Case Report

# Aqueductal Web Causing Triventricular Hydrocephalus in Neurofibromatosis-1

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A B S T R A C T

**Introduction:** Triventricular obstructive hydrocephalus in neurofibromatosis 1 occurs in 1-5% of cases. Cause of obstruction is brainstem glioma, periaqueductal and tectal hamartoma, aqueductal web and superior vellum medullary synechiae. Aqueductal web is a rare cause of hydrocephalus in NF1. Thin T2 (CISS) sequence in the sagittal plane is needed for its diagnosis.

**Case report:** We report a case of 12- year old male presenting with frontal headache and blurring of vision with a family history of NF1. MRI of brain showed triventricular hydrocephalus with normal sized fourth ventricle with periventricular CSF ooze. Focal hamartomas were noted in right globus pallidus, pons and medulla. Sagittal thin T2WI (CISS) sequence revealed a thin hypointense web in the inferior portion of the aqueduct of Sylvius.

**Conclusion:** Hamartomas are seen as focal areas of high signal intensity in up to 93% of cases of NF1. These are usually seen in basal ganglia, brain stem and cerebellar peduncles.

**Keywords:** Neurofibromatosis, Hydrocephalus, Hamartoma, Aqueduct, Web.

## INTRODUCTION

Neurofibromatosis-1 clinically presents as skin lesions, increase in size of head, learning disabilities, visual disturbances, bone deformities with short stature. Neurofibromatosis-1 is a multi-system affection. Neuroimaging is required to diagnose complications like hydrocephalus visual disturbances, gliomas and hamartomas.

1 to 5% cases of neurofibromatosis-1 show obstructive hydrocephalus. Cause of obstruction is brainstem glioma, periaqueductal and tectal hamartomas, aqueductal web and superior vellum medullary synechiae.[1]

Aqueductal web is a rare cause of hydrocephalus in neurofibromatosis 1.[2] High resolution thin sections of the aqueduct are needed in cases of obstructive hydrocephalus to detect aqueductal web. Hamartomas do not need surgical treatment. Endoscopic third ventriculostomy can treat obstructive hydrocephalus. Diagnosis by imaging followed by early treatment can increase life expectancy as neurofibromatosis can reduce life expectancy up to 15 years.[3] Differential diagnosis is aqueductal stenosis and superior vellum medullary synechiae.

## CASE REPORT

A 12- year old male presented with frontal headache and blurring of vision since 15 days . There was history of 4 episodes of non- projectile vomiting for 2 days. On examination, the patient had neck rigidity. He had multiple cafe au-lait spots on the trunk. There was history of neurofibromatosis in his father. Developmental history was normal. The patient complained of pain in the left calf region since one month, gait instability since 6 months and swelling over left calf since 10 years, which was progressively increasing.

MRI brain showed mild to moderate dilatation of both lateral ventricles and the third ventricle with periventricular CSF ooze. Fourth ventricle was not dilated. A thin hypointense web was noted in the inferior portion of the aqueduct of

