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Non Neurofibromatosis Child Diagnosed as Scalp Diffuse Neurofibroma with Bony Defect: Case Report and Literature Review

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Abstract

A child of 13-year-old with unremarkable past medical history presented with long history of painless scalp swelling noticed since early childhood over the left occipitomastoid area with slow rate of increase in size and diffuse ill-defined edges. Clinical assessment of the swelling revealed presence of bony defects palpable through the swelling with the rest of systemic assessment was unremarkable. Patient scenario and presentation was vague to spot a diagnosis and so patient is investigated systematically (CT brain, RI brain, DSA). histopathology revealed diagnosis of scalp diffuse neurofibroma which is considered a surprise for us. neurofibroma is uncommon diagnosis seen in neurosurgery and associated bony defect seen in our case is considered rarer event in scalp neurofibroma, add to that our case is not a case of neurofibromatosis in which the neurofibroma is commonly associated. A lesson to learn from such case is that do not under estimate small lesion with long insignificant history and vague diagnosis and always follow systematic assessment and approach to reach the right diagnosis.

Introduction

Neurofibromas are nerve sheath lesions and may arise from Schwann cells, perineural cells, or fibroblasts [1]. they considered a benign tumor of the nerve and of rare malignant type. They can arise from

the nerve anywhere in the body [1]. They commonly occur at age of 20-40 years and commonly seen at the head and neck region of the body [2].

Neurofibroma is seen in different form; plexiform neurofibroma which arises from several nerve bundles; diffuse neurofibroma which is uncommon form and seen commonly in children and young adults and the localized type [2]. Neurofibromas are commonly seen clinically as swelling Mostly of small size and solitary which make them easily to be suspected as differential diagnosis. the Diffuse type is rare and commonly seen in adults [3] and presented as infiltrative, poor defined lesion and involving the skin and subcutaneous tissues with boggy sensation on palpation and of slow growth nature.

Such uncommon clinical presentation makes the clinical suspicious of such diagnosis is less ranked or even not included. The possible underlying causes mentioned in the literature behind such type of tumor are either to be seen as idiopathic form or due to genetic cause which is the neurofibromatosis type 1(about 10%.) [5].

Case Presentation

13-year-old boy quit well presented with a long history of scalp swelling for about 4 years back in time, of slow increasing in size since then, painless and not associated with any other significant symptoms. Recently and at the present age, swelling becomes more noticeable, bumpy in feeling and concerning for the parents who decided to seek medical advice .at presentation, patient is quite well, fully conscious and cooperative and has no any neurological deficit and this is confirmed by the neurological assessment which was unremarkable for any relevant findings and also was the assessment of the other systems enquiry and assessment.

On local assessment of the presenting complaint ;there was a noticeable scalp swelling in the left occipital post auricular area of the head, measures about 6x8 cm with diffuse nature and ill-defined borders ,no overlying skin changes seen (taking into consideration that patient is a black colored skin) (figure13).the swelling is painless ,has boggy sense on palpation with palpable bony two defects (figure3)and negative trans illumination test.patient has no any sign of neurofibromatosis type 1 on clinical assessment.

Initially the diagnosis was not so clear and so proper assessment with imaging is planned considering the other relevant possible diagnosis .so patient investigated accordingly with CT brain (figure1-2-3) which revealed subcutaneous diffuse lesion and seen more in the MRI brain. MRA and MRV also done(figure4-9). patient is prepared for DSA (with both conventional and selective type done) to assess the vascularity of the lesion and to identify the relation of it to the underlying transverse sinus (figure10-12).

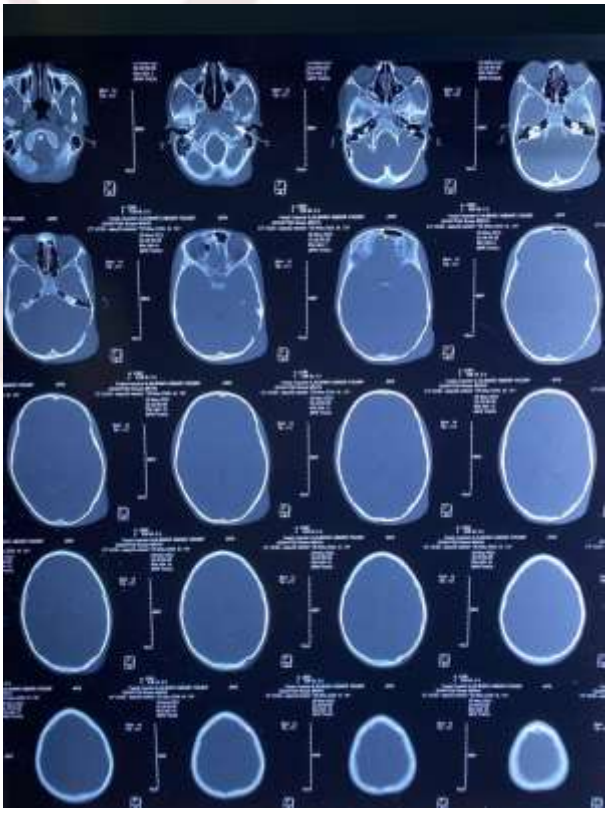

Figure (1): CT brain axial cut bone window showing the bony defect at the left occipito mastoid area

