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Case Report

Spheno-Orbital Meningo-Encephalocoele due to Absence of Sphenoid Wing in NFM 1 - A Case Report

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ABSTRACT

Background: Orbital - facial manifestations are relatively rare in neurofibromatosis 1. Encephalocoeles are defined as herniation of meninges with brain matter. Absence of sphenoidal wing leads to Spheno orbital encephalocoele due to protrusion of brain tissue and meninges through superior orbital fissure in orbital cavity. Plexiform neurofibromatosis can involve 1st division (orbital) of the trigeminal nerve. Osseous lesions of neurofibromatosis 1 are conventionally believed to be dysplasia (primary bone defect), recent reports postulate that these could be secondary to presence of an adjacent plexiform neurofibroma, or other ocular neoplasms like optic nerve glioma, neurofibroma and schwannomas.
Our Case: Presentation of a clinical case of 20 years old male patient of neurofibromatosis type 1 with right Spheno-orbital meningoencephalocele presenting with diffuse swelling on right side of face and head since 10 years. We describe role of imaging modalities (X-ray, USG, CT, 3D- CT and MRI) in diagnosis of Spheno-orbital meningoencephalocele. X-ray showed bare orbit due to absence of innominate line. USG, CT and MRI showed absence of sphenoid wing with resultant right Spheno-orbital meningoencephalocele with resultant inferior displacement of right eye ball. Associated plexiform neurofibroma was noted in extra-cranial soft tissues in right fronto-temporo-parietal region. Our case is unique as there was complete absence of sphenoid wing rather than dysplasia.
Conclusion: Sphenoid dysplasia seen in NFM 1 can be primary due to absence of sphenoid wing or due to interaction between neurofibromas and sphenoid bone during skull development. Spheno-orbital meningoencephalocele can result due to sphenoid dysplasia.

Keywords: Encephalocele (MeSH unique ID: D004677), Neurofibroma, Plexiform (MeSH unique ID: D018318), Neurofibromatosis 1 (MeSH unique ID: D009456), Sphenoid Bone (MeSH unique ID: D013100).

INTRODUCTION

Encephalocoeles are defined as herniation of meninges with brain matter and are seen in rate of 1 in every 4000 live births. Absence of sphenoidal wing leads to Spheno orbital encephalocoele due to protrusion of brain tissue and meninges through superior orbital fissure in orbital cavity. The pulsation of the brain in intraorbital field leads to the pulsation of the globe through the defect. Basal encephalocoeles are relatively rare, constituting 1.5 % of all cases. [1]

Depending on the site, encephalocoeles are classified as occipital, of the cranial vault (inter frontal, anterior fontanel, inter parietal, posterior fontanel, temporal), frontoethmoidal (nasofrontal, nasoethmoidal, temporal), frontoethmoidal (nasofrontal, nasoethmoidal, naso-orbital),

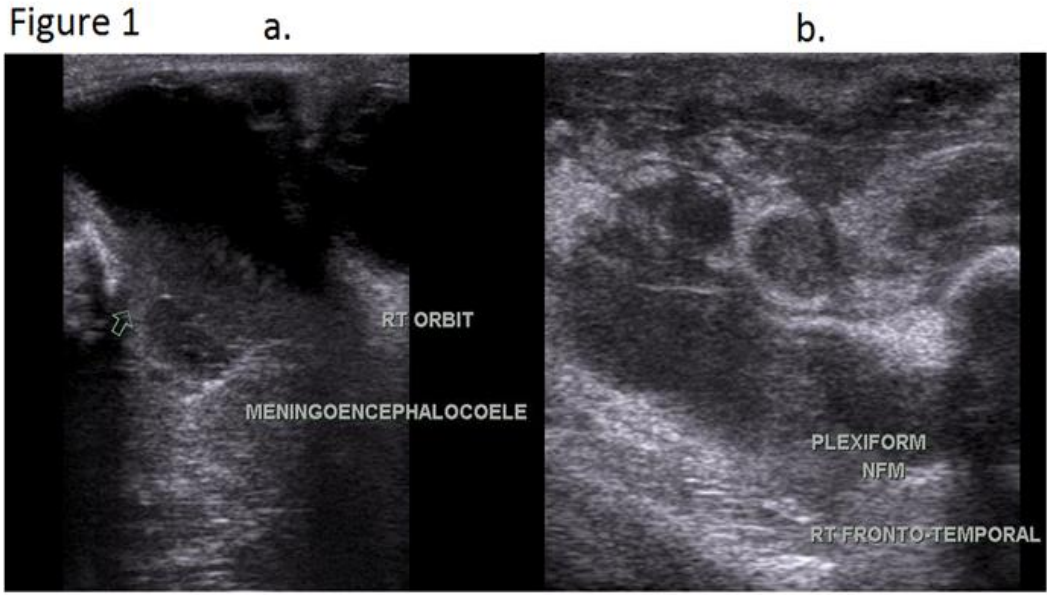
basal (transethmoidal, transsphenoidal, transorbital, sphenoethmoidal, sphenoorbital) and cranioschisis. [2]

Spheno ethmoidal meningo-encephalocoele can be seen in NFM 1 due to sphenoid dysplasia.

