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A Congenital Purple Plaque, a Case Report with the Review of Literature

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Abstract: Dermatofibrosarcoma protruberans (DFSP) is an uncommon, locally aggressive tumour arising. it accounts for 0.1% of all malignancies. Although it can present any time from the neonatal period onwards, paediatric cases are rare, and account for 6% of all DFSP. The cause of dermatofibrosarcoma protuberans is not known but, injury to the skin in the affected location could be a risk factor. It can occur within pre-existing scars and tattoos. Clinical presentation is typically as a solitary plaque or nodule most often on the trunk or extremities. The initial lesion is a skin coloured, slow growing plaque which develops a red /brown colour and nodular surface. but tends to recur after excision. It rarely spreads to other sites beyond the skin. The article presents an overview of epidemiology, distinct clinical evolution, histopathological characteristics, gene involvement and differential diagnosis of DFSP. Management should be multi-disciplinary, with a view to maximising tumour clearance while minimising tissue loss. We present A 3-year-old girl was referred to dermatology for a slow-growing painless lesion on her left lower abdomen. The lesion had been present since birth. At ten months-of-age a paediatrician made a clinical diagnosis of infantile haemangioma. Examination of the skin of the left lower abdominal quadrant revealed a 5×3 cm ill-defined pink plaque containing multiple discrete violaceous nodules. Histology revealed a dermal infiltrate of monomorphic spindled cells arranged in a storiform pattern with no cytological atypia, Immunohistochemistry staining was positive for CD34. Following multi-disciplinary review, she was referred to plastic surgery for wide local excision. The indolent behaviour of early DFSP can lead to a delay in diagnosis. Also, they are commonly misdiagnosed as vascular malformations children. It is recommended that patients undergo surveillance for local recurrence for longer than five years following primary excision.

Keywords: Dermatofibrosarcoma Protruberans, DFSP, Plaque, Fibroblastic, Immunohistochemistry, Moh's Micrographic Surgery, Spindle Shape Cells, CD34

1. Introduction

Dermatofibrosarcoma protruberans (DFSP) is an uncommon, locally aggressive tumour of fibroblastic origin from the dermis. The initial lesion is a skin coloured plaque which develops a red /brown colour and nodular surface. It grows slowly but tends to recur after excision. It rarely spreads to other sites beyond the skin, the indolent behaviour of early DFSP can lead to a delay in diagnosis. Usually unexpected pathological results of presumably benign skin mass prompt towards correct diagnosis. [7]

2. Clinical Case

A 3-year-old girl was referred to dermatology for a slow-growing painless lesion on her left lower abdomen. She had no past medical history nor relevant family history. The lesion had been present since birth. At ten months-of-age a paediatrician made a clinical diagnosis of infantile haemangioma. Recorded dimensions at this time were 3.5×1.5cm. Over the ensuing two years the lesion grew slowly, prompting a dermatology referral.

Examination of the skin of the left lower abdominal quadrant revealed a 5×3 cm ill-defined pink plaque

containing multiple discrete violaceous nodules. (Figure 1). Dermoscopic appearance was unremarkable. There was no change in skin texture or skin atrophy. Diagnostic punch biopsy was performed.

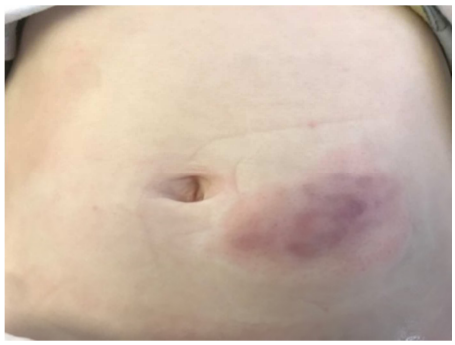

Figure 1. *The left lower abdominal quadrant revealed a 5×3 cm ill-defined pink plaque containing multiple discrete nodular violaceous components.*

