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# Case Report

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## Brachium Pontis Stroke Revealing Neurofibromatosis Type-2

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### ABSTRACT

NF2 is an autosomal dominant disorder with neuroectodermal dysplasia. Most patients present with characteristic clinical tumors during or beyond the adolescent age group. The diagnosis is mainly clinical. Vasculopathy is rarely associated with NF2. Vascular complication as the presenting symptom in NF-2 is unknown. We report a case of a 2-year-old child with no prior family history of neurofibromatosis presenting with ataxia and brain-stem stroke.

**Keywords:** Neurofibromatosis type 2, vasculopathy, brain stem stroke.

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### Introduction

NF1 and NF2 are autosomal dominant disorders characterized by dysplasia of neuroectodermal tissues and mesodermal dysplasia.<sup>1</sup> Multiple cases of vasculopathy associated with NF1 including moya moya disease and infarct as well as renal artery stenosis have been reported. However, vascular insult associated with NF2 is extremely rare and infarct as the presenting symptom in a child under 5 years has never been reported. Our case demonstrates the role of complete imaging workup and magnetic resonance (MR) spectroscopy in the early diagnosis of NF2 given the bizarre clinical presentation and unusual conglomeration of imaging findings.

### Case Report

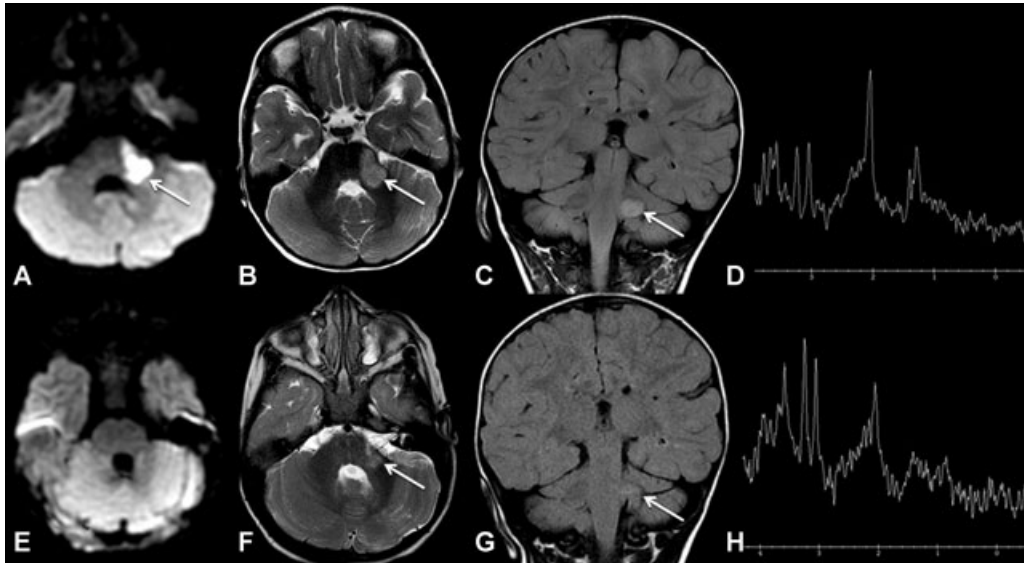
A 2-year-old girl presented with a history of fever and cough for 1 week and acute onset of ataxia for 2 days. Her parents reported that she had several falls at home. On examination, the patient was irritable but interactive. She had significant ataxia on exam. The skin examination revealed several café au lait spots over her left lateral chest wall, around 2 mm each. MR brain disclosed calcified lesions in the left cerebellar hemisphere and an intraaxial T2 hyperintense tumor-like lesion in the left brachium pontis (Figs 1A–D). Of note is the presence of restricted diffusion in this lesion (Figs 1A and 2). An elevation of lactate was noted on MR spectroscopy (Fig 1D). Findings were highly suggestive of an infarct. MR angiography was unremark-

able and had no evidence of dissection or arterial narrowing. MR spine revealed a cervical cord lesion with a focus of enhancement (Fig 3A) likely representing an ependymoma. Multiple lumbar nerve roots were enlarged being most prominent at L3–L4 (Fig 3B). L3 spinal lesion was subsequently biopsied, revealing schwannian proliferation with disruptive nerve fibers, felt to be representative of schwannoma.

Based on these spinal and brain lesions as well as café au lait spots on the skin, a diagnosis of NF1 with likely brain stem infarct was suggested. Genetic testing for NF1 mutation was unremarkable. Genetic testing for NF2 revealed a heterozygous truncating mutation in the NF2 gene (c.169 C>T), thus confirming diagnosis of NF2. At 2.5-month follow-up, the brain-stem lesion had been replaced by a focus of gliosis as expected for an infarct (Figs 1E–H). Spinal cord lesion is under regular surveillance and a decision was taken not to biopsy it presently.

