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STATUS EPILEPTICUS AS A RARE CLINICAL PRESENTATION OF NEUROFIBROMATOSIS 2

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ABSTRACT

Background: Neurofibromatosis 2 (NF2) is a rare genetic disorder of the nervous system usually diagnosed based on clinical and radiological findings. NF2 patients usually have positive family history with one or more 1st degree relative affected. Patients most commonly present with difficulty in hearing due to Vestibular Schwannomas. **Presentation**: Here we highlight an interesting case of 22 year old male who presented to Emergency Department in status epileptics and on further investigation was found to have multiple tumours (Bilateral Schwannomas, meningiomas, neurofibromas) and posterior subcapsular cataract consistent with NF2. Electroencephalogram (EEG) showed left focal epileptiform discharges. He was managed with antiepileptics and planned for surgical intervention for remaining tumours. **Conclusion:** Most patients of NF 2 have positive family history and symptoms appear at earlier age and are mostly related to Vestibular Schwannomas which is the most common tumour. However our patient had atypical initial presentation and combined with absence of family history and late age of presentation and absence of other symptoms made this a diagnostic challenge and a rare presentation.

KEYWORDS: Neurofibromatosis 2, Schwannomas, Meningiomas, Posterior subcapsular cataract, status epilepticus.

BACKGROUND

The neurofibromatosis including Neurofibromatosis 1 and Neurofibromatosis 2 comprises a group of genetically distinct disorders of the nervous system characterised by predisposition to nerve sheath tumours.

NF 2 is diagnosed based on a Manchester diagnostic criteria which includes (1)
• Bilateral vestibular schwannoma OR
• First-degree family relative with NF2 and either:
– Unilateral vestibular schwannoma or
– Two of the following: meningioma, schwannoma, glioma, neurofibroma, juvenile posterior subcapsular lens opacity.

Besides vestibular schwannomas other common tumours in NF2 include meningiomas, ependymomas, cataracts, and epiretinal membranes.[1] The most common presentation in adults is progressive bilateral sensorineural hearing loss due to vestibular Schwannomas.[3] Others may present with decreased visual acuity due to cataract, damage in the optic pathways, macular hamartomas, or corneal opacity.[4] The current incidence of seizures in NF2 patients is unknown and most of the seizures are due to leptomeningeal tumours (meningiomas, meningioangiomatosis).[5] NF2 presenting as status epilepticus is an extremely rare phenomenon with very few cases reported worldwide.[6]

CASE PRESENTATION

A 22 year old male presented with chief complaints of multiple episodes of generalised tonic clonic movements of the limbs associated with clenching of teeth and tongue biting, involuntary passage of urine, frothing from mouth followed by loss of consciousness lasting for 30 mins following which he was brought to ED. The EEG of the patient showed epileptiform discharges from the left cerebral hemisphere (figure 1). The patient was treated with Lorazepam 5 mg and became symptomatically better and regained consciousness. Furthermore he was loaded with Sodium valproate 1000 mg followed by maintenance dose of 500 mg twice daily. CSF study was done and it was within normal limits (Total cell count – 5, all monomorphs, protein – 46, sugar – 78). Rest of the biochemical parameters were normal, including bacterioscopic and virological tests; alcohol and drug intake was excluded. CEMRI Brain with whole spine screening was done which showed bilateral vestibular Schwannomas (figure 2), Left lobe meningiomas (figure 3) and intraspinal lesion suggestive of ependymomas. Slit lamp examination of the patient

showed right eye posterior subcapsular cataract, fundus examination was within normal limits. Pure tone audiometry revealed the presence of mild Sensorineural hearing loss in both the ears though the patient never had any symptoms. Abdominal examination revealed a mass in the epigastric region extending to the right hypochondrium and lumbar region which was non tender with smooth margins non mobile with respiration and it was suggestive of Neurofibroma arising from the Right kidney (figure 4) in MRI Abdomen. Thus he was diagnosed to have NF 2. Review of the history revealed the seizure episode was preceded by aura in from of auditory hallucinations, impaired taste and smell sensation and felling of pin pricking sensation in the limbs consistent with the imaging findings. The patient was completely asymptomatic before this episode with no history of similar illness in any other family members.

TREATMENT AND FOLLOW UP

The patient remained seizure free for the rest of his stay in the department was discharged and was then transferred to neurosurgery for surgical resection of the Schwannomas. He was planned for regular follow up in in surgery department for Right kidney neurofibroma and for the posterior subcapsular cataract in ophthalmology department.

