A multidisciplinary approach to sphenoid wing dysplasia presenting with pulsatile proptosis in neurofibromatosis Type 1: A rare case report

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Neurofibromatosis (NF) with sphenoid wing dysplasia is a rare clinical entity. Herewith, we present a case of NF with sphenoid wing dysplasia which presented with pulsatile progressive proptosis. Other ocular symptoms or visual disturbances were absent. Diagnosis of the condition was not easy and the management was a challenging task which needed multidisciplinary approach as there were ocular, neurological, orthopedic, and dermatological manifestations. With neurosurgical intervention, reconstruction of the sphenoid wing was possible. Proptosis was corrected without any disturbance of vision.

Key words: Lisch nodule, neurofibroma, neurofibromatosis, pulsatile proptosis, sphenoid wing dysplasia

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Neurofibromatosis type 1 (NF1), also known as Von Recklinghausen's disease, is a relatively common autosomal dominant hereditary neurodermal dysplasia that affects multiple systems of the body with an incidence of around one in 2500–3000.[1] The major defining features of NF1 are café-au-lait spots, peripheral neurofibromas, and Lisch nodules.[2] All structures of the eye except the lens can be involved.[3] Sphenoid wing dysplasia is one of the characteristics of NF1 affecting 5%–10% of the cases.[4]

Case Report

A 38-year-old female patient presented to our institute with proptosis (pulsatile reducible) of the right eye of 4–5 months duration which was gradually increasing in size, associated with watering. She had multiple painless nodular swellings all over the body since birth, which increased in number and size over time. There was positive family history with father and sister having similar swellings.

General examination revealed multiple café-au-lait skin spots over the arms and trunk. On ocular examination, the best corrected visual acuity was 6/6 in both eyes and color vision was normal. Right eyeball had pulsatile proptosis (eye was pushed forward, downward, and inward) which reduced on external pressure. Anterior segment examination revealed multiple Lisch nodules on iris [Fig. 1]. Posterior segment of both eyes were normal. Extra-ocular movements were normal, angles were open on gonioscopy, no refractive error on retinoscopy, patent lacrimal syringing, and normal A-scan.

The clinical diagnosis of NF 1 was made, and radiological investigations were done. X-ray of the skull revealed bare orbit sign suggestive of the absence of lesser wing of sphenoid

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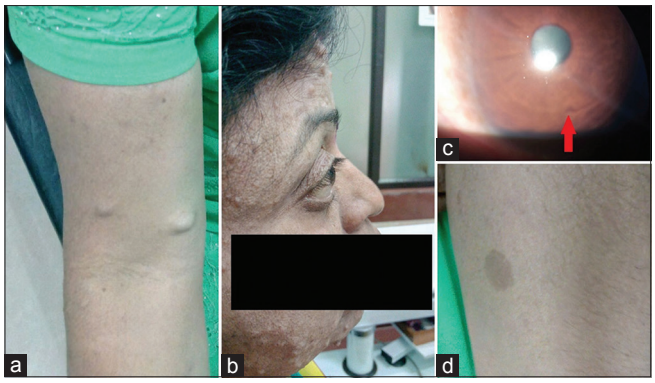

Figure 1: Photograph of the patient. (a) Neurofibromas. (b) Proptosed right eye. (c) Lisch nodule. (d) Café-au-lait spots

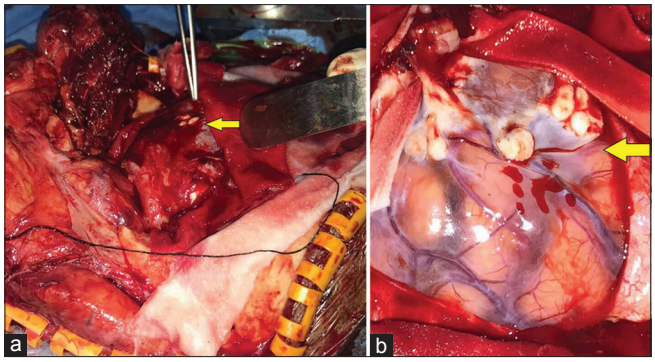

Figure 3: Surgical findings. (a) Intracranial dissection exposing orbital defect. (b) Dura excised exposing herniated enlarged temporal lobe with intact arachnoid

which was confirmed with computed tomography (CT) scan. MRI revealed expansion and (antero-posterior) enlargement of middle cranial fossa with herniation of right temporal lobe with dura into posterior aspect of the right orbit causing compression and medial displacement of right optic nerve [Fig. 2].

