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Case Report

Spontaneous Intracranial Bleed in a Young Non-hypertensive Patient of Neurofibromatosis Type 1

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ABSTRACT

Neurofibromatosis type 1(NF1), is an autosomal dominant inherited disorder characterized by numerous cutaneous neurofibromas, intertriginous freckling, dermal and plexiform neurofibromas and cafe-au-lait spots1.Cerebrovascular disease in the setting of NF 1 is a rare entity. Among the rare cerebrovascular abnormalities, the most common is occlusion of the small cerebral arteries leading to infarcts. Intracranial aneurysms are quite uncommon with only few cases reported worldwide. It is hypotheised that the pathogenesis may be attributable to the proliferation of Schwann cells and the subsequent degeneration in the adjoining vessel wall. We hereby report an uncommon case of NF1 associated with massive intracerebral haemorrhage most likely caused by vessel wall rupture.

KEYWORDS: Cafe-au-lait spots, Lisch nodule, Plexiform neurofibromas

CASE REPORT

A 44 year old normotensive and nondiabetic male patient diagnosed case of Neurofibromatosis type 1 presented in our casualty with sudden onset right sided weakness and speech difficulty associated with severe headache. On admission, the patient was fully conscious and oriented with motor weakness of right half of body and right-sided facial weakness. There was no history of similar event in past nor was any non modifiable risk factor for the same. No visual or sensory symptoms were present. Widespread cutaneous neurofibromas and angiomas were found along with axillary freckling and cafe –au-lait spots over trunk (Figure 1). Ophthalmological evaluation showed presence of Lisch nodules (Figure 2). Noncontrast CT of brain revealed a significant amount of bleed in left gangliocapsular region with slight oedema. Surprisingly the blood pressure and fundoscopy was also normal for the patient. He did have a family history of NF1 with his son also a diagnosed case of NF1 with Moya Moya disease. On subsequent investigations routine blood parameters were normal and further MR angiography of intracranial vessels revealed similar findings without any aneurysm or malformation. The patient was managed conservatively for the raised intracranial pressure and responded well to treatment. Neurodeficit improved partially with time and is still under follow up. MR angiogram did not reveal any intracranial aneurysm of blockage of arteries which leads us to a conclusion of source of bleed to be either a vascular malformation or episodic hypertensive bleed.

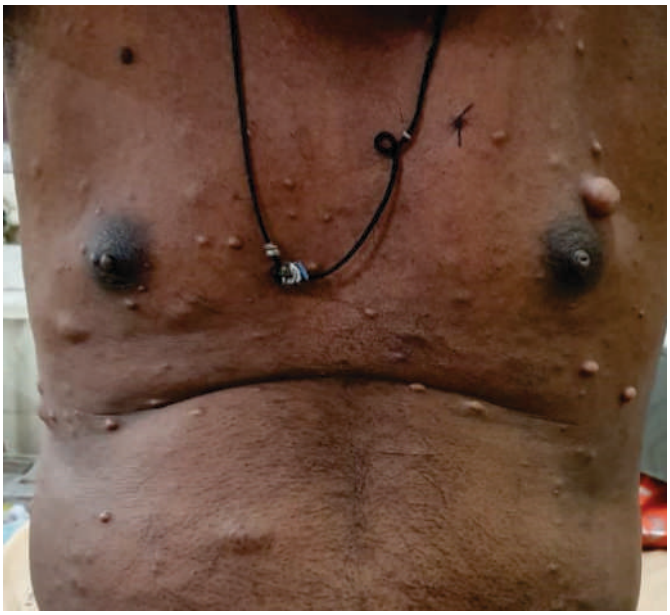

Figure 1: Multiple Neurofibromas and plexiform neuromas

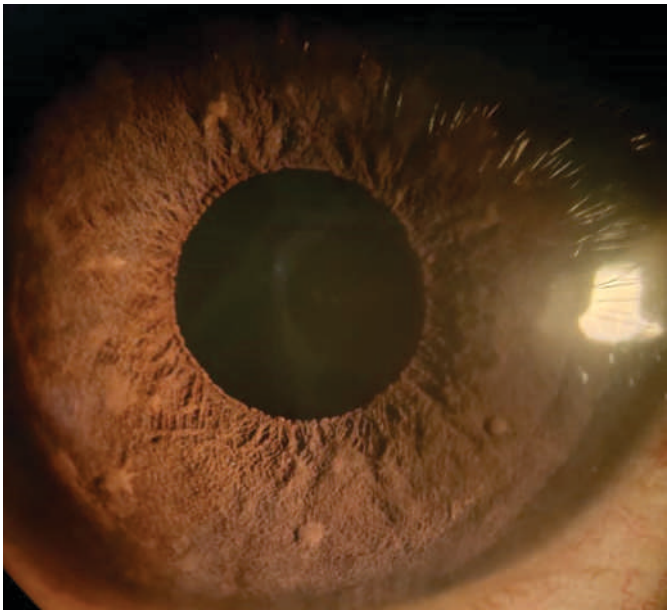

Figure 2: Lisch nodules over iris as seen on slit lamp examination

DISCUSSION

Neurofibromatosis type I (NF-1) is a complex multi-system disorder caused by the mutation of NF1 gene on chromosome 17 which is responsible for production of protein neurofibromin. NF-1 causes tumors of the nervous system which can grow anywhere in the body. It is an autosomal dominant disease and one of the most common mendelian disorders[2]. Clinical hallmarks include hamartomas on iris called as Lisch nodules, benign skin tumors called neurofibromas, multiple café au lait spots and large benign tumors of nerves called plexiform neurofibromas. Central nervous system symptoms include scoliosis, learning difficulties, visual symptoms and rarely seizures. NF-1 affected individuals also have a much higher incidence of

