Congenital desmoid fibroma of the soft tissues

in a 6-month-old girl

Abstract :

Desmoid fibroma are a group of clinically heterogeneous deep fibrous tumors grouped under the term "desmoid fibromatosis". They are classified according to their biological behavior into three groups: sporadic, associated with familial adenomatous polyposis, and the familial or multicentric form. The infantile form is sometimes described as a fourth group.

We report the first case in the world of congenital desmoid fibroma in the soft tissues of the anklein a 6-month-old female who benefited of an excision, with a good evolution over 6-year follow-up.

Through this observation, the authors suggest to make a review of the literature.

Introduction:

Desmoid fibroma is a rare benign fibrous tumor, characterized by local malignity and tendency to recurrence, originating from aponeurotic structures and fascias.

In the limb, it mainly affects the root. The foot and the hand are rarely affected, with variable clinical aspects. The clinical and radiological signs are not specific, then the diagnosis is histological but sometimes difficult. More than 75% of the cases occur during the first three decades; the neonatal or congenital form has never been described in the literature.

We report the first worldwide case of congenital desmoid fibroma in the soft tissues of the ankle in a 6-month-old female. Through this observation we suggest to review the clinical, therapeutic and evolutionary aspects of desmoid fibroma.

The case:

It was a 6 months old girl, from a well monitored pregnancy, father died from an unlabeled digestive neoplastic pathology. Apgar at birth 10/10. Examination of the right foot found a mass in the ankle, with a hard consistency and regular contours, 5 cm in diameter, ulcerated in places, bleeding during dressing change (figure 1), with gradual increasing in size.

A first consultation was made at the age of one month where an MRI was performed which objectified signs in favor of hemangioma (figure 2), then a treatment based on beta blockers was conducted for three months without improvement.

Removal of the mass was performed in front of the non-improvement and the anemic Syndrome due to bleeding of the mass. The surgical procedure revealed a whitish mass with 6 cm long axis, which appears to have a clear cleavage plane, and sends septums to the subaponeurotic plane. The resection was complete, including the fascia. The closure was done in the transverse plane given the presence of a slight excess of skin without using skin graft or a flap.

The histological study came back in favor of desmoid fibroma. Genetic complement was performed showing a normal karyotype. Control every six months was done, then annually with good evolution over a period of 6 years (figure 3: 3-yearfollow-up).

Discussion:

Desmoid tumors occupy an important but uncommon place between fibromatosis and fibrosarcoma's. Desmoplastic fibroma appears to be a very rare tumor. According to Taconis et al [1] and Vaz et al [2]. It represents 0.3% of benign bone tumors, with local malignancy and no metastasis or malignant transformation.

Desmoplastic fibroma occurs often twice in women. More than 75% of cases were noted in the first three decades. The incidence is high between 20 and 40 years old. Contrary to our patient who presented a neonatal mass and therefore congenital [2- 3] which led to confusion with hemangioma and thus a delay in diagnosis.

The discovery mode is not specific, It's a very slow-growing tumor. The most frequent symptoms are pain and swelling [3].

The etiology still unknown, and several hypotheses have been proposed. The role of hormonal impregnation has been noted by many authors suggesting that desmoid tumors are estrogen-dependent. The incidence is higher in women in their reproductive years, during pregnancy or after childbirth, but also when taking oral contraception. The role of trauma, caused by previous surgery, has also been suggested [4]. Nevertheless, their existence as reactive lesions seem to be questioned at present, given the preponderant role of genetic factors [5]. Genetic factors have been well incriminated in case of association with PAF which is an autosomal dominant disease linked to the mutation of the adenomatous polyposis coli tumor suppressor gene (APC). The hypothesis retained in our patient was hormonal impregnation probably associated with genetic factors given that the father died of non-specified digestive neoplasia.

The macroscopic appearance of desmoplastic fibroma's has been well described by Mazabraud et al [6]: it's a whitish or grayish homogeneous tissue with firm or elastic consistency, rubber-like, the sectional slice of the tumor appears shiny with fasciculations. In our case, it's was a whitish, elastic but ulcerated mass bleeding on contact, which led to confusion with hemangioma, the diagnosis was later rectified. Sometimes there is an impression ofrelatively clear cleavage between the tumor area's and the adjacent bone tissue.

In Microscope, there is a large histological similarity between the bone desmoid fibroma and the soft tissue desmoid fibroma's (abdominal or extra-abdominal desmoid tumors). It's a connective tissue very rich in collagen. These collagen fibers are arranged in large parallel bundles or, on the contrary, in fine wavy fibers without a clear fasciculate arrangement. There is a moderate cellular richness, even weak. The cells are spindle-shaped and small. The nuclei are small, regular, round or oval, with fine chromatin, mitoses are rare. There is no atypical mitosis. The desmoplastic fibroma has moderate vascularization without bony or cartilaginous component.

Their benign histological structure and poor metastatic potential contrast with a high local aggressiveness and a high rate of recurrence after surgical excision. This justify an extra-lesional removal with an intact margin of peri-tumoral bone tissue to limit the risk of local recurrence.

Analysis of the treatment of desmoplastic fibroma's in the literature is difficult. Indeed, the therapeutic approach is based on the experience of each author.

Postoperative surveillance must be prolonged. Indeed, the average delay of recurrence is three years [3]. Recurrences have been described up to ten years after the initial treatment. Surveillance should be clinical and radiological.

The natural evolution of desmoplastic fibroma after treatment is marked by high risk of tumor recurrence.

The prognosis is good in terms of survival, it determines the modalities of treatment which must remain conservative in the majority of cases. Wide surgical excision, without mutilation, ensures the best chances of recovery without sequels. When It's not possible, radiotherapy is a therapeutic alternative and leads to high rate of complete regression [7].

