Myoid differentiation in dermatofibrosarcoma protuberans and its fibrosarcomatous variant: 10 years' experience in a local tertiary hospital

Lo CH, United Christian Hospital

Introduction

Dermatofibrosarcoma protuberans (DFSP) is a relatively rare, locally aggressive, dermal based fibroblastic tumour, with an incidence of 2-4 new cases per million per year. It usually affects young to middle-aged adults, and is most commonly found on the trunk and proximal extremities. It typically presents as a slow growing nodular or multinodular cutaneous mass. Rapid enlargement may occur during pregnancy or due to tumour progression to fibrosarcomatous DFSP. Other than fibrosarcomatous DFSP, a few histological variants such as pigmented DFSP, myxoid DFSP and DFSP with myoid differentiation are also described. The usual emphasis is on fibrosarcomatous DFSP as it acquires metastatic potential. Myoid differentiation is rare, and more often found in fibrosarcomatous DFSP. In this study, we try to characterize the immunostaining pattern regarding myoid differentiation in DFSP, and discuss the potential pitfall in making the diagnosis.

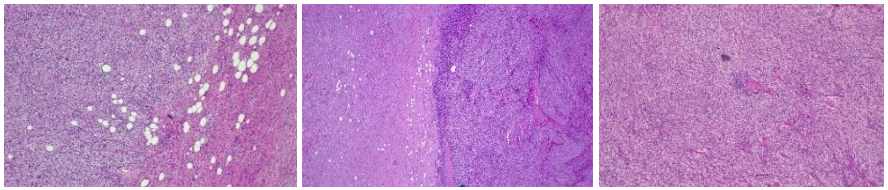
Material and methods

A total of 17 cases of DFSP were found in the past ten years in our hospital, 7 of them were excluded as those were biopsy or re-excisional specimen from same tumour or patient. The details of the remaining 10 cases are listed in the table as below. All cases are reviewed to confirm the diagnosis of DFSP and assessed for myoid differentiation. Myoid differentiation is defined as tumour cells with brightly eosinophilic cytoplasm, well-defined cytoplasmic margins and vesicular nuclei.

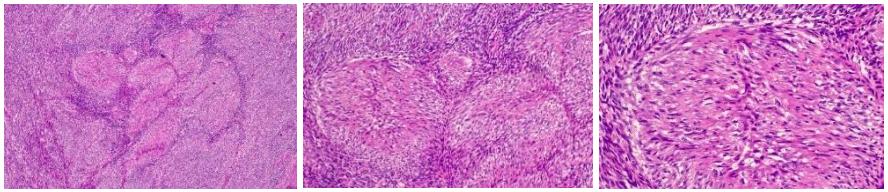
Case	Age	Sex	Location	Fibrosarcomatous variant	FISH for COL1A1-PDGFB fusion
1	16	F	abdominal wall	no	not done
2	25	F	breast	no	yes
3	25	M	thigh	no	yes
4	28	F	thigh	no	not done
5	41	F	abdominal wall	no	not done
6	47	M	back	yes	not done
7	48	M	abdominal wall	no	not done
8	58	M	back	no	not done
9	67	M	back	no	yes
10	75	M	back	no	yes

Results

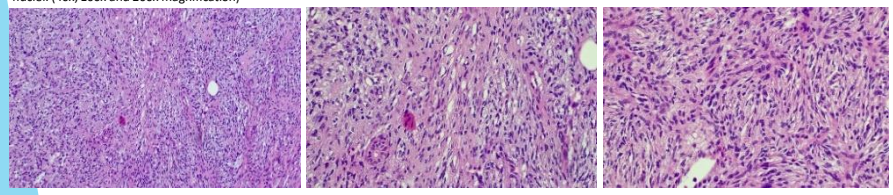
Around 10% of the tumour area in case 6 (the only case of fibrosarcomatous DFSP) and around 5% of the tumour area in case 8 are found to have myoid differentiation.