After the diagnosis was confirmed, the patient was taken up for the surgical intervention to prevent any visual disturbance or other complication due to hemimegaencephaly. Right fronto-zygomatico-orbital craniotomy was done to delineate the defect in orbital roof and posterior wall. After exploration temporal polar dura was excised, herniated temporal lobe was repositioned. The orbital roof and lateral wall reconstruction was done along with separate dural repair leaving arachnoids intact. Polymethyl methacrylate mould was fashioned in shape of orbital defect. While preparing mould, nonabsorbable suture, silk was passed through it and kept secured. It was fixed in place with surrounding available bone using drills and silk suture 2-0. After installment, polymethyl methacrylate (PMMA) was washed with cool water thoroughly to prevent heat damage to the adjacent brain tissue. Bone flap was replaced and wound closed [Fig. 3].

Histopathological examination of right dural tissue revealed features suggestive of neurofibroma. Postoperatively, the patient had regressing proptosis with no diminution of vision. Repeat CT scan revealed bone graft *in situ* in the right sphenoid region [Fig. 4].

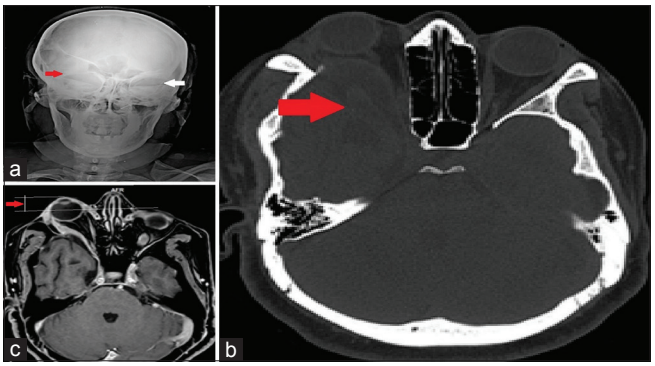

Figure 2: (a) Skull anteroposterior radiograph showing the absence of right innominate line (red arrow). (b) Axial computed tomography in bone window confirming the absence of right-side innominate line. (c) Axial T1-weighted image demonstrating right-sided proptosis with herniation of temporal lobe

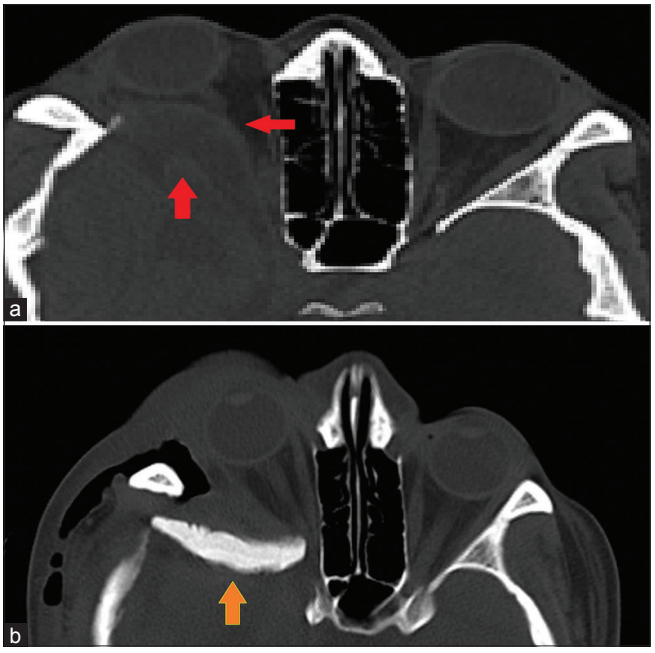

Figure 4: Axial computed tomography scan images in the bone window (a) preoperative computed tomography shows right eye proptosis with heniation of temporal lobe compressing optic nerve. (b) Immediate postoperative computed tomography showing reduced herniation with polymethyl methacrylate graft *in situ* with soft-tissue edema

Discussion

NF is a common genetic disease that belongs to a group of disorders called phacomatosis. It encompasses two rare diseases type 1 and 2. Both are autosomal dominant multisystem disorders with phenotypic overlap. Earliest evidence of NF1 appeared through a paper published by Friedrich Daniel Von Recklinghausen.[5] It is caused due to mutation in NF1 gene located on chromosome 17 that encodes for protein neurofibromin (a tumor suppressor); leading to uncontrolled cell proliferation.[5]

People with NF1 can develop skeletal abnormalities such as osteopenia, scoliosis, sphenoid wing dysplasia, congenital tibial

