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Caso aislado

Dermatofibrosarcoma protuberans in an infant: Report of a case with features of giant cell fibroblastoma in two recurrences*

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RESUMEN

Objetivos: Describimos un caso de dermatofibrosarcoma protuberans en un lactante, que presentó rasgos histológicos focales de fibroblastoma de células gigantes en dos recidivas. Material y métodos: Lactante de tres meses de edad que fue visto en consulta por una masa tumoral en la pierna derecha. Dicha lesión estaba presente desde el primer mes de vida y ha recidivado hasta el momento en tres ocasiones. Resultados: Las primeras muestras consistían en una tumoración densamente celular con un neto patrón "estoriforme". La segunda y tercera recidivas presentaban, de manera focal, rasgos de fibroblastoma de células gigantes. Conclusiones: Este caso pone de manifiesto la estrecha relación entre el dermatofibrosarcoma protuberans y el fibroblastoma de células gigantes, y subraya la necesidad de llevar a cabo una extirpación amplia de estas lesiones como tratamiento inicial. El dermatofibrosarcoma protuberans puede darse en lactantes y niños pequeños y debe, por tanto, incluirse en el diagnóstico diferencial de los tumores de la edad pediátrica. **Rev Esp Patol 1999; 32(1): 71-75.**

Palabras clave: Dermatofibrosarcoma protuberans - Fibroblastoma de células gigantes - Lactante

SUMMARY

Objectives: We report on a case of dermatofibrosarcoma protuberans in an infant, which showed focal histological features of giant cell fibroblastoma in two recurrences. Materials and methods: A 3-month-old male infant was examined because of a tumor mass in his lower right leg. The lesion was first noticed at the age of 1 month and has so far recurred three times. Results: The initial material showed a densely cellular tumor with a distinct storiform pattern. The second and third recurrences had focal features of giant cell fibroblastoma. Conclusions: This case exemplifies the close relationship between the dermatofibrosarcoma protuberans and giant cell fibroblastoma and underlines the need for wide surgical margins at the time of the initial excision. Dermatofibrosarcoma protuberans can occur in infancy and childhood and should, therefore, be included in the differential diagnosis of tumors in the pediatric age. **Rev Esp Patol 1999; 32(1): 71-75.**

Key words: Dermatofibrosarcoma protuberans - Giant cell fibroblastoma - Infancy and childhood

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INTRODUCTION

Dermatofibrosarcoma protuberans (DFSP) is a relatively common tumor of low grade malignancy, considered to be of fibroblastic origin (1). It is usually seen in young adults, and is well known for its tendency to recur. On the other hand, giant cell fibroblastoma (GCF) is a distinctive, rare tumor in childhood with benign biologic behavior (2). These two entities share a number of characteristics, and their close relationship is becoming increasingly evident (3), to the point that GCF is widely regarded as a juvenile variant of DFSP. However, DFSP can also occur in childhood and, more rarely, in infancy (4). We report herein a case of DFSP diagnosed in infancy, in which two recurrences showed definite foci of GCF.

CASE REPORT

A three-month-old male infant was first seen at the Hospital Niño Jesús (Madrid) in February of 1991 because of a mass in his right lower leg. He had been born to a 29-year-old woman after an uneventful pregnancy. The lesion had first been noticed at 1 month of age as a lump on his lower inner right leg, and had been growing steadily ever since. At the time of initial examination, the mass measured 5.0 cm in diameter and appeared well delimited, without adherence to the underlying tissues; it was covered by skin with a slight violaceous discoloration. A punch biopsy was followed by local excision. The specimen consisted of an irregularly ovoid, rubbery, tan to whitish mass measuring 4.5 × 3.5 × 3 cm. Microscopic examination showed involvement of dermis and subcutaneous tissue by a spindle cell neoplasm with a distinct storiform pattern, minimal cytologic atypia and rare mitotic figures (Fig. 1). Several surgical borders appeared to be involved by tumor. The lesion was interpreted as an atypical variant of fibrous histiocytoma (dermatofibroma).

The first recurrence took place in May, 1992. The received fragments of tissue measured in aggregate 5 × 4 × 3.5 cm and had the same gross appearance as the initial material. The histological appearance was almost identical, but there were several discrete microscopic foci with numerous giant cells lining what appeared to be sinusoidal spaces (Fig. 2); retrospective evaluation of these areas at the time of the second recurrence led to their recognition as GCF. Once again, several surgical margins were involved.

