Expanding the Spectrum of Lesions with COL1A1::PDGFB Fusion: three unusual myofibroblastic lesions

Gerardo Cazzato MD[1] , Joseph Khazen MD[2], Damiano Arciuolo MD[3], Grazia Nucci MD[1], Sonia Maniglio MD[1], Sabina Barresi[4], Maria Elena Errico MD[6], Alessandra Borzillo MD[6], Carmelo Lupo[5], Rita Alaggio MD[4]

[1]Unit of Anatomical Pathology , University of Bari "A. Moro" , IT ; [2]Unit of Anatomical Pathology , La Sapienza University of Rome, IT ;[3]Pathology Institute, Catholic University of Sacred Heart, Rome, IT; [4]Pathology Unit, Bambino Gesù Children's Hospital, IRCCS, Rome, IT; [5]Diapath Spa; [6]Pathology Dept "AORN Santobono Pausilipon", Pediatric Hospital, Naples, I

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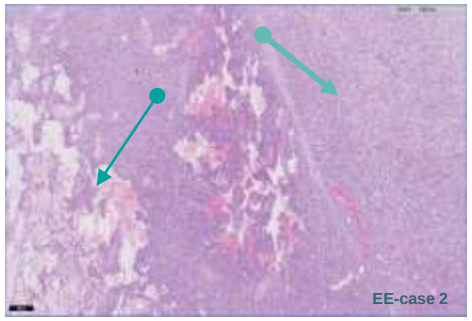
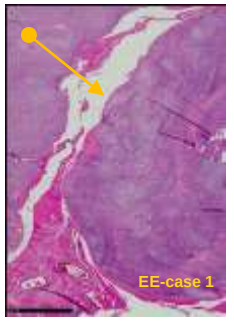
BACKGROUND

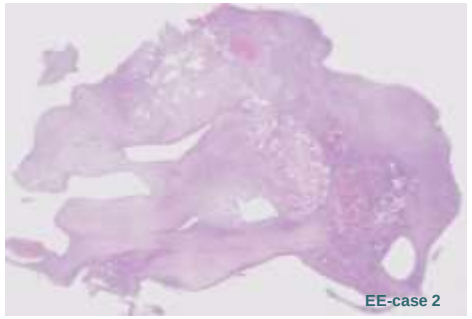
Dermatofibrosarcoma protuberans (DFSP) is a low-grade, locally aggressive CD34-positive myofibroblastic neoplasm characterized by the presence of the COL1A1::PDGFB fusion. The recent extensive molecular characterization of mesenchymal neoplasms is continuously expanding the morphological spectrum of lesions ultimately diagnosed as DFSP. We describe three unusual myofibroblastic lesions in which a COL1A1::PDGFB fusion was identified using RNA sequencing two of which were only diagnosed as DFSP after molecular analysis.

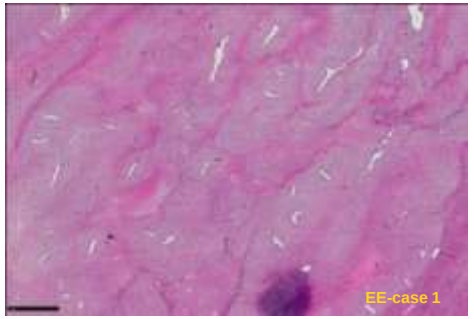
DESIGN

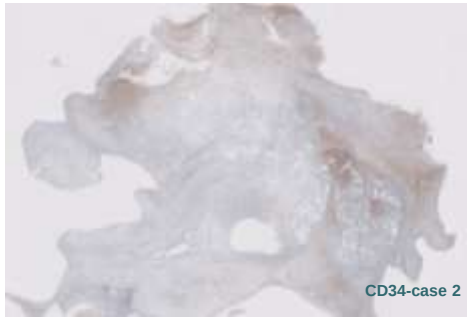
CASE	AGE (y.o.)	SEX	SITE	SIZE (cm)	MORPHOLOGY	IHC CD34	MUTATION
1	3	M	Thoraco-lumbar	2	spindle cell proliferation deeply infiltrating the subcutis and the adnexa	positive	COL1A1::PDGFB
2	15	F	Anterior region leg	3	Two different components made up of GCF and DFSP	focally positive	COL1A1::PDGFB
3	47	M	Left lower leg	25	Spindle and epithelioid cells, with plexiform and perivascular pattern	positive	COL1A1::PDGFB











CASE 1 subcutaneous thoracolumbar nodule in a 3 year-old male which consisted of a CD34-positive spindle cell proliferation deeply infiltrating the subcutis and the adnexa in a lipofibromatosis-like fashion. Molecular analysis showed a COL1A1::PDGFB fusion and margin widening revealed a thin rim of residual lesion with more classical DFSP features.

CASE 2 lesion involved the anterior leg of a 15 year-old female and corresponded to a CD34-negative proliferation with focal features of giant cell fibroblastoma (GCF) and DFSP-like areas. A diagnosis of DFSP was made only after demonstration of the COL1A1::PDGFB fusion.

CASE 3 large plaque on the lower leg of a 47 year-old male composed of CD34-positive spindle and epithelioid cells arranged in a plexiform perivascular pattern. After molecular analysis and due to the extremely unusual immunomorpholgic features, a diagnosis of myofibroblastic lesion with COL1A1::PDGFB fusion was made.

CONCLUSION

These cases emphasize the fact that single molecular alterations are associated with an increasingly growing morphological spectrum. Particularly, in the first two cases occurring in children, we have shown after integration with molecular characteristics that DFSP may present with unique, previously undescribed patterns. In the third case, it is unclear whether the lesion represents a unique variant of DFSP or a different entity, the latter alternative suggesting that the spectrum of myofibroblastic lesions carrying a COL1A1::PDGFB fusion might be wider than previously thought.