CASE REPORT

A 20 years old male patient presented with diffuse swelling on right side of face and head since 10 years. It was painless. History of surgery for the swelling 10 years back - known case of neurofibromatosis. He had café au lait spots on the trunk. He was referred for USG of orbits and local swelling.

USG of both orbits was done with a high resolution linear probe (7-10 MHz). Left orbit appeared normal. A well-defined anechoic cystic lesion measuring approx. 6.3 (AP) x 3.9 (T) x 2.5 (CC) cm was noted in right orbit compressing and displacing right eyeball inferiorly (Figure 1a). Postero-lateral bony wall of right orbit was not seen. Local USG swelling showed diffuse soft tissue lesion of heterogeneous echotexture in right fronto-temporal region without significant vascularity or cystic areas or calcifications (Figure 1b).



USG: Sagittal USG Right orbit. **USG Local Swelling**

Figure 1 a - USG Right orbit (Sagittal section) showing meningo-encephalocoele superior to right eyeball with inferior displacement of eyeball. Figure 1 b - USG showing diffuse mixed echoic solid lesion in extra-cranial soft tissues in right fronto-temporal region.

CT scan (Figures 2, 3, 4) and MRI (Figures 5, 6) of the head, orbits and maxillofacial region revealed absence of lesser and greater wings of the sphenoid on right side with resultant anterior herniation of right temporal lobe. A large CSF filled space measuring approx. 6.3 (AP) x 3.9 (T) x 2.5 (CC) cm was noted herniating anteriorly through this defect superior to right eyeball causing extrinsic compression and inferior displacement of superior rectus and eyeball in right orbit - suggestive of meningo-encephalocoele. As a result, right Sylvian fissure and temporal horn of the right lateral ventricle were displaced anteriorly. A diffuse heterogeneous soft tissue density lesion of measuring approx. 8.5 (L) x 3.5 (T) x 11.2 (CC) cm was noted in extra-cranial soft tissues in right fronto-temporal region extending in right infra-temporal fossa causing scalloping of the outer cortex of right fronto-temporo-parietal bone - suggestive of plexiform neurofibromatosis. It was of heterogeneous signal intensity on T2WI and iso- to hypointense to muscle on T1WI on MRI. Sulcal spaces of right cerebral hemisphere showed mild prominence. The rest of the brain parenchyma reveals normal appearance.

Figure 2 (a-d) - CT scan of brain and orbits showing absence of right greater and lesser wing of sphenoid with Spheno-orbital meningo-encephalocoele and plexiform neurofibroma in scalp in right fronto-temporal region.

Figure 3 (a-d) - CT scan of brain and orbits showing absence of right greater and lesser wing of sphenoid with Spheno-orbital meningo-encephalocoele and plexiform neurofibroma in scalp in right fronto-temporal region.

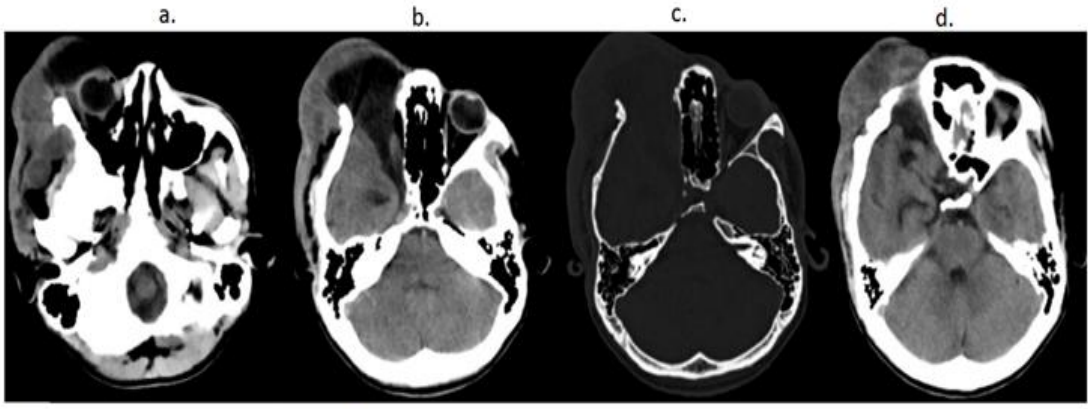


Figure 2: Axial plain CT Brain and Orbits in Soft tissue and bony windows.

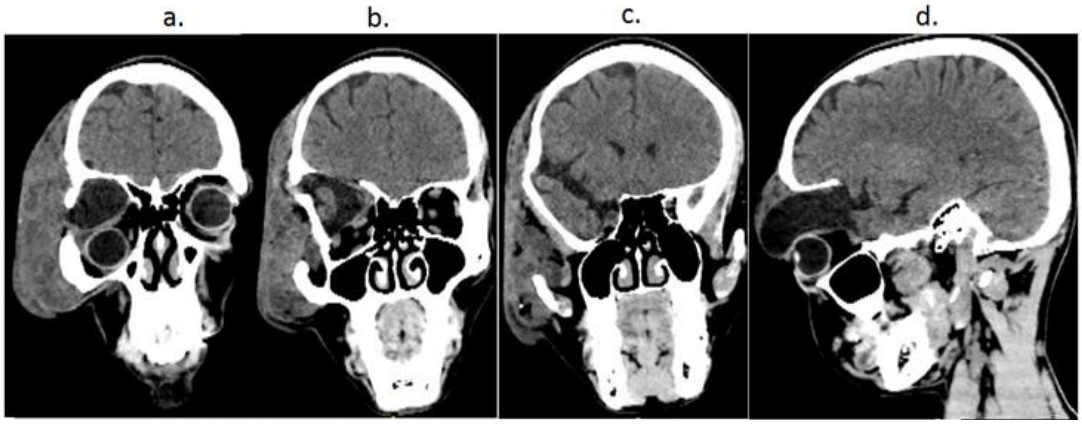


Figure 3: Coronal and Sagittal Plain CT scan of orbits

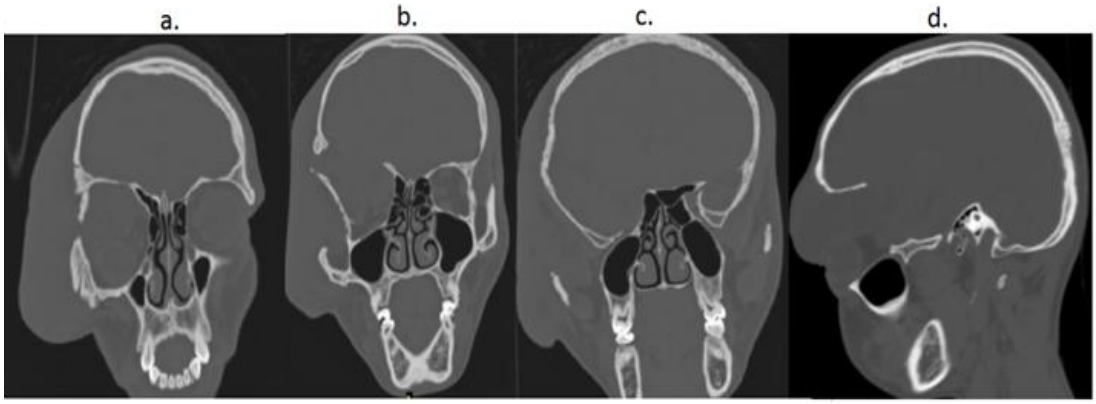


Figure 4: Coronal and sagittal CT scan of orbits in bony window

Figure 4 (a-d) - CT scan of brain and orbits showing absence of right greater and lesser wing of sphenoid with widened right orbit.

Figure 5 (a, b) - MRI brain and orbits showing absence of right greater and lesser wing of sphenoid with Spheno-orbital meningo-encephalocoele and plexiform neurofibroma in scalp in right fronto-temporal region.

