20 QUESTIONS ANSWERED

Cerebral Cavernous Malformation
Cerebral Cavernous Angioma
Cavernoma

A PATIENT'S GUIDE

ALLIANCE TO CURE CAVERNOUS MALFORMATION
AllianceToCure.org

WELCOME

You received this booklet because your doctor diagnosed you or a loved one with at least one *cavernous malformation* in your brain or spinal cord. Your doctor may have used the terms *cerebral cavernous malformation (CCM), cavernoma, or cavernous angioma* for your diagnosis. These are all names for the same blood vessel abnormality. We will use the terms *cavernous malformation* and *lesion* in this booklet.

Chances are this is the first time you've heard of a cavernous malformation. In this booklet, we'll be offering information to help you make informed decisions about your care. You may also want to share this booklet with the important people in your life to help them gain an understanding of cavernous malformation.

There is no typical way in which cavernous malformations affect people. The symptoms, the course of the illness, and its severity are different from person to person. While this booklet offers general information and resources, everyone is unique and all medical advice, diagnosis, and treatment options should be discussed with your physicians. We base our information on clinical care guidelines written by our Scientific Advisory Board Clinical Experts Panel. Our website at AlliancetoCure.org contains extensive additional information.

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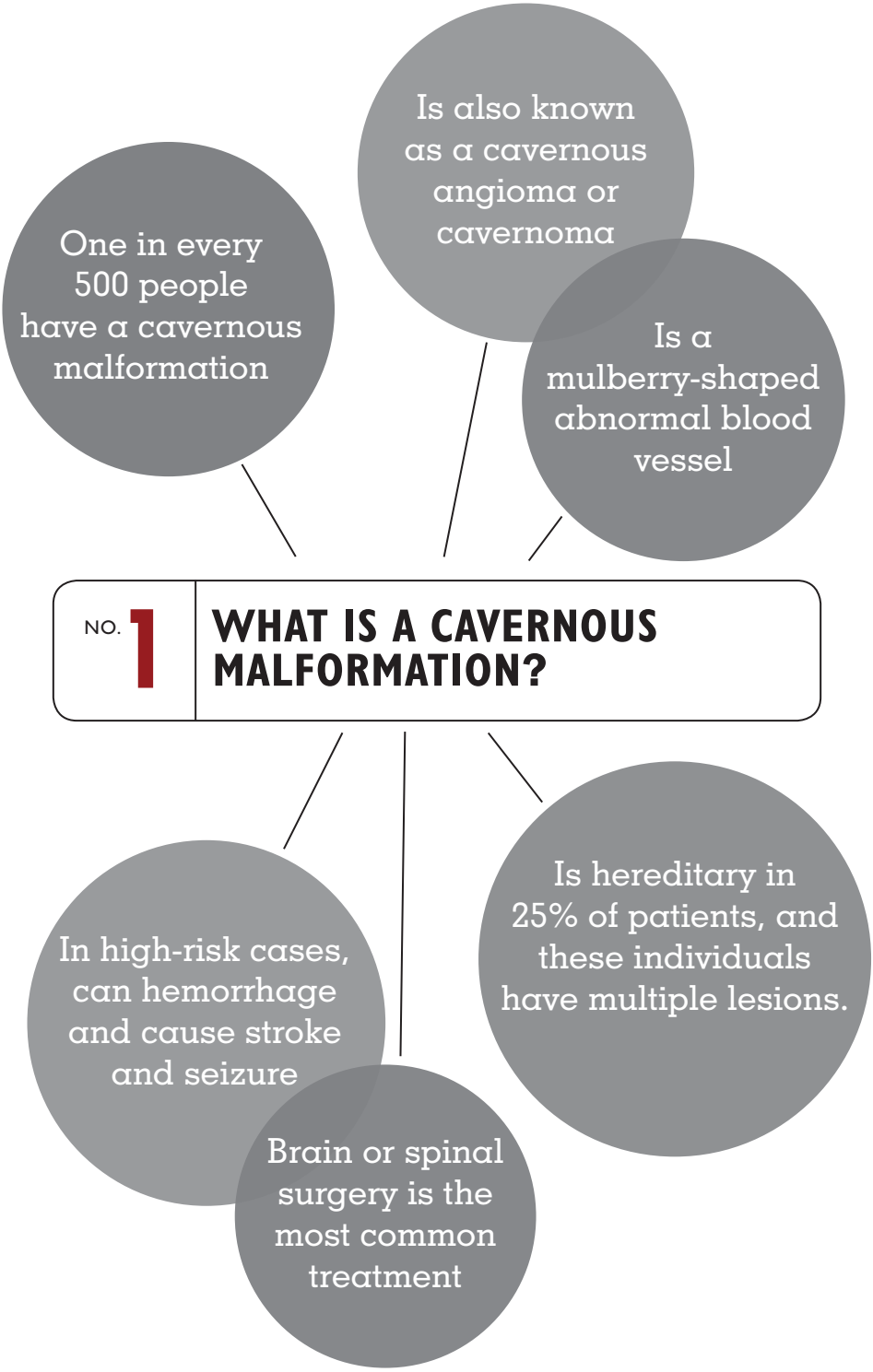
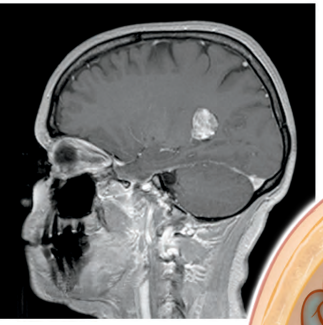
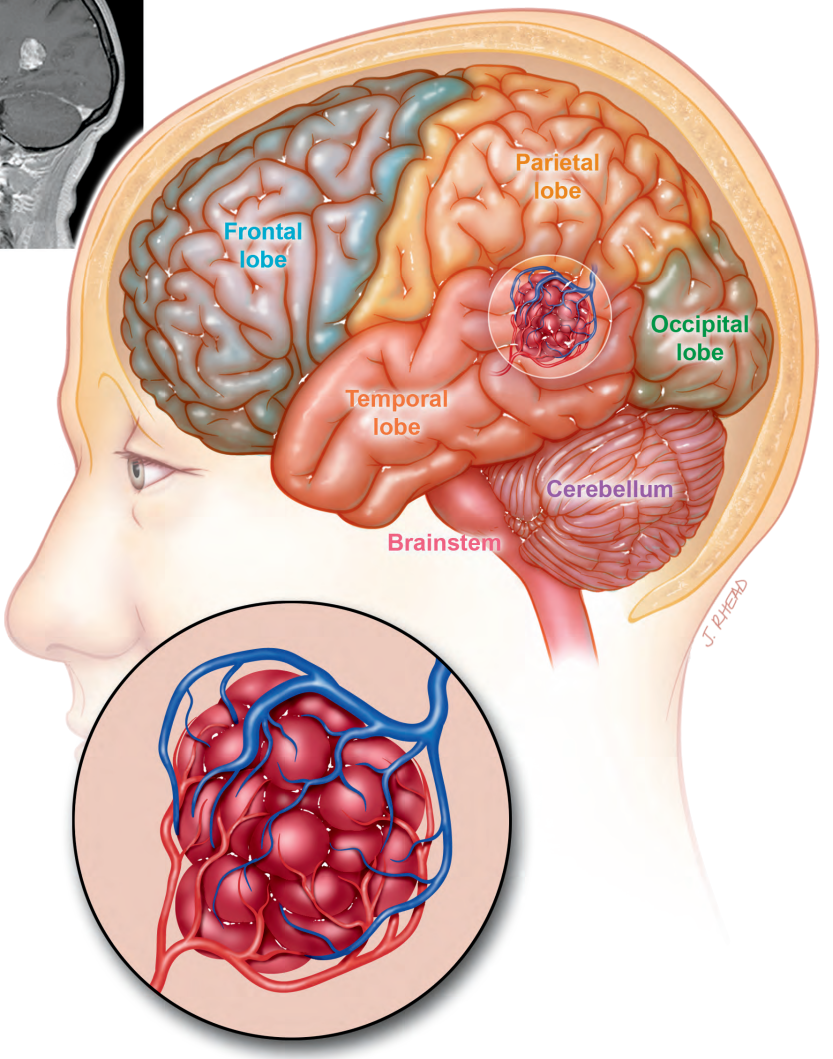

Is also known as a cavernous angioma or cavernoma
One in every 500 people have a cavernous malformation
Is a mulberry-shaped abnormal blood vessel
NO. 1
WHAT IS A CAVERNOUS MALFORMATION?
In high-risk cases, can hemorrhage and cause stroke and seizure
Is hereditary in 25% of patients, and these individuals have multiple lesions.
Brain or spinal surgery is the most common treatment

Cavernous malformations are made of abnormal blood vessels. They are not cancerous, but are sometimes called vascular tumors. Cavernous malformations are made of the smallest blood vessels, called capillaries. They resemble mulberries and have very thin walls compared with other blood vessels. Blood moves through them very slowly. The thin vessel walls can be leaky and can allow blood into the surrounding brain or spinal cord tissue. We believe about one in every 500 people have a cavernous malformation, but most people never have symptoms. Of the people who have a cavernous malformation, we believe many will never have a symptomatic hemorrhage.

