Segmental neurofibromatosis: An unusual association with ocular, skeletal, and cerebral anomalies

Sir,

Segmental neurofibromatosis (SNF) is an uncommon variant of neurofibromatosis that is characterized by café au lait macules and freckles or neurofibromas limited to one region of the body.[1] It is unilateral in majority with right side of the body being more commonly affected. Cervical and thoracic regions are usually involved. It affects all age groups with slight male preponderance.[2] Riccardi included it in his classification as neurofibromatosis type 5.[3] Roth *et al*. further classified SNF into four subtypes: true segmental, localized cases with deep involvement (nonfamilial), hereditary segmental (no deep involvement, familial), and bilateral segmental (no deep involvement, nonfamilial).[4] Systemic involvement is rare in SNF.[3]

Herein we describe a 43-year-old woman presented with asymptomatic raised skin lesions on left side of face since 3 years. Lesions were gradually increasing in size and number. On examination she had apparent facial asymmetry with left side of the face being less prominent than right side [Figure 1]. Eight skin-colored and hyperpigmented papules and nodules varying in size from 0.3 to 1 cm were present on left preauricular, retroauricular, and mandibular areas [Figure 2]. They were discrete, smooth, sessile, and soft in consistency.

Left eye revealed irregular pupil [Figure 3] and reduced visual acuity. Slit lamp examination showed pear-shaped pupil and absence of iris in temporal region. A part of

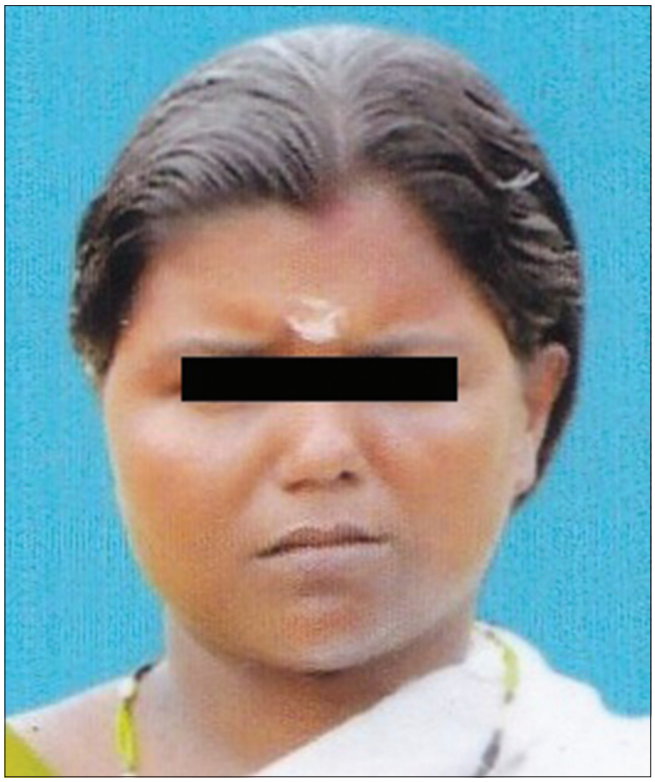

Figure 1: Asymmetry of face with hypoplasia of left side. The photo was taken after excision of neurofibromas