### Discussion

NF2 is inherited as an autosomal dominant disorder, caused by inactivating mutations of the NF2 gene in chromosome 22q12. Because detection of tumors at an early stage is effective in improving the clinical management of NF2, presymptomatic genetic testing is an integral part of the management of NF2 families. The disorder occurs in 1:25,000; however, disease prevalence is on the rise due to earlier diagnosis.<sup>2</sup> The



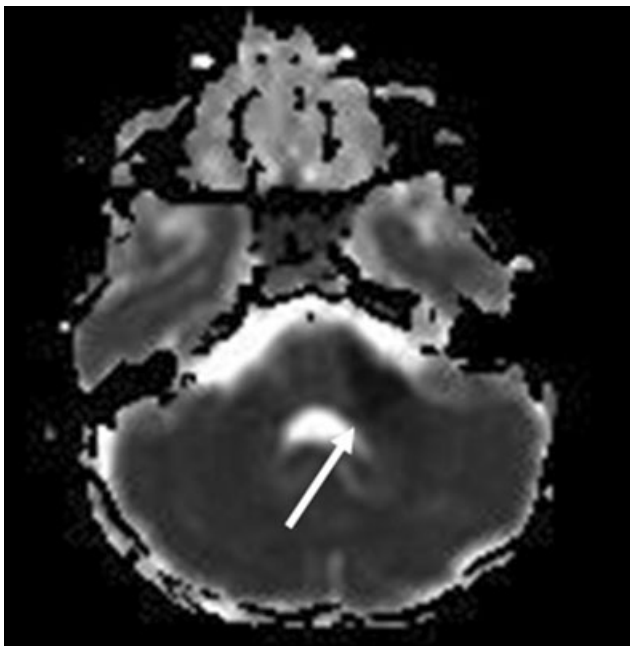
**Fig 1.** (A–D) A lesion demonstrated in the left brachium pontis shows restricted diffusion (A, arrow), and T2 prolongation (B, C, arrows). Elevated lactate peak and decreased NAA peak are seen on MRS (D). (E–H) 2.5-month follow-up. Resolution of the restricted diffusion (E), and almost complete resolution of T2 and FLAIR prolongation with residual gliosis (F, G, arrows) are noted. Follow-up MRS shows myo-Inositol (ml) and Cho peaks with decreased n-Acetyl aspartate (NAA) confirming gliosis.

original National Institute of Health (NIH) criteria have been expanded to include patients with no family history, who have multiple schwannomas or meningiomas, but who have not yet developed bilateral eighth nerve tumors.<sup>3</sup> Although bilateral vestibular schwannoma is classical, more than 50% of patients present with other symptoms. Varied presenting features are more common in children.<sup>4-6</sup>

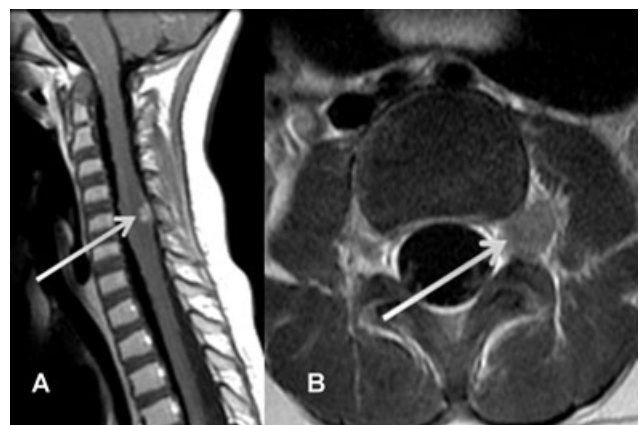
Hence, there is a need for modification of the criteria for diagnosis and a high index of suspicion in the pediatric age group.

### Manifestations in Children

The pattern of presentation and the natural history of NF2 in childhood differ from adulthood in many aspects.<sup>7</sup> The most common initial symptoms in adult onset NF2 are usually attributed to eighth cranial nerve dysfunction. However, in the youngest NF2 age group subtle skin tumors, small posterior capsular cataracts or neurological signs secondary to other than vestibular schwannomas and/or brain-stem or spinal cord compression are more common and manifest long before



**Fig 2.** ADC Map at the level of the lesion shows decreased ADC values consistent with cytotoxic edema.



**Fig 3.** Postcontrast T1-weighted sagittal image through cervical spinal cord shows cord expansion with tumor showing enhancing focus posteriorly (A, arrows). Postcontrast T1-weighted axial image through lower lumbar spine shows enhancing L3/L4 nerve root schwannoma (B).

dysfunction of cranial nerve VIII. Thus, the first sign of more severe multitumor disease in early childhood is often a non-eighth cranial nerve tumor. Vestibular schwannomas account for 15-30% of initial presentation in pediatric NF2.<sup>7</sup> Our case also had calcification in the cerebellar hemisphere, which although unusual has been reported with NF-2 previously.<sup>3</sup>

### Vasculopathy in NF-2

Vasculopathy is known in NF1. However, vasculopathy in NF2 is rare and only five cases have been described so far. Vascular complication as the presenting symptom of NF2 has never been described to the best of our knowledge. The cases of vascular dysplasia reported include a case of left middle cerebral artery stroke,<sup>1</sup> brain stem stroke in a known NF2 adolescent, aneurysm of middle meningeal artery, and renal artery dysplasia.<sup>1</sup>

### Role of MRI

Based on the conglomeration of findings the diagnosis of possible neurofibromatosis was made. The lesion in the brain stem was the stumbling block and was difficult to differentiate from a glioma purely based on the standard imaging findings. Inclusion of diffusion weighted imaging and magnetic resonance spectroscopy interrogation of the lesion in the regular MR protocol helped in the confident diagnosis of stroke over tumor.

### Conclusion

Our case being the first reported case of infarct as presenting symptom in NF2 makes it intriguing and highlights the importance of a complete neuroradiological evaluation in the pediatric age group. In view of our case and previous reported cases of NF-2 associated with vasculopathy, this rare complication of NF-2 should be kept in mind, particularly in the pediatric age group.

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