The rarest place for a symptomatic cavernous malformation is in the spinal cord (nerves). A spinal cord cavernous malformation is not the same as a spinal hemangioma, which is located in the vertebra (bone). Cavernous malformations may also occur in the area of the brain called the brainstem. Spinal cord and brainstem cavernous malformations can cause very severe symptoms and complications when they hemorrhage.





CEREBRAL CAVERNOUS MALFORMATION

Also known as Cavernous Angioma or Cavernoma

PATIENT STORY: Stacie

Sporadic Cavernous Malformation *with Seizure*

Stacie awoke in the middle of the night, just like many other nights. Only this time, as she tried to walk across the room, she became unsteady and fell into the wall. The dizziness happened again the next morning. A 36-year-old mother of four, Stacie sought medical help, but after numerous tests, the doctors were unable to provide her with a diagnosis.

One evening a year later, Stacie experienced what she later would learn was a series of focal onset seizures that included involuntary movement, a loss of awareness, and difficulty speaking. Her husband rushed her to the local hospital where a CT scan revealed an unidentified mass deep in her right temporal lobe.

Stacie was transferred to a larger hospital with more advanced diagnostic capabilities where she received an MRI. Her brain mass was diagnosed as a cavernous malformation, and she was discharged on anti-seizure medication. Despite this, the seizures continued and she sought a surgical consultation. The doctors informed her that removing her lesion would be difficult because of its deep location, and surgery could cause additional harm. Stacie needed to accept her new normal.

It has been a year since her diagnosis. After trying a few different anti-seizure medications to find the best fit, Stacie's seizures are now under control. She says she has learned to leave her high heels in the closet because she still experiences dizziness. Sometimes she has trouble finding the right words. "It's like playing charades with my kids," laughs Stacie. "Eventually they figure out what I mean." She is happy to be driving again and able to take care of her family.

Says Stacie, "My brain hemorrhage changed my life, but I know there's a reason. I will use it as a strength and not a weakness."

NO. 2

WHO MIGHT BE INVOLVED IN MY CARE?

If you have symptoms, you may have many medical professionals involved in your care.

NEUROLOGIST
manages symptoms like seizure and pain

NEURO-OPHTHALMOLOGIST
cares for brain-related vision problems

NEUROSURGEON
evaluates for surgery

REHABILITATION SPECIALISTS
improve functioning after brain or spine event

DERMATOLOGIST
cares for vascular skin lesions in hereditary illness

GENETICIST
evaluates for hereditary form of the illness

ORTHOPEDIST
monitors spine in CCM3 patients (see Genetics)

NO. 3

WHAT IS AN *INCIDENTAL* CAVERNOUS MALFORMATION?

Your doctor may have discovered your cavernous malformation when you had brain or spinal imaging for a reason not related to your cavernous malformation.

For example, you may have received a CT scan or an MRI after a car accident or concussion. In this case, doctors will call your cavernous malformation an *incidental* finding. Research has shown that incidental cavernous malformations that have not had a previous hemorrhage have a very small chance of ever becoming problematic. While it is advised that you follow the precautions listed in Question Number 16 about lifestyle changes in this booklet and on our website at AllianceToCure.org, you may not need repeat imaging unless instructed by your doctor or if you develop symptoms.

If you have a familial diagnosis, it is wise to have an occasional follow-up doctor's visit to maintain a relationship with a care provider should you ever develop symptoms. For a solitary incidental cavernous malformation, ongoing care should be discussed with your diagnosing doctor. If the lesion is in an area of the brain known to be more prone to hemorrhage such as the brainstem, it may be wise to maintain a care provider relationship. For others, the decision may be based on comfort level. Surgery is never recommended for an asymptomatic cavernous malformation that has not hemorrhaged.

WANT TO LEARN MORE?

Our **PATIENT EXPERT COURSE** is a great resource:

NO. 4

WHAT IS *SPORADIC* CAVERNOUS MALFORMATION?

