Head and Neck Plexiform Neurofibromas: Case Discussion and Literature Review

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Abstract

Introduction: Plexiform neurofibromas (PN) are a rare benign neoplasm with an approximately 5% lifetime risk of malignant transformation which may arise in association with neurofibromatosis type 1 (NF-1). The aim of this study is to characterize the diverse phenotypic manifestations of PNs within the head and neck, including tumor subsite and clinical presentation.

Methods: A PubMed and Scopus databases were queried using a comprehensive keyword search in accordance with PRISMA guidelines. All English-only texts reporting any patient-specific data were included. Patient demographics, clinical presentation, and treatment approach data were collected.

Results: Of the 89 studies included in this investigation, all were retrospective. Sample size ranged from one to 63 patients, and a total of 186 patients were reported. The age at diagnosis ranged from two days to 76 years old, with a mean of 27.0 (± 13.3) years. 53.8% of the included patients were male, and 64.7% had a concurrent diagnosis of NF-1. PNs were observed in a myriad of head and neck subsites, with the most commonly reported being the superficial soft tissues of the neck (30.4%), followed by the parapharyngeal space (22.8%) and periauricular region (16.1%). Three patients developed malignancy at the site of their plexiform neurofibroma (malignant peripheral nerve sheath tumor, n = 2; angiosarcoma, n = 1). Most patients were offered definitive or subtotal surgical resection depending on disease extent, although sclerotherapy and radiofrequency ablation were also reported.

Conclusions: The presentation and distribution of head and neck PNs demonstrates significant phenotypic heterogeneity. An association with NF-1 is less common when the first diagnosis arises in adulthood. Due to the rarity of head and neck PNs and potential morbidity associated with definitive resection, future investigations are warranted to further characterize variants of this disease and appropriate management.

Background

- ❖ Plexiform neurofibroma (PN) is a rare, benign tumor originating from peripheral nerve sheath.
- ❖ PNs may arise sporadically but are most commonly observed in patients with neurofibromatosis type 1 (NF-1).
- ❖ 20-50% of patients with NF-1 develop PNs, among other tumors subtypes.
- ❖ PNs may affect any nerve however approximately 50% arise in the head and neck.
- ❖ Depending on their size and location within the head and neck, the morbidity of PNs may range from minimal disfigurement and mild pain to life-threatening airway obstruction.
- ❖ The primary management strategy for PN is surgical resection however approach may be limited by the extent of disease and patient morbidity.
- ❖ The heterogenous presentations of head and neck PN arising sporadically have not been well-characterized.

Methods

Case Presentation

An 18-year-old female with a history of a right facial and orbital plexiform neurofibroma diagnosed in childhood and who underwent two prior incomplete resections presented with right vision loss and a large facial mass. Imaging demonstrated a heterogenous, hypervascular mass surrounding and entering the right orbit with extension to the right masticator space and infratemporal fossa **(Figure 1).** Angiography revealed mass to receive primary vascular supply from the facial and ophthalmic arteries. She passed right carotid balloon occlusion testing and underwent preoperative embolization of the right ophthalmic artery, middle meningeal artery, and external carotid branches.

She underwent radical resection of the tumor including right orbital exenteration, partial maxillectomy, subtotal parotidectomy, infratemporal fossa, and pterygopalatine fossa resection followed by latissimus dorsi free flap reconstruction with Otolaryngology and Neurosurgery. She continues to do well postoperatively without evidence of disease recurrence.

Literature Review

- ➢A literature review was conducted using the PubMED/MEDLINE, and Scopus databases. Search terms employed were “plexiform neurofibroma” AND “head”; “neck”; “oral cavity”; “oropharynx”; “larynx”; ”hypopharynx”; “nasal”; “sinus”; “salivary”; OR “cutaneous”.
- ➢Database queries included publications from date of inception to December 2022.
- ➢Review was conducted in accordance with PRSIMA guidelines.

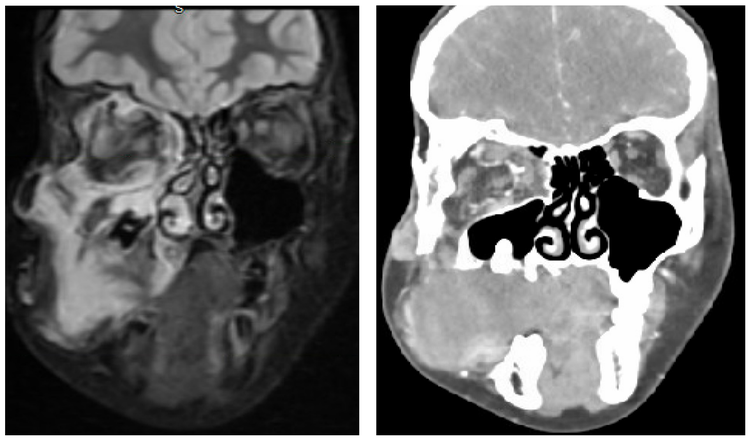

Figure 1.
Coronal T1 MRI and CT images of large right facial plexiform neurofibroma.

Results

- ❖ 186 patients with diagnosed PN identified through literature review.
- ❖ 64.7% of reported patients with NF-1
- ❖ Demographics:
  - **Age:** range 2 days to 76 years old
    mean 27 (± 13.3) years
  - **Sex:** 53.8% male
- ❖ Tumor subsites:
  - **Cutaneous most common (30.4%)**
  - Parapharyngeal space (22.8%)
  - Periauricular soft tissue (16.1%)
- ❖ **1.6% diagnosed with malignancy at tumor subsite**
  - Malignant peripheral nerve sheath tumor (n=2)
  - Angiosarcoma (n=1)
- ❖ Vast majority of patients underwent partial or complete surgical resection to relieve tumor burden
- ❖ Tracheostomy required if tumor associated with upper airway obstruction
- ❖ Other reports therapies included sclerotherapy, radiofrequency ablation

Discussion

- ❖ PNs of the head and neck display a vast spectrum of presentations by subsite and extent of disease, and tumors can develop at any age.
- ❖ Patients with NF-1 tend to develop PN at younger age than those with sporadic disease.
- ❖ Surgery is the first-line therapy however curative treatment may not be possible depending on associated morbidity.
- ❖ The case highlighted demonstrates a highly morbid presentation of plexiform neurofibroma involving multiple head and neck subsites treated successfully through a multidisciplinary surgical approach.

Conclusions

PNs present heterogeneously within the head and neck, and an individualized approach must be curated according to disease subsite and goals of care.

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