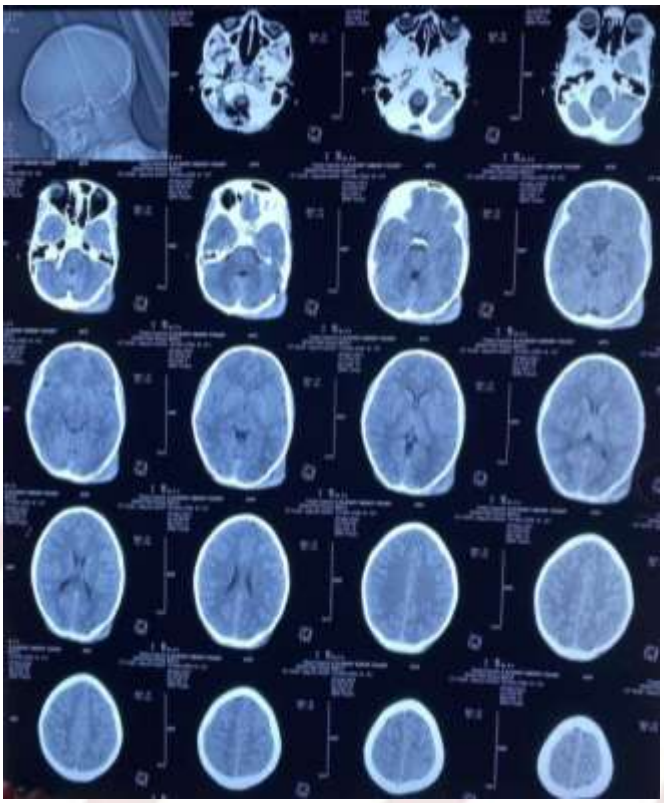

Figure (2): Non contrast CT brain axial cut soft tissue window shows the scalp swelling over the left occipito mastoid area which is diffuse with the bony defect seen but without intracranial extension of the lesion.

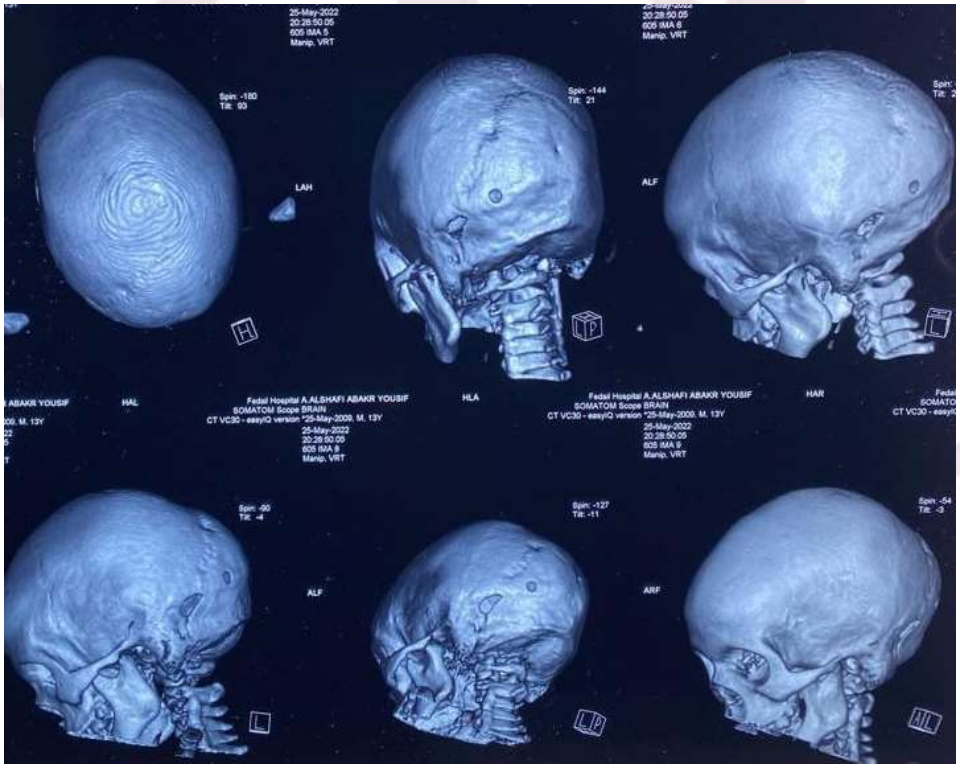

Figure (3): CT brain 3D reconstruction shows two bony defect over the squamous occipital and occipito mastoid area of the skull.

