Neurofibromatosis type 1: a rare cause of parotid swelling in a child

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Von Recklinghausen disease, also known as neurofibromatosis type 1, is an autosomal dominant disorder that presents as neurocutaneous syndrome. These patients have increased chances of developing other tumors such as plexiform neurofibromas. Plexiform neurofibromas are a proliferation of Schwann cells in the nerve sheath. Affliction of the parotid gland in a young child is a rare presentation of these tumors. We present the management of one such case. ***Ann Pediatr Surg*** **13:155–156 © 2017 Annals of Pediatric Surgery.**

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Introduction

Neurofibromatosis constitute a group of autosomal dominant neurocutaneous disorders affecting the nervous system, the bone, soft tissue, and the skin. Von Recklinghausen disease is a type 1 neurofibromatosis (NF1). NF1 is characterized by cutaneous neurofibromas, café au lait spots, Lisch nodules, and freckling in non-sun-exposed areas with increased risk for nervous system neoplasm. Occasionally the patient may develop plexiform neurofibromas of the salivary gland. Here we present the clinical and radiological presentation of one such patient.

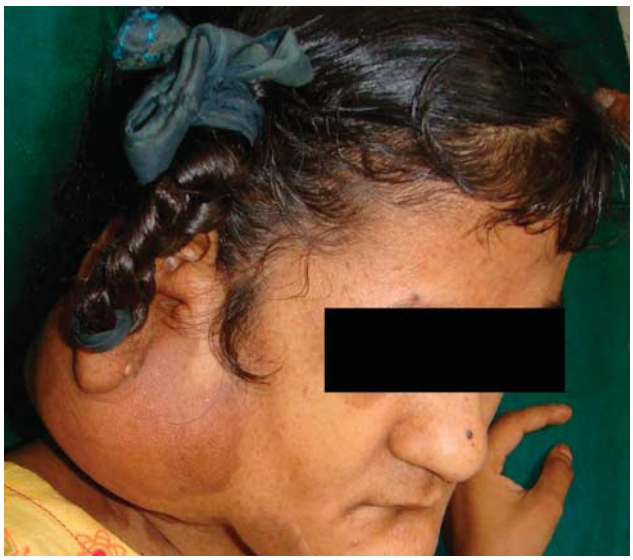
Case report

A 14-year-old female child was brought to the outpatient department by her parents with a complaint of abnormal pigmentation all over her body since childhood and recent development of painless right-sided facial swelling. The swelling was initially small in size but rapidly grew in size over the past 6 months and extended behind the right ear and descended to involve the neck with development of some tenderness. On examination, a swelling of 12×15 cm in size was found to be present in the right parotid region pushing the ear outwards (Fig. 1). It was slightly tender, hard in consistency, free from skin, but fixed to underlying structures. There were around 20 café au lait macules all over the buttocks, torso, and legs. Kyphoscoliosis was also noted. A diagnosis of von Recklinghausen disease was made on clinical examination. The patient's father was also found to be affected with abnormal pigmentation and multiple swellings involving the head and neck region.

Chest radiography showed thoracic scoliosis with deformity in the left clavicle with thinning of ribs and mediastinal widening. ECG showed left ventricular hypertrophy with a strain pattern. Contrast-enhanced computed tomography (CT) showed a heterogenous soft-tissue mass with extensive central hypodensity necrosis with involvement of the right parotid region, extending into the ipsilateral parapharygeal space, masticator space, and pterygoid fossa, involving the pharyngeal mucosal space, medially causing luminal compromise and posteriorly abutting and compressing the ipsilateral internal carotid and internal jugular vein (Fig. 2). The subsequent MRI confirmed the findings of the CT scan with multiple bilateral neuromas in the region of the face and neck appearing isointense to hypointense on T1-weighted scans and hyperintense on T2 with strong postcontrast enhancement. The lesion on the right side involving the right parotid region appeared to extend widely into the masticator, parapharyngeal, and pharyngeal mucosal space with large areas of necrosis. However, there was no evidence of invasion of the skull base or intracranial extension.

Perioperatively the tumor was found to involve the superficial lobe of the parotid gland with some infiltrations of the deep lobe as well. The tumor was completely dissected with preservation of the facial nerve. The tumor probably involved the anterior branch of the great auricular nerve, which is the other nerve traversing the parotid gland substance. Histopathologic examination showed diffuse infiltration of spindle cells in a myxoid background with

Fig. 1



Clinical photograph showing the parotid swelling.