Figure 1. The bottom of the initial punch biopsy shows a neoplastic growth with a distinct storiform pattern involving the subcutis.

The second recurrence was evaluated in July, 1994. This time the fragments of tumor tissue measured in aggregate 7 × 5 × 4 cm. The greater part of the lesion showed a classical storiform pattern, but the mitotic index was between 5 and 7 mitotic figures per 10 high-power fields (Fig. 3). This time the histological pattern, together with the immunohistochemical findings (see below) were interpreted as corresponding to DFSP. Only one discrete microscopic focus of GCF was identified in a total of 15 sections examined. No fibrosarcomatous areas were seen. Once again the tumor was present at the surgical margins.

The third and latest recurrence took place in May of 1995; this time the surgical procedure consisted of wide local excision, and only a 3 cm tumor nodule with features of DFSP was found. The margins of excision were clear by at least 2 cm. No further recurrence has been noted as of August of 1998.

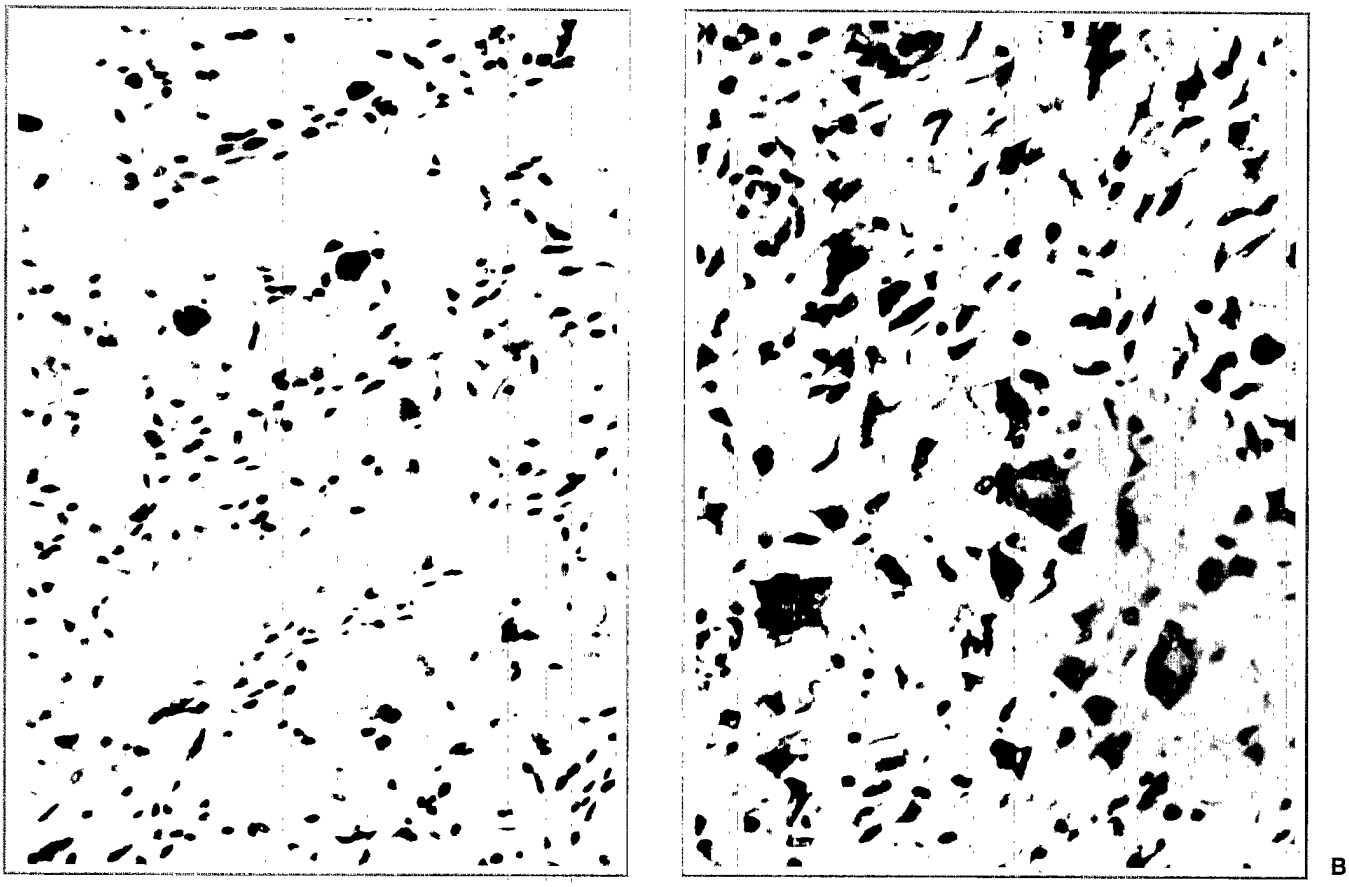

Figure 2. A) and B) Areas with features of giant cell fibroblastoma in the first recurrence.

Immunohistochemical study

At the time of the second recurrence, the immunohistochemical study of the lesion (not done initially on the previous biopsies, and subsequently performed, see below) showed intense, diffuse positivity for vimentin (Dako, monoclonal, dilution 1:70) and CD34 (Imico, monoclonal, 1:30) in the DFSP component, and only rare cells positive for CD34 in the GCF areas. All other immunohistochemical studies were negative. These comprised desmin (Dako, monoclonal, prediluted); muscle-specific actin (HHF-35, Dako, monoclonal, 1:50); smooth muscle actin (Dako, monoclonal, 1:40); S-100 (Dako, policlonal, prediluted); neuron-specific enolase (Dako, monoclonal, prediluted) and factor XIII-a (Behring, P, 1:1,000). The retrospective immunohistochemical evaluation of the archival tissue (initial punch biopsy, initial excision and first recurrence) showed identical features.

DISCUSSION

Although it occurs predominantly in the trunk of young adults, DFSP has occasionally been reported in infancy (4), and a handful of congenital cases are on record (5-8). The difficulty of the diagnosis in this setting lies not so much in its histological appearance (classically consisting of a storiform pattern with poor circumscription), but in the lack of awareness of the existence of this