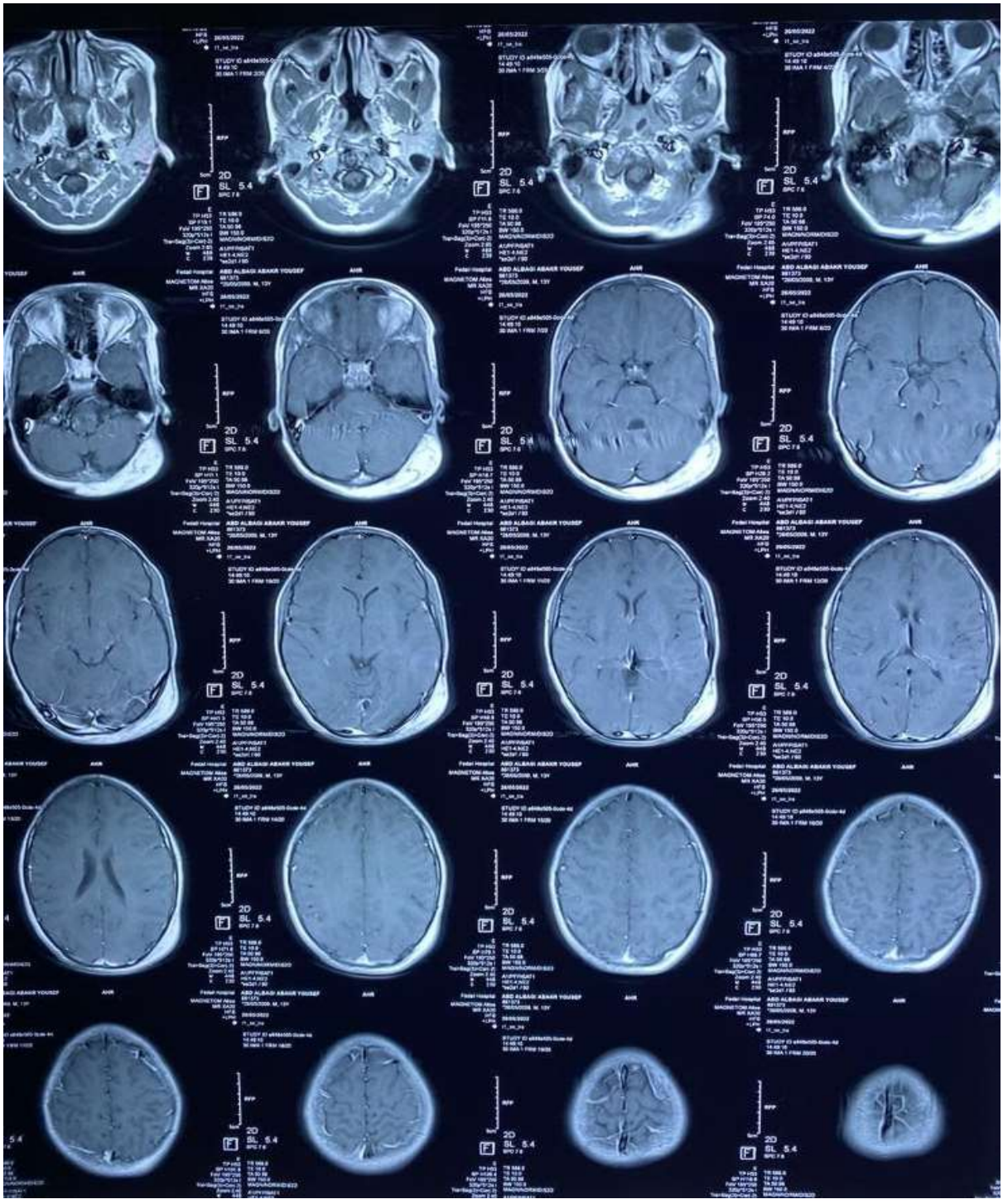

Figure (4): MRI brain with contrast axial cut shows the lesion enhancing intensily and purely extra cranially.

