Title: A Case of Recurrent Head and Neck Dermatofibrosarcoma Protuberans

Authors and Affiliations: Kathryn Dillman, BS; Nicholas Culotta, MD; Christopher Haas, MD; LSU School of Medicine New Orleans and LSU Department of Dermatology

Patient Initials: DF **Personal Data:** 68 y.o. M

History of Present Illness:

Patient is a 68-year-old male with a history of dermatofibrosarcoma protuberans (DFSP) of the left cheek who presented to dermatology clinic for follow-up skin exam. He noted a few concerning lesions on the trunk and the face that he would like examined, but denied any bleeding, ulceration, enlarging, or non-healing lesions.

He initially reports a diagnosis of DFSP of the left cheek in 1987. He had multiple excisions including a previous pec-flap with failure and required left orbital enucleation. In 2008, he had repeat re-excision with pathology positive for DFSP and negative surgical margins. He had follow-up with otolaryngology (ENT) in December 2013 with an enlarging nodule to the left cheek in previous area of excision. In January 2014, the patient had excision of the lesion with multiple positive margins. The patient then elected for clinical monitoring and refused further surgery. In December 2019, the patient was seen by ENT and had no evidence of recurrence on exam. He was then seen by tele-dermatology and referred for in-patient evaluation but did not note any concerning lesions at that time.

Past Medical History: Hypertension, nephrolithiasis, Guillain-Barre syndrome, DFSP, actinic keratoses

Medications: Allopurinol, tamsulosin, hydroxyzine, pantoprazole, tramadol

Allergies: Iodine and Iodide containing products (results in anaphylaxis)

ROS: As mentioned in history of present illness. No additional ROS.

Physical Exam: Flesh colored firm nodule to left scalp with polymorphous vessels.

Laboratory: None.

DDx: DFSP vs pseudo-lymphoma vs leiomyoma vs fibrosarcoma vs dermatofibroma vs other

Biopsy: Left scalp shave biopsy consistent with DFSP involving both the peripheral and deep margins.

Clinical Course: The biopsy performed on his left scalp nodule was consistent with DFSP involving both peripheral and deep margins. The patient was referred to ENT for further intervention. He underwent wide local excision of this lesion with one centimeter margins sending both circumferential and deep margins for frozen section. Ultimately, both margins showed positive results for DFSP. These positive margin results were followed by mapping punch biopsies and an MRI face with and without contrast. His mapping punch biopsies came back negative for DFSP. Because of previous positive margins after surgical excision, he underwent re-excision again sending frozen sections for both circumferential and deep margins.

His circumferential margin ultimately came back positive again for DFSP. The current plan is to refer him to both medical and radiation oncology. Tyrosine kinase inhibitor testing for imatinib eligibility is also being considered. His MRI has not yet been completed.

Discussion: DFSP, a soft tissue tumor that typically involves the dermis and subcutaneous fat, is a rare tumor (incidence rate of 0.8 to 4.5 cases per million person-years) that usually presents as a asymptomatic, slow-growing plaque. The plaque is typically skin-colored or red-brown and firm/indurated. It most-commonly presents on the trunk of young adults with the most common sites being the pelvic region and the shoulder, but it can also be seen on the extremities, head, and neck. While it normally involves the dermis and subcutaneous fat, it may also involve the muscle and fascial layers in rare cases. There are two variants: the pigmented Bednar tumor and the fibrosarcomatous variant. Currently, it is thought that a chromosomal translocation between chromosomes 17 and 22 results in a COL1A1-PDGFB fusion protein, which causes excess production of platelet-derived growth factor (PDGF). This specific translocation accounts for 90% of cases with the remaining 10% still involving overexpression of the PDGFB gene. Other risk factors include injury to the skin, with occurrences previously noted in pre-existing scars and tattoos. Histopathologically, DFSP display tumor cells in a storiform or intersecting pattern running parallel to the epidermal surface. Spindle cells and mitoses are also present and typically surrounded by a collagenous stroma with hyaline or myxoid changes. For diagnostic purposes, CD34 is positive in 80-100% of cases on an immunohistochemical panel. Diagnosis is confirmed with an incisional or excisional biopsy, and treatment involves surgical removal through Mohs micrographic surgery (MMS) or wide local excision. The prognosis for DFSP is good with an overall 10-year survival rate of 99.1%. Recurrence is common with DFSP; specifically, recurrence rates for wide local excision are at 7.3% compared to MMS at 1%. Despite a low risk of metastasis with high recurrence rates, clinical follow-up of these patients is encouraged. Other treatments include imatinib mesylate, a chemotherapy agent, or an oral form of a tyrosine kinase inhibitor. Common differential diagnoses include cutaneous melanoma, keloid scarring, epidermal inclusion cyst, dermatofibroma, dermatologic metastatic carcinoma, and morphea.

