fibrous lesions of the oral cavity. J. Oral. Path. 5 (1976) 241

Melrose, R. J., A. M. Abrams: Juvenile fibromatosis affecting the jaws. Report of three cases. Oral Surg. 49 (1980) 317

Reich, R. H.: Die lokalisierte kongenitale Fibromatose – ein Beitrag zur Differentialdiagnose maligner Tumoren im Kindesalter. Dtsch. Zahnärztl. Z. 36 (1981) 729

Reihl-Ehlers, B.: Zur juvenilen diffusen Fibromatose im Kiefer-Gesichtsbereich. Probleme der Klassifikation und Therapie. Dtsch. Z. Mund-, Kiefer-, Gesichts-Chir. 4 (1980) 54S

Rodu, B., D. R. Weathers, W. G. Campbell Jr.: Aggressive fibromatosis involving the paramandibular soft tissues. A study with the aid of electron microscopy. Oral. Surg. 52 (1981) 395

Soule, E. H., D. J. Pritchard: Fibrosarcoma in infants and children. A review of 110 cases. Cancer 40 (1977) 1711

Stout, A. P.: Juvenile fibromatoses. Cancer 7 (1954) 953

Stout, A. P.: Fibrosarcoma in infants and children. Cancer 15 (1962) 1028

Takagi, M., G. Ishikawa: Fibrous tumour of infancy. Report of a case originating in the oral cavity. J. Oral. Path. 2 (1973) 293

Teng, P., M. J. Warden, W. L. Cohn: Congenital generalized fibromatosis (renal and skeletal) with complete spontaneous regression. J. Pediatr. 62 (1963) 748

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