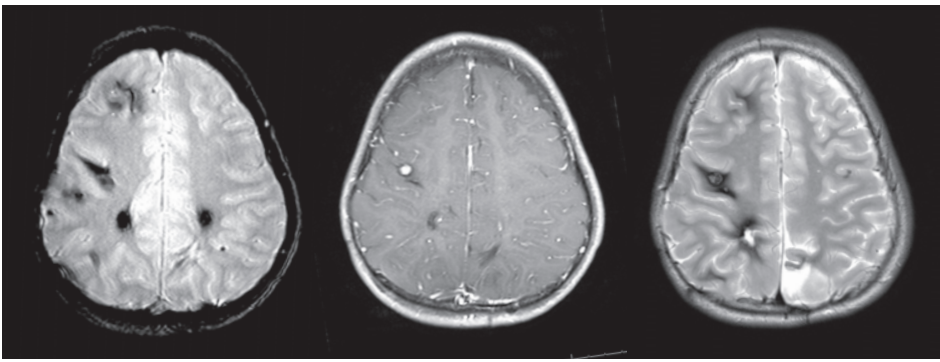
Sporadic cavernous malformation means that you are the only one in your family to have a cavernous malformation, and you do not have a genetic mutation that would cause your children to inherit the condition. Usually, individuals with the sporadic form of the illness have just one cavernous malformation with no other visible blood vessel abnormalities or medical history to explain the lesion. Sometimes, individuals have a different kind of blood vessel abnormality called a developmental venous anomaly (DVA) or they have a history of brain radiation for cancer. A DVA or brain radiation can lead to the development of one or more cavernous malformations (see What Causes Multiple Cavernous Malformations?). To be certain you have the sporadic form of the illness, you will need to have an MRI with some special images called *susceptibility weighted imaging* or SWI. Ask your doctor if this was part of your original MRI.

See Stacie and Vern's Patient Stories in the this booklet to learn more about sporadic cavernous malformation with and without a DVA.

ARE YOU WONDERING: DO I HAVE A SPORADIC CAVERNOUS MALFORMATION?

This **DECISION TREE** might help.

NO. 5 WHAT CAUSES *MULTIPLE* CAVERNOUS MALFORMATIONS?



MRI with multiple cavernous malformations as seen on SWI (T2*), TI, and T2 sequences.

Some people have more than one cavernous malformation. This can happen for several reasons:

- Most commonly, people with more than one cavernous malformation have a hereditary form of the illness. Heredity is the transmission of genetic characteristics from parents to their offspring. This hereditary form typically results in the development of additional cavernous malformations over time.
- Some people may have a second kind of abnormal blood vessel called a developmental venous anomaly (DVA). Your physician may also call this a venous malformation. This dilated blood vessel rarely causes symptoms on its own. However, it may create conditions that make it more likely for cavernous malformations to form. This is not hereditary.
- Radiation treatments or radiosurgery (also known as Gamma Knife) to the brain or spine for cancer or other conditions can cause cavernous malformations to form many years later. In people with the hereditary form, radiosurgery may cause more lesions to occur. You can find out more information about radiosurgery in the Other Treatments section of this booklet.

PATIENT STORY: Taylor

Brainstem Cavernous Malformation

In January 2015, three-year-old Taylor's parents, Elizabeth and Pasco, grew concerned when they noticed changes in their daughter's behavior. Taylor's walking was unsteady, her speech was impaired, and her face was drooping. MRI confirmed that Taylor had a large cavernous malformation that had hemorrhaged in the pons region of her brainstem.