Capsule Summary: This is an interesting case of recurrent head and neck dermatofibrosarcoma protuberans (DFSP) that has been affecting the patient on-and-off for at least thirty-seven years. After multiple surgical excisions, the patient continues to have positive margins. Future treatment plans include pursuing tyrosine kinase inhibitor therapy with the involvement of both medical and radiation oncology. DFSP is a rare soft tissue tumor that typically involves the dermis and subcutaneous fat with infrequent extension to the muscle and fascia. Clinically, it presents as an asymptomatic plaque that is firm and flesh-colored or red-brown. It is thought to be due to a chromosomal translocation (involving chromosomes 17 and 22) resulting in the COL1A1-PDGFB fusion protein. Diagnosis is confirmed with biopsy, and treatment involves both surgery and possible medical treatment with chemotherapy or oral tyrosine kinase inhibitors. Common differential diagnoses include melanoma, keloids, epidermal inclusion cysts, dermatofibromas, cutaneous metastases, non-melanoma skin cancer, and pseudo-lymphoma.

References:

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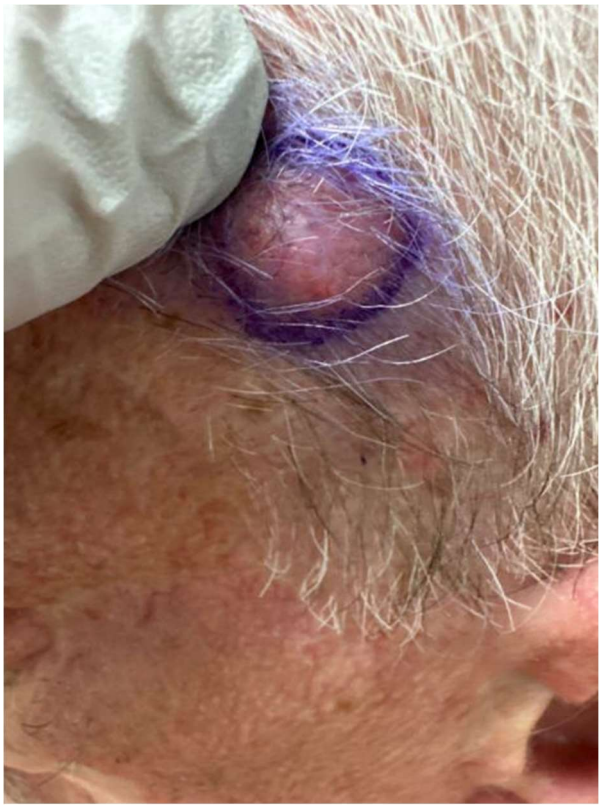
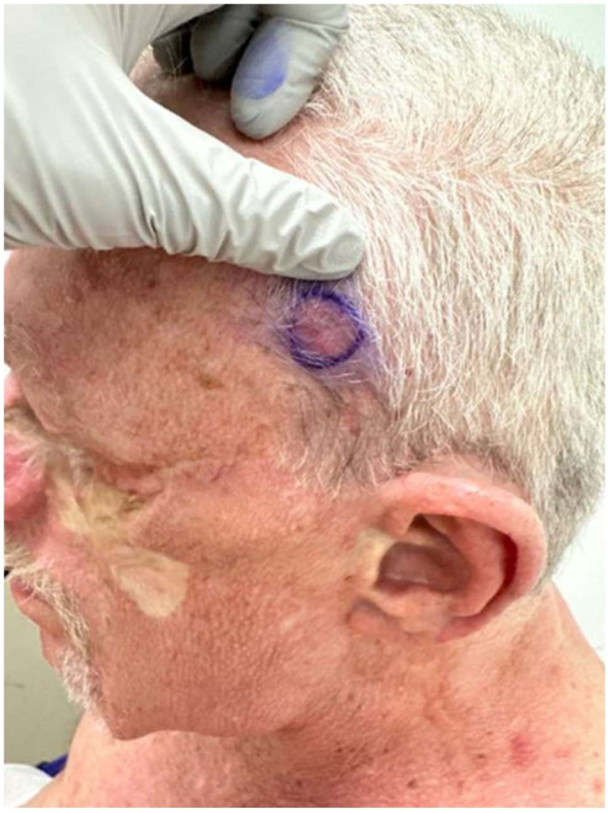
- Huis In 't Veld EA, van Coevorden F, Grünhagen DJ, Smith MJ, van Akkooi ACJ, Wouters MWJM, Hayes AJ, Verhoef C, Strauss DC, van Houdt WJ. Outcome after surgical treatment of dermatofibrosarcoma protuberans: Is clinical follow-up always indicated? Cancer. 2019 Mar 1;125(5):735-741. doi: 10.1002/cncr.31924. Epub 2019 Jan 15. PMID: 30644528.
- Neff R, Collins R, Backes F. Dermatofibrosarcoma protuberans: A rare and devastating tumor of the vulva. Gynecol Oncol Rep. 2019 Jan 25;28:9-11. doi: 10.1016/j.gore.2019.01.006. PMID: 30733992; PMCID: PMC6357686.

Clinical Images:





Pathology Images: