A Case of Neurofibromatosis with Seizure Disorder

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Abstract

According to studies, NF1 is associated with an increased prevalence of seizures that varies between 4% and 10%. The increasing occurrence of epilepsy in this cohort is mostly caused by brain tumours associated to NF1, according to recent evidence from individuals with NF1 and seizures. Multiple hyperpigmented cutaneous macules first appeared throughout childhood, marking the beginning of the condition. We report an unusual case of a Seizure disorder with features of neurofibromatosis like several café au lait macules over body,Lisch nodules and neurofibroma with hereditary history Neurofibromatosis. The Boy has diffused swelling that is 4 cm × 3 cm in size as well as coffee-colored patches all over his body. The NF-1 diagnosis was made using the diagnostic criteria established by the National Institutes of Health Consensus Development Conference; two or more criteria were needed to establish a diagnosis.

Keywords: Café au lait spots, neurofibroma type 1, von Recklinghausen disease

1. Introduction

An extremely prevalent hereditary condition is neurofibromatosis (NF). The two genetically separate subtypes of this phacomatosis, It is passed down from parent to offspring in an autosomal dominant fashion and is characterised by many skin lesions and tumours in various parts of the nervous system. Neurofibromatosis type 1 (NF1), sometimes called Recklinghausen's disease, affects around 1 in 3500 persons and causes a variety of cutaneous and peripheral nerve system disorders. Café-au-lait macules (CALMs), freckling of the axillae and/or inguinal regions, Lisch nodules of the iris, neurofibroma, and bone dysplasia are among symptoms associated with NF1. Optic pathway gliomas are a particularly dangerous kind of brain tumour that are more common in people with NF1 (OPG). In addition, reports have been made of cerebrovascular illnesses including Moya-Moya syndrome (MMS). Known as a benign neural tumour, neurofibroma is made up of a varied combination of fibroblasts, perineurial-like cells, and Schwann cells that originates from the peripheral nerve sheath. Neurofibromas in the oral cavity are not unusual, despite the fact that they are the most prevalent skin lesion. With common locations including the tongue, gingiva, major salivary glands, and jaw bones, neurofibromatosis (NF) often affects the fifth cranial nerve and higher cervical nerves. Furthermore, lips and mucosa have both seen it. Clinically, distinct, submucosal neurofibromas appear in the oral cavity. It may also manifest within the bone in very rare circumstances. Type 1 neurofibromatosis, also known as von Recklinghausen disease, and type 2 neurofibromatosis are the two forms of neurofibromas.

Frederich von Recklinghausen first identified Neurofibromatosis Type 1 in 1882. Neurofibromatosis type 1 (NF-1), sometimes called von Recklinghausen disease, is an autosomal dominant disorder that affects one in every 3,000 live births and is characterised by a fundamental deformity in the undeveloped brain peak cells that lead to ectodermal and mesodermal subsidiaries. Both NF-1 and NF-2 treatments aim to manage the consequences and reduce the symptoms.

2. Case report

A 23 yrs old male patient brought to casualty with Complaints of 1 episodes of seizure since morning

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and on arrival also, he has 1 episode of active seizure.

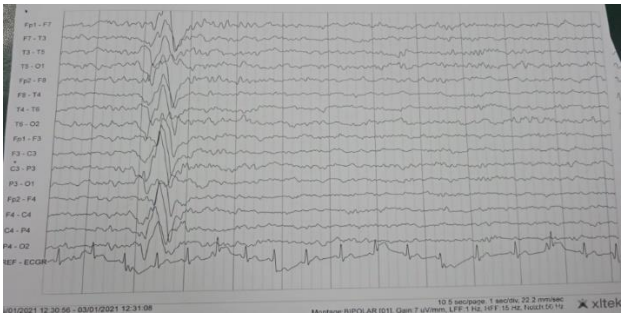
History of Present illness

Patient was apparently normal while he was in casualty under observation, He has 1 episode of seizure for 15 sec, Involve Upper limb and lower limb, Ass. With uprolling of eye, frothing from mouth, Tongue bite +, N/h/o Loss of consciousness.

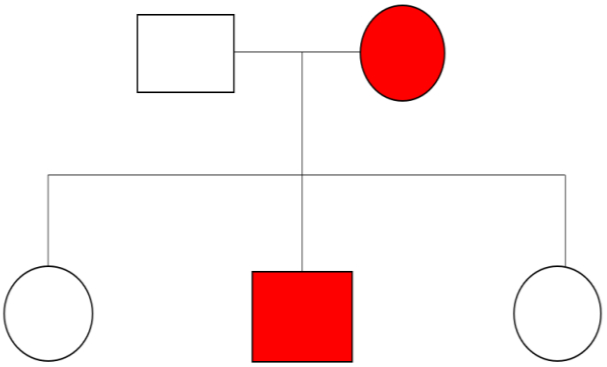
N/K/C/O Seizure disorder/HTN/T2DM

Family History Neurofibromatosis in Mother.

