

Objectives

- 1) the importance of molecular analysis in diagnosis and management of atypical vascular neoplasm
- 2) targeted adjuvant medical therapies in combination with surgery can optimize the outcome of treatment.

Case Presentation

A 69-year-old Caucasian female presented with a rapid enlarging violaceous, crusting, pulsatile vascular nodule on the nasal bridge. The lesion first appeared as a small vascular papule and recurred, growing rapidly soon after initial excisional biopsy. The lesion was friable and bled with minimal trauma but was not associated with pain.

Laboratory studies: unremarkable.

Medical and Family History: noncontributory.

Physical Exam: Left nasal dorsum with erythematous violaceous exophitic vascular mass, with surrounding telangiectasia and induration extending to nasal sidewall margin. Left nasofacial sulcus with palpable pulsation.

Histopathology: dermal proliferation of irregularly dilated vessels ramifying throughout the dermis, lined by a single layer of cytologically bland endothelial cells and surrounded by pericytes. Within the deep dermis are fibrin thrombi adjacent to multiple papillary fronds and proliferation of bland endothelial cells. Initial immunohistochemistry staining was WT1(+), indicative of vascular neoplasm. Additional staining with HHV8, PROX1, and GLUT1 were negative. There was no evidence of malignant transformation. Subsequent molecular analysis revealed PIK3CA mutation.

Diagnosis and Treatment

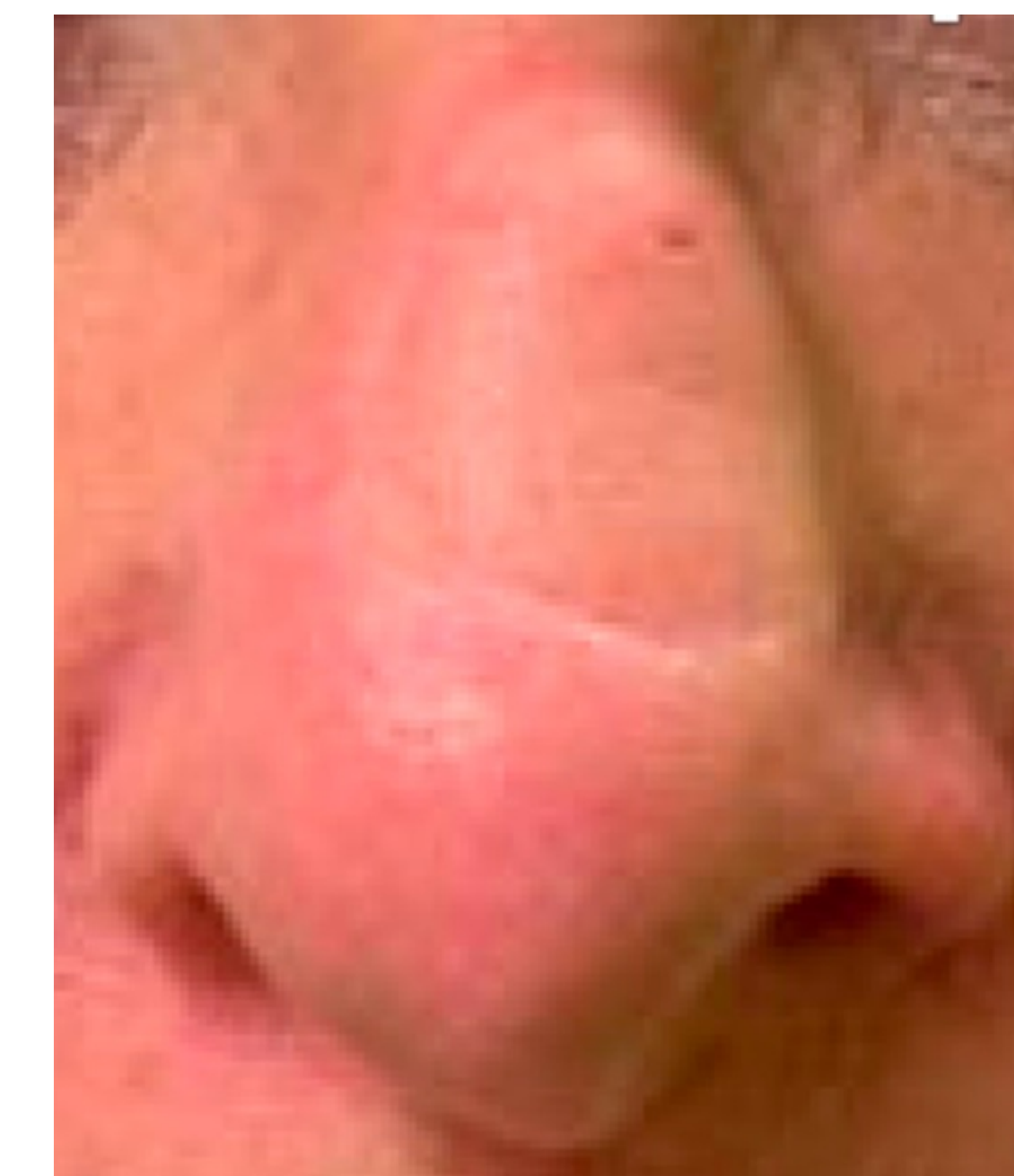
Diagnosis: Vascular Tumor with PIK3CA mutation

Treatment: clobetasol, propranolol, and sirolimus in combination before undergoing complete excision and forehead flap reconstruction subsequently. The patient has remained disease free for more than two years.

Clinical Images



A: Vascular tumor located on the nose prior to excision



B: After excision and flap reconstruction

Discussion and Conclusion

Sirolimus, a mTOR inhibitor, was used as an off label targeted adjuvant therapy to reduce vascular proliferation prior to surgical intervention and continued after excision to prevent local recurrence. It has been found in clinical trials to significantly reduce the growth of vascular tumors but because of the potential for severe adverse effects, patients must be monitored closely. Although PIK3CA mutations are commonly associated with solid tumors in adults and with vascular anomalies in children, de novo formation of a vascular tumor with PIK3CA mutation in adults is rare. Additionally, diagnosis can be challenging because of the varying phenotypes and tissue mosaicism that may present with PIK3CA mutations. The use of molecular analysis to capture genetic variants in rare tumor can facilitate proper management and optimize surgical outcome.

References

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Disclosures

The authors have no relevant disclosures.