CEREBRAL CAVERNOUS MALFORMATION

SCHOOL OF MEDICINE
NEUROLOGY

Cerebral cavernous malformation called CCM1 (the common Hispanic mutation) is more prevalent in New Mexico than anywhere else in the world.

What is cerebral cavernous malformation (CCM)?

Low flow vascular malformations (mulberry shaped) often called cavernomas, cavernous angioma, or cavernous malformations. It is an autosomal dominate mutation where each child of an affected parent has a 50/50 chance of developing CCM lesions.

Why should I care about CCM?

New Mexico is the home to a founder mutation in CCM1 of the KRIT1 gene. There is a higher incidence of CCM1 (the common Hispanic mutation) in New Mexico than anywhere else in the world stemming from descendants from a Spanish settler family in the 1600s. It is under-diagnosed, under-reported, and often goes without management until symptomatic exacerbations.

How can I diagnose CCM in my patient?

Diagnosis is made through genetic testing or imaging by an MRI. Three generation family history helps to identify individuals who may necessitate genetic testing or MRI scanning however prior authorization may be required for the testing or scan. Genetic testing is ordered by completing the requisition form and obtained prior authorization from the insurance company, if necessary. A list of genetic testing companies can be found at https://alliancetocure.org/. if the insurance company does not have contracts with a genetic testing company.

FOR MORE DETAILS AND INFORMATION ON CCM:

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What is the diagnostic code for CCM1?

ICD9 747.81, ICD10 D18.02, and ICD11 8B22.41 for cerebral cavernous malformation related to CCM1 mutation.

Where can I get more information and resources?

https://www.alliancetocure.org/
https://hsc.unm.edu/medicine/departments/neurology/

What symptoms come with CCM1?

The most prevalent symptoms with CCM1 presentation are headaches, seizures, weakness, and numbness and tingling. Due to the neurological nature of this disease, there are a myriad of others symptoms that can present and is why a three-generation family history is vital.

Can my patient take aspirin (any NSAIDS) or antithrombotics?

If aspirin (NSAIDs) or anthithrombotics are needed, your patient should be monitored for new neurological deficts or new onset headaches. Aspirin and antithrombitics should be used for the least amount of time clinically indicated.

Is female hormone therapy allowed?

Increased hemorrhage risk has been identified in individuals taking hormone therapy whether for contraception, menopausal therapy, or other medical reasons. Female hormone therapy should be avoided or used with extreme caution, and again, the patient would need to be monitored for new neurological deficits or new onset headaches.

How do I treat their conditions that are not related to CCM1?

Treat the patient's medical conditions as indicated by standards of best practice however for any infections, if found to be gram positive, antibiotics need to be prescribed for the minimum amount of time necessary to treat the condition. The growth of gram-negative bacteria may trigger inflammation through a leaky gut and promote lesion growth and/or leakage.

Resources:

Flemming, K. D., Smith, E., Marchuk, D., & Derry, W. B. (2003). Familial Cerebral Cavernous Malformations. In M. P. Adam (Eds.) et. al.,GeneReviews®. University of Washington, Seattle.

Flemming, K. D., & Lanzino, G. (2020, September). Cerebral cavernous malformation: what a practicing clinician should know. InMayo Clinic Proceedings(Vol. 95, No. 9, pp. 2005-2020). Elsevier.