cardiovascular disease than the population in general. The three most common cardiovascular manifestations of NF1are vasculopathy, hypertension, and congenital heart defects[3].

The primary neurologic involvement in NF-1 is of the peripheral nervous system, and rarely that of the central nervous system. Sørensen *et al* found that cerebrovascular accidents often occurred at a younger mean age than other patient[4]. Hypertension is significantly associated with higher mortality, and the mean age of death among NF1 patients is almost 14 years younger than expected[5]. Cerebrovascular accidents in NF1 patients usually arise from occlusions of the internal carotid, middle, or anterior cerebral artery[6]. In rare incidences telangiectatic vessels form around the area of the stenosis and appear as a "puff of smoke" ("Moya-Moya") on cerebral angiography which is the case with the only child of this patient. Intracranial aneurysms and arteriovenous fistulae may also be seen in NF1 patients but are very rare and mostly in elderly population[7].

NF1 patients who present with a neurological deficit of sudden onset at any age should be evaluated promptly for cerebrovascular disease either occlusive of haemorrhagic. Cerebral angiography is appropriate when the history and Computed tomography is indicative of vascular lesion. It is noteworthy that many reported cases of neurofibromatosis with cerebral occlusive disease reveal stenotic or telangiectatic involvement of mostly middle or anterior cerebral arteries[8]. Some anecdotal reports also describe aneurismal bleeding in these patients[9]. Hypertension is a common cause of cerebrovascular mortality in NF1 patients but we could not find any evidence of high BP in this patient or any family history of same. This leads us to a conclusion of possibility of any vascular malformation or agenesis leading to deep cerebral bleed in this case. Treatment of in individuals with NF1 is identical to that in other CVApatients including both surgical and medical options. This opens up further possibilities of preliminary diagnostic screening in such patients to rule out any high risk lesion in both cardiovascular and cerebral vasculature and correction of the same medically or surgically.

CONCLUSION

Neurofibromatosis patient are prone to cerebrovascular complications in form of both vasoocclusion and parenchymal bleeding leading to neurodeficit or coma at a relatively young age. Active screening with neuro-imaging along with strict blood pressure monitoring should be the protocol to avoid morbidity and mortality in these patient groups.

CONFLICT OF INTEREST: None

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REFERENCES

1. Friedman JM, Gutmann DH, MacCollin M, Riccardi VM. *Neurofibromatosis: phenotype, natural history, and pathogenesis*, 3rd ed. Baltimore: Johns Hopkins University Press, 1999.
2. Rasmussen SA, Friedman JM. NF1 gene and neurofibromatosis 1. American journal of epidemiology. 2000; 151(1):33-40.Friedman JM. Epidemiology of neurofibromatosis type 1. *Am J Med Genet* 1999; 89: 1–6.
3. Friedman JM, Arbiser J, Epstein JA, Gutmann DH, Huot SJ, Lin AE, Mcmanus B, Korf BR. Cardiovascular disease in neurofibromatosis 1: report of the NF1 Cardiovascular Task Force. Genetics in Medicine. 2002; 4(3):105-11.
4. Sørensen SA, Mulvihill JJ, Nielsen A. Long-term follow-up of von Recklinghausen neurofibromatosis: survival malignant neoplasms. *N Engl J Med* 1986; 314: 1010–1015.
5. Zöoller M, Rembeck B, Åkesson HO, Angervall L. Life expectancy, mortality prognostic factors in neurofibromatosis type 1: a twelve-year follow-up of an epidemiological study in Göoteborg, Sweden. *Acta Derm Venerol* 1995; 75: 136–140.
6. Zochodne D . Von Recklinghausen's vasculopathy. *Am J Med Sci*1984; 287: 64–65
7. Schievink WI, Michels VV, Piepgras DG. Neurovascular manifestations of heritable connective tissue disorders. A review. *Stroke* 1994; 25: 889–903.
8. Jat, Kana Ram, et al. "Neurofibromatosis type 1 with intracranial haemorrhage and horseshoe kidney." *Paediatric neurology* 2008; 39 (4): 295-297.
9. Sasaki, J., et al. "Neurofibromatosis associated with multiple intracranial vascular lesions: stenosis of the internal carotid artery and peripheral aneurysm of the Heubner's artery; report of a case." *No shinkei geka. Neurological surgery* 1995; 23 (9): 813-817