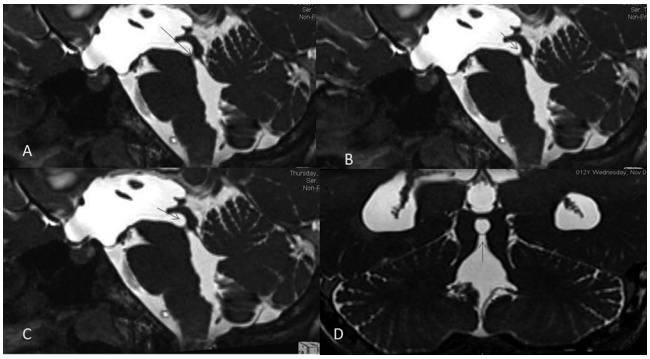


**Figure-1:** Thin T2 (CISS) sagittal (A-C), coronal (D) showing thin hypointense web in aqueduct.

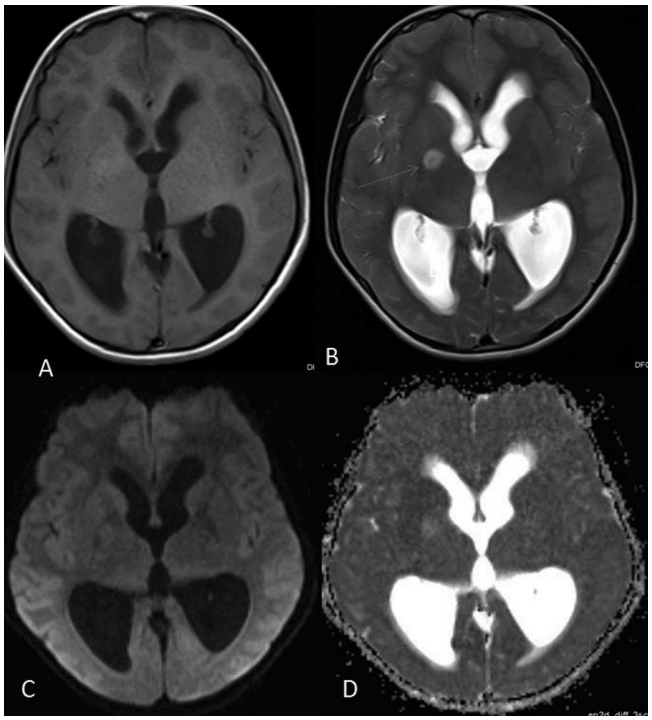


**Figure-2:** MRI brain showing hamartoma in right globus pallidus appearing isointense on T1WI (A), hyperintense on T2WI (B), not showing restriction on DWI(C,D).

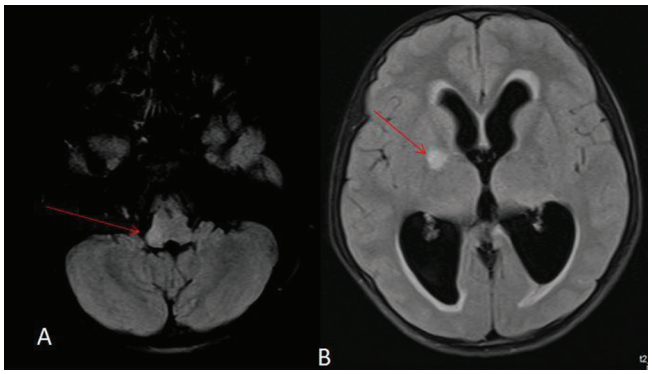


**Figure-3:** MRI brain showing hamartoma in medulla on right side (A) and right globus pallidus(B) appearing hyperintense on FLAIR.

Sylvius, best appreciated on 3D CIS images with dilatation of superior part of aqueduct. A focal hyperintense lesion was noted in right Globus pallidus on T2 and FLAIR showing no diffusion restriction and post contrast enhancement. Subtle T2 and Flair hyperintense signal without diffusion restriction and enhancement were noted in the medulla on right side and in dorsal pons. The findings were suggestive of aqueductal web causing proximal obstructive hydrocephalus and hamartomas in medulla, pons and right globus pallidus.

Local USG of left calf showed multiple fusiform shaped solid lesions of variable sizes in subcutaneous fat planes on postero-lateral aspect of the left leg. These appeared heterogeneous in echotexture along the course of cutaneous nerves and within calf muscles. Sciatic nerve in both thighs was thickened and nodular.

MRI of left thigh showed thickening of subcutaneous fat

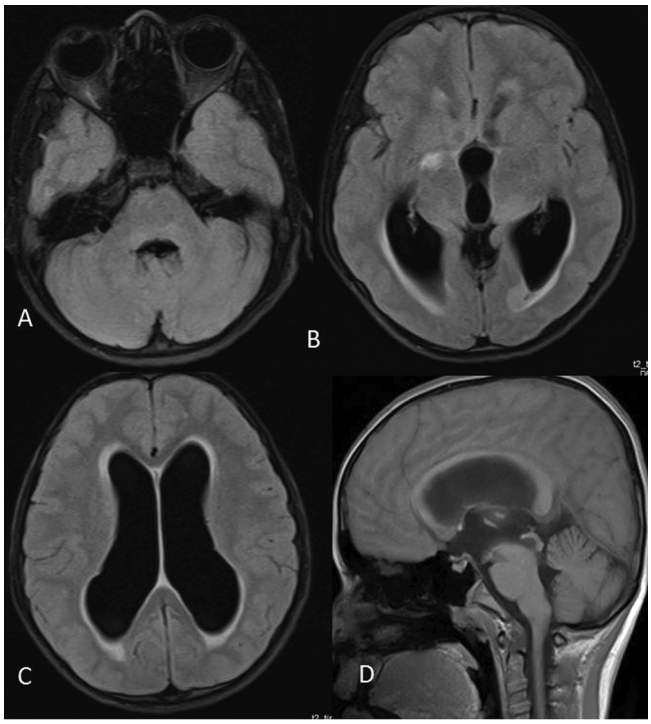


**Figure-4:** MRI brain (Axial FLAIR: A-C, Sagittal T1- D) showing normal sized fourth ventricle(A), dilated third (B) and both lateral ventricles(C) and dilated superior portion of aqueduct due to web in inferior portion of aqueduct (D).

