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Congenital infantile digital fibromatosis: a case report and review of the literature

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Abstract

Infantile digital fibromatosis (IDF) is a rare benign fibroproliferative tumor of early childhood. IDF preferentially affects the fingers and the toes. Malignant transformation or metastases have never been reported. Surgical treatment has been advocated previously but local recurrences were frequently observed. Recent literature supports clinical surveillance without any medical or surgical intervention as spontaneous regression usually occurs after two to three years. A six-month-old Caucasian girl with IDF on the left fourth digit is presented here. The tumor progressively increased in size after birth. Topical imiquimod cream and diflucortolone valerate cream, both displaying antifibrotic properties, had no effect on tumor growth. Currently, the lesion size remains stable without any treatment. Early recognition of IDF is important in order to avoid unnecessary surgical intervention that may prove to be potentially aggravating, unless serious functional or cosmetic concerns intervene. Parents should be reassured concerning the benign nature of IDF and be informed that spontaneous involution of IDF might be expected.

Introduction

Infantile digital fibromatosis (IDF) is a rare benign tumor of fibromatous origin. IDF preferentially occurs during early childhood and is present at birth in about 30% of the cases.[1] Predilection sites include the distal phalanx of the fingers and the toes. Initially IDF was considered as a potentially malignant tumor, leading to the amputation of the affected digits.[1-3] More recently several case reports of spontaneous regression suggest a benign biological behavior.[3-5] Malignant transformation and metastases have never been reported.[1,6] Histopathology often reveals paranuclear inclusion bodies within proliferating spindle-form cells.[7] Currently conservative treatment is recommended according to the benign nature of IDF, the tendency to spontaneous regression, and the frequent recurrences after surgery.[6,8]

Case Report

A six-month-old Caucasian girl presented after birth with three reddish, confluent, small, painless, indurated, ill-circumscribed nodules on the fourth digit of the left hand, which progressively increased in size (Figure 1). Previous medical history was unremarkable and there was no history of trauma or inflammation. No allergic history was reported. The other hand and both feet were unremarkable. None of the family members had experienced similar lesions. There was no functional impairment and the size of the lesion did not appear to bother the child in daily life. No signs of scratching were present. Further clinical examination was normal and Rontgen examination revealed the cutaneous and subcutaneous location of the lesion without further bone or joint involvement. Six months later a second examination showed no differences and the developmental stage of the child was normal according to age.

A 3-mm punch biopsy was performed under local anesthesia, and the histopathology revealed a fibromatosis affecting the reticular and adventitial dermis, arranged as sheets and interlacing bundles of eosinophilic myofibroblasts set in a collagenous background. Some cells demonstrated eosinophilic cytoplasmic inclusions (Figure 2). No mitotic figures and no atypical cells were observed. The overlying epidermis was unremarkable. A final diagnosis of congenital IDF was suggested.

As the data in the current literature recommend avoiding surgical intervention, non-invasive treatments exhibiting anti-fibrotic properties were sought. A topical treatment with imiquimod cream (Aldara cream), applied three times a week during four weeks, failed to reduce the tumor size and was associated with significant skin irritation, forcing the interruption of the treatment. Subsequently, topical applications of diflucortolone valerate significantly reduced the inflammation although no effect on the lesion size was observed. Eight months later the tumor size remained identical. As no functional concerns were revealed, conservative management is being followed.

Discussion

IDF is a rare tumor usually affecting one or more digits.[1] Numerous synonyms describe IDF, including Reye's tumor,[9] multiple hyaline fibromatosis, infantile dermal fibromatosis, subdermal fibromatosis tumor of infancy,

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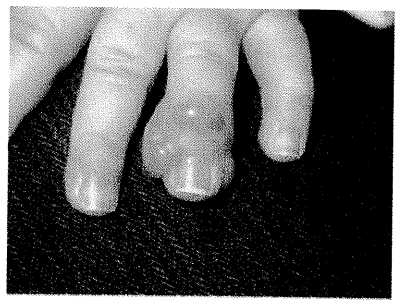

Figure 1. Infantile digital fibromatosis affecting the fourth digit.

