A 26-year-old Patient with Neck Pain while Weight-Lifting: A Case of Familial Cerebral Cavernous Malformation Syndrome

Oliver Tamis[1]; Johnnie Woodson[2]; Mike Hansen, MD/MBA[3]; Angela Thomas, MD[4]

MOUNTAIN VIEW HOSPITAL
A Sunrise Health System Hospital

Introduction

A 26-year-old male with unremarkable medical history presented with upper back pain for one year that worsened with activity. X-rays showed mild degeneration. The patient was diagnosed with osteoarthritis and myofascial tightness, and physical therapy was ordered. He was agreeable to follow up in 6 months. Upon follow-up, patient endorsed continued pain with activity.

Due to continued pain, imaging was ordered with unexpected results, which required neurosurgical evaluation. Patient's family history was also explored in more detail given the new findings.

Background

Familial Cerebral Cavernous Malformation Syndrome (FCCM) is a hereditary disorder in which multiple cavernous malformations, or cavernomas, are found in the brain and/or spinal cord. Cerebral cavernous malformations (CCM) can show up as early as infancy, but typically become most evident between the second and fifth decades when they are accompanied by seizures, focal neurological deficits, nonspecific headaches, and cerebral hemorrhage. Yet, despite these possible symptoms, up to 50% of people with FCCM experience no symptoms throughout their lives. This case highlights how relatively unnoticed this disease may be and possible steps towards diagnosis and management.[1]

References

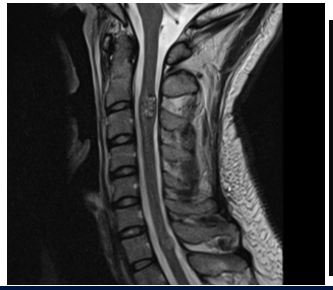
1. Morrison L, Akers A. Cerebral Cavernous Malformation, Familial. 2003 Feb 24 [Updated 2016 Aug 4]. In: Adam MP, Ardinger HH, Pagon RA, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993-2021. Available from: https://www.ncbi.nlm.nih.gov/books/NBK129

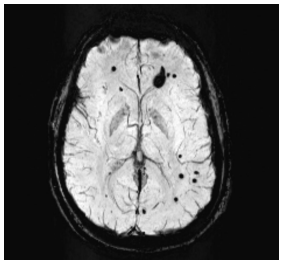
Methods

Non-contrast MRI of cervical spine was ordered due to persistent pain in a young patient. Imaging demonstrated two intramedullary/cord lesions suggestive of cavernomas (pictured below, left). Radiology recommended further evaluation with contrast-enhanced MRI of brain, cervical, thoracic, and lumbar spine to rule out additional lesions.

Imaging of the thoracic and lumbar spine did not reveal additional lesions. However, MRI of the brain revealed multiple scattered cavernous lesions, located throughout the brain (the most prominent of which are pictured below, right, although many more lesions were noted). Multiple cavernous lesions located in both brain and spinal cord are highly suggestive of familial cerebral cavernous malformation syndrome.

The patient was unaware of any family history of FCCM, though he did note a paternal family history of stroke at a young age in multiple uncles and previous generations. As cavernomas may rupture, leading to hemorrhage, this further strengthens the likelihood of FCCM.





Results

Cervical MRI:

2 intramedullary/cord lesions which have a bubbly T2 appearance. No significant edema. They have imaging features suggestive of cavernoma, but reports of multiple spinal cord cavernomas are rare. Neoplasm (including hemangioblastoma associated with VHL) also on the differential. Lack of edema makes metastasis or infection less likely.

Brain MRI:

Scattered areas of gradient susceptibility effect, larger lesions with a popcorn-like appearance as described in the body of the report involving both the supra and infratentorial brain. Given the patient's young age and the appearance on MR, this likely represents a case of familial cerebral cavernous malformation syndrome. There are far greater than 5 lesion seen in this patient. Correlate for other family members with similar brain imaging findings.

Conclusion

Cavernous malformations are often incidental findings due to many patients being asymptomatic or having mild/vague symptoms as in this patient. Recognition of this disease is important because close neurological monitoring is warranted due to potentially devastating late effects of the condition.

In general, symptoms such as nonspecific headache, seizure, or focal neurologic deficits warrant additional investigation if not properly explained by other disease processes or noxious stimuli. In the case of FCCM, these symptoms may be indicative of hemorrhage or cavernous enlargement requiring surgical intervention. Additionally, functional and/or cognitive decline is likely in these individuals, making physiatry an essential resource in FCCM. Control of comorbidities including blood pressure is imperative. Avoidance of contact sports is required for these patients and family members who have not been evaluated for FCCM.

This particular patient was referred to UCLA for expert surgical neurosurgical evaluation.

Author Information

1. MS4, Touro California School of Medicine, Class of 2021.
2. MS4, UNLV School of Medicine, Class of 2021
3. PGY3, Sunrise Health Consortium GME, Department of Physical Medicine and Rehabilitation, HCA
4. Southern Nevada VA, Department of Physical Medicine & Rehabilitation, Traumatic Brain Injury Chair

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