Upon examination, we discovered around seven café au lait macules over the body, as well as a scattered swelling that measured about 4 cm × 3 cm. On Slit lamp examination shows Lisch nodules. We have started patient on Anti epilieptic medication Tab.Levipill 500 mg and advised patient to follow up.



FIGURES 1 EEG: Generalized Tonic Clonic Seizure



Figures 2 eeg: family pedigree chart

Figures 3 : Café-au lait spots

One of the seven essential NF1 diagnostic indicators is the café-au-lait macule. The typical lesion is uniform in appearance and well defined, with smooth margins (like the California coast). Although it may vary from tan to dark brown, the hue is often somewhat similar to that of its namesake. Patients must meet this condition by having six or more >5 mm (pre-puberty) or >15 mm (post-puberty). More than two are present in less than 1% of infants under 5 who do not have NF1, and this is strongly indicative of NF1.

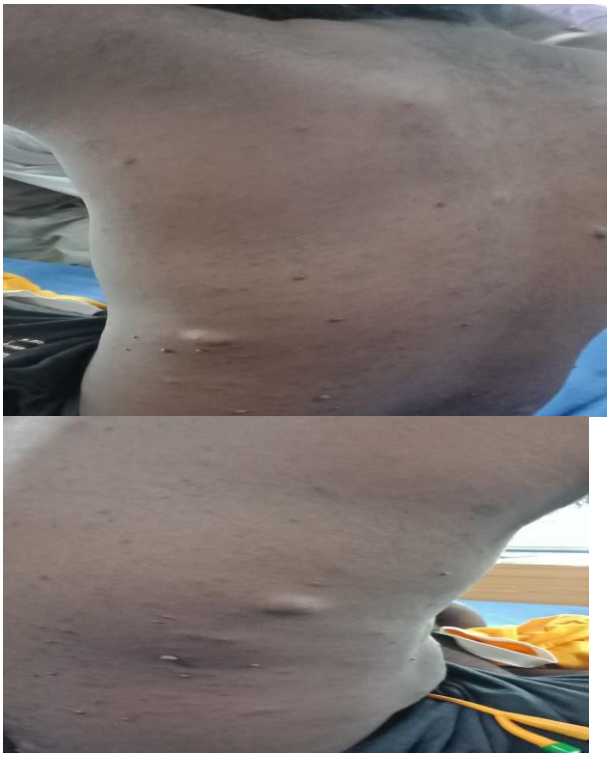
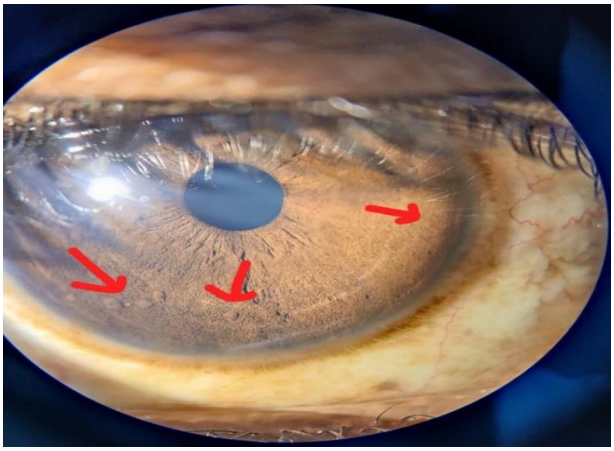


Figure 4: neurofibroma

One other distinguishing feature of NF1 is the neurofibroma. There is a vast range in the size and appearance of neurofibromas, which may appear anywhere on the body. The terminology has changed and, at times, proven a little unclear. Tumors located "subcutaneously" are solid and nodular, whereas those located on the skin's surface, known as the "dermal" or "dermal," are often dome-shaped, soft, fleshy, and skin-colored or somewhat hyperpigmented. Adolescence is the typical onset of cutaneous neurofibromas, and in certain cases the tumours may grow and spread even into adulthood.



Figures 5: Lisch nodules

Lisch nodules are taken into account when two of the diagnostic criteria for type 1 neurofibromatosis are met. They are regarded as melanocytic hamartomas of the iris, often seen before the age of six, which grow in quantity and frequency as people mature. They often come in shades of light or dark brown, and lighter-colored eyes make them more prominent.