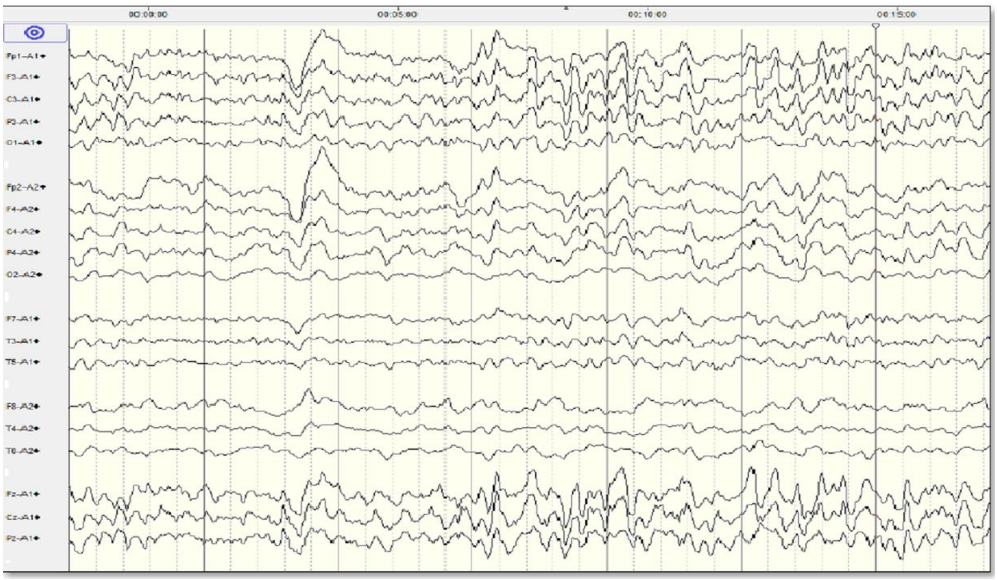

Figure 1- EEG showing left focal epileptiform discharges.

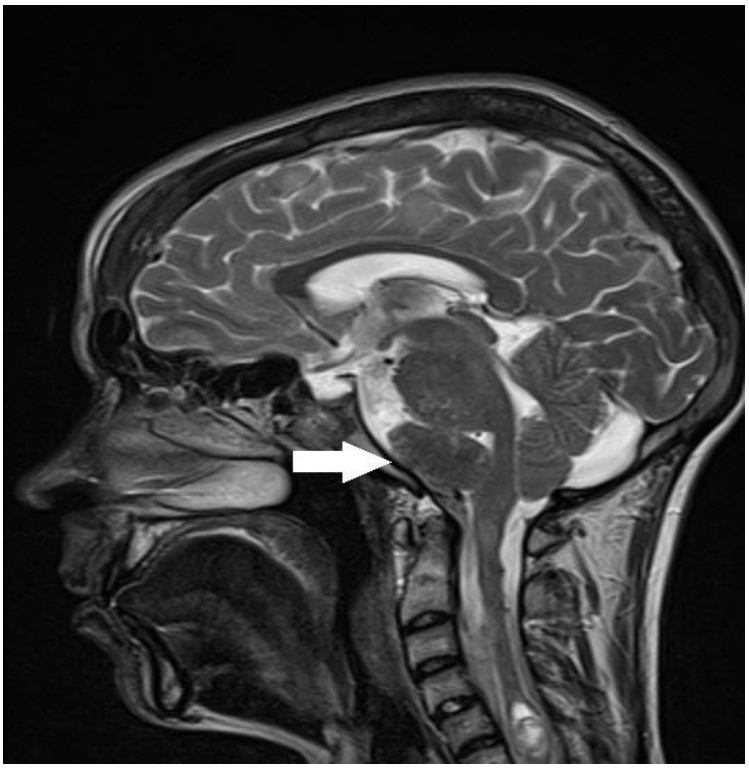

Figure 2- T1 sagittal image showing Cerebellopontine angle tumour suggestive of vestibular Schwannomas.

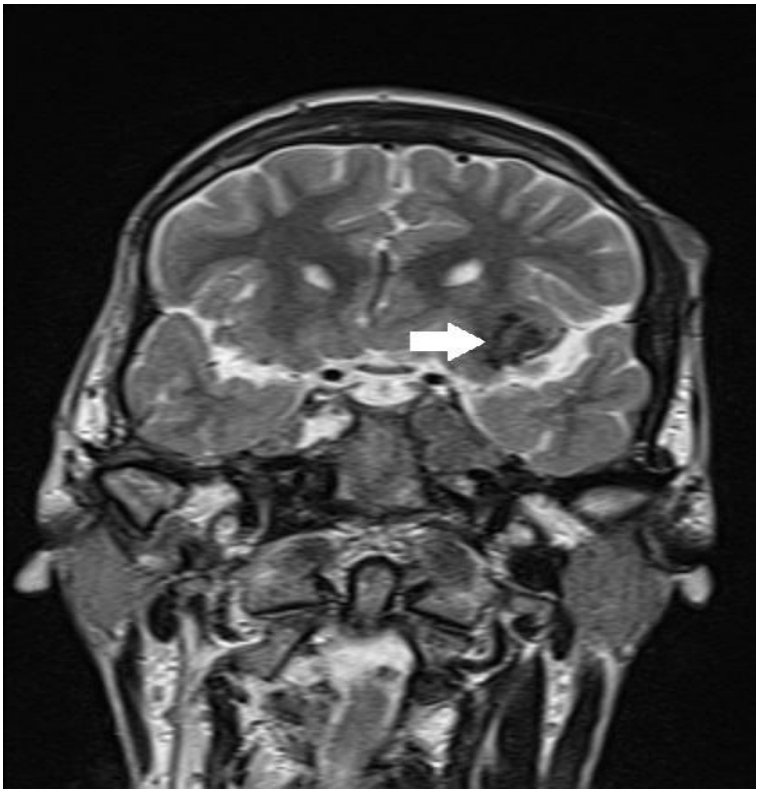

Figure 3 - T2 coronal image showing lesion with post contrast enhancement suggestive of meningioma.