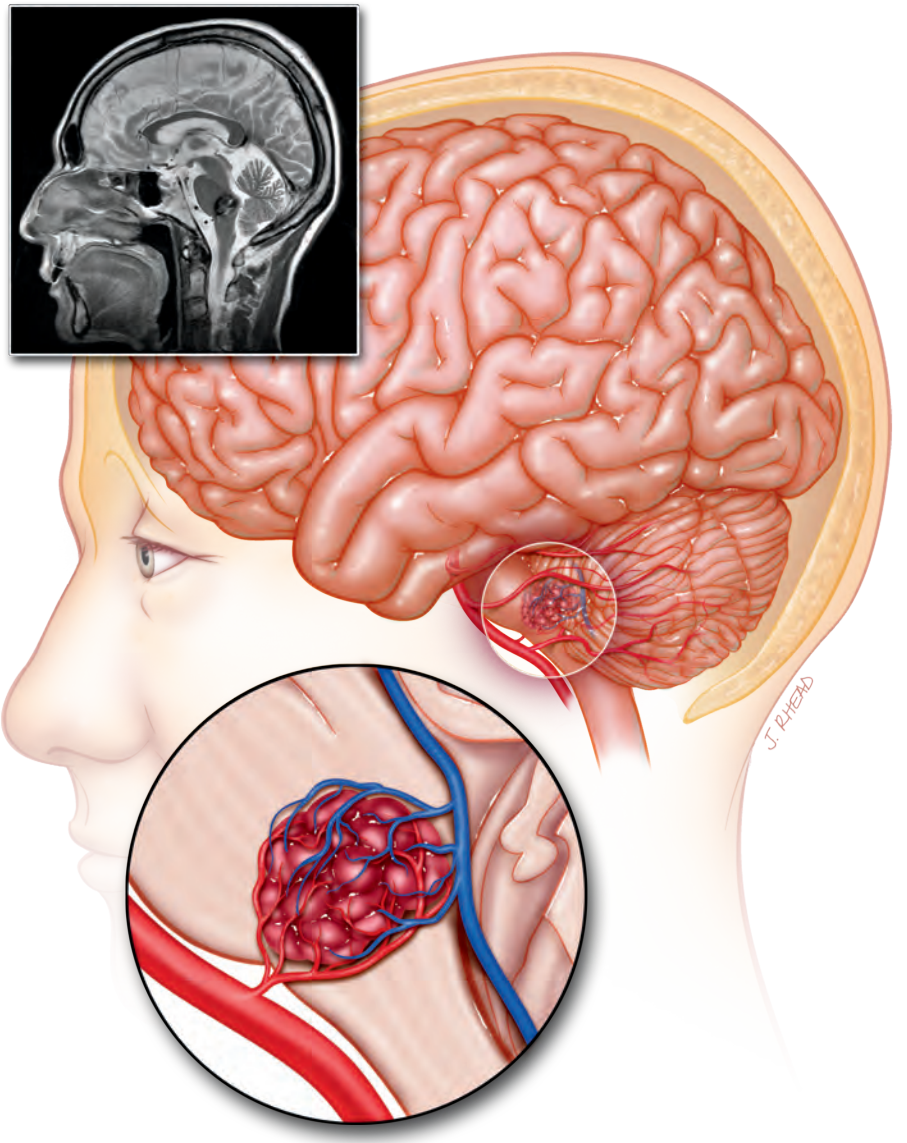
Taylor's parents faced a difficult decision. Surgically removing the cavernous malformation could result in additional deficits. However, another hemorrhage could be life threatening. Elizabeth and Pasco obtained multiple surgical opinions and decided to move forward with surgery at a hospital on the other side of the country. Nine months after the onset of her symptoms, Taylor underwent the delicate operation.

Taylor emerged from the surgery with left-sided facial paralysis, not uncommon with trauma to the pons. She had difficulty coordinating her movements, which affected her standing and walking. Since her surgery, Taylor has undergone intensive physical rehabilitation and made great improvements in her ability to get around. Her family is researching options to bring movement back to her face.

Because she had additional cavernous malformations, Taylor received genetic testing. Testing revealed a mutation of the CCM3 gene, which tends to lead to a more severe form of cavernous malformation illness and can cause scoliosis and other kinds of benign brain tumors. Taylor is the first in her family to have a CCM3 mutation.

Taylor's parents are grateful for the positive surgical outcome, but they still worry. "Unfortunately, always in the back of mine and her daddy's mind is the anxiety of not knowing what is going on in there," said Elizabeth. "Is there any new activity, anything we should know about?"

Elizabeth and Pasco treasure Taylor and her resilience. "She is beautiful, smart, charming, witty, stubborn, and strong. She still amazes us every day with things she does."



BRAINSTEM CAVERNOUS MALFORMATION
Located at the junction of the pons and the medulla oblongata

NO. 6

WHAT IS A *BLEED* OR *HEMORRHAGE*?

All cavernous malformations have some oozing of blood in the area of the lesion. This is what gives them their typical appearance on MRI. Oozing is different from the more significant symptomatic hemorrhage or bleed.

A symptomatic hemorrhage is new bleeding in or around the cavernous malformation and is often associated with new symptoms. Symptomatic hemorrhage is the most serious complication of cavernous malformation and is the most common reason for surgery. The hemorrhage may cause new symptoms or an increase in symptoms.