3. Discussion

In the current instance, the patient reported a positive family history in addition to café au lait spots and Lisch nodules.Recent research has linked NF1 with an increased risk of seizures, with estimates ranging from 4-to 10%.Epilepsy is more common in those who have NF1 and have seizures, and this is mostly attributable to the presence of brain tumours caused by NF1.Both focal cortical dysplasia and hemimegalencephaly have been linked to NF1, which may have an impact on the development of epilepsy.The central nervous system is affected by neoplastic and non-neoplastic conditions in 15–20% of NF-1 patients. Cortical abnormalities in the cerebral hemisphere and the cerebellum, as well as vasculopathy, have been documented in NF1..NF1 is characterised by a number of abnormalities in the brain, including transmantled cortical dysplasia, a periventricular band of heterotopic dim matter with pachygyria of the cerebral cortex, and perisylvian polymichrogyria.It has been found that individuals with hemimegalencephaly and NF1 had excellent seizure control. Common MRI abnormalities seen in patients with drug-resistant epilepsy include localised cortical dysplasia and polymicrogyria, both of which are developmental malformations of the cerebral cortex.About portion of NF1 people with epilepsy don't show a primary irregularity on X-ray, and X-ray anomalies in these patients are not generally connected with epileptiform releases on EEG. They pondered whether the genetic disorder contributed to brain overstimulation, seizure susceptibility, and other persistent abnormalities that result in epilepsy.Neurofibromatosis is a frequent MRI lesion but has not been associated to seizures, which are often caused by lesions in the brain such tumours or cortical dysplasia.Studying the magnetic resonance imaging (MRI) of 172 NF1 patients (23 with epilepsy and 149 without), the researchers found that NBOs (neurofibromatosis brilliant articles) were available in 16 (69.6%) of the epilepsy patients and 108 (72.5%) of the patients without epilepsy.There was no relationship between the number or area of these cerebrum sores and epilepsy in our gathering. Of the 11 NF1 patients with intracranial tumours, 4 (36.36%) had seizures, whereas only 19 (11.8%) of the 161 NF1 patients without tumours did.And last, this research related the onset of epileptic seizures in NF1 patients to intracranial tumours rather than NBOs.In NF1, many forms of seizures and syndromes have been described. The majority of seizures in people with NF1 are focal and secondary generalised. The several focal lesions of NF1, which include tumours and abnormalities of cortical development, are likely to be the source of the seizures in people with the condition. West syndrome (infantile spasms) and febrile seizures were more common in NF1 individuals than in the general population.As much as a quarter of people with NF1 have abnormal EEGs. Lennox-Gastaut syndrome is characterised by anomalies in the electroencephalogram (EEG), including spike and slow spike wave buildings at 2 Hz, as well as expected to central or multifocal spike waves. The most well-known sort of EEG irregularity is a central condition.As a result, even if previous neuroimaging was normal, seizure formation necessitates neuroimaging. Although the link between NBOs and seizures is debatable, most research have determined that NBOs are not linked to seizures. Seizures in people with NF1 are usually manageable with one or more anti-epileptic medicines (AEDs); in some cases, the terminating lesions are surgically removed. The most effective treatment for temporal lobe gliomas is surgery.Recently, the possible pathogenic involvement of UBOs in seizures has been assessed and ruled out. It has been proposed that NF1 maternal inheritance is a seizure risk factor.The mammalian target of rapamycin (mTOR) remains an intriguing potential epilepsy and epileptogenesis factor. The mTOR pathway may even be detrimental to epilepsy development, according to recent results in animal studies.Patients with tuberous sclerosis complex, a phacomatosis in which mTOR is constitutively activated, have less seizures while using mTOR inhibitors. It's interesting to note that mTOR is constitutively active in NF1.Loss of neurofibromin causes RAS to become hyperactive, which in turn causes the RAF/MEK/ERK, phosphoinositide 3-kinase (PI3K), and mTOR pathways to become hyperactive. NF1 brains may thus be naturally hyperexcited.

4. Conclusion

In conclusion, when an NF1 patient exhibits seizures, the clinician should rule out not just brain tumours but also other less common NF1-related presentations as hydrocephalus, stroke, and vasculopathy. To evaluate whether NF1 per se could predispose to epilepsy, further research on people with NF1 and non-structural seizures is needed. In the event that the patient initially does not meet the diagnostic criteria, careful follow-up and symptom monitoring are required. In order to direct the patient to the proper experts for a diagnostic workup and therapy, a multidisciplinary team approach is suitable for patients with NF1 and seizures. For future pregnancies, genetic testing and prenatal counselling are also advised.

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