Plexiform neurofibroma encasing vital organs

Sir,

Neurofibromatosis (NF) is one of the most common neurocutaneous syndromes characterized by varied

clinical manifestations including café-au-lait macules and nerve sheath tumors. A century ago, von Reckinghausen described neurofibromatosis in his patients- Marie Kientz and Michael Bar.[1] Diagnosis of NF is based on the criteria established by NIH (National Institutes of Health Consensus Conference), which classified neurofibromatosis into eight subgroups.[2] Four types of neurofibromas have been described, namely, cutaneous neurofibromas, subcutaneous, nodular and diffuse plexiform neurofibroma.[3] Plexiform neurofibroma is an uncommon type of neurofibromatosis, commonly seen along the branches of trigeminal nerve.[4] We herewith report a case of large, diffuse, plexiform neurofibroma in a child.

A four-year-old female child was brought with large hyperpigmented patch involving the trunk since birth, associated with underlying swelling on the left side of the abdomen. There was no positive family history. Cutaneous examination revealed a large café-au- lait macule with well-defined margins measuring 25 × 30 cm, involving the left side of the anterior abdominal wall extending to the back. There was a diffuse subcutaneous swelling underneath the café-au-lait macule measuring 10 × 12 cm. Swelling was firm in consistency, nontender and not compressible. Overlying skin showed hyperpigmentation and hypertrichosis, with follicular prominence. On the basis of these clinical findings, the patient was diagnosed with plexiform neurofibroma. Ultrasonography of the abdomen revealed soft tissue swelling. MRI of the spine showed multiple, soft tissue masses in the paraspinal region extending anteriorly into the retroperitoneum encasing the aorta and its branches and the pancreas, extending along the inferior aspect of lower ribs. MRI diagnosed it as neurofibroma. Biopsy taken from the subcutaneous swelling along with the skin confirmed the diagnosis of plexiform neurofibroma, which showed spindle cells in bundles consisting of nerve cells and fibroblasts surrounded by connective tissue stroma.

Plexiform neurofibroma (PFN) is considered to be a hamartoma than a typical tumor.[4] It may be nodular plexiform or diffuse plexiform. Diffuse plexiform neurofibroma accounts for about 5% of neurofibromatosis type I and is always congenital and pathognomonic of neurofibroma type I.[3] It is highly vascular and may involve all the layers of skin, adjacent fascia and deeper elements at times even replace the muscle and may erode the bone and infiltrate viscera. PFN is usually asymptomatic but can cause pain, impairment of function and disfigurement. Plexiform neurofibroma is commonly seen along the branches of trigeminal nerve. Orbital and periorbital regions of the face are the most common sites of involvement.[5] The risk of malignancy is 5% to 10%.[3] Tonsgard *et al*., have reported a case of plexiform neurofibroma, which involved retroperitoneum, mediastinum and paraspinal region.[6] The case under discussion is comparable with the case report by Tonsgard *et al*. Plexiform neurofibromas involving gastrointestinal tract is rare. These patients may present with epigastric pain, motility disorders, dyspepsia, anemia, hematemesis, intussusception, volvulus, intestinal perforation or bowel obstruction.[7] Zacharia *et al*., while reviewing MRI findings of the abdominopelvic neurofibromatosis type I, found that the abdominopelvic wall was the most common site followed by lumbosacral plexus and retroperitoneum.[8]

Surgical resection is the treatment required in this case. However, resection may not be advisable at this stage as the tumor involved vital structures and there are no pressure symptoms. The child has been under follow-up for the last two years and has no pressure symptoms. Prognosis is not good in this case as the tumor is likely to increase in size and cause pressure symptoms in future. The patient under study presented with large PFN with the involvement of vital organs. Hence, it is mandatory to suspect and investigate for internal organ involvement in all cases of PFN.

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REFERENCES

1. Von Recklinghausen FD.Ueber die in multiplen Fibrome der Haut and ihre Beziehung zu den multiplen Neuromen. Berlin: Hirschwald; 1882.
2. Stumf DA, Alksne JF, Annergers JF. Neurofibromatosis: Conference statement, National Institute of Health development conference. Arch Neurol 1988;45:575-8.
3. Pivnick EK, Riccardi VM. The neurofibromatosis. In: Freedberg IM, Eisen AZ, Wolff K, Austen KF, Goldsmith LA, Katz SI, *et al*. editors, Fitzpatrick's dermatology in general medicine. 5th ed, vol. 2, New York: McGraw Hill; 1999. p. 2152-8.
4. Sengupta SP. Tumours and cysts. In: Long and short cases in surgery. 1st ed, Calcutta: New Centre Book Agency Publications; 1996. p. 39-75.
5. Riccardi VM. von Recklinghausen neurofibromatosis. N Engl J Med 1981;305:1617-27.

6. Tonsgard JH, Kwak SM, Short MP, Dachman AH. CT imaging in adults with neurofibromatosis I: Frequent asymptomatic plexiform lesions. Neurology 1998;50:1755-60.
7. Hochberg FH, Dasilva AB, Galdabini J, Richardson EP. Gastrointestinal involvement in von Recklinghausen's neurofibromatosis. Neurology 1974;24:1144-51.
8. Zacharia TT, Jaramillo D, Poussaint TY and Korf B. MR imaging of abdominopelvic involvement in neurofibromatosis type I: A review of 43 patients. Pediatr Radiol 2005;35:317-22.