Infantile digital fibromatosis: case based review and first reported dermoscopic picture

Paschalis Konstantinou¹
Eliana Eliadou²
Theodoros Lyssiotis³
Dimitris Moniatis⁴
Justin Weir⁵

¹ Dermatologist-Venereologist, Member of Dermatology Venereology Society of Cyprus (CDVS), Ex member of Scientific Committee of CDVS, Cyprus
² Pediatric Surgeon, ‘Ygeia Polyclinic’, Limassol, Cyprus
³ Pathologist, ‘Dr. T. Lyssiotis’, Histopathology and Cytology Center, Cyprus
⁴ Special Radiologist, ‘Ygeia Polyclinic’, Limassol, Xray Department, Cyprus
⁵ Charing Cross Hospital, Histopathology Department, ICH-NHS, London, UK

Address for correspondence:
Paschalis Konstantinou
3 Grigori Afxentiou str, Appt 202, Mesa Geitonia
4005 Limassol, Cyprus
E-mail: pascalinou@gmail.com

Summary

Infantile Digital Fibromatosis (IDF) is a relatively not so rare benign tumor of soft tissue origin that occurs mostly in infancy. In this study we present the case of an 11-month-old male infant diagnosed with a solitary IDF of the 4th finger of right hand. The clinical, dermoscopic, surgical, ultrasound and histopathologic features of the disease are presented; the differential diagnoses, therapeutic procedures and prognosis all are described through a short literature review. In this case report we present for the first time the dermoscopic features of IDF followed by ultrasound correlation. An excisional biopsy for diagnostic and therapeutic reasons was performed which resulted in an improvement of the functionality of the finger.

KEY WORDS: infantile digital fibromatosis; benign tumor infancy; Reye’s tumor.

Introduction

Infantile Digital Fibromatosis (IDF) was first described in 1965 by Reye as a recurrent fibrous tumor of the digits (1-4). Since then approximately 250 cases are reported worldwide (2) most of them coming from Pediatric, Surgery and Pathology Departments. IDF is a benign fibrous tumor of myofibroblast origin that occurs mostly on the digits of hands and feet 3:1 (5); it has never been reported affecting the thumb or 1st toe. It is present at birth in about 30% (6, 7) and is diagnosed until the 1st year of life in 80% of the cases (3). In adults it is extremely rare; an IDF lesion was reported once in a female patient, with an age of onset at 52 years (7, 8). It appears as a non tender, fixed, skin colored or pink-red, hard, dome shaped papule, nodule or elongated plaque few millimeters to 1-2 cm long or it can manifest as a larger tumor covering the whole surface of the digits. Most of the cases, approximately 90% (9), present as solitary lesions. Other rare localizations of IDF are the dorsal and lateral surfaces of hands and feet.

The cornerstone of the diagnosis remains the histopathologic picture with the presence of the characteristic eosinophilic paranuclear inclusion bodies in the dermal myofibroblasts.

The current approach is conservative treatment for small lesions, as many of them show spontaneous regression and surgical excision for the larger ones that impede the functionality of the finger.

Case report

An 11-month-old male infant presented with a 5-month non involuting, round, hard mass on the skin of the medial dorsal phalanx of the 4th right hand’s finger (Figure 1).

The mother reported that it started as a slightly raised red spot that ‘looked like an insect bite’. She noticed a rapid growth during the next 2 months and afterwards the progression was very slow. She has applied different local products including moderate-potency cortisone creams without any change. Lately, frequent traumas and discomfort of the child while stretching the finger were noticed. The family was stressed about the benign or malignant nature of the lesion.

The clinical examination showed a healthy child with normal physical and mental growth. There was a hard, fixed, dome shaped, pinkish to skin-colored dermal papule, measuring around 8 mm, with a smooth shiny surface and well defined round borders. The overlying skin was well attached.

The dermoscopy performed revealed a homogenous ivory-white color with the presence of red dots and linear vessels on the periphery of the lesion. The brown ring on the top of the lesion represents a trauma after a self-bite (Figure 2).
An ultrasound confirmed the dermoscopic findings, showing a homogenous sonority with the presence of a thin vasculature at the periphery of the lesion. A decision for an excisional biopsy was undertaken for diagnostic and therapeutic reasons. The lesion was almost completely excised, except of its lateral branches that were infiltrating the finger’s deeper structures. The wound healed completely by primary intention few weeks later without the need of a skin graft.