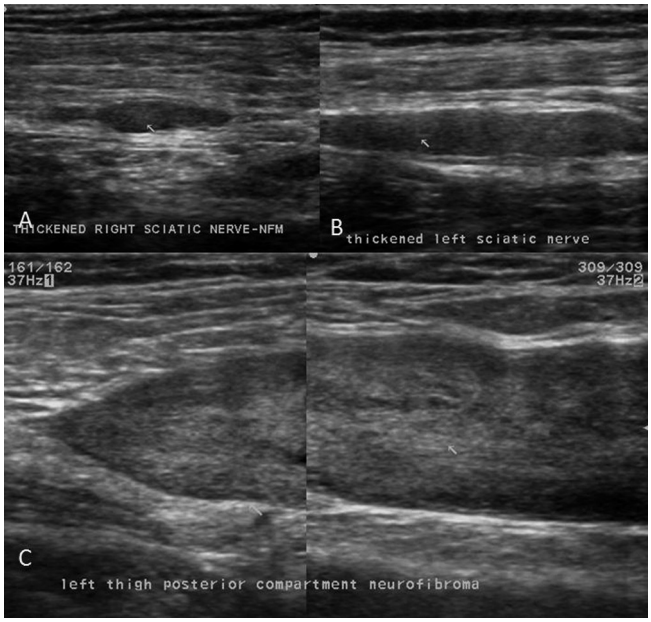


**Figure-5:** (A-C): High resolution ultrasound of posterior compartment of both thighs showing thickening of sciatic nerves due to mixed echoic neurofibromas.

on postero-lateral aspect of the left leg with overlying skin thickening. Multiple lesions of varying sizes appearing hypointense on T1 WI and hyperintense on T2WI and STIR were noted along the cutaneous nerves of left lower limb predominantly along the postero-lateral aspect of the leg in subcutaneous fat plane suggestive of cutaneous neurofibromas. Left sciatic nerve was markedly thickened and nodular appearing hypointense on T1 and hyperintense


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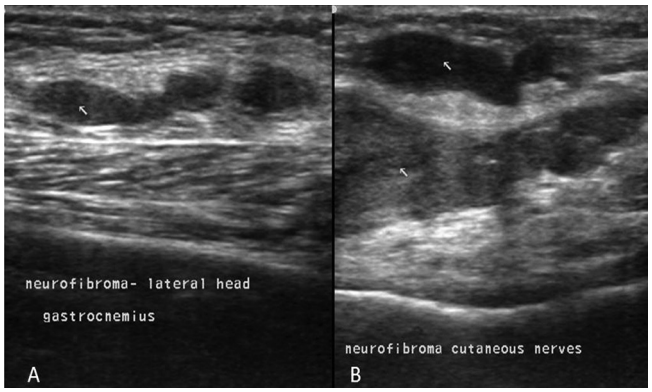


**Figure-6:** (A,B): High resolution ultrasound of both legs showing thickening of cutaneous and muscular nerves due to mixed echoic neurofibromas.

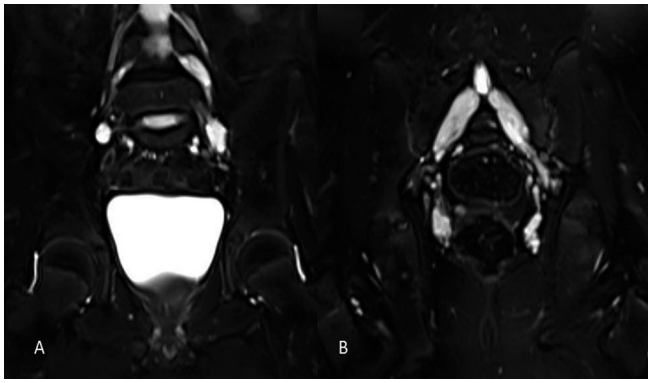


**Figure-7:** (A,B): Coronal STIR lumbosacral plexus showing thickening of lumbar and sacral nerves due to neurofibromas on either side.

