Spontaneous near fatal hemorrhage in neurofibromatosis type 1 of the scalp

Edwin Stephen*, Rajeev Kariyattil, Alok Mittal, Faisal Al-Azri, and Khalifa Al-Wahaibi

Senior Consultant, Vascular Surgery, Department of Surgery, Sultan Qaboos University Hospital

Received: 23 June 2020
Accepted: 20 October 2020

***Corresponding author:** *edwinmay2013@gmail.com*

DOI 10.5001/omj.2022.02

Abstract
Type 1 Neurofibromatosis (von Recklinghausen's disease) is an autosomal dominant disorder characterized by café-au-lait spots, pigmented hamartomas of the iris and multiple neurofibromas. Patients can present with hemorrhage secondary to trauma or rarely with spontaneous hemorrhage, both of which can be lethal and life-threatening. We report a 14 year old girl with a rapidly expanding hematoma in a rare presentation of spontaneous bleed into her NF1. She needed emergency coiling of the left maxillary artery and branches, to successfully arrest bleeding and resuscitate her from hemorrhagic shock.

Introduction
Neurofibromatosis is also known as Von Recklinghausen's disease [VRD] and usually involves organs of ectodermal origin – the nervous system, eyes and skin. [1]There are three known variants – Type 1 Neurofibromatosis [NF1], Type 2 Neurofibromatosis, and schwannomatosis. [2]. Bleeding into a NF-1 lesion can be spontaneous or secondary to trivial trauma and can be fatal if not diagnosed and managed expeditiously.

Case summary
A 14 year old girl presented to our emergency department in the early hours of the morning with a rapidly expanding swelling in the left hemi-scalp /face, of her previously diagnosed NF1- Figure 1. There was no definitive history of trauma. She was fully alert and oriented in time, place and person. A spontaneous bleed into the NFI was suspected and an emergency CT angiography [CTA] performed [Figure-2]. About 2 hours after her presentation, the swelling suddenly increased in size, she became hypotensive and hemoglobin dropped from 11.7 gm% to 6.6 gm%. An urgent digital subtraction angiography [DSA] was performed via a right trans-femoral artery access, while resuscitation measures were ongoing with the anesthesia team in the hybrid operating room. Following coiling of the left maxillary artery and branches [Figure 3] there was no further increase in the size of the swelling. She was discharged three days later with an appointment to see the plastic surgery team to assess possibility of excision of the NF1.

Figure 1 Neurofibromatois left hemi-scalp and face

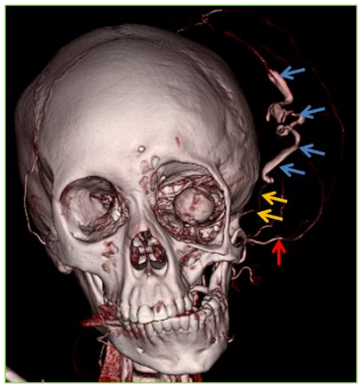

Figure 2 CTA 3D volume rendered CTA image reveals active arterial contrast extravasation seen as jet of contrast (Blue arrows) from left maxillary artery branch (Yellow arrow) . Note that there is significant displacement of left sided superficial temporal artery (red arrow) along with galeal tissue.

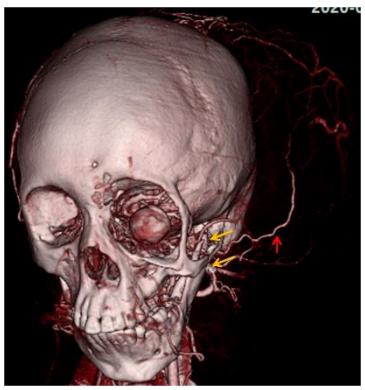

Figure 3 3D volume rendered CT Angiographic image acquired after angioembolization revealed absence of active arterial contrast extravasation with metallic coils (Yellow arrows) along the course of maxillary artery branch, however; persistent displacement of left sided superficial temporal artery (red arrow) and galeal soft tissue noted.

Discussion

NF1 is a rare autosomal dominant neurogenic disease characterized by café-au-lait spots, pigmented hamartomas of the iris, skeletal deformities and multiple neurofibromas. The incidence ranges from 1 in 2500 -3500 individuals with about fifty percent of these patients having a family history of the disease. The genetic mutation is of the NF1 tumor suppressor gene located on chromosome 17q11.2.Pathogenesis is uncertain with no sex or racial predilection. Learning disability and malignant transformation are known to be associated,as well. [2, 3, 4, 5] A friable vascular tree is thought to be secondary to invasion of the vessel wall by the neurofibroma and or arterial dysplasia. The neurofibromatous tissue itself has thin walled ectatic blood vessels which lie within the loose neural stroma that has replaced the normal adipose tissue. [5] Bleeding is seen usually in the intrathoracic, gut (GIST), or subcutaneous and less commonly in the brain or retroperitoneal tissue. Vascular lesions are usually diagnosed when patients have pain or a hemorrhage which at times can be fatal. [6, 7]