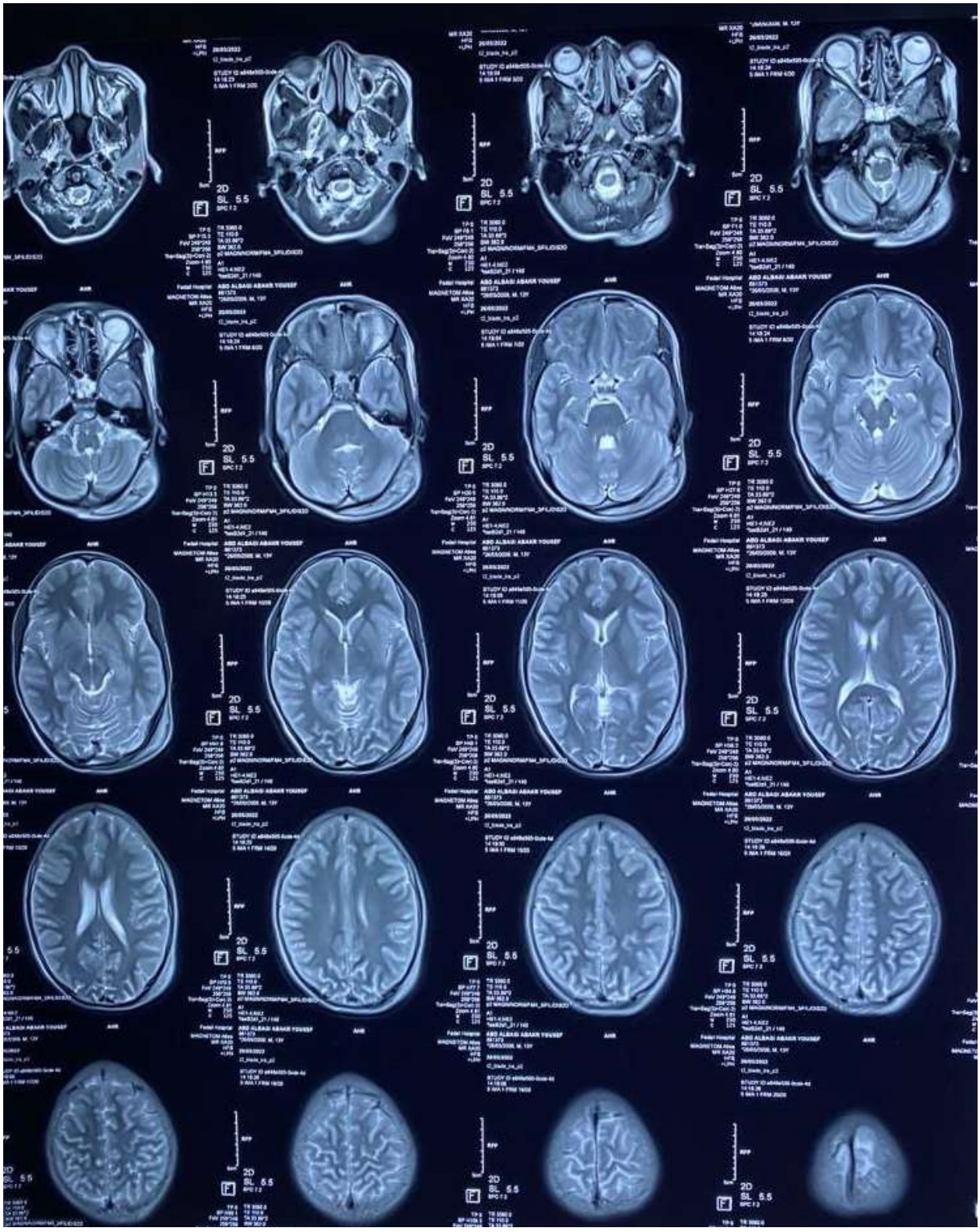

Figure (5): MRI brain T2 weighted image axial cut of the lesion

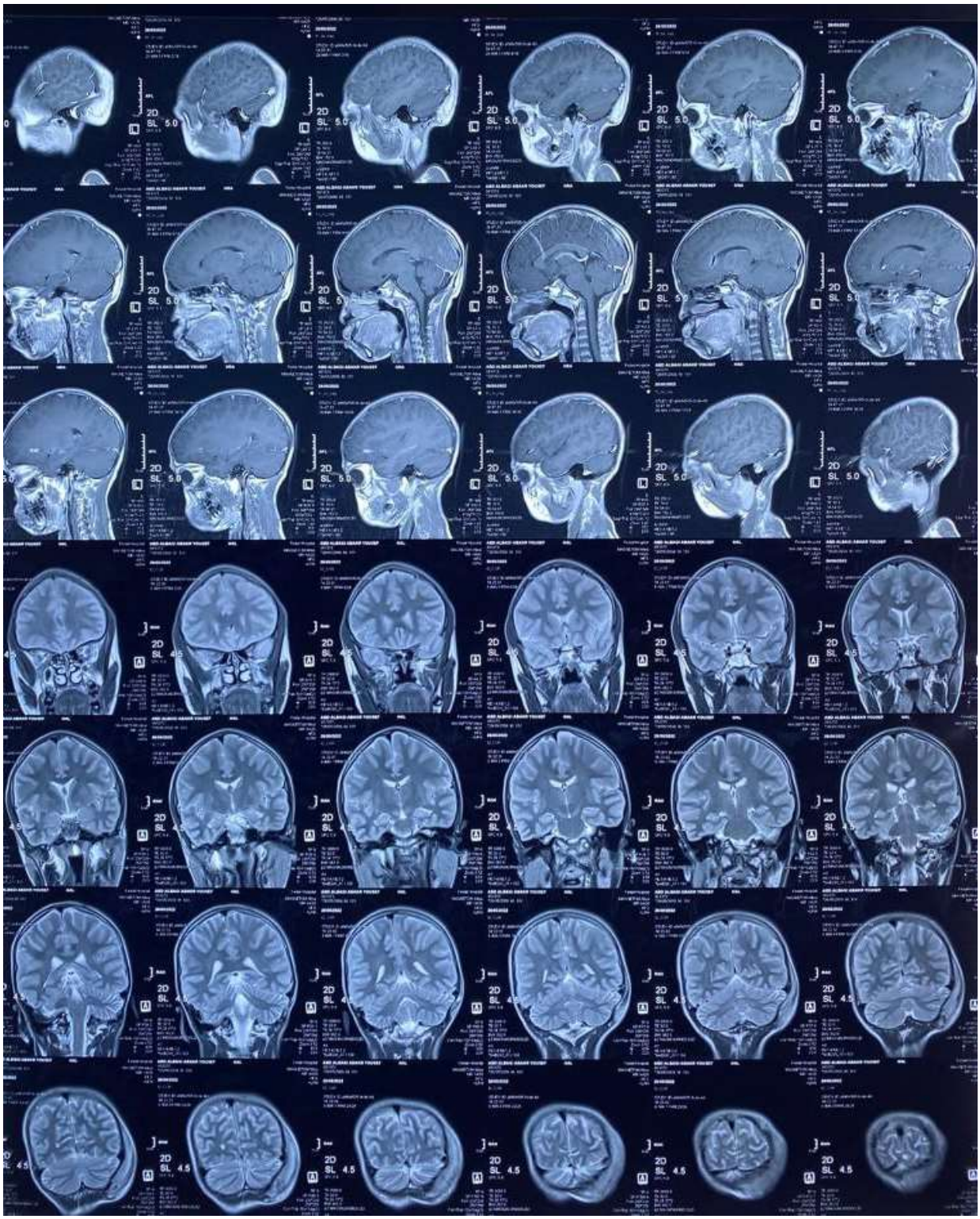

Figure (6): MRI brain of the lesion with different cut views, sagittal T1 weighted image with contrast, T2 weighted image coronal cut.

Figure (7): MRV of the lesion shows the draining nature of the lesion

