Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder associated with increased incidence of neoplasms. The incidence of NF1 is estimated to be 1/4000. The gene has been localized to 17q11.2, which encodes the protein neurofibromin, known to be important in cell growth and regulation. Previously, estimates of head and neck tumors in patients with NF1 have ranged from 25–30% [1–3]; however, the incidence of cervical soft tissue tumors and the value of screening MR for children with NF1 are not known.

Purpose. The purposes of this study were to determine the incidence and clinical significance of cervical tumors seen on MR imaging in children with NF1.

Materials and methods. A retrospective review of the brain and orbit MR with cervical images obtained on 95 children who meet the NIH consensus criteria for NF1 and who are followed at our neurofibromatosis clinic was carried out.

Results. Cervical tumors were found on MR imaging in 21 of 95 (22%) children. Of 21 children with cervical tumors, 14 children were determined to be surgical candidates. In nine children, MR imaging altered the clinical management by demonstrating tumors for which surgery was indicated, but the tumors were not suspected prior to MR imaging.

Conclusion. Cervical tumors are commonly seen in children with NF1. MR imaging may demonstrate a significant number of tumors that require surgery, but were not suspected prior to MR imaging.

Introduction

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Abstract

Background. Children with neurofibromatosis type 1 (NF1) are at increased risk of developing plexiform neurofibroma throughout the body, including the cervical soft tissues. However, the incidence of cervical soft tissue tumors and the value of screening MR for children with NF1 are not known.

Purpose. The purposes of this study were to determine the incidence and clinical significance of cervical tumors seen on MR imaging in children with NF1.

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in this study. All abnormal mass lesions greater than 2 cm in maximum diameter demonstrating enhancement with intravenous contrast were considered to be tumors (Fig. 1). Children who had other known malignancies, lymphoproliferative disorders, or causes for significant lymphadenopathy that may present as cervical mass, such as lymphoma, were excluded from the study.

The clinical significance of MR detection of cervical tumors was determined by review of the clinic and hospital charts. If the findings of MR imaging altered the course of planned treatment prior to the MR examination, the findings were considered to be significant. For the purpose of surgical management, if the tumor did not disfigure the patient or compromise the neuromuscular, respiratory, or vascular function so that surgical debulking was not considered necessary by the patient or craniofacial reconstructive surgeon, the tumor was categorized as an incidental tumor.

**Results**

There were 96 children who were newly diagnosed as having NF1 between 1990 and 1996 and followed at our neurofibromatosis clinic. One child also had lymphoma and was excluded from the study. The 95 children’s ages ranged from 1 month to 17 years (mean, 6 years) at the time of the initial MR examination.

Twenty-one children (22 %) had MR evidence of tumors in the cervical soft tissue. There was no significant difference in the age of the children with cervical tumors (1–14 years, mean age = 5.5 years) compared to the study group. All lesions were identified along the carotid artery and were visible on the postcontrast T1 fat-saturation coronal images as enhancing masses (Fig. 2). None of the tumors demonstrated areas of cystic degeneration or hemorrhage. None of the children had visible hypertrophy of the adjacent bony structures. All tumors were visible on the initial MR examination. Significant growth was seen in one child on a follow-up MR examination at 1 year. Subsequent to the second MR examination, surgical debulking was considered necessary.

Fourteen children (15 %) required surgical debulking of the cervical tumors, while seven children had small cervical tumors that were not resected. The average size of the tumors that required debulking was 5 cm, while those that did not require debulking were 2.5 cm. The children who required surgical debulking were slightly older (2–14 years, mean 5.5 years) than those who did not (1–12 years, mean 3.5 years). The majority of children with tumors that required surgical de-

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**Table 1** NIH consensus diagnostic criteria for neurofibromatosis type 1 [1]

The diagnosis of NF1 requires two or more of the following:

1. Six or more café au lait macules over 5 mm in greatest diameter in prepubertal individuals and over 15 mm in greatest diameter in postpubertal individuals
2. Two or more neurofibromas of any type or one plexiform neurofibroma
3. Freckling in the axillary or inguinal regions
4. Optic glioma
5. Two or more Lisch nodules (iris hamartomas)
6. A distinctive osseous lesion such as sphenoid dysplasia or thinning of long bone cortex, with or without pseudarthrosis
7. A first-degree relative (parent, sibling, or offspring) with NF1 by the above criteria

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**Fig. 1** One-year-old child with incidental cervical tumor. Coronal fat-saturated enhanced T1-weighted image (TR 600, TE 25) demonstrates a homogeneously enhancing tumor (arrow) coursing along the right carotid artery. This tumor measured 3 × 2 × 1 cm in diameter

**Fig. 2** Three-year-old child with a large cervical plexiform neurofibroma. Coronal fat-saturated enhanced T1-weighted image (TR 600, TE 25) of the cervical soft tissues demonstrates multiple enhancing tumors (arrows) that extended inferiorly into the thorax, but were not clinically suspected prior to MR examination. The tumor extended along the carotid and jugular vessels and the trachea. Due to the extent and impending compromise of the airway, the tumor was surgically debulked.