Conclusion:

Neonatal forms of desmoid fibroma of the extremities represent a new clinical entity of desmoid tumors; their locally invasive character is not yet demonstrated. The definitive surgical treatment is difficult.

References:

1- TACONISWK, SCHUTTEHE, VAN DERHEULRO: Desmoplastic fibroma of bone: a report of 18 cases. Skeletal Radiol,1994, 23,283-288.

2- G. Vaz, A. Richard, O. Guyen, J. Bejui-Hugues, J.-P. Carret. Le fibrome desmoplastique ou fibrome desmoïde osseux. À propos de 2 nouveaux cas d'une tumeur osseuse rare. Revue de chirurgie orthopédique 2005, 91, 782-787

3- Böhm P, Kröber S, Grescniok A, Laniado M, Kaiserling F. Dermoplastic fibroma of the bone. Cancer 1996; 78:1011–23.

4- Reimato J, Scheinin T, Hayvry P. The desmoid syndrome. Am J Surg 1986; 151:230–7.

5- Fletcher CDM. Soft Tissue Tumors. 2nd Ed. In: Fletcher CDM, editor. Diagnostic Histopathology of Tumors, vol. 2. Philadelphia: Churchill Livingstone; 2000. p. 1473–539

6- MAZABRAUD A: Fibrome desmoïde. In:Anatomie pathologique osseuse tumorale. 1994, Springer Verlag France, Paris, p. 167-172.

7- DEQUANTER D., gebhart M, TUMEURS DESMOIDES J Chir., 2002, 139 (4), pp. 236-239

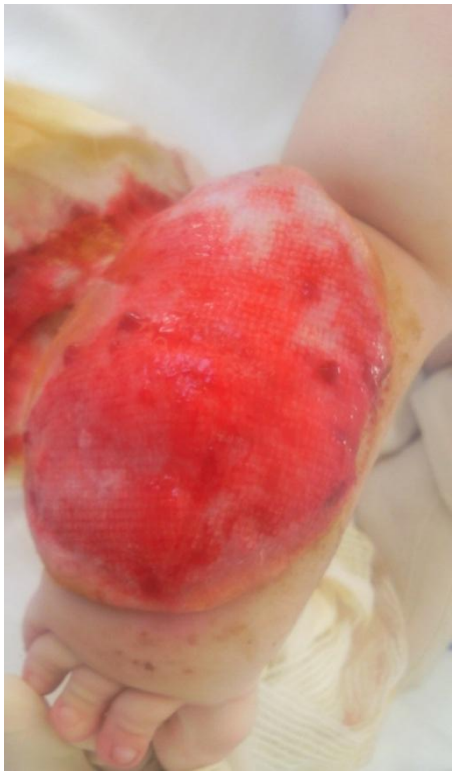

Figure 1: clinical aspect of the mass with an ulcerated surface

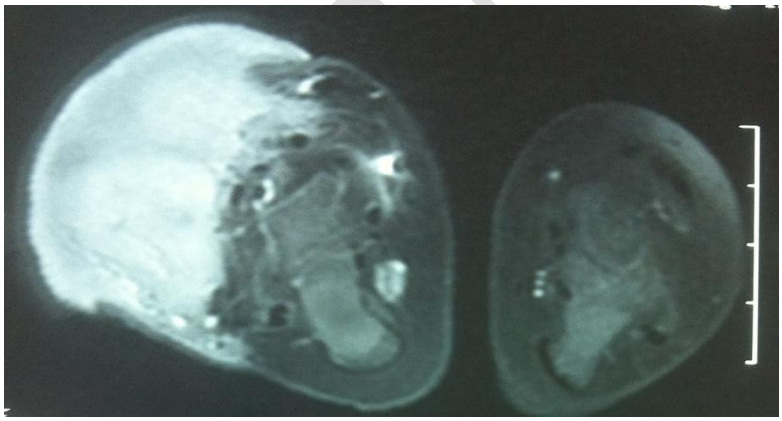

Figure 2: MRI aspect of the mass which sends partitions in under aponeurotic

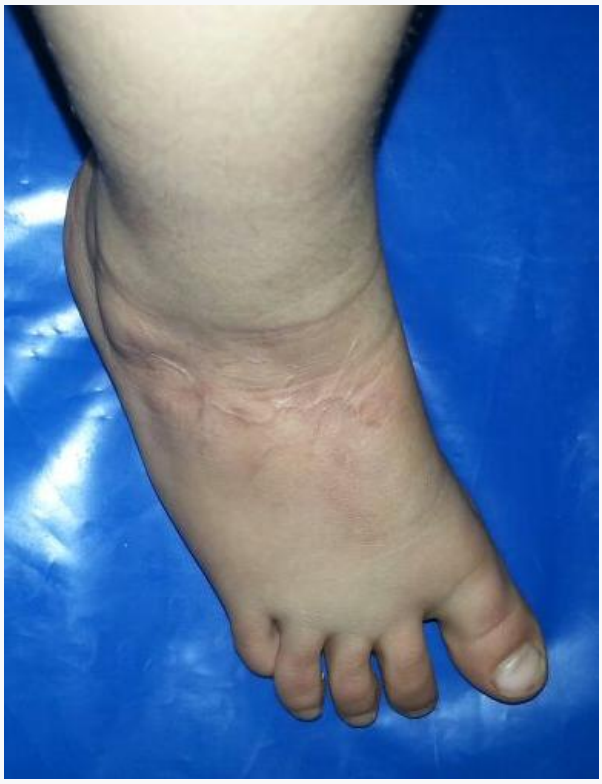

Figure 3: current appearance of the foot and the scar (3-year follow-up)