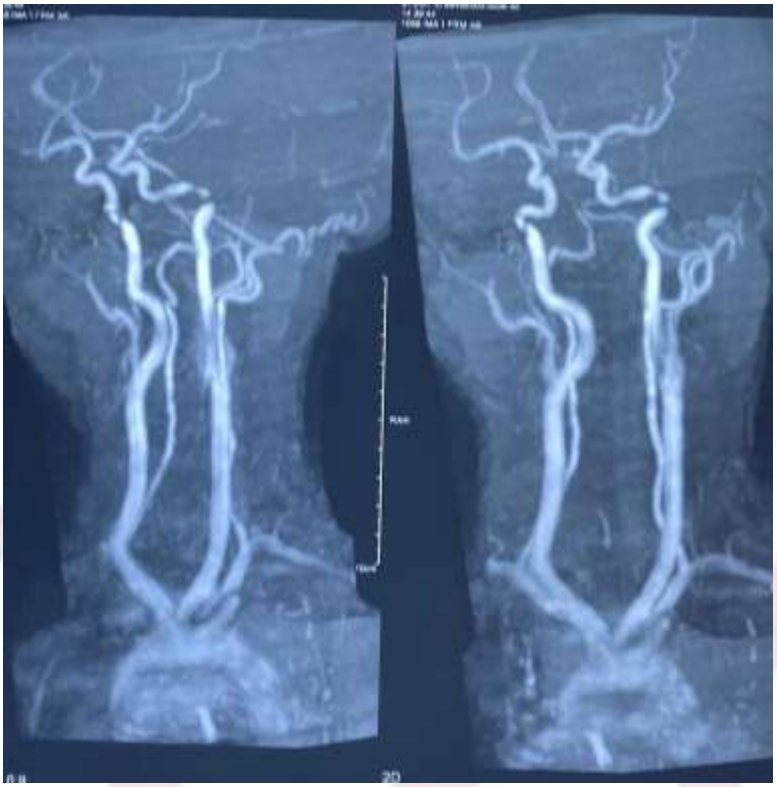

Figure (8): MRA of the lesion done from the arch of the aorta shows the nature of the blood supply of the lesion which is mainly derived from the occipital artery.

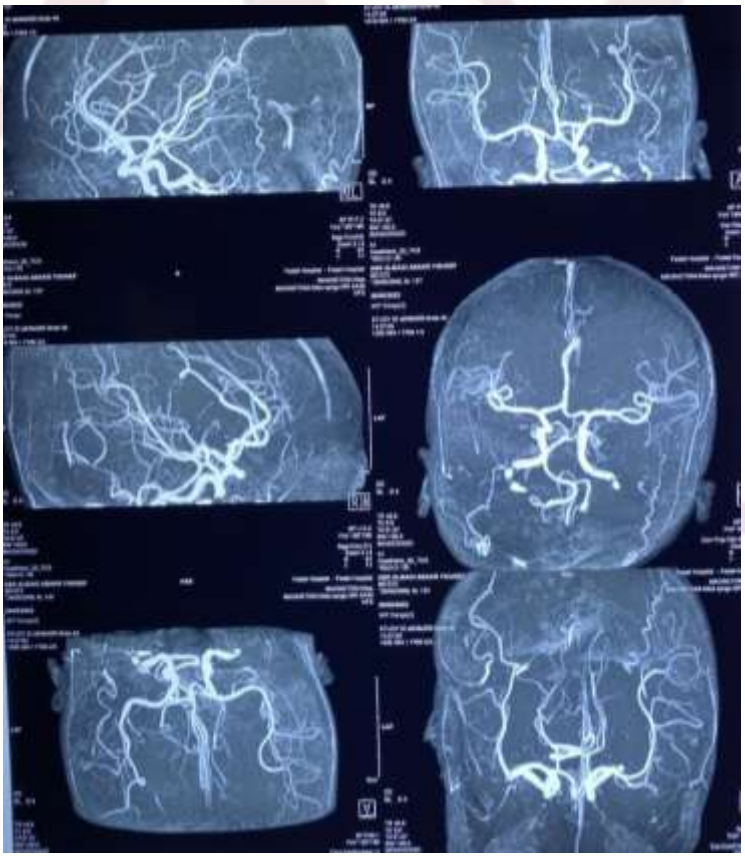

Figure (9): MRA 3D reconstruction view of the lesion.