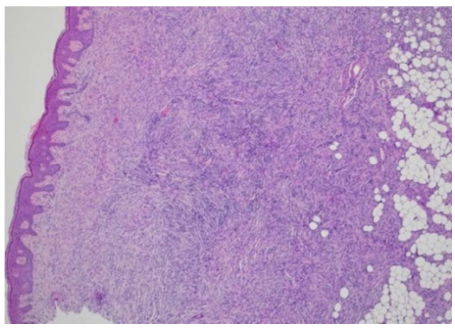

Figure 2. *Proliferation of spindle-shaped cells in the deep dermis with later lesions infiltrating the subcutaneous fat. There is superficial sparing of epidermis.*

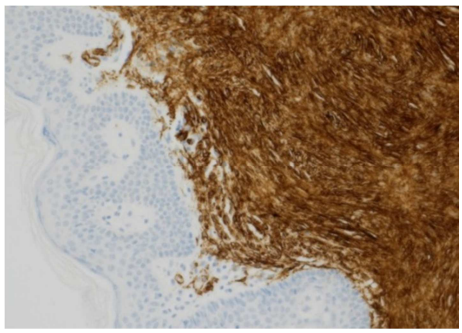

Figure 3. *Immunohistochemistry staining, positive for CD34.*

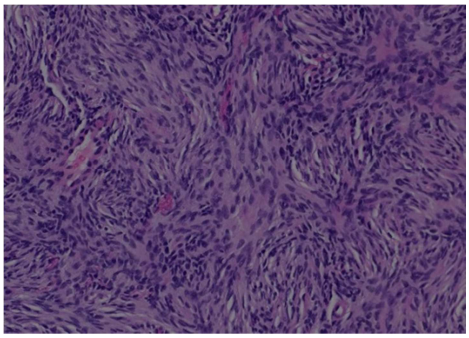

Figure 4. *A dermal infiltrate of monomorphic spindled cells arranged in a storiform pattern with no cytological atypia. Spindled tumour cells are quite uniform in apprearance with elongated nuclei.*

Histology revealed a dermal infiltrate of monomorphic spindled cells arranged in a storiform pattern with no cytological atypia, nor significant mitotic activity. There was deep infiltration of subcutaneous fat in a lacy pattern (Figure 2). There was epidermal sparing. (Figure 3) Immunohistochemistry staining was positive for CD34 (Figure 4). The COL1A1-PDGFB gene fusion product was detected by next generation sequencing, further supporting the diagnosis.

Following multi-disciplinary review, she was referred to plastic surgery for wide local excision.

3. Discussion

Dermatofibrosarcoma protruberans (DFSP) is a rare tumour, first described in 1924 as invasive, progressive dermatofibroma [7]. It is locally-aggressive fibrohistiocytic tumour that accounts for 0.1% of all malignancies. [1] Age and gender are not a risk factor for DFSP and its recurrence. [7] Although it can present any time from the neonatal period onwards, [2] paediatric cases are rare, and account for 6% of all DFSP. The age-adjusted incidence is 1 per million in patients under 20 years old. [3, 4] The cause of DFSP is unknown, however The mesenchymal tumour is thought to originate from a dermal stem or undifferentiated mesenchymal cell with fibroblastic, neurological and muscular features. [9] 90% of DFSP harbour the t (17, 22) translocation which causes the *COL1A1-PDGFB* fusion gene which predicts response to tyrosine kinase inhibitors. Indeed, imatinib mesylate has been used as a successful adjunctive targeted therapy in some paediatric DFSP patients. [2] The chromosomal translocation leads to fusion of platelet derived growth factor beta polypeptide gene (PDGFB) and collage type 1A1 (COL1A1). The gene rearrangement up regulates PDGFB, resulting in over production of platelet derived growth factor (PDGF), continuous autocrine activation of platelet drive growth factor receptor beta (PDGFRb), cellular proliferation leading to tumour formation. In cases without the COL1A1/PDGFB translocation, a different chromosomal translocation has been demonstrated involving PDGFB on chromosome 22. CD34 is commonly positive (80–100% of cases), while FXIIIa, SMA, desmin, S100, and keratins are negative. [9] Typical Presentation is as a solitary plaque or nodule most often on the trunk or extremities. [4, 5] Majority of DFSP are on trunk (50%) followed by extremities (35%) and then Head and Neck (15%). [9, 10] lesions of head and neck tend to show higher recurrence. [7] During pregnancy DFSP can show accelerated growth. Clinically there are two distinct phases. Early-stage lesions are slow growing range in colour from skin-coloured to red or blue. [4] Later-stage lesions grow more rapidly, and becoming protuberant and nodular. [7] They are commonly misdiagnosed as vascular malformations or tumours in children. [4] DFSP is benign but if not completely excised it can cause high burden morbidity. [7] distant metastasis is rare in 1% to 4 % and usually only occurs after multiple local recurrences**.** The most common site of metastasis via hematogenous spread is lungs. The