The specific symptoms a person experiences will depend on the location and size of the cavernous malformation, as well as the amount of blood that has leaked outside the lesion. Some lesions have slow oozes that produce mild or no symptoms. Eventually, the blood breaks down and leaves behind an iron deposit called hemosiderin.

The risk for hemorrhage is greater for those with multiple cavernous malformations, but it is impossible to predict which of the many lesions might bleed.

Many lesions never hemorrhage. However, once a cavernous malformation has had one symptomatic hemorrhage, it is at significantly greater risk of bleeding again. Lesions that are not in the brainstem have about a 1 in 6 chance of bleeding again. Brainstem lesions have about a 1 in 3 chance of bleeding again. Most of these second hemorrhages will take place in the first two years after the original hemorrhage.

At five years after a hemorrhage, the risk of another hemorrhage gradually returns to less than 2% per year, similar to the risk from a lesion that has never bled.

NO. 7

WHAT CAUSES *HEMORRHAGE*?

Hemorrhage is the most serious potential consequence of cavernous malformation. About 25% of diagnosed cavernous malformation patients have had a symptomatic hemorrhage. You may be concerned about taking blood thinners and other medications and wonder what effect they may have on your risk for hemorrhage. Recommendations regarding blood thinners have been evolving. Recent research indicates blood thinners do not appear to increase the risk of hemorrhage. This is an important discussion that you should have with your providers. For the most up-to-date information on this topic, and because there are many related subtopics, we recommend that you frequently visit the Cavernous Malformation In Depth – Hemorrhage page on our website. Researchers are examining the risk of hemorrhage related to the use of hormonal agents, Botox use, and probiotics, as anecdotal evidence suggests a possible relationship. As always, if you have questions about your risk of hemorrhage or any other concerns, it is strongly advised that you consult your physician.

While we cannot yet determine exactly what caused a lesion to bleed,research has found that the following activities do not increase the likelihood of a bleed: aerobic activity and noncontact sports, vaginal childbirth, air travel in a commercial aircraft with normal cabin pressure, and consuming alcohol and caffeinated beverages in moderation.

WANT TO LEARN MORE?

Visit the **CAVERNOUS MALFORMATION IN DEPTH - HEMORRHAGE** section of our website for more information

NO. 8

WHAT *SYMPTOMS* CAN A CAVERNOUS MALFORMATION CAUSE?

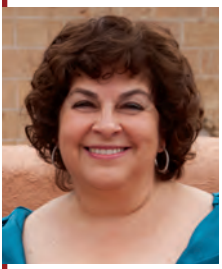
The symptoms of a hemorrhage from a cavernous malformation depend upon its location and size.

- *Seizures are one of most common symptoms of cavernous malformation. Brainstem and spinal cavernous malformations don't cause seizures. Seizures fall into two general groups: focal seizures are local to one area of the brain and generalized seizures involve both sides of the brain. All cavernous malformation seizures begin as focal seizures but some progress to generalized seizures. Neurologists use anti-epilepsy medications to control seizures. However, neurosurgeons have had good results in decreasing or eliminating seizure with brain surgery if they are able to pinpoint which cavernous malformation is causing the seizures. Surgery is most successful when it occurs within two years of a first seizure.*
- *We know people with cavernous malformation experience more frequent headaches than other people. A headache does not necessarily mean a new hemorrhage. For the most part, we can't distinguish a cavernous malformation headache from any other kind of headache. A headache may be related to your cavernous malformation if it is unlike one you have ever experienced previously or if it is in the general location of your lesion.*

- *Lesions in the frontal, parietal, or temporal lobes, or in the cerebellum. can cause attention, memory, social, mood, and learning problems. This is particularly true for individuals with many lesions throughout the brain.*
- *Cavernous malformations in many parts of the brain and spinal cord can lead to weakness or numbness in a person's arms or legs. Some can also cause pain, such as those located in the thalamus. A cavernous malformation in the brainstem may lead to coordination problems, referred to as ataxia, and facial paralysis may occur, usually on one side.*
- *Cavernous malformations may also lead to vision problems. There are two kinds of vision problems: those caused by lesions in the occipital lobe of the brain, which affect how visual information is processed, and those caused by lesions in the brainstem, which affect how the eyes work.*
- *Cavernous malformations can cause hearing problems, including loss of hearing and tinnitus, dizziness, or nausea, particularly if located in or near the cerebellum.*
- *A cavernous malformation in the medulla, the lowest part of the brainstem, can lead to spasms of the diaphragm, which resemble hiccups that don't go away. More rarely, these can cause swallowing or breathing problems.*
- *Cavernous malformation hemorrhages in the brain may result in fatigue. Individuals may complain of fatigue for months to years after a major hemorrhage or brain surgery.*
- *Spinal cord cavernous malformations can result in numbness, weakness, paralysis, tingling, burning, or itching. The location and extent of the symptoms depend on the level of the spine affected. Spinal cord lesions can also cause difficulty with bladder and bowel control.*