The histopathology revealed a hypocellular, non-encapsulated lesion composed of spindle shaped fibroblasts with some multinucleated forms set within a collagenous stroma. There are intracytoplasmic spherical inclusions within the majority of the fibroblasts (Figure 3). These findings were consistent with the first clinical diagnostic hypothesis of infantile digital fibromatosis.

Few months post surgery we have noticed the growth of small, hard, fixed, dome shaped, dermal papules at the distal periphery of the previous lesion and 1 year post surgery the papules are stable with no further growth; there is neither functional impairment nor discomfort (Figure 4).

Discussion

Infantile digital fibromatosis is an interesting benign fibrous tumor of infancy, because of its special clinico-
Infantile digital fibromatosis: case based review and first reported dermoscopic picture

Pathological characteristics, its course with frequent spontaneous regression and its high recurrence rate after surgery. Soft tissue tumors represent 25% of all neoplastic diseases in infancy and 85% of them is benign (10, 11). Fibromatoses represent approximately 9% of all soft tissue tumors in children and IDF covers the 2% of them (12).

The differential diagnosis is quiet interesting including: amelanotic melanoma, xanthomas, solitary mastocytomas, dermatofibromas, multicentric reticulohistiocytosis, subcutaneous granuloma annulare, rheumatoid nodules, sarcoidosis, pseudolymphomas and other tumors of fibrous origin such as fibroma of the tendon sheath, juvenile aponeurotic fibromatosis, fibrous hamartoma of infancy and infantile fibrosarcoma (one of the most common malignant fibrous tumors of infancy) (10, 11). Clinically, the fibroma of tendon sheath could resemble IDF, once it often presents as a solitary hard, subcutaneous mass at the fingers, but it mostly involves the thumb, the overlying skin is not adherent and the age of onset is usually in childhood or adolescence (10). Solitary lesions of infantile myofibromatosis and digitocutaneous dysplasia syndrome are quite similar but these diseases have many other clinical manifestations that easily differentiate them from IDF (10, 13).

The clinical case presented here is the first reported case where dermoscopic findings are described as an IDF lesion. Further studies are needed in order to better establish the dermoscopic characteristics of IDF, thus helping further the diagnostic approach.

Few therapeutic approaches have been tried so far other than surgery (7, 14, 15). Intra-lesional steroids tried in 7 patients, cleared 6 lesions and 1 recurred vs 5/10 treated with surgery (14). Intralesional fluorouracil cleared completely a lesion in 1 patient with good cosmetic results (15). Topical steroids and imiquimod (6) had no therapeutic effect with the latter provoking significant skin irritation leading to its interruption.

Initially the surgical procedures were very aggressive leading to amputation of digits, as there were questions about the benign or malignant nature of the disease (16). Currently after several years of experience and follow up the approach is conservative, as many cases show spontaneous regression after 1-10 years (median 2 years) (17) and post surgery recurrences are as high as 50-90% (18-23). If there is no functional impairment then a small incisional biopsy is needed in order to establish the diagnosis as the histopathology is the gold standard. A regular follow up is then imperative as it is important to rule out further evolution and secondary joint restrictions. A complete surgical excision would be necessary in case of large lesions that impede the function of the finger and perhaps it would be useful to try intralesional steroids before deciding to proceed to surgical intervention of larger tumors.
There is a growing interest during the last years in the research and the design of small molecule drugs that interfere with growth factors, interleukins, cytokines, etc., as we begin to know more on the pathophysiologic pathways of different diseases. Transforming growth factor beta family (TGF-β) is known that plays an important role in the pathogenesis of sclerotic plaques in systemic sclerosis; recently it was shown that there is a TGF-β dependent pathway inducing fibrosis in DLE (24) and the hypotheses about IDF presume a central role of TGF-β in fibrosis in DLE (24) and the hypotheses about IDF presume a central role of TGF-β superfamily (2, 20, 25). Thus molecules that target TGF-β could be a promising field in the research of treatment modalities for fibrotic tumors and especially IDF. In conclusion, dermatologists, pediatricians and surgeons should be aware of this benign fibrous tumor of infancy, study its special features and apply the best diagnostic and therapeutic approaches according to the characteristics of each case.

Conflict of interest

Authors declare no conflicts of interest, commercial associations or financial gain regarding this article.

References