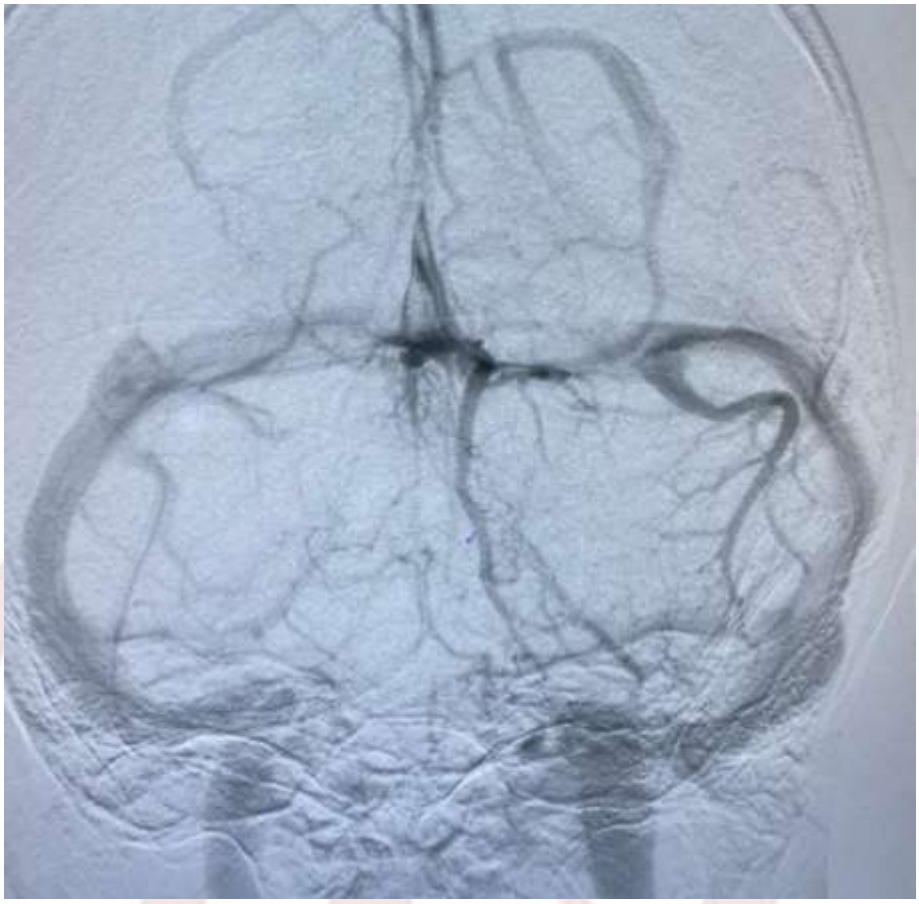

Figure (10): DSA of the lesion shows the abnormally venous draining and communication with the transverse sinus of the lesion.

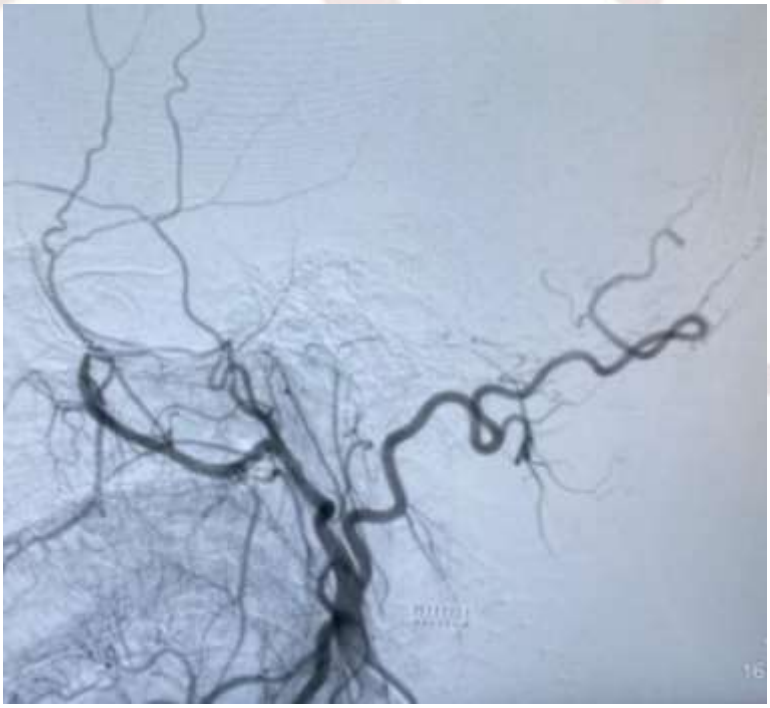

Figure (11): Selective DSA study of the occipital arterial supply of the lesion.