PATIENT STORY: Joyce

Spinal Cavernous Malformation

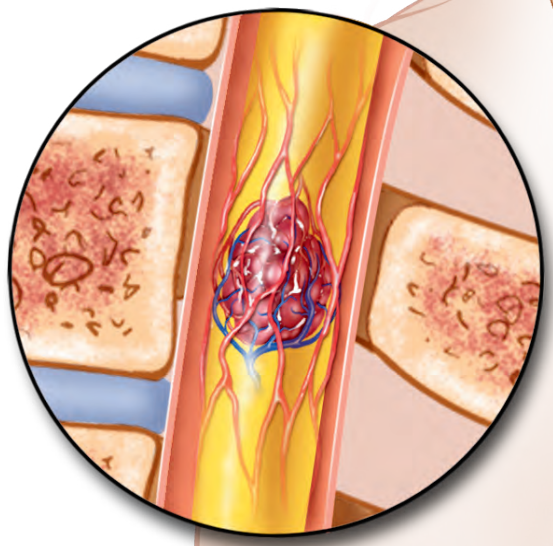


"I had just put my left arm down on the kitchen table when I experienced this intense pain from the tips of my fingers to my elbow," Joyce told the New York Times in a 2007 interview. "It felt like my whole arm was burning."

It would be years before doctors accurately diagnosed Joyce with a cavernous malformation in her cervical spinal cord. By that time, the pain had spread to her right arm. Her surgeon told her the lesion was accessible, and Joyce had successful surgery in 2004. The surgery helped to resolve some of Joyce's pain and to prevent any further escalation.

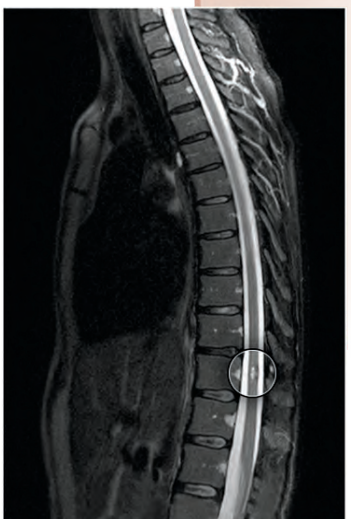
Joyce also has multiple cavernous malformations in her brain. This is because of a hereditary genetic change known as the Common Hispanic Mutation. While anyone of any ethnicity can have a hereditary form of cavernous malformation illness, thousands of people who trace their heritage to the original Spanish settlers of New Mexico share this specific genetic mutation. A single ancestor connects them all, and many of these families have remained in New Mexico, including Joyce. No other place in the world has as many affected people.

Joyce has been active in advocacy and in raising awareness for the illness throughout New Mexico. In addition to the New York Times interview, Joyce was central in drafting legislation in New Mexico that created a mandate to educate more doctors. Joyce has presented at patient conferences and worked with media around the state to share her story and the genealogy she has researched. Joyce was happy to discover that neither of her children inherited the illness. In Joyce's family, the Common Hispanic Mutation has stopped with her, though her work toward further research and a cure continues.



SPINAL CAVERNOUS MALFORMATION

Small lesions can cause significant problems in a tight space



NO. 9 HOW OFTEN DO I NEED AN *MRI*?

This is an important discussion to have with your physician. Some advise their patients to have repeat MRIs on a specific schedule. Others suggest waiting until there are additional symptoms. Experts suggest more frequent imaging for those who may not be able to report symptoms, like young children or those with intellectual or communication problems. These individuals may also need sedation for MRI. You and your doctor will weigh the risk of sedation against the benefit of imaging.

