Midline Carcinoma with NUT Rearrangement in a 23-year-old male: A Case Report

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ABSTRACT

Nuclear protein in testis (NUT) midline carcinoma (NMC) is a newly recognized, poorly-differentiated and rapidly progressive carcinoma most commonly involving mid-line structures. The majority of the cases involve the upper aerodigestive tract and mediastinum, but cases have been reported to involve the lung, eyelid, bone, skin and stomach.1

Although rare, the recent frequency of NMC has not been defined.2 A review of the National Cancer Institute’s Surveillance, Epidemiology and End Results Program (SEER) database from 1973-2008 demonstrated an average annual incidence of 0.3 per 100,000 of the US population.2

We report the clinicopathologic features of a 23-year-old Chinese male with rapidly progressive NUT midline carcinoma that required emergent tracheotomy and died within 3 months of presentation. This case illustrates the importance of identifying the NUT translocation.7 Diagnosing this translocation can be accomplished by a variety of methods including karyotyping, fluorescent in situ hybridization (FISH) and reverse transcription polymerase chain reaction (RT-PCR).

INTRODUCTION

NMC is a relatively newly described disease which may frequently go unrecognized by unfamiliar pathologists in favor of diagnoses such as squamous cell carcinoma, Ewing Sarcoma, Sinonasal undifferentiated carcinoma,6 and small-cell neuroendocrine carcinoma.7 NMC demonstrates the cytologic features of a poorly or undifferentiated malignancy and is named for and defined by DNA mutation of the NUT gene on chromosome 15q14. The clinical course is relatively equally affected.4 To our knowledge frequency by race or ethnicity has not been described. The average survival is 9.5 months, despite aggressive treatment with chemoradiation. Presently, there is no well-defined, viable treatment protocol.

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To date, all but one described case of NMC have been identified as rapidly progressive and fatal. The case of this 23 year old Chinese male confirms this clinical progression. This disease entity should be considered as a differential in all young and middle-aged, non-smoking patients with aggressive, poorly differentiated and undifferentiated pathology of midline structures. Viable treatment options for this difficult malignancy may also improve patient survival.

CASE PRESENTATION

A 23-year-old Chinese male presented with a 10-day history of enlarging nontender cervical lymphadenopathy, dysphagia and dysarthria. On fiberoptic exam he had a large hypopharyngeal post-cricoid submucosal mass. CT of the neck revealed an enhancing hypodense mass. CT of the chest revealed multiple bilateral lung nodules. PET-CT performed on 1/28/12 demonstrated multiple bilateral enlarged soft tissue attenuation masses in the neck and mediastinum.

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REFERENCES


DISCUSSION

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CONCLUSIONS

To date, all but one described case of NMC have been identified as rapidly progressive and fatal. The case of this 23 year old Chinese male confirms this clinical progression. This disease entity should be considered as a differential in all young and middle-aged, non-smoking patients with aggressive, poorly differentiated and undifferentiated pathology of midline structures. Viable treatment options for this difficult malignancy may also improve patient survival. Staging properly for diseases based on tumor mass and clinical stage, with aggressive treatment, may improve our ability to characterize and evolve to our understanding of disease epidemiology and viable treatment options.

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