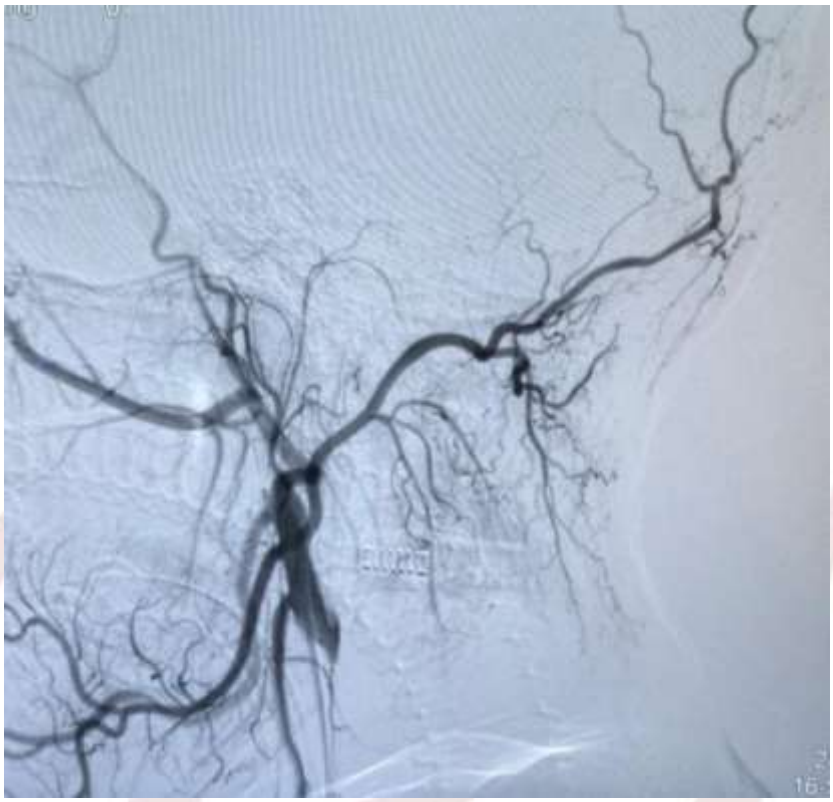

Figure (12): Selective DSA more arterial phase of the lesion.

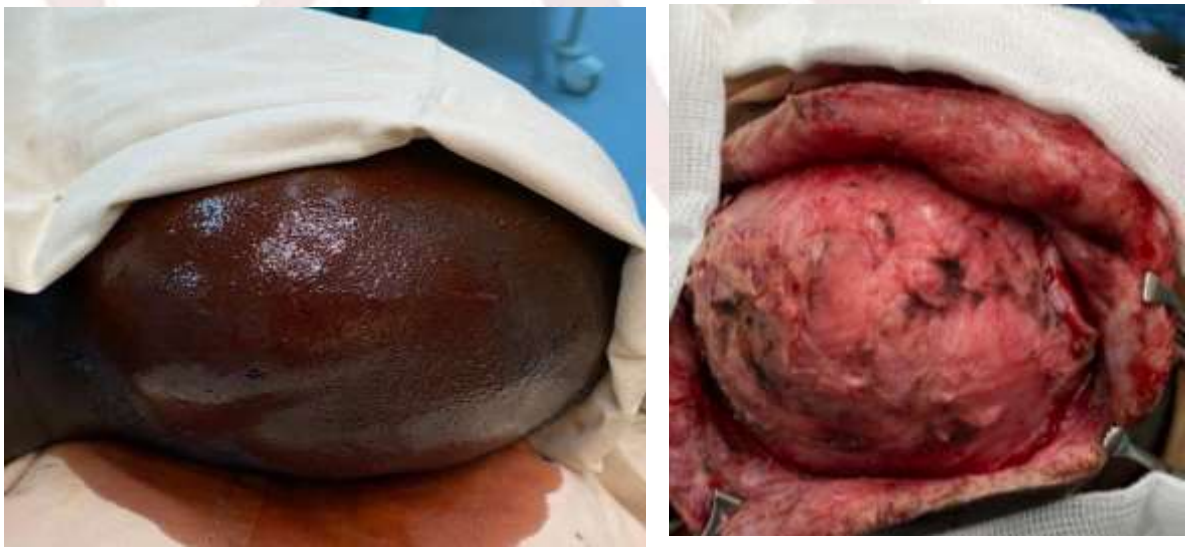

Figure (13): Intraoperative photo of the lesion before skin opening and after the opening showing the enclosed diffuse nature of the lesion in the scalp