lesion in children (4). As exemplified by this case report, failure to make a precise diagnosis initially can lead to suboptimal surgical treatment, with an increased risk of recurrence. Other possible complications of DFSP include the development of metastases in less than 0.5% of cases (mainly to lymph nodes) and the presence of areas of fibrosarcoma in the recurrences, with an alleged metastatic potential of up to 15% (9, 10).

GCF, a rare pediatric tumor of presumed fibroblastic origin, was delineated as a pathological entity by Shmook-

Figure 3. A somewhat denser spindle cell proliferation of cells with plump nuclei and abundant mitotic figures was seen in the latest recurrence. No fibrosarcomatous areas were present.

ler and Enzinger in 1982 (2). In a 1985 review, Chung was the first author to suggest that GCF could represent a juvenile variant of DFSP (11). Four years later, Shmookler *et al.* presented clinical and histological evidence of such a relationship (12). There is further evidence in the medical literature of the close link between both lesions. There are reports of hybrid lesions (13-15), examples of GCF recurring as DFSP (16, 17) and vice versa (18). Our case is yet another variation on the same subject: foci of clear-cut GCF were present in two of the recurrences studied. In contrast to the report by Beham and Fletcher (13), in our case there was a sharp separation between the two patterns.

There are also features in common between these two entities at the immunohistochemical level. The human progenitor cell antigen CD34 has been found to be so consistently positive in the cells of DFSP that it is routinely used, along with factor XIIIa, in the differential diagnosis of fibrohistiocytic lesions of the skin with a storiform pattern (19, 20). Recently, it has been shown that GCF shares this same phenotype (15).

The close relationship between both entities has also come to light thanks to cytogenetics and gene cloning. DFSP regularly shows the presence of a supernumerary ring chromosome (21-23) that contains sequences of both chromosomes 17 and 22 (24). Only two reports regarding the cytogenetics of GCF are on record, and in both a t(17;22) has been identified (25, 26). These chromosomal abnormalities apparently result in the dysregulation of platelet-derived growth factor B (PDGFB), with the overproduction of a mature PDGFB protein that would have oncogenic properties.

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