

# PRIMARY NON-NEOPLASTIC AQUEDUCTAL STENOSIS ASSOCIATED WITH VON RECKLINGHAUSEN'S DISEASE

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## SUMMARY :

*Primary non-neoplastic aqueductal stenosis associated with von Recklinghausen's disease (neurofibromatosis) is a rare entity, aetiology of which is still unknown. About 19 similar cases have been reported in the literature. We report two additional cases presenting with this rare association. Our two patients had a good neurological recovery by ventriculo-peritoneal shunt procedure (follow up 4-6 months).*

## KEY WORDS :

*Aqueductal stenosis, Computed tomography, Neurofibromatosis*

## INTRODUCTION

Primary non-neoplastic aqueductal stenosis with von Recklinghausen's disease has rarely been described (1,2,3,5,7,8,14,15,16,18,19). The first report of this entity was by Pennybacker (4). Since then 20 cases have been reported in the literature (4,18,19). The aetiology of aqueductal stenosis in neurofibromatosis is unclear but according to some authors has been considered to be a result of peri-aqueductal gliosis, aqueductal anomaly or peri-aqueductal slow developing tumours (4,6,9,15,16,18,19,20).

The purpose of this paper is to present our two additional cases and discuss this clinical entity with a review of the literature.

## CASE REPORT

### Case 1.

A 12-year-old boy was admitted in February 1986 to our clinic, complaining of unsteady gait and headache that had been progressively increasing over 1 year. Especially in last 4 months the degree of headache and gait disturbance was severe.

On examination the patient showed multiple cutaneous well defined café au lait spots and subcutaneous nodules (fibroma molloscum) in various parts of the body, especially on the trunk (figure 1). His father also had multiple similar pigmented spots and subcutaneous nodules over his body. Neurological examination revealed bilateral mild papilledema, ataxic gait, bilateral cerebellar test abnormality.

Plain skull films showed signs of raised intracranial pressure. Computed tomography revealed markedly enlarged lateral and 3rd ventricles without

enlargement of the 4th. After enhancement no abnormal density area was recognised (figure 2). A ventriculo-peritoneal shunt with a medium pressure valve was inserted. The post-operative course was uneventful. Four months after the operation the patient's neurological responses returned to normal limits.

### Case 2.

An 18-year-old boy was admitted to our clinic complaining of gait disturbance for 1 year and intermittent headache for 4 months. There was no familial history of neurofibromatosis. On examination typically nodules were found. Neurological examination revealed bilateral papilledema, ataxic gait and left increased patella and achilles reflexes. Babinski sign was found bilaterally.

Plain skull films showed signs of raised intracranial pressure. Thoracolumbar x-rays revealed right-sided thoracolumbar scoliosis. CT of the patient revealed a triventricular hydrocephalus with marked enlargement of the lateral and 3rd ventricles, the 4th ventricle was normal in localisation and size (figure 3).

A ventriculo-peritoneal shunt with a medium pressure valve was inserted. The post-operative course was uneventful 6 months later. On follow-up neurological examination, there was no papilledema and gait disturbance was minimal.

## DISCUSSION

Neurofibromatosis is a congenital and autosomal dominant disease involving tissues of neuroectodermal and mesenchymal origin (3,4,12,18,19). It is characterized clinically by various manifestations i.e.,



Fig 1 : Café au lait spots on the trunk

café au lait spot, subcutaneous nodules, scoliosis, pseudoarthrosis, intracranial tumours, endocrine abnormalities (3,4,9,12,18,19). Aqueductal stenosis can develop due to intracranial neoplasms, as acoustic schwannomas, brain stem gliosis or deep localized tumours (4,12,13,17).

Primary non-neoplastic aqueductal stenosis with neurofibromatosis is a rare entity, the first was illustrated by Pennybaker. Since then 20 cases have been reported in the literature (4,18,19).

Spadora et al reported three cases of their own primary non tumoural aqueductal stenosis associated with neurofibromatosis disease and 16 similar cases collected from the literature (18,19).

Table I-II summarizes 20 cases reported in the literature. Among these only 8 had pathological examination. Some authors proposed the following classification of the pathogenetic mechanism responsible for primary aqueductal stenosis, simple stenosis, forking septum formation and peri-aqueductal gliosis. In cases collected from the literature pathological findings were septum formation in three cases and peri-aqueductal gliosis in two (6,16,18,19,20).



Fig 2 : Markedly enlarged lateral and 3rd ventricles without enlargement of the 4th ventricle



Fig 3 : Triventricular hydrocephalus noted in Case 2.

In the literature neurological symptoms are collected under four main headings; headache, gait disturbance, ocular disturbances and seizures (18,19). Our cases showed similar symptoms.

For diagnosis of this entity CT is important but nevertheless has no significant value in determining the pathogenetic mechanism. Triventricular hydrocephalus without any other intracranially, especially brain stem, pathology is specific (3,4,10,11,18,19). Among the collected cases CT was performed on only four (19).

Today the aetiology of aqueductal stenosis in neurofibromatosis is still unknown. Some authors noted the possibility of a neoplasm in the region of the aqueduct that may not have been detected in spite of CT. To detect such tumours sagittal magnetic resonance is or will be useful (4,19).

Today the best choice of treatment of this entity

TABLE : 1

Case	Author	Age-Sex	Diagnostic Tests	Surgical Procedure	Pathological Findings
1	Pennybacker	—	—	—	—
2	Ressul	11-F	—	O	Periaque. gliosis
3,4,5	Jequier et al.	31-F	—	O	Forking
6,7,8	Turnbull, Drake	19-F	PEG-Ventr	Perforation of aqueductal membrane	Septum Formation
		9-F	PEG-Ventr		
		6-M	Ventr.		
9	Loiseau	21-M	—	Ventricle cisternostomy	—
10	Pou-Serradel et al.	9-M	Ventr.	VA Shunt	—
11	Descuns et al.	10-M	Ventr.	VA Shunt	—
12	Salvolini	—	CT-Ventr.	Ventriculocisternostomy	Periaque. gliosis
13,14	Lapras	17-F	PEG	Intra-aqueductal tube	—
		45-M	Ventr.		
15	Jacoby	13-M	PEG-CT	—	—
16	Lean	23-M	Ventr.-PEG	Ventriculocisternostomy	—
17	Radhakrishnan	28-M	CT	VA Shunt	—
18,19,20	Spadora et al.	10-M	PEG-Ventr.	VA Shunt	—
		11-M	CT		
		9-M	CT		

TABLE : II Results of Surgery (Total 20 cases).

Surgical	Number of Cases	Results		
		Exitus	Improvement	No follow-up
Perforation of aqueductal memb.	2	1	2	—
Ventriculocisternostomy	3	—	2	2
VA Shunt	7	—	7	—
Intra-aqueductal tube placement	2	—	2	—
No Surgery	6	—	—	—

seems to be to insert ventriculo-atrial or ventriculo-peritoneal shunt. Periodic CT control of the patient is necessary.

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