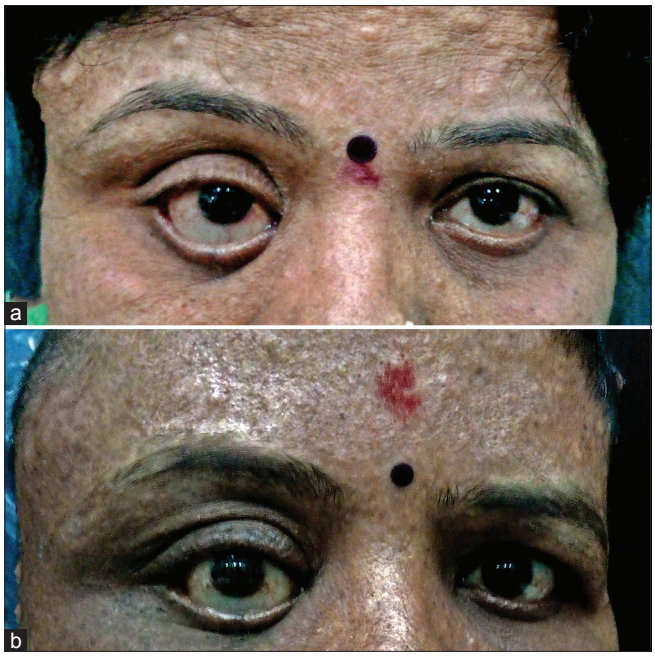

Figure 5: (a) Right eye proptosis preoperatively. (b) Postoperative day 7 on follow-up proptosis reduced

dysplasia, and pseudoarthrosis.[1] These defects are usually seen after a careful physical examination in asymptomatic individuals, with one eye appearing asymmetric (proptotic or sunken).[1] Exophthalmos, when present, is either associated with a mass lesion in the orbit or a buphthalmic globe due to congenital glaucoma. Exophthalmos accompanied by pulsating protrusion of the eye without bruit indicates a major congenital defect in the posterior wall of the orbit (sphenoido-orbital encephalocele).[6]

At present, the diagnosis of NF1 is made using established clinical criteria developed by the National Institutes of Health Consensus Conference in 1987 [Table 1].[7]

While no medical therapies are currently available, trials are ongoing to discover and test medical treatments for the various manifestations of NF1, primarily plexiform neurofibromas, learning disabilities, and optic pathway gliomas, which are a significant cause of morbidity in these patients.[5]

Orbital abnormality is seen rarely in NF1. Orbitotemporal manifestations in NF1 have been classified into 3 groups: (1) orbital soft-tissue involvement only with a seeing eye; (2) orbital soft-tissue and significant bone involvement with a seeing eye; and (3) orbital soft-tissue and significant bone involvement with a blind or absent eye.[8] Bone involvement mostly consist of sphenoidal dysplasia.

Surgical treatment is complicated due to high rate of pseudoarthrosis seen in NF1 patients. Surgical goal includes dural closure and restoration of the orbital cavity. This can be performed through intracranial approach or lateral orbital approach. Intracranial approach affords easier retraction, better exposure of bony defect, and preservation of optic nerve.[9] Orbital defect can be tackled with split bone graft or synthetic bone cement (PMMA) or titanium mesh. Acrylic has some advantages above metal substances; it is easy to shape, lighter in weight, radiates less heat, and radiolucent. Animal experiments revealed that acrylic adheres to the dura mater with no reaction to other underlying layers.[10]

Table 1: National Institutes of Health diagnostic criteria*[7]

Six or more café-au-lait macules >5 mm in greatest diameter in prepubertal individuals and >15 mm in greatest diameter in postpubertal individuals
Two or more neurofibromas of any type or one plexiform neurofibroma
Freckling in the axillary or inguinal regions
Optic glioma
Two or more Lisch nodules (iris hamartomas)
A distinctive osseous lesion such as sphenoid dysplasia or tibial pseudoarthrosis; or
A first-degree relative with neurofibromatosis 1 as defined by the above criteria

*Diagnosis is made when two or more criterias are present

Our patient was diagnosed with the help of clinical criteria. Clinical findings correlated with radiological findings. It belonged to group 2 who had sphenoidal dysplasia with herniating hemimegaencephaly [Fig. 5]. Surgical intervention was done for the functional improvement of the patient with preservation of vision. Intracranial approach with PMMA graft was preferred for promising field of exposure during intra-operative navigation and cost-effective for the patient. PMMA bone cement is easily moldable and comes considerably cheaper compared to titanium implants which are usually premolded and very expensive.

Conclusion

Multidisciplinary approach provides safe and optional cosmetic and functional outcome in patients with NF with sphenoid wing dysplasia, who need orbitocranial reconstruction.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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