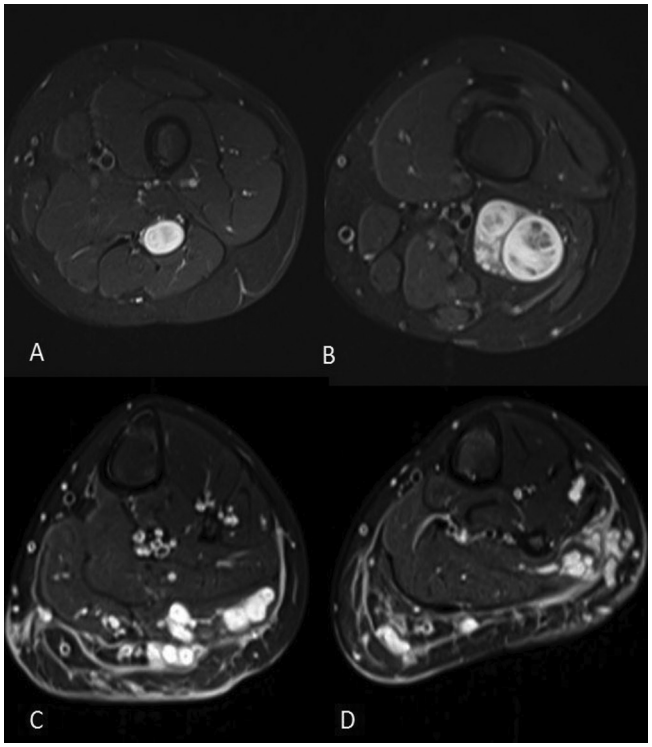


**Figure-8:** Axial STIR - (A, B): left thigh showing thickening of sciatic nerve due to neurofibromas. (C, D): left leg thickening of cutaneous and muscular nerves due to multiple neurofibromas.

on T2WI and STIR. Lower dorsal, lumbar and sacral nerves were thickened and nodular at places. Muscles and bones appeared normal.

## DISCUSSION

Neurofibromatosis occurs in one in every 2000 to 3000 births.[4] CNS manifestations of neurofibromatosis are cranial nerve schwannoma, meningiomas, hamartomas, gliomas, pilocytic astrocytoma ependymomas and angiomas.[4-8]

Both superficial and deep neurofibromas can occur in neurofibromatosis 1. Distribution and characterization of superficial neurofibromas differ from deep neurofibromas. Superficial neurofibromas extend through the skin surface are usually unilateral and asymmetric and tend to be more diffuse rather than nodular and fascicular. The nodules and fascicles are smaller. The signal characteristics can be homogeneous or heterogeneous without targets. They have smaller average lesion volume than their deeper counterpart and tend to have slower growth. Deep neurofibromas have target like appearance on MRI. Microscopically plexiform neurofibroma consists of nerve fascicles or nerve distended by tumor cells embedded in a rich myxoid matrix. Longitudinal bundles of residual nerve fibres are centrally situated in the neurofibroma. This results in central T2 dark nerve fibres and peripheral T2 bright myxoid appearance of target like lesions.

Superficial neurofibroma histologically shows spindle cells infiltrating around normal structures like skin adnexa, adipose tissue and blood vessels.[9]

Hamartomas are seen as hyperintense areas on T2WI. These are seen in 93% of patients with neurofibromatosis 1.[10] No mass effect or edema or contrast enhancement is noted. These are usually seen in basal ganglia, brain stem and cerebellar peduncles. The exact nature of hamartomas is unknown. Hamartomas adjacent to the cerebellar aqueduct or the fourth ventricle inlet can cause hydrocephalus in the course of neurofibromatosis 1. Hydrocephalus in neurofibromatosis 1 has an incidence of 1-5%.[11-15]

Brainstem or cerebellar hamartomas seen adjacent to the aqueduct of Sylvius or fourth ventricle inlet in the absence of hydrocephalus should be followed up at regular intervals for up to 12 years due to the proliferative nature of hamartomas in neurofibromatosis 1. Presence of triventricular hydrocephalus in neurofibromatosis 1 should be evaluated by conventional sequences and thin T2 (3D CISS). Particular attention should be given to the cerebellar aqueduct, fourth ventricle, brain stem and cerebellum. The causes of hydrocephalus in neurofibromatosis 1 are- 1. Periaqueductal or tectal plate hamartomas, hamartomas in cerebellum or pontine tegmentum filling the fourth ventricle, brain stem gliomas, cerebral aqueduct web and superior vellum medullary synechia.[10] The causes of obstruction can occur in isolation or combination.

## CONCLUSION

Though it is not unusual to detect triventricular obstructive hydrocephalus in neurofibromatosis 1, presence of the web in aqueduct is a rare cause of hydrocephalus. A careful examination should be performed in sagittal 3D

CISS sequence for diagnosis of web after ruling out hamartomas in periaqueductal region, brain stem and cerebellum.

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