CRTC1::TRIM11-fusion tumor with rare sinonasal presentation and locally aggressive disease course

Adam Bedeir[1], Anthony Crymes[2], Jack Reid[3], Matthew J. Oberley[4], Mark Swanson[2], Mark G. Evans[4]

[1]Basis Phoenix High School, Phoenix, AZ, [2]Keck School of Medicine, University of Southern California, Los Angeles, CA, [3]City of Hope National Medical Center, Duarte, CA, [4]Caris Life Sciences, Phoenix, AZ

Introduction

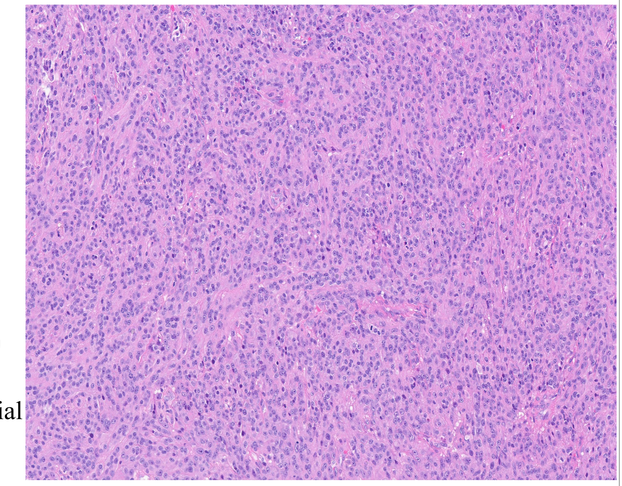
- Cutaneous melanocytic tumor with *CRTC1::TRIM11* fusion (CMTCT) is a rare dermatological neoplasm that has not yet been formally recognized by the World Health Organization.
- At present, our knowledge of this disease is limited to single reports and a series of 41 cases with limited follow-up information.
- It is typically described as a skin lesion expressing melanocytic makers, often leading to its misdiagnosis as melanoma or clear cell sarcoma.
- CMTCT is regarded as an indolent tumor with infrequent recurrence after complete surgical excision.
- However, we present a case with uncommon sinonasal presentation and metastasis after multiple resections.

Clinical History

- A 23-year-old woman presented with a right nasal vestibule mass, requiring partial rhinectomy in a foreign country, where she subsequently received chemoradiation. Her tumor at the time was diagnosed as a mucosal melanoma.
- Eleven years following initial presentation, she demonstrated a recurrent right nasal tumor, as well as neck mass, cervical lymphadenopathy, and lung nodules.
- The patient then underwent a nasal endoscopic resection and modified neck dissection.
- The patient is currently alive and undergoing post-surgical chemoradiation.

Histological Findings

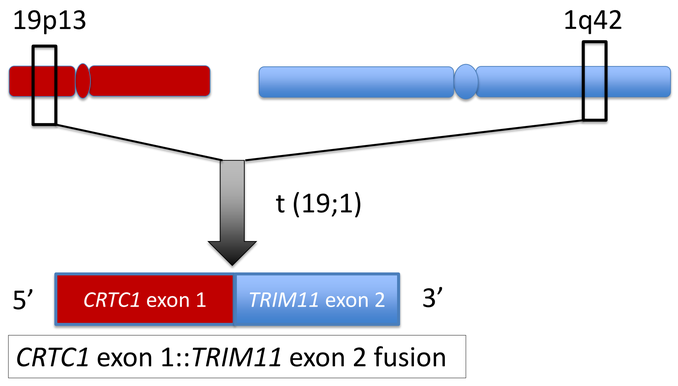
- The sample from the nasal endoscopic resection demonstrated a tumor that was composed of intersecting nests and bundles of cells.
- The tumor cells showed abundant eosinophilic cytoplasm and epithelioid, ovoid, and spindle morphology. The nuclei were vesicular and frequently included a single prominent nucleolus.
- Mitotic activity was low (<5 mitoses per 10 HPFs). Atypical mitoses were rare.
- No tumor necrosis was present in the material evaluated.



Molecular Results

Whole exome and whole transcriptome sequencing identified:

- *CRTC1* exon 1::*TRIM11* exon 2 fusion.
- Pathogenic *TERT* c.-146C>T alteration.



CRTC1 exon 1::*TRIM11* exon 2 fusion

Conclusions

- This case expands our understanding of CMTCT and emphasizes its potentially aggressive behavior.
- The sinonasal presentation contrasts the tumor’s typical appearance on the skin and could mimic mucosal melanoma.
- Of note, our patient developed recurrent disease and distant metastases, thereby emphasizing the possible role of systemic therapy and suggesting that simple surgical excision may not be curative in all cases.
- Our case illustrates that CMTCT is an uncommon and potentially misidentified neoplasm, but that its proper diagnosis is essential for treatment selection and follow up.

References

- Cellier L, Perron E, Pissaloux D, et al. Cutaneous melanocytoma with *CRTC1-TRIM11* fusion: report of 5 cases resembling clear cell sarcoma. Am J Surg Pathol. 2018;42:382–391.
- Bontoux C, Baroudjian B, Le Maignan C, et al. *CRTC1-TRIM11* fusion in a case of metastatic clear cell sarcoma: are *CRTC1-TRIM11* fusion-bearing tumors melanocytomas or clear cell sarcomas. Am J Surg Pathol. 2019;43:861–863.
- Kashima J, Motoi T, Nishimaki M, et al. A case report of cutaneous melanocytoma with *CRTC1-TRIM11* fusion: Is CMCT distinct from clear cell sarcoma of soft tissue?. Pathol Int. 2019;69:496–501.
- Hanna J, Ko JS, Billings SD, et al. A cutaneous melanocytic tumor with *CRTC1::TRIM11* translocation: an emerging entity analyzed in a series of 41 cases. Am J Surg Pathol. 2022;46(11):1457—1466.

Contact

Mark G. Evans, MD (mevans@carisls.com)

M.J.O. and M.G.E. disclose a financial association with Caris Life Sciences (full-time employment, travel/speaking expenses, stock/stock options).