

Plexiform neurofibroma in a 16-year-old girl

Reena Anand, DNB; D.S. Yadav, DNB; Vikram Yadav, MS; Deepali Yadav, MD; and Deepak Bhatia, MD

Plexiform neurofibroma is a rare, poorly defined benign tumor of the peripheral nerve sheath. It spreads out just under the skin, or deeper in the body, and occurs exclusively in patients with neurofibromatosis type I. Facial plexiform neurofibroma may produce various degrees of cosmetic and functional deformities in the head and neck region. It is a virtually pathognomonic and often disabling feature of neurofibromatosis type I. We present a case of plexiform neurofibroma in an 18-year-old female.

Case report

An 18-year-old female visited our diagnostic center with a chief complaint of a swollen area on the right side of the face. The patient first noticed the swelling ten years ago; it began insidiously and then grew slowly to attain its present size (Figs. 1A, 1B).

After an asymptomatic phase of nine years, the patient began to experience mild, intermittent, dull, aching pain in the swollen area, which continued over the past year. There was no history of any regression in size of the swelling or any discharge from the swollen area. The patient had never noticed similar swelling elsewhere in the body. Medical, surgical, and dental histories were unremarkable. Family history did not reveal any similar complaints in immediate or distant relatives.

The patient underwent several imaging procedures. Computed tomography (CT) showed overgrowth of the skin and subcutaneous tissues, with invasion and encasement of the muscles and soft tissues of the right side of the face (Figs. 2-4). There were dysplastic changes and deformity involving the right side of maxilla, mandible, and upper cervical vertebrae. There was scoliosis of the cervical spine.

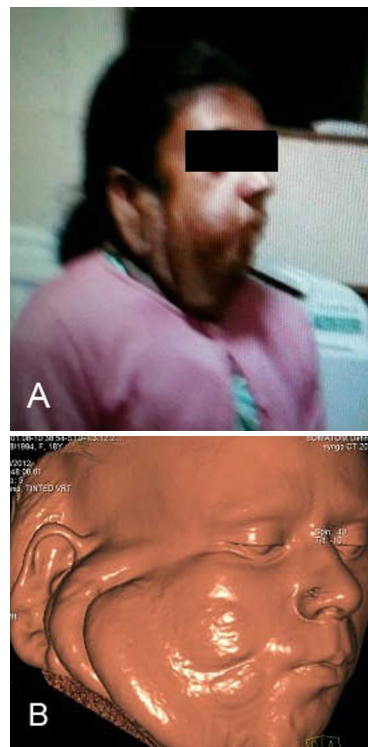


Figure 1. 18-year-old female with plexiform neurofibroma. A. Photograph. B. three-dimensional, volume-reformatted CT image of the face, showing swelling on the right side.

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The authors are all at the Modern Diagnostic and Research Centre, Gurgaon, India. Contact Dr. Anand at anand.reena@yahoo.com.

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Extension of the lesion was seen posterior to the back of neck and up to the right paravertebral region.

MRI showed overgrowth of the skin and subcutaneous tissue that was hyperintense on T2-weighted images (Figs. 5, 6).

Incisional biopsy was performed under general anesthesia. The histopathological sections showed an ill-defined lesion present diffusely in the dermis and subcutaneous

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Figure 2. 18-year-old female with plexiform neurofibroma. Axial CT image shows dysplastic changes and deformity involving right side of maxilla, mandible, and the upper cervical vertebrae.

tissue, enveloping the normal structures, consistent with a plexiform neurofibroma.

Discussion

The term neurofibromatosis describes a group of genetic disorders that primarily affect the cell growth of neural



Figure 3. 18-year-old female with plexiform neurofibroma. Coronally reformatted CT scan image extension of lesion posteriorly to the back of neck and up to the right paravertebral region.

tissues. At least eight forms of neurofibromatosis have been recognized, the most common form being neurofibromatosis type I (NF-I), also known as von Recklinghausen's disease, which accounts for about 90% of the cases. NF-I is estimated to occur in one of every 3,000 births. There is no sex predilection. It is an autosomal dominant disorder caused by a spectrum of mutations in the NF-I gene. It has one of the highest spontaneous mutation rates among genetic diseases. Only 50% of these patients have a positive family history of the disease; the other half represent spontaneous mutations (1).

Other possible abnormalities that may be seen include central nervous system tumors, macrocephaly, mental defi-

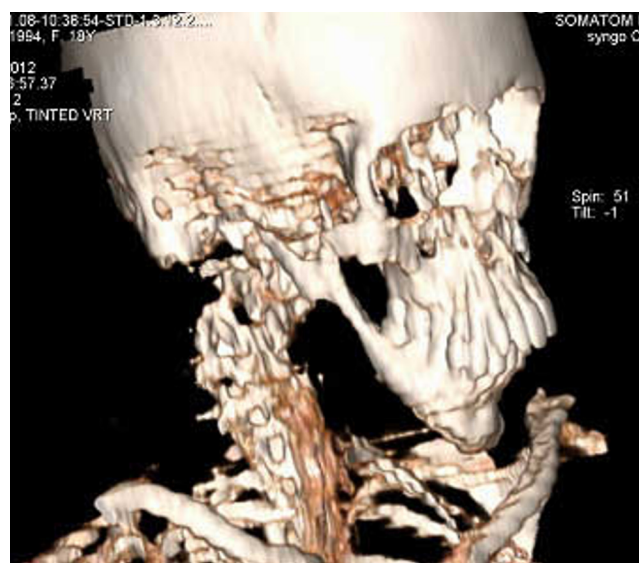


Figure 4. 18-year-old female with plexiform neurofibroma. Three-dimensional, volume-reformatted CT image of the skull, cervical spine, and upper chest demonstrating a dysplastic right mandible.

ciency, seizures, and short stature (2). Sexual precocity is seen in 3-5% of affected children. Our patient had numerous café au lait spots and one plexiform neurofibroma.