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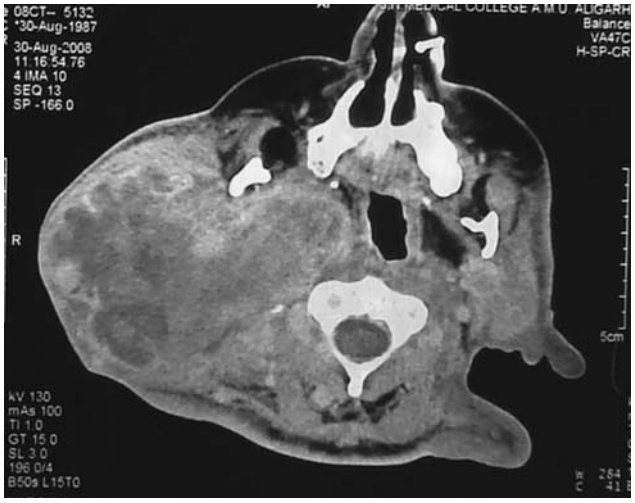
Fig. 2



Contrast-enhanced computed tomography showing extensive a heterogeneous soft-tissue mass with involvement of the right parotid region, extending into the ipsilateral parapharygeal space, masticator space, and pterygoid fossa.

areas of dense encapsulations of spindle cells in between. These features were consistent with plexiform neurofibromas. In the postoperative period the patient recovered well, with no evidence of facial nerve involvement.

Discussion

Two distinct variants of NF1, also known as von Recklinghausen disease, and type 2 (NF2), or bilateral acoustic neurofibromatosis. Both have distinct presentations. NF1 accounts for more than 90% of all cases of neurofibromatosis [1]. Besides cutaneous neurofibroma, plexiform neurofibromas are frequently seen in patients with NF1. Histologically they are characterized by a proliferation of Schwann cells in the nerve sheath, and by mast cells and perineurial and endoneurial fibroblasts in a myxoid matrix.

They are locally infiltrative benign tumors that tend to grow slowly [2]. Although they may grow any time, growth spurts are particularly seen in early childhood and during puberty or pregnancy. These tumors arise in various regions of the body, including the trunk, limbs, head, and neck [3,4].

These tumors may remain asymptomatic for many years but can cause cosmetic abnormalities, pain, and functional deficits as well [2–4].

Cross-sectional imaging including CT and MRI are the investigations of choice. On CT they appear hypoattenuated, whereas on T1-weighted MRIs they are isointense to hypointense. These tumors may show a 'target sign' on T2-weighted images with peripheral hyperintense signal intensity and central isointense to hypointense signal intensity. Contrast enhancement may be heterogeneous [4]. Tissue biopsy confirms the diagnosis.

If the tumor is causing cosmetic disfigurement or there is a sudden increase in size causing pain or nerve dysfunction then excision of the tumor, as much as possible, should be considered [5]. A recurrence rate of 20% is reported in the patients who have undergone complete resection while the figure reaches 44% with subtotal resection [6]. The risk factors for recurrence are young age, tumors of the head and neck region, and incomplete resection. Success of surgical intervention is limited by the infiltrative nature of the tumors. Malignant peripheral nerve sheath tumors are reported to occur in 2–5% of patients with NF1 [7].

Conclusion

Presentation of plexiform neurofibroma in association with NF1 is rare. Imaging, particularly MRI, can aid in diagnosing and planning the subsequent operative approach and follow-up. Because of its infiltrative and aggressive nature it presents a surgical challenge. Surgery is recommended to relieve cosmetic and compressive symptoms.

Conflicts of interest

There are no conflicts of interest.

References

1 Ferner RE. Neurofibromatosis 1 and neurofibromatosis 2: a twenty first century perspective. *Lancet Neurol* 2007; **6**:340–351.
2 Tonsgard JH. Clinical manifestation and management of neurofibromatosis type 1. *Semin Pediatr Neurol* 2006; **13**:2–7.
3 Imazu M, Nakamura Y, Nakatani H, Kaneda H, Okamura K, Sato O, Hayase Y. Cervicothoracic malignant peripheral nerve sheath tumor in a 12-year-old girl with neurofibromatosis type 1. *Eur J Pediatr Surg* 2006; **16**:285–287.
4 Losty P, Hu C, Quinn F, Fitzgerald RJ. Gastrointestinal manifestation of neurofibromatosis in childhood. *Eur J Ped Surg* 1993; **3**:57–58.
5 Lin J, Martel W. Cross-sectional imaging of peripheral nerve sheath tumors: characteristic signs on CT, MR imaging, and sonography. *Am J Roentgenol* 2001; **176**:75–82.
6 Needle MN, Cnaan A, Dattilo J, Chatten J, Phillips PC, Shochat S, *et al.* Prognostic signs in the surgical management of plexiform neurofibroma: the Children's Hospital of Philadelphia experience, 1974–1994. *J Pediat* 1997; **131**:678–682.
7 Ferner RE, Gutmann DH. International consensus statement on malignant peripheral nerve sheath tumors in neurofibromatosis 1. *Cancer Res* 2002; **62**:1573–1577.