Metastatic Melanoma of Unknown Primary in a 28-year-old patient

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Introduction

Melanoma is an aggressive skin cancer that occurs due to mutations in pigment-producing cells known as melanocytes.\(^1,2\) It has a tendency to metastasize beyond its primary site with a median survival of only 6-9 months after metastasis.\(^3\) Melanoma of Unknown Primary (MUP) is a rare subtype of melanoma wherein a primary lesion is absent making early detection before metastasis difficult. We report a case of metastatic MUP in a 28-year-old patient.

Case Report

A 28-year-old woman from Ethiopia with no pertinent past medical history presented with transient hemiparesis and hemiparesis involving her left upper and left lower extremities. The patient reported no associated diplopia, facial droop, or leg weakness. She experienced a similar episode four months prior, which lasted 30 seconds, and for which she did not seek medical care. The patient worked as a nurse and had frequent contact with patients with active pulmonary tuberculosis. She did not smoke.

On exam, the patient had normal motor and sensory function, balance, and mentation. Her skin exam showed no lesions, and she had palpable lymphadenopathy.

On further evaluation, blood counts were normal, an HIV test was negative and an Interferon-Gamma Release Assay was positive. Chest x-ray showed multiple well-circumscribed pulmonary nodules of varying sizes. A non-contrast CT of the head showed multiple lesions near the gray-white junction. A brain MRI showed numerous T2-hyperintense intra-axial lesions with adjacent vasogenic edema. A contrast-enhanced CT of the chest/abdomen/pelvis revealed a hypodense lesion in the left hepatic lobe (5.1 cm x 4.5 cm). At this point, both malignant and infectious causes were considered, including \textit{M. tuberculosis}. A biopsy of the liver lesion was conducted due to its lower morbidity as compared to a brain or lung biopsy. It did not reveal any acid-fast bacilli, and an \textit{M. tuberculosis} amplification test was negative. Immunohistochemical studies from the biopsy were S100 and SOX10 positive, suggesting metastatic melanoma. A follow-up exam did not reveal any skin lesions suggesting a primary site. She was offered treatment for metastatic melanoma.

Discussion

Melanoma of unknown primary (MUP) occurs in only 3.2\% of all melanoma cases.\(^4\) There are two main theories to its pathogenesis. The first is the complete spontaneous regression of a primary lesion.\(^5,6\) Partial regression of cutaneous melanoma is well-documented and thought to occur in 23\%-58\% of cases.\(^4\) The second hypothesis is that MUP develops within lymph nodes due to migration of melanoblasts to those sites.\(^4,6\)

MUP is more common in men than in women and more prevalent in Caucasian patients compared to any other racial demographic.\(^6,7\) It is usually diagnosed in the 4\textsuperscript{th} or 5\textsuperscript{th} decade of life.

A diagnosis of MUP is made based on detection of metastatic disease on imaging and subsequently confirmed by biopsy in the absence of a clear primary site. The prognosis is similar or slightly better for MUP than for a melanoma with a known primary site.\(^4,7\)
In conclusion, melanoma of unknown primary is a rare form of melanoma, often found late due to lack of an easily visible skin lesion. It is diagnosed based on clinical symptoms, imaging, and histopathological analysis of biopsied tissue. The overall prognosis is similar or slightly better than patients that have a known primary site.