Neurofibromas are the most common benign tumors of NF-I. These can develop at any point along a nerve and often form by late adolescence. They are nodular and may be brown, pink, or skin-colored and soft to firm to touch. Three subtypes of neurofibromas exist: cutaneous, subcutaneous and plexiform. Cutaneous and subcutaneous varieties are not specific for neurofibromatosis, whereas the plexiform variety is specific for the disease.

Roughly 20-30% of patients with neurofibromatosis have axillary freckling, known as Crowe's sign. Both axillary freckling and inguinal freckling may develop during puberty. Areas of freckling and regions of hypertrichosis occasionally overlay plexiform neurofibromas. This freckling is not seen in patients without neurofibromatosis. Nodules (Lisch nodules) and café au lait macules of the iris bilaterally

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ally have been described as characteristic of neurofibromatosis in the ophthalmological literature (3).

Plexiform neurofibromas are benign tumors that spread out either just under the skin or deeper in the body. They originate from nerve-sheath cells or from subcutaneous or

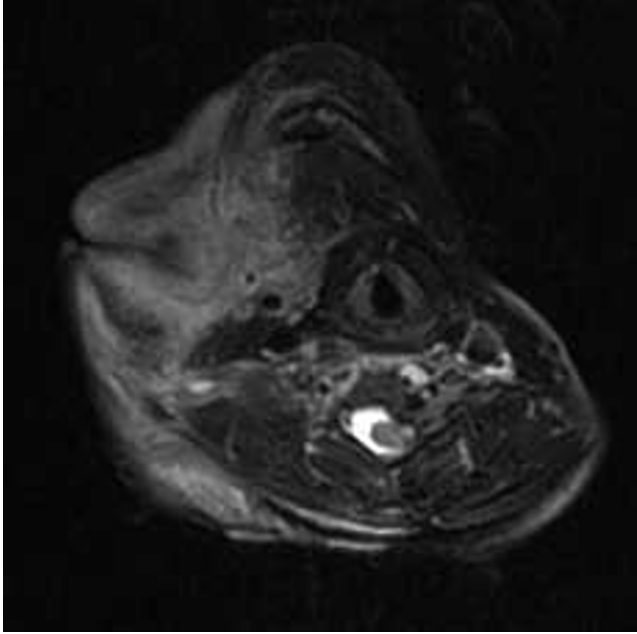


Figure 5. 18-year-old female with plexiform neurofibroma. T2-weighted axial MRI showing overgrowth of the skin and subcutaneous tissue of the right side of the face and neck with encasement of muscle, soft tissue, and superficial blood vessels.

visceral peripheral nerves and can involve multiple fascicles. The term "plexus" refers to a combination of interlaced parts or a network. Plexiform neurofibromas are uncommon and occur almost exclusively in about 5-15% patients with neurofibromatosis-I. Two types of plexiform neurofibromas have been recognized: (a) a diffuse type/ elephantiasis neurofibromatosa and (b) a nodular type (4, 5).

These plexiform neurofibromas can arise anywhere along a nerve and have poorly defined margins. They may appear on the face, legs, or spinal cord and frequently involve the cranial and upper cervical nerves. The cranial nerves most commonly involved in plexiform neurofibromas are the fifth, ninth, and tenth. These masses can be quite disfiguring, and hemifacial hypertrophy can occur (6). These tumors are known to cause symptoms ranging from minor discomfort to extreme pain. The consistency of the lesion has been compared to that of a "bag of worms" because of the presence of soft areas interspersed with firm nodular areas. This consistency was well appreciable in the lesion seen in our patient. These lesions sometimes demonstrate a vascular nature, and they may cause dangerous bleeding and complicate surgical procedures. The size of these tumors may increase during puberty and pregnancy.



Figure 6. 18-year-old female with plexiform neurofibroma. T2-weighted coronal MRI showing extension of the lesion up to the right paravertebral region. Note the cystic lesion measuring 30 x 15 mm in the base of the right neck.

Deep plexiform neurofibromas typically present with a target-like appearance on T2-weighted MR images, with central low-signal intensity and peripheral high-signal intensity. Superficial plexiform neurofibromas do not typically possess the target-like appearance seen in their deep counterparts and can be easily mistaken for other entities, such as venous malformations. Deep plexiform neurofibromas consist of a nerve or nerve fascicle distended by tumor cells, embedded in a rich myxoid matrix. Longitudinal bundles of residual nerve fibers are often centrally situated in the neurofibroma. This architecture could account for the centrally T2 dark (nerve fibers) and peripherally T2 bright (myxoid) appearance of these target-like lesions. The absence of a target-like appearance does not rule out a lesion being a neurofibroma, particularly if the lesion has a superficial location.

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