Left: H&E section from case 8, showing the typical storiform growth pattern and infiltration of subcutaneous fat in a honeycomb appearance. (100x magnification)
Middle and right: : H&E section from case 6, showing the abrupt transition from classical area to fibrosarcomatous differentiation, featuring cellular spindle cell fascicles with a herringbone appearance. (20x and 40x magnification)



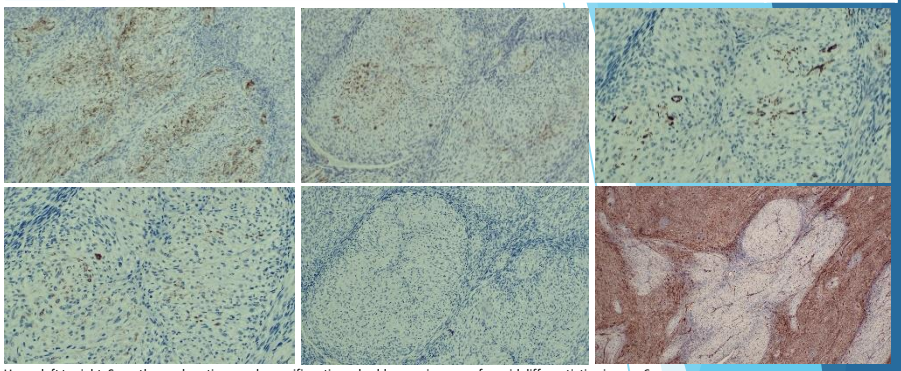
H&E sections showing areas of myoid differentiation in case 6. The cells contain brightly eosinophilic cytoplasm, well-defined cytoplasmic margins and vesicular nuclei. (40x, 100x and 200x magnification)



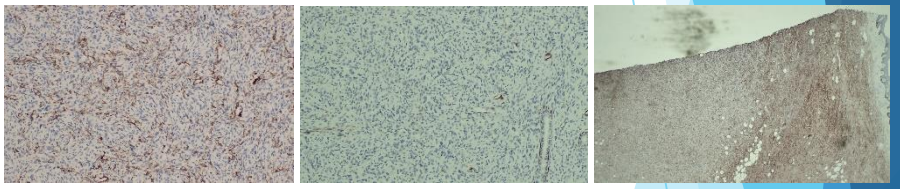
H&E sections showing areas of myoid differentiation in case 8. (40x, 200x and 200x magnification)

Immunostains for smooth muscle actin have been performed on all 10 cases. Those 8 cases without myoid differentiation are negative, while the 2 cases with myoid differentiation (case 6 and 8) show positivity in the myoid areas. Further immunostains are performed in these 2 cases, including CD34, muscle specific actin, caldesmon, calponin and desmin. The staining pattern in myoid areas are listed in table below.

case	SMA	Muscle specific actin	caldesmon	calponin	desmin	CD34
6	positive	patchy	a few positive cells	a few positive cells	negative	negative
8	positive	negative	negative	negative	negative	weak



Upper left to right: Smooth muscle actin, muscle specific actin and caldesmon in areas of myoid differentiation in case 6
Lower left to right: Calponin, desmin and CD34 in areas of myoid differentiation in case 6



Left and middle: Smooth muscle actin and muscle specific actin in areas of myoid differentiation in case 8
Right: CD34 shows weaker staining in myoid area (left side) compare to the classical area (right side) in case 8

Discussion

Myoid differentiation is rare in DFSP, and more often found in fibrosarcomatous DFSP. In our study, 2 out of 10 cases show focal myoid differentiation, including the only case of fibrosarcomatous DFSP. Since the diagnosis of DFSP usually relies on the classical morphology and CD34 positivity, recognition of myoid differentiation is important because it may cause diagnostic difficulties with smooth muscle tumours or myofibroblastic tumours (e.g. leiomyoma, leiomyosarcoma, myofibroma), especially in small biopsy samples. The myoid areas are morphologically different from classical DFSP, and can show positive staining (albeit patchy to focal) for smooth muscle markers, such as smooth muscle actin, muscle specific actin, caldesmon and calponin. Staining for CD34 can be weak or even negative, complicating the diagnosis. In challenging cases, molecular study is helpful as large majority of DFSP show characteristic chromosomal translocation involving the collagen type 1 alpha 1 (COL1A1) gene on chromosome 17 and the platelet-derived growth factor β (PDGFB) gene on chromosome 22. The detection of COL1A1-PDGFB fusion by fluorescence in situ hybridization can aid the diagnosis.

Conclusion

DFSP is an uncommon, locally aggressive, dermal based fibroblastic tumour. Fibrosarcomatous DFSP is an important variant as it acquires metastatic potential. Myoid differentiation in DPSP is rare, and more often found in fibrosarcomatous DFSP. The recognition of myoid differentiation is important as it may cause diagnostic difficulties with other tumours, especially in small biopsy samples. In histologically challenging cases, detection of COL1A1-PDGFB fusion by fluorescence in situ hybridization is helpful.

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