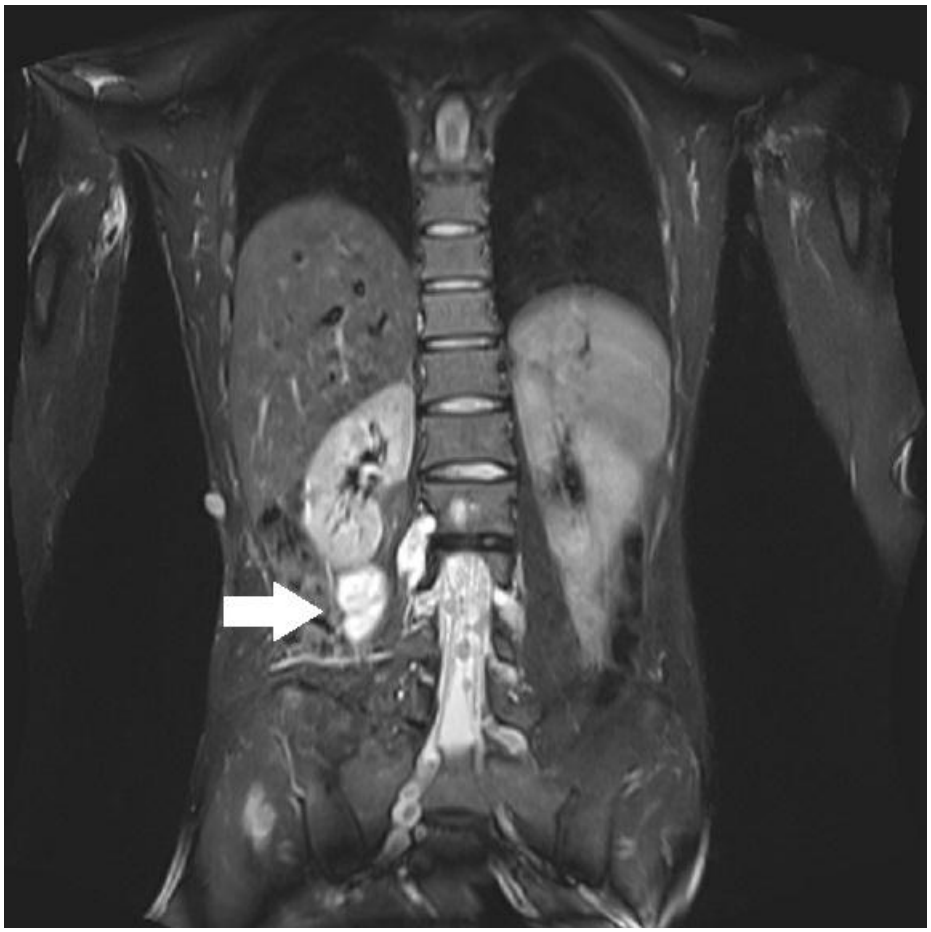

Figure 4- T2 coronal view showing neurofibroma adjacent to the right kidney.

DISCUSSION

The diagnosis of Neurofibromatosis is based on Manchester criteria. Around 50% of the patients of NF2 have no family history.[7] The NF2 gene has been mapped to chromosome 22. It is a tumour suppressor gene of size 110 kb and its protein product, named merlin or schwannomin, mediate communication between the extracellular milieu and cytoskeleton.6–8. Loss of heterozygosity of chromosome 22q (LOH 22q) has been detected in many specimens of NF2-related tumors, including acoustic schwannoma and meningioma.[8,9,10] Mean age-at-onset in 58 individuals is around 20.3 years; initial symptoms resulted from vestibular schwannomas (44.4%), other CNS tumours (22.2%), skin tumours (12.7%), and ocular manifestations including cataracts and retinal hamartomas (12.7%).[11] Only few case report of NF2 as status epilepticus at onset or during the course of the disease are present. As such seizure is not a hallmark of the disease neither is required for the diagnosis of the same which may be surprising since intra cranial tumours are invariably present in all patients. Some studies have shown that the frequency of seizures may be underestimated in these patients[12] and further studies are required to estimate the true prevalence and characteristics of seizures in these patients. EEG as a tool may be quite helpful in this regard. As this case show despite the absence of any other symptoms at initial presentation further investigations showed multisystem involvement consistent with the disease and it re-emphasizes the need for detailed evaluation of NF2 patient.

CONCLUSION

1. Status epilepticus as initial presentation of NF2
2. Absence of family history
3. Presence of tumours and lesions in multiple sites consistent with NF2 despite obvious absence of any symptoms in the patient.

Conflict Of Interest: None.

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