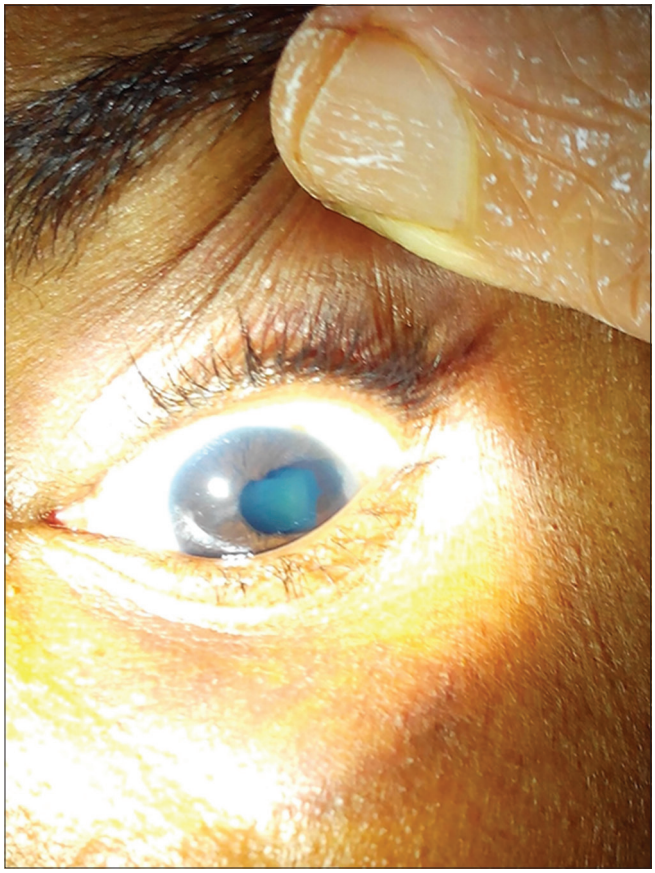

Figure 3: Irregular left pupil

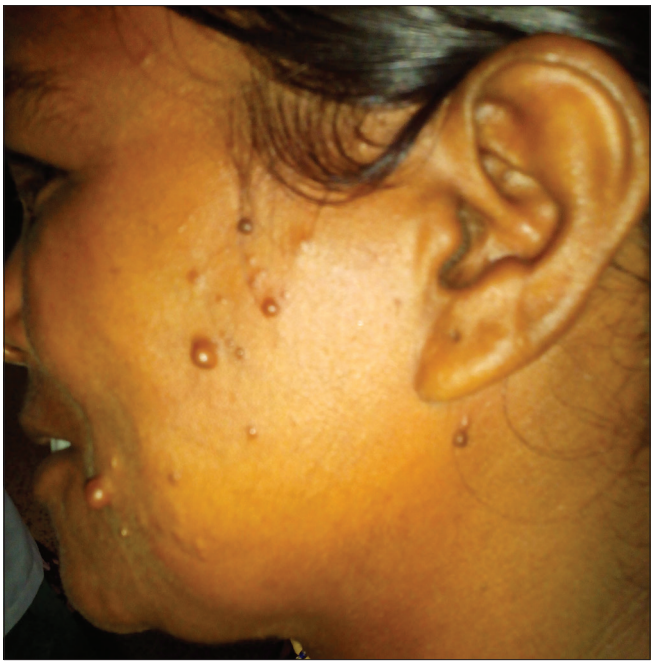

Figure 2: Neurofibromas over left side of face

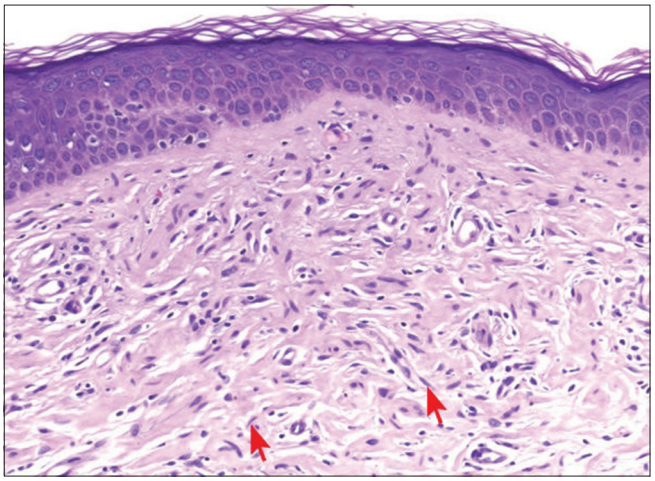

Figure 4: Histopathology of neurofibroma showing Schwann cells with wavy nucleus (H and E, ×40)

the iris showed forward extension into anterior chamber and was attached to the cornea giving a colobomatous appearance. Fundus examination showed deep optic disc cup with optic atrophy. Gonioscopy revealed poorly developed anterior chamber angle. Applanation tonometry showed raised intraocular pressure. Right eye was normal.

Other systems were within normal limits.

Histopathology of skin lesions showed a well-circumscribed neoplasm composed of spindle-shaped cells with wavy nuclei and moderate eosinophilic cytoplasm in dermis consistent with neurofibroma [Figure 4].

Plain computed tomography (CT) scan of head showed hypoplasia of maxillary sinus, alveolar arch, and inferior

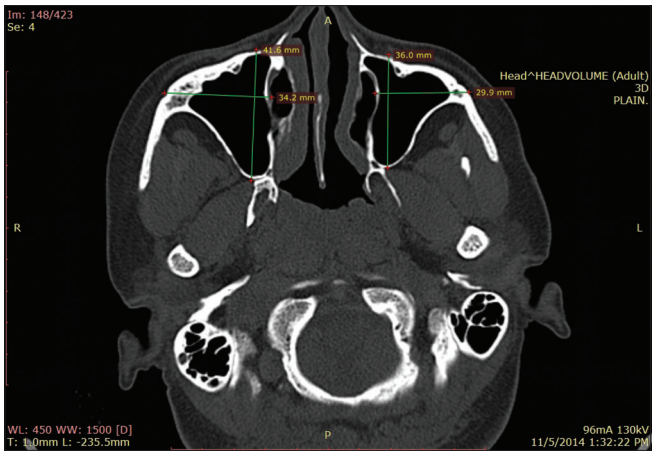

Figure 5: Axial computed tomography scan of head–bone window showing hypoplasia of left maxillary sinus

turbinate on left side [Figure 5]. Hypodense areas were seen in parietal white matter bilaterally. On T2-weighted sequence in magnetic resonance imaging (MRI) scan of brain, hyperintense lesions were identified in these areas suggestive of unidentified bright objects (UBO). The patient was diagnosed as having left facial SNF with facial skeletal hypoplasia, eye anomalies (colobomatous iris, angular dysgenesis) and bilateral UBOs in brain. Our final diagnosis was localized type of SNF with deep involvement. Neurofibromas were excised and she is taking treatment for glaucoma.

Systemic complications including ophthalmological and neurological involvement are rare in SNF.[3] Rare associations reported are optic pathway gliomas, peripheral nerve sheath tumors, ipsilateral renal agenesis, facial asymmetry due to asymmetric enlargement of right side of mandible, and agenesis of corpus callosum.[2] A case of SNF with ipsilateral thoracic skeletal hypoplasia has been reported.[5] UBOs in brain MRI is a common finding in NF1, it is thought to represent areas of abnormal myelination. It is associated with cognitive impairment if present in thalamus.[6]

SNF affecting the face with associated changes in bone, eye, and brain has not been reported. Knowledge of rare associations may help to know more about the pathogenesis of the disease. Detailed evaluation of cases of SNF may be required to rule out systemic involvement.

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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