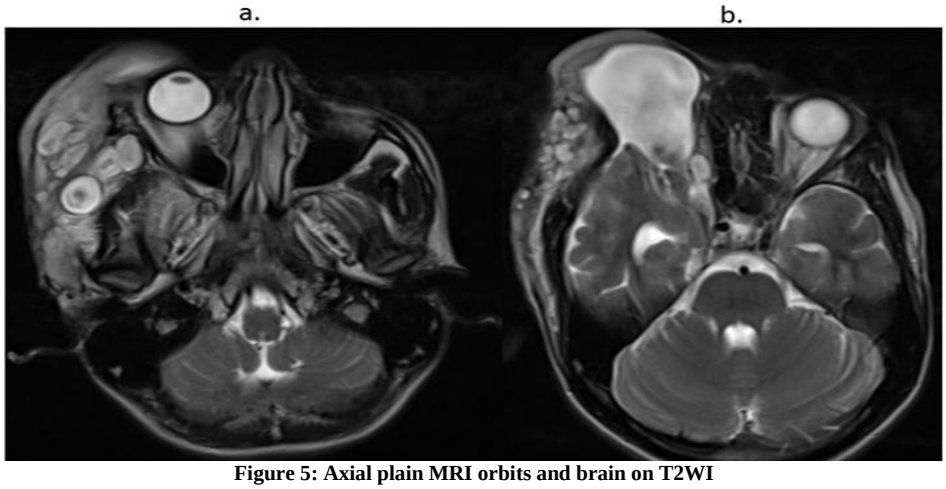


Figure 5: Axial plain MRI orbits and brain on T2WI

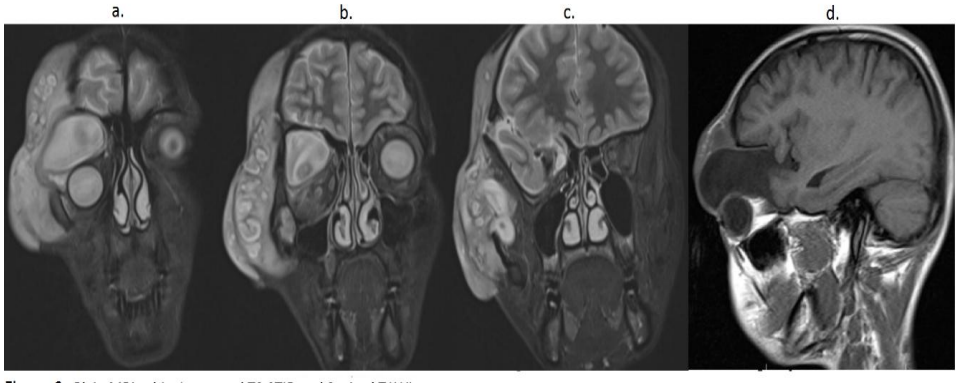


Figure 6: Plain MRI orbits in coronal T2 STIR and Sagittal T1WI

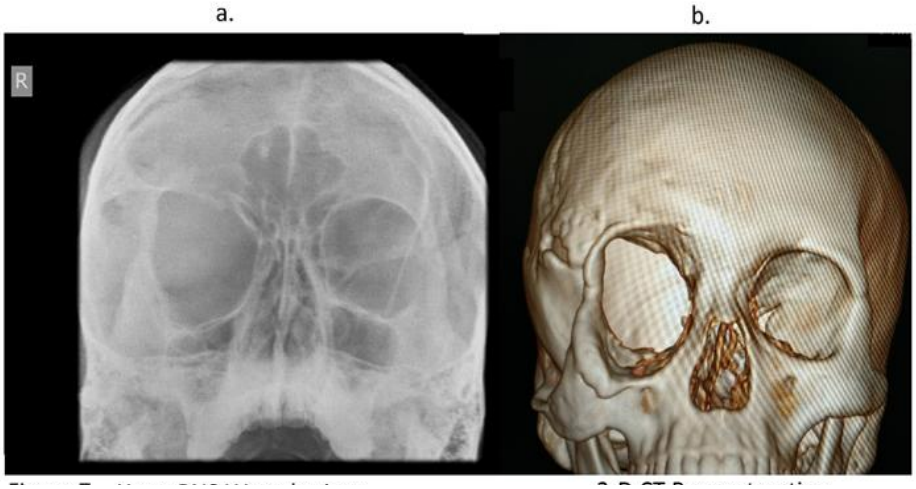


Figure 7- Bare orbit on Water's view (a) and on 3-D CT reconstruction (b).

Figure 6 (a-d) - MRI orbits showing absence of right greater and lesser wing of sphenoid with Spheno-orbital meningo-encephalocoele with inferiorly displaced right eyeball and plexiform neurofibroma in scalp in right fronto-temporal region.

X-ray Water's view (Figure 7a) showed absence of in nominate line on right side - suggestive of bare orbit, confirmed on 3D CT (Figure 7b). A diagnosis of right Spheno orbital meningo-encephalocoele with plexiform neurofibromatosis in extra-cranial soft tissues in right fronto-temporo-parietal region in neurofibromatosis I was made.