fibrosarcomatous variant of dermatofibrosarcoma protuberans has a higher risk of local recurrence (14% to 52%) and distant metastases (8% to 29%). Dermatofibrosarcoma protuberans has a good prognosis. In rare instances the main complication the rare instances is metastasis. Other complications could be centered around post-surgical cosmesis and scarring. Soft tissue construction should be performed immediately after oncology clearance.

The clinical evolution of DFSP is reflected in its histology. In the early phase, the characteristic spindle-shaped fibroblasts show minimal atypia, and few mitotic figures. [5] In the later nodular stage, the spindle cells are arranged in a whorled or storiform pattern, (figure 4) and show greater rates of atypia and mitosis. [5]. It has various forms; the rare variants are atrophic and pigmented forms. Pigmented DFSP, also called Bednar tumours, usually present as a black appearance or dark blue bruise colour which are due to melanic containing dendritic cells in addition to other typical histological findings. [7] The Atrophic form which clinically presents as a flat plaque dermal lesion, histologically it has dermal plaque like histological features with dermal layer thinning. both pigmented and atrophic types have spindle cells with vimentin and CD34 positive. [7] Other variants are myxoid, giant cell, and fibrosarcomatous variant. The latter is the rarest but most aggressive and greater potential of metastasis.

Differential diagnosis for DFSP lesions is, dermatofibroma, cutaneous neurofibroma, epidermal inclusion cyst, cutaneous melanoma, dermatologic metastatic carcinoma, intradermal spindle cell lipoma, schwannoma and solitary fibrous tumour. Histologically cellular fibrous histiocytoma /dermatofibroma, negative for CD34 and positive FX13a. Solitary fibrous tumour, CD34 positive, spindle cell lipoma is S100 positive. angiosarcoma positive for CD31 and ERG. Spindle cell melanoma that expresses S100, MART1/MELAN A. [9]

Diagnosis is established by skin biopsy for histopathological and immune histochemical tests. while MRI and fluorescence in situ hybridisation in addition to reverse transcriptase is helpful for screening and choosing the best treatment. [7]

DFSPs develop microscopic finger-like projections that, if inadequately excised in the first attempt, lead to recurrence over a period of years. [4, 5] Thus optimal management is via wide local excision followed by Moh's micrographic surgery (MMS). MMS is preferred as it is tissue-sparing, yet still optimises tumour clearance. [1, 5] Although it DFSP is locally invasive, distant metastasis is rare. [5] complete surgical excision is the gold standard however neoadjuvant therapy with imatinib and radiotherapy is recommended for the cases with incomplete resection. [6, 13] In children adjuvant radio therapy should be avoided as there is long-term risk of secondary malignancies and potential for growth disruption. [8] A prospective study of 46 paediatric cases found a local recurrence rate of 6.5%, and no distant metastasis. [2] Management should be multi-disciplinary, with a view to maximising tumour clearance while minimising tissue loss. It is recommended that patients undergo surveillance for local recurrence for longer than five years following primary excision. [1, 5] In cases where factors are suggestive of low risk and the location of the disease makes it difficult to remove, it may be kept left alone under strict surveillance in view of the difficulty of treating it vs good prognosis. [7]

4. Conclusion

This case highlights that congenital DFSP may be mistaken as a vascular tumour. Having been misdiagnosed as an infantile haemangioma early on, the persistence and growth of the lesion over time prompted dermatology referral, and ultimately histological diagnosis. Our patient underwent a wide local excision and is now undergoing clinical surveillance in line with best practice. A multi-disciplinary approach and good communication between team is crucial and close collaboration with the pathologist who is familiar with the sectioning technique is paramount. [8]

Conflict of Interest Statement

The authors have no conflict of interest to declare.

Consent Statement

Written consent has been obtained from the patient's parents, a copy of which will be uploaded with this submission.

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