Multidimensional imaging using 3 / 2D ultrasound and CTA / DSA plays a crucial role in assessing the source of bleed and planning intervention. [8] Neuroimaging findings in patients with NFI include focal areas of increased signal intensity on T-2 weighted imaging, called “unidentified bright objects” and may disappear in adulthood; increased brain volume and abnormal cerebral vessels in 2 -6% of patients with NF1.[9]

Spontaneous hemorrhage into the scalp, face and skull base, as in our case is rare. There is a need to expedite diagnosis and intervention to avoid airway compromise and prevent compression of vascular and orbital structures. [10] Open surgical repair is likely to be catastrophic due to accompanying skeletal deformities. [3]

In our patient the sphenoid bone was absent on the affected side, the maxillary artery through this bony defect was bleeding, compounded with the fragility of the vasculature. Hence, achieving proximal control would be difficult /challenging with open surgery. She was shifted to the hybrid operating room and via a right trans-femoral access, the left maxillary artery and its branches emoblized with coils, successfully.

The hardware used for the endovascular procedure was - 5F arterial sheath (Radiofocus, Terumo Corporation, Japan) ; 0.035” Hydrophilic guidewire (Radiofocus, Terumo Corporation, Japan); 5F, 100 cm shaft H1 guiding catheter (Cook Medical, USA);
2.8 F, 130 cm shaft, Progreat Microcathter with (Terumo Corporation, Japan);
3x30 mm, 0.018’ Hilal embolization microcoils (Cook medical, Denmark).

She required blood transfusions during and after the procedure. At discharge, day 3, post procedure, she was to see the plastic surgery team for suitability of excision of the lesion as a combined effort with the neurosurgical and vascular surgery team/s.

Conclusion

NF1 is a rare disorder that can present with catastrophic bleeds due to the fragile nature of the vasculature. Spontaneous scalp bleeds are rare in this subset of patients. Urgent imaging in the form of a CTA and or DSA with emolization can be lifesaving.

Acknowledgement – The authors thank the parents for permitting use of the clinical picture for purposes of teaching and publication.

Conflict of interest - None

References

1. Jouhilahti, E. M.; Peltonen, S.; Heape, A. M. & Peltonen, J. The pathoetiology of neurofibromatosis 1. Am. J. Pathol. 2011; 178(5):1932-9
2. Korf, B. R. Neurofibromatosis. Handb. Clin. Neurol. 2013; 111:333-40
3. Qin H, Chen W, Kong J, Chen F, Wen X, Li Z, Zhang L. Tratamiento quirúrgico de un neurofibroma masivo de cabeza y cuello en un paciente con neurofibromatosis tipo 1: Reporte de un Caso. Int. J. Morphol. 2018;36(1):194-200
4. Zhu, L.; Shi, L.; Wang, B.; Bi, M.; Pu, J.; Zhang, L.; Zhang, Y.; Wang, X. & Zhang, G. Novel mutations in one allele in a Chinese family with neurofibromatosis type 1: Including a complex insertion-deletion mutation. J. Dermatol. 2016; 43(11):1332-5
5. Sung HB, Ji HK, Jun SK, Seung BH, Jung SC, Yong HY, Lucia K. Recurrent massive subcutaneous haemorrhage in neurofibromatosis type 1: A case report. J Korean Med Sci. 2007 Aug;22(4):728-730
6. Moerbeek, Patrick & Van Buijtenen, Jesse & Heuvel, Baukje & Hoksbergen, Arjan. (2015). Fatal Retroperitoneal Bleeding Caused by Neurofibromatosis: A Case Report and Review of the Literature. Case reports in medicine. 2015. 965704. 10.1155/2015/965704. [Research gate, accessed 09 June 2020]
7. G. S. Oderich, T. M. Sullivan, T. C. Bower et al., "Vascular abnormalities in patients with neurofibromatosis syndrome type I: clinical spectrum, management, and results, Journal of vascular surgery. 2007;46(3): 475–484
8. Ximena W, Nelson L, Rodrigo DP, Laura C. Multidimensional ultrasound and computed tomography imaging support in bleeding plexiform neurofibromatosis of the scalp: A case report and literature review. Indian J Dermatol. 2015 Jul-Aug; 60(4): 421-424
9. Korf BR, Lobbous M, Metrock LK. Neurofibromatosis type1 (NF1): Pathogenesis, Clinical features, and diagnosis. June 03,2020 [Accessed on 5th October 2020]
10. Svider PF, Patel CR, Lamba S, Gandhi C, Baredes S, Jyung RW. Massive, spontaneous facial hematoma in a neurofibromatosis type 1 patient. International Journal of Otolaryngology and Head & Neck Surgery, 2013, 2, 17-20