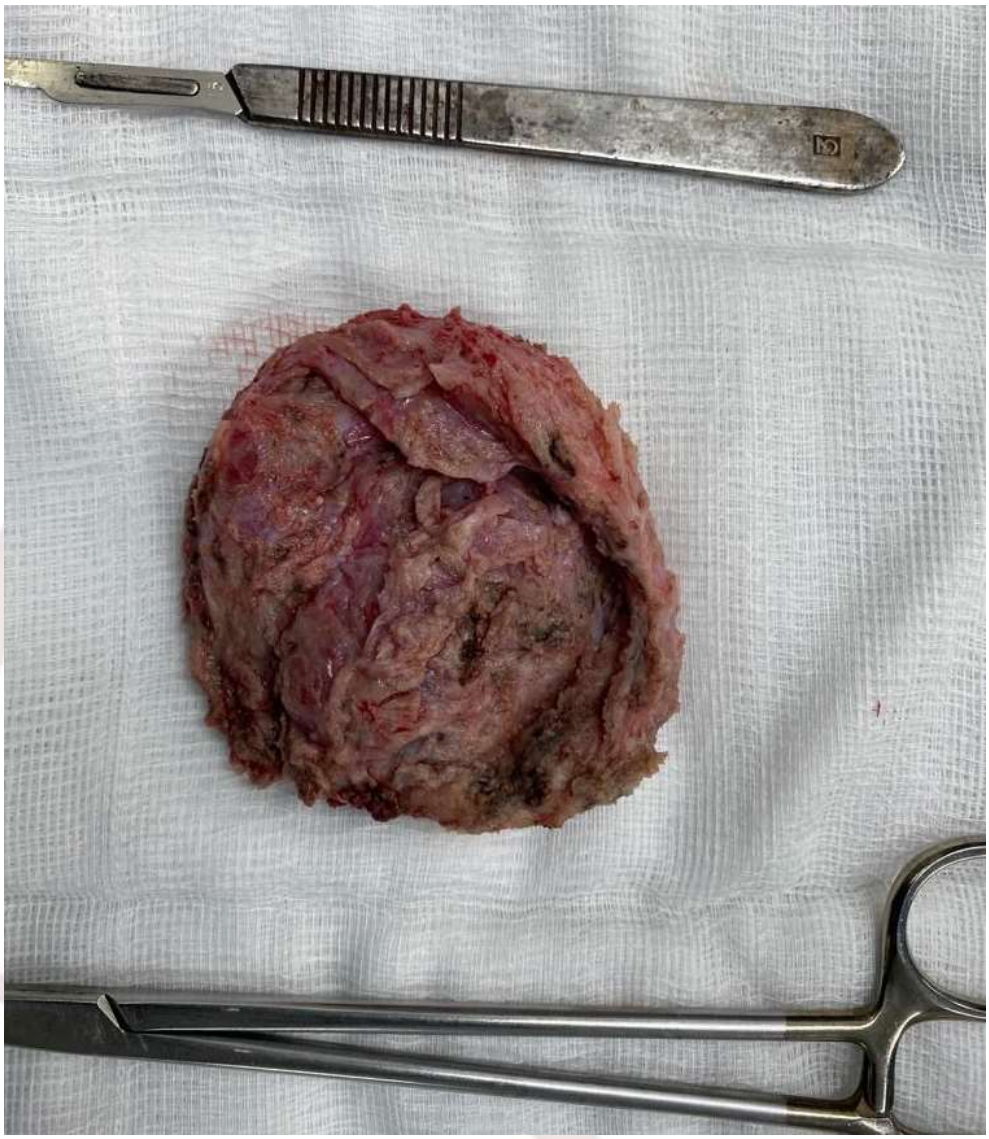

Figure (14): The lesion with its capsule after complete excision

Discussion

Neurofibroma as a type of tumor is not common and the diffuse type is even more uncommon [3] like in our case which is a child with diffuse type. bony defect in neurofibroma is considered a rare event especially in the absence of NF 1 [3] and again our case was non fibromatosis case and showed bony defect at the presentation.

The bony defect is a common finding in the cases of neurofibromatosis type 1 in which Over 50% of those patients and not all show osseous defects [1]. Sphenoid wing dysplasia and orbital defects are the most common calvarias anomalies in patients with NF-1 [5].in our case the bony defect was seen as two small bony holes over the left occipital mastoid region and the occipital region a little pit up and more medially (figure3) which were palpable through the swelling during clinical assessment.

Surgery was done in a meticulously hemostatic way focusing to deal carefully and early with blood supply and venous draining of the lesion (figure13-14).no intracranial extension is found which is reported in the literature to be seen in case of the plexiform neurofibroma [6] and so the pericranium is left to cover the bony defects and accompanying cranioplasty is not considered because we lack the definitive diagnosis and the defects were small .literature mentioned a risk of the recurrence [7] and there is lack of consensus about the role and timing of cranioplasty in such cases [3].

Case reported in the literature are not so many especially the diffuse type with bony defect and most reported case are adult especially the giant solitary type without neurofibromatosis [3]. neurofibroma has insignificant clinical presentation and it represents a challenging diagnosis and Even proper history taking, physical examination, ophthalmological, and radio diagnostic investigations may fail in clinching the diagnosis in the absence of neurofibromatosis. and the answer for its definitive diagnosis is always by the histopathology and this is noticed in reported cases in the literature. This exactly what happened in our case which was a vague case preoperatively with few differentials ranked and the highly suspect one was scalp angioma and the scalp neurofibroma was not considered as one of the differential but the surprise came from the result of histopathology. so surgical treatment should be suited separate to every case alone [8].

Conclusion

Scalp neurofibroma is not a common type of tumor seen in neurosurgery, and the diffuse type with bony defect is even more uncommon presentation and more confusing one to diagnose.

Proper assessment with relevant investigations and imaging taking into consideration the differential diagnosis is required. add to that the reported cases of such diagnosis in literature are not so many; so such cases need to be reported to increase awareness for diagnosis of such conditions with proper assessment and plan of management.

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