Intramuscular Myxoma of the Neck in Mazabraud Syndrome

Yi-Hsuan Emmy Wu MD1; Jagdish Dhingra MD1,2
1. Department of Otolaryngology-Head & Neck Surgery, Tufts Medical Center, Boston, MA
2. ENT Specialists, Brockton, MA

INTRODUCTION

- Mazabraud syndrome (MS) is a rare disease characterized by the combination of skeletal fibrous dysplasia and intramuscular myxoma. The association of soft tissue myxomas with fibrous dysplasia was recognized by Henschen in 1926.

- Mazabraud et al. proposed the existence of a syndrome described by intramuscular myxoma and fibrous dysplasia in 1967.

- Approximately 80 cases of MS have been reported, with most myxomas occurring in the thigh.

- A case of MS with an intramuscular myxoma in the head and neck region has not been reported previously. Here we present a new case of MS in a patient with a myxoma in the neck and a history of fibrous dysplasia.

CASE PRESENTATION

- A 46 yo male with a history of polyostotic fibrous dysplasia presented with a slowly enlarging, but otherwise asymptomatic neck mass of 6 weeks duration, with no response to oral antibiotics.

- Physical examination revealed a discrete, round, firm, non-tender deep neck mass with limited mobility measuring 2.5 cm across in the left supraclavicular region.

- Fine needle aspiration showed loose clusters of spindle cells without lymphocytes in the specimen (figures 1a, 1b).

- CT of the neck and chest with contrast showed a 2.7 cm well-demarcated homogeneous solid lesion with minimal contrast enhancement (figure 2).

- The mass was excised and histological examination revealed fibromyxoid features with nodular myxoid separated by collagenous areas. Immunohistochemistry was positive for vimentin and CD 34, and negative for MUC4. A diagnosis of juxta-articular intramuscular myxoma was made (figures 3, 4, 5).

DISCUSSION

- Myxoma is a benign neoplasm that is usually diagnosed in the sixth to seventh decades of life. Most patients present with a painless mass of long duration due to relative lack of symptoms.

- In MS, myxomas are frequently multiple and appear in close proximity to the region of the most severely affected dysplastic bone. Females are affected twice as often as males.

- Myxomas are more common with the polyostotic form of fibrous dysplasia.

- Myxomas appear as a well-defined mass on MRI, with low signal intensity on T1 weighted sequences and high signal intensity on T2 weighted images.

- McCune-Albright syndrome (MAS) is defined by the combination of fibrous dysplasia, brown pigmented areas of skin (café au lait spots), and endocrine disorders. MAS, monostotic fibrous dysplasia, and polyostotic fibrous dysplasia have the same mutation in the GNAS1 gene located on chromosome 20q13.2-q13.3 encoded to a Gs(α) protein. Mutations increase protein activity and abnormal proliferation.

- A major clinical overlap exists between MS and MAS. GNAS1 mutations also commonly occur in intramuscular myxomas, and MAS has been reported to be associated with 26 of 80 cases of MS reported as of January 2011.

- Recommended therapy for myxoma is excision if pain or pressure symptoms develop. Awareness of MS, especially when the myxoma is solitary, can prevent misdiagnosing a malignancy and avoid unnecessary wide excision and radiation.

- There is a greater risk of malignant transformation of fibrous dysplasia into sarcoma in patients with MS, versus patients with fibrous dysplasia alone. A high level of suspicion and long-term follow-up for patients with MS is necessary.

CONCLUSIONS

MS is a rare disease characterized by the combination of FD and intramuscular myxoma. Ours is the first reported case of MS with myxoma presenting in the neck. Patients with MS have a higher risk of malignant transformation of FD into sarcoma and require long-term follow-up.

REFERENCES