DISCUSSION

Neurofibromatosis (Von Recklinghausen disease) is a hereditary condition due to neuroectodermal and

mesodermal dysplasia named after the researcher who reported it in 1882. It is transmitted as an autosomal condition; however 50 % cases occur as spontaneous mutations with no gender predilection. It is a group of heterogeneous diseases with two distinct types - neurofibromatosis 1 and neurofibromatosis 2. Neurofibromatosis 5 (segmental NF) is a rare form of neurofibromatosis in which cutaneous and neural changes are confined to one region of the body. [3]

Eight separate forms of neurofibromatosis (1-8) have been described. More than 90 % of all neurofibromatosis cases are NF 1. Family history of NF 1 is elicited in 52-60 % cases. Its incidence is 1 in 3000/4000. [4]

The diagnostic criteria of NF 1 include more than 2 of these findings: 6 or more 5 mm or larger café au lait spots, 1st degree relative with NF 1, axillary or inguinal freckling, 2 or more pigmented iris hamartomas (Lisch nodules), either 1 plexiform NF or 2 or more NF, optic nerve glioma and the presence of characteristic bone lesion (dysplasia of the greater sphenoid wing, pseudoarthrosis). [4]

Plexiform neurofibromatosis are diffusely infiltrating multiple tortuous worm like masses arising along axis of major nerves and are poorly delineated. The 1st division (orbital) of the trigeminal nerve is commonly involved with associated sphenoid wing dysplasia, middle cranial fossa arachnoid cyst or prominent sub-arachnoid space. [3]

25 % of patients have head and neck manifestations; however orbital - facial manifestations are relatively rare. These manifestations occur due to either one or combination of plexiform neurofibromatosis (PNF), orbital osseous dysplasia, orbital neoplasms and buphthalmos (congenital glaucoma). Buphthalmos (globe enlargement) occurs due to raised intra ocular pressure due to obstructed aqueous outflow seen as enlargement of eyeball in all dimensions as compared to contralateral normal eyeball. Plexiform neurofibromatosis can involve the orbital, temporal region and/or the face. It can involve any motor or sensory nerve, para-sympathetic or sympathetic nerves of head and neck. The 1st division of the trigeminal nerve at orbital apex is most frequently involved seen as irregular soft tissue lesion enlarging and deforming the muscular anatomy of temporalis fossa, eyelid and recti. [5]

It can affect nearby bones and invade all orbital structures. Sphenoid wing dysplasia is a classic bone lesion of NF 1 seen in 5-10 % cases. It presents as pulsatile exophthalmos due to herniation of temporal lobe into the orbit. On frontal skull radiograph, it is seen as “bare orbit sign “due to absence of innominate line which is projection of greater wing of the sphenoid bone.

Though, osseous lesions of neurofibromatosis 1 are conventionally believed to be dysplasia (primary bone defect), recent reports postulate that these could be secondary to the presence of an adjacent plexiform neurofibroma, or other ocular neoplasms like optic nerve glioma, neurofibroma and schwannomas which are common lesions seen in neurofibromatosis 1. [6]

Typical manifestations of neurofibromatosis 1 include buphthalmos, sphenoid wing dysplasia, plexiform neurofibroma and enlarged ipsilateral middle cranial fossa. Atypical findings are the atypical involvement of the temporal bone with less involvement of the adjacent sphenoid wing and lateral herniation of temporal lobe into a deformed and convex outwardly bulged temporal bone rather than classical intra-orbital herniation of the temporal lobe. [5,6]

Though sphenoid dysplasia occurs in 5-10 % cases of NF 1, absence of sphenoid wing is very rare. Its radiological characteristics describe defect in greater sphenoid wing with enlargement of middle cranial fossa. [7]

Imaging of cranio-facial abnormalities in NFM 1 can be done with

USG, CT and MRI. USG is real time, easily available, cheaper and can show proptosis, pulsatile exophthalmos, Spheno-orbital meningo-encephalocoele, anterior herniation of the temporal lobe due to absence of sphenoid wing.

The advantage of MDCT over MRI- Though MDCT has risk of radiation; it can scan large area in less time. MDCT shows skeletal abnormalities better than MRI. Reconstructions and 3D images demonstrate detailed bony information and image quality. It allows isotropic volume data for 3D imaging. With MDCT, good axial and 3D images of the sphenoid bone and the surrounding area can be obtained. [8]

MRI has multiplanar imaging ability with no radiation and can detect Spheno-orbital meningoencephalocele with sphenoid dysplasia and associated plexiform neurofibromatosis.

CONCLUSION

Sphenoid dysplasia seen in NFM 1 can be primary due to absence of sphenoid wing or due to interaction between neurofibromas and sphenoid bone during skull development. Spheno-orbital meningo-encephalocoele can result due to sphenoid dysplasia.

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