NEUROIMAGING HIGHLIGHT

Rare Neuroimaging Findings in an Adult Neurofibromatosis Type 1 Patient

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Neurofibromatosis type 1 (NF1) is a rare neurocutaneous disorder characterized by nervous system tumors and dermatologic lesions.[1] Osseous lesions such as scoliosis and sphenoid dysplasia[2] may also be present, but calvarial defects are rarely seen in NF1.[1–3] We present a case of an adult NF1 patient with a large occipital calvarial defect and other associated findings.

The patient is a 25-year-old woman from the rural Philippines with a congenital right upper back mass. The gradual increase in size of the mass prompted consult. There was no family history of NF1.

On examination, there was a large, pigmented, pedunculated mass located at the right posterior neck, shoulder, and upper back (Figure 1(A)). There were also multiple papules and hyperpigmented patches on the face, trunk, and extremities. Her neurological examination was normal.

Cranial CT scan showed an 8.6×9.2 cm right occipital calvarial defect (Figure 1(B)–(C)). Contrast cranial MRI showed an occipital encephalocoele containing dysplastic cerebellum, a jugular foramen meningocele, and ectasia of the cerebellum and medulla (Figure 1(D)). The encephalocoele extended inferiorly to the midcervical area (Figure 1(E)–(F)). There was also a large, fungating, multilobulated, mildly enhancing soft tissue lesion that involved the cutaneous–subcutaneous right hemicalvarium, right auricular–periauricular, and right neck, shoulder, and upper back regions.

The patient underwent pre-operative embolization and subtotal excision of the mass. We decided to treat the calvarial defect and encephalocoele conservatively, since the patient was neurologically intact and asymptomatic from these lesions.

The diagnosis of NF1 was made based on the presence of more than six cafe-au-lait macules and one plexiform neurofibroma.[1] What makes this case rare is the presence of a calvarial defect and the associated encephalocoele, meningocele, and medullary and cerebellar ectasia. Moreover, to our knowledge, this is only the fourth reported case of NF1 with a jugular foramen meningocele.

Only 28 cases of calvarial defects in NF1 have been reported, and they commonly involve the parietal and occipital bones.[2–4] It was speculated that the mutation of neurofibromin, which has a role in bone metabolism, may lead to downregulation of osteoblastic and upregulation of osteoclastic activity, favoring bony dysplasia.[4]

Even rarer are the presence of meningoceles and cephaloceles, which were most likely formed as a consequence of a calvarial defect.[5] They are not typically seen as patients usually consult at a younger age, at an earlier period in the disease process.[4,5] The calvarial and neural ectasia in our patient may have progressed over time, resulting in the large extent and severity at present, since she did not seek consult until adulthood.

Calvarial defects and accompanying lesions in NF1 were usually treated with cranioplasty using autologous material or titanium with or without duraplasty or excision of dysplastic neural tissues.[1,4] In our patient, conservative treatment was advised because she was asymptomatic and had no neurologic deficits. This may be due to the long-standing nature of her condition, allowing the posterior fossa structures to gradually adapt to the new environment.

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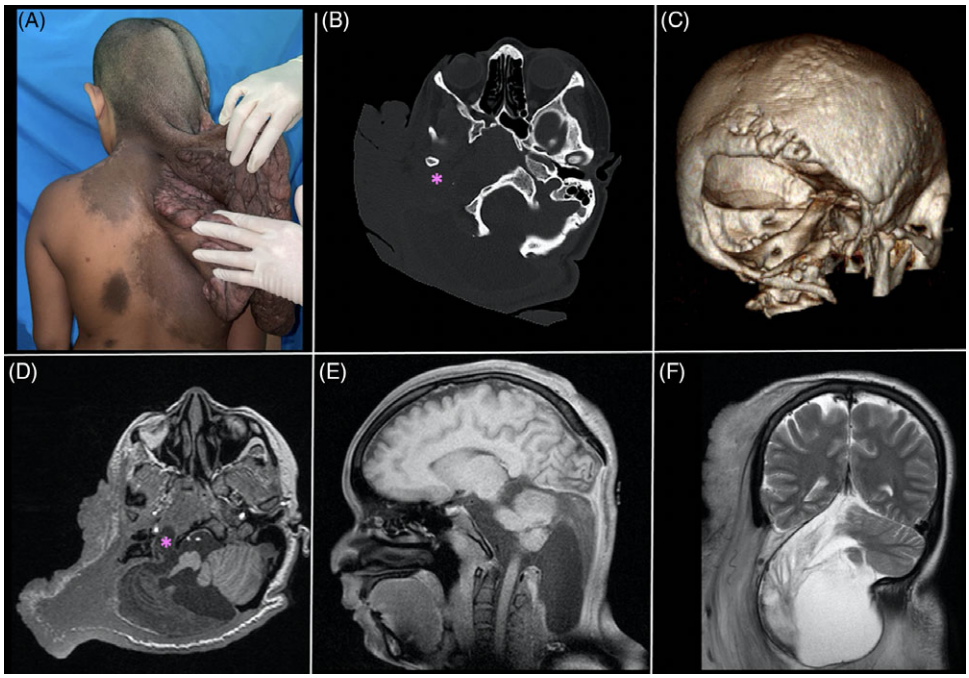

Figure 1: *(A) View of the patient from the back, showing a large cutaneous–subcutaneous lesion on the right posterior neck, shoulder, and upper back; also seen are hyperpigmented patches in the posterior trunk; (B) cranial CT, bone window, showing a calvarial defect in the right occipital area and obliteration of the right external auditory canal and adjacent bony structures (asterisk); (C) 3D reconstruction of the cranial CT showing the occipital calvarial defect and intact upper cervical vertebrae; (D) axial sections of contrast cranial MRI, T1 sequence, showing the dysplastic right cerebellar hemisphere herniating through the calvarial defect, along with dysplastic parts of the medulla and middle and inferior cerebellar peduncles. There was also a right jugular foramen meningocele extending to the right carotid space (asterisk); (E) sagittal sections of cranial MRI, T1 sequence, showing the inferior extent of the encephalocoele down to the soft tissues at the C4 spinal level; (F) coronal sections of cranial MRI, T2 sequence, showing the dysplastic right cerebellum within the encephalocoele. These cranial imaging findings were seen in the background of a large, fungating, multilobulated, mildly enhancing soft tissue lesion in the cutaneous–subcutaneous right hemicalvarium, right auricular–periauricular, right lateral, anterior, and posterior neck regions, right shoulder, and right upper back.*

Conflict of Interest

The authors declare no conflicts of interest.

Statement of Authorship

MUH: conceptualization; resource; data curation; writing – original draft preparation; writing – review and editing. JGP: conceptualization; resource; data curation; writing – original draft preparation; writing – review and editing. KPC: conceptualization; writing – original draft preparation; writing – review and editing. EBB: resource; data curation; writing – original draft preparation; GDL: writing – review and editing; supervision. KOK: writing – original draft preparation; writing – review and editing; supervision; project administration.

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