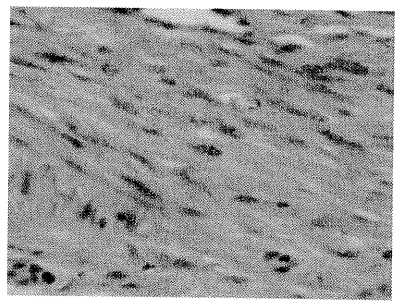

Figure 2. High-power histological presentation of the characteristic perinuclear cytoplasmic cellular inclusions found in infantile digital fibromatosis. (Hematoxylin and eosin stain)

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fibroma durum multiplex, recurrent digital fibroma, and juvenile dermatofibroma. IDF typically develops during the first year of life and the majority of cases are sporadic. Congenital onset, although rare, is described also,[7-10] as observed in our patient. Occurrence of IDF in adulthood has been reported only once in a 52-year-old woman with a tumor on the dorsum of the proximal nail fold of the right second toe.[11]

Clinically, IDF presents as smooth, round, indurate, confluent nodules up to 2 cm in diameter. The lesions are single or multiple.[7,8] The color is pink to reddish. Initially IDF exhibits an indolent progression followed by a rapid growth phase during several months. Subsequently, the tumor size remains stable until spontaneous regression without scarring occurs.[8] IDF is painless and not pruritic. It is localized usually on the lateral and dorsal aspects of the fingers and/or toes sparing the thumb and the great toe. Ulceration may occur. Functional impairments or deformities are rare.

Histopathology reveals a proliferation of myofibroblasts often displaying characteristic paranuclear eosinophilic inclusion bodies.[1] They are typically juxtanuclear and sometimes indent the nucleus.[6,7] Mitotic figures are rare. Usually the tumor cells immunohistochemically express vimentin, calponin, desmin, and α-smooth muscle actin, whereas results for keratin and muscle actin are negative.[1] The positive actin staining suggests the presence of contractile filaments. Masson trichrome histochemical staining reveals a red staining and a purple coloration with phosphotungstic acid-hematoxylin.

The etiology of IDF remains uncertain but the following hypothesis is proposed currently. The differentiation from fibroblasts to myofibroblasts is a cornerstone process in wound healing and tissue repair. Myofibroblasts may create high contractile forces that are beneficial for physiological tissue remodeling but harmful for tissue function, especially when it becomes excessive such as in hypertrophic scars, in nearly all fibrotic diseases, and during stroma reactions to tumors.[12] The intracellular inclusions suggest a viral origin. However, this etiopathogenic hypothesis was invalidated as human papillomavirus HPV DNA types 6, 11, 16, and 18, and herpes simplex virus DNA type I and II could not be demonstrated in IDF.[13] IDF may be associated with articular alterations and dysfunction.[6,8,14] The etiopathogenesis of IDF joint deformities is not clear. Given the presence of contractile proteins within the pathological cells, as suggested by histochemical, immunohistochemical, and ultrastructural studies, they may intervene in the progressive contracture, especially when the tumor occurs adjacent to a joint.[8]

The differential diagnosis of IDF includes keloids, hypertrophic scar tissue, terminal osseous dysplasia and pigmentary defects, and juvenile aponeurotic fibroma.[6] The diagnosis is clinical essentially but histological confirmation confirms the diagnosis and prognosis. The current management of IDF recommends avoiding surgical intervention, as spontaneous involution of IDF is the rule.[6] In fact, wide local surgical excision has been associated with a recurrence rate of up to 60%.[1,3] If required by functional impairment, Mohs micrographic surgery is recommended.[11] An patient with IDF was treated effectively by Mohs surgery without any residual functional impairment or recurrence of the tumor.[15] However, even Mohs surgery is regularly associated with recurrences.[12] Consequently, surgical treatment should be avoided unless severe dysfunction is observed. Several medical approaches have been evaluated. A 7- year-old boy was successfully treated with five monthly injections of fluorouracil.[16] Topical imiquimod, an imidazoquinoline amine, acts as a toll-like receptor (TLR) 7 and 8 agonist and stimulates the innate and cell-mediated immune system through induction of interferon-α (IFN-α), IFN-γ, and interleukin-1, 6, 8, and 12 production. Beside the antiviral and antitumor properties, imiquimod also inhibits human fibroblast collagen production by INF-α and INF-γ.[17] Topical and intralesional corticosteroids also display anti-fibrotic properties, and are used for treating keloids and hypertrophic scars.[17] Despite these anti-fibrotic properties, our case failed to respond to topical imiquimod therapy and was associated with significant adverse events. Topical corticosteroid applications reduced only the inflammatory aspect of IDF without reducing the lesion size.

In conclusion, IDF is a rare benign childhood tumor, which is important to recognize to avoid unnecessary surgery unless serious functional concerns intervene. Parents should be informed of the benign nature of IDF. This represents a strong argument for a conservative approach until spontaneous involution occurs.

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