Surgical Management of an Unresectable Trigeminal Nerve (V2) Plexiform Neurofibroma

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Introduction

Neurofibromatosis is an autosomal dominant condition which affects 1 in 4,000 births, known to be caused by a genetic defect on chromosome 17. Clinically, this disease process is characterized by numerous cutaneous or subcutaneous neurofibromas, optic gliomas, and café au lait spots. Plexiform neurofibromas are slow growing, infiltrative tumors associated with Type 1 Neurofibromatosis (von Recklinghausen’s disease). Plexiform neurofibromas are most often noted in the head, neck, and face while less commonly seen in the spine, mediastinum, and abdomen. They are general slow growing tumors, however can undergo rapid growth during periods of hormonal changes including infancy, puberty as well as pregnancy. Plexiform neurofibromas are histologically characterized by a proliferation of Schwann cells within a nerve sheath and involve multiple nerve fascicles. They are known to be locally invasive benign tumors, however have the potential to undergo malignant change. Approximately 5% of these tumors undergo transformation into a malignant peripheral nerve sheath tumor (MPNST). The management of plexiform neurofibromas is surgical excision, as these infiltrating tumors are radioresistant and given their slow growth, chemotherapy is not beneficial. Unfortunately, because these tumors are infiltrating, sacrificing vital structures may be required in order to improve function and prevent further progression of this debilitating disease process.

Methods

We present a case of a 9 year-old male evaluated for a previously diagnosed plexiform neurofibroma presenting as a left mid-facial mass with palatal involvement. The patient had developed worsening function with oral competence, oral consumption of food, nasal obstruction, and ptosis of the left lower eyelid. [Figure A] Pre-operative imaging showed a poorly defined mass in the distribution of the second branch of the trigeminal nerve (V2) with extension into the hard and soft palate, soft tissues of the middle and nose, pterygopalatine fossa, and orbital apex. [Figure B] Radiologically, lesion was suspected to originate from the foramen rotundum. In light of the functional compromise caused by this lesion, it was felt this child would benefit from surgical debulking.

Results

The patient underwent tumor debulking with an infrastructure maxillectomy, cervicofacial advancement flap and reconstruction to address his ptosis. Intraoperatively, dilation of the infraorbital foramen was noted. [Figure C] He subsequently underwent second stage procedures which included endoscopic transnasal debulking and medial orbital decompression. His subsequent surgical procedures focused on maintaining oral function, preservation of orbital function and debulking of facial and periorbital skin redundancies that compromised function and improved appearance. He also underwent oral rehabilitation with maxillary obturator construction. Histopathology displayed a characteristic plexiform pattern of intertwined spindle-shaped cells grouped in twisted cords surrounded by connective tissue fibers. [Figure D] Immunohistochemistry demonstrated tumor cells staining positive for S-100 and Mart-1. The patient was discharged home post-operatively and seen in follow-up on multiple occasions two years after his initial debulking procedure. He currently receives close observation.

Conclusions

We present a case illustrating the complexity of the surgical management seen with unresectable tumors. It is vital to understand the association of plexiform neurofibromatosis with neurofibromatosis type 1 (von Recklinghausen’s disease) as well as its infiltrative nature. One must also appreciate the clinical assessment, radiologic presentation and histopathologic findings seen in NF-1 patients with plexiform neurofibroma. Finally, one must be able to recognize the clinical issues that present with surgical management of unresectable diseases processes. Although benign in nature, plexiform neurofibromas can be functionally debilitating. The management of unresectable disease is challenging but can provide functional benefit for selected patients when the goals of treatment are well-defined.

References