CT scan is another kind of imaging that is sometimes used. CT scan is much faster than MRI, but the images are not as clear, and CT scan involves exposing a patient to radiation. You may have a CT scan in an emergency when MRI is not available. People with multiple cavernous malformations should limit their exposure to CT scan as much as possible because it is not clear whether the radiation exposure can cause the development of more lesions.

A doctor may suggest a cerebral angiogram also known as a cerebral arteriogram. This procedure allows the doctor to see the arteries and veins in your brain. A cavernous malformation is not visible on angiogram, but the test is done when another type of blood vessel lesion, called an arteriovenous malformation, is suspected. If a cavernous malformation has a typical appearance on MRI, it does not require a cerebral angiogram as part of routine care.

NO. 10 DO I NEED *SURGERY*?

If your cavernous malformation is causing symptoms, it is advised that you have an indepth discussion about surgical options with your neurosurgeon. The decision to have surgery always involves weighing risks and benefits.

Open surgery, also known as craniotomy when performed through skull or laminectomy when performed on the spine, remains the most common treatment for symptomatic cavernous malformation. The Alliance to Cure Cavernous Malformation Clinical Care Consensus Guidelines provide guidance on clinical decision-making around surgery. These guidelines are available on our website. Most commonly, surgery is appropriate after a second cavernous malformation hemorrhage or for lesions that are causing epilepsy. For epilepsy, some craniotomies are being replaced by minimally invasive procedures.

There are neurosurgeons who specialize in cerebrovascular (brain blood vessel) and skull base (brainstem) surgery. It is important to gain as much information as possible regarding your neurosurgeon's experience and skill level with cavernous malformations. A second opinion, ideally from a CCM Center of Excellence or Clinical Center, is often helpful. These institutions are recognized by the Alliance to Cure Cavernous Malformation as providing high-quality interdisciplinary care for both sporadic and familial cavernous malformation patients.

NO. 11 ARE *OTHER TREATMENTS* FOR CAVERNOUS MALFORMATION AVAILABLE?

Doctors have treated cavernous malformation with *stereotactic radiosurgery*, also known as gamma knife, linear accelerator, X-knife, Brainlab, or CyberKnife, for many years, but its effectiveness is not clear. In stereotactic radiosurgery, focused radiation is directed at the cavernous malformation without opening the skull. Experts now recommend that radiosurgery be considered only with individuals who have a single symptomatic lesion that is in an area of the brain where the risks of traditional surgery would be too high. Radiosurgery should not be used for treating cavernous malformations that do not cause symptoms or for cavernous malformation in people with the hereditary form of the illness, as the radiation itself might trigger new cavernous malformations to form.

Researchers are working to find ***medications*** *to treat cavernous angiomas. It may take more than one medication to treat every situation. Medications could be useful to:*

- Stabilize the cavernous malformation so it does not hemorrhage. Patients could use medications even before a first hemorrhage. This is particularly true for people with hereditary forms of the illness.
- Stabilize the cavernous malformation after a hemorrhage to reduce the risk of the lesion bleeding again. Remember, there is a high-risk time in the first years after a hemorrhage. During this time, it might make sense to use a stronger temporary medication.
- Slow or stop the development of more lesions in people who have multiple cavernous malformations.
- Shrink or destroy existing cavernous malformations. This is the ultimate goal.

While surgery is currently the only treatment for the lesions themselves, there are some medications that may be helpful for treating some symptoms such as seizures and spasticity. Therapy can also be helpful for treating symptoms, including physical therapy, occupational therapy, vision therapy, vestibular therapy, and mental health therapy. Medical specialists on your team may include a geneticist, a neuro ophthalmologist, a physiatrist (a physical medicine specialist), or an orthopedic specialist.

NO. 12 SHOULD I *GET* GENETIC TESTING?

Most people with a cavernous malformation do not need genetic testing. We believe only 25% of individuals with a cavernous malformation have a hereditary form of the illness. The best way to determine whether you need genetic testing is to have an MRI with a sequence called susceptibility-weighted imaging (SWI). If you have just one lesion on the SWI sequence, you do not need testing. If there is more than one lesion but all of the lesions are close to a DVA, you also do not need testing. You have the sporadic form of the illness, and you can't pass this to your children.

People who have multiple cavernous malformations that are not associated with a developmental venous anomaly should request genetic testing. Sometimes people are the first in their family with a hereditary form. You do not need other affected family members to justify testing.

NO. 13 SHOULD I HAVE CONCERNS ABOUT *PREGNANCY* WITH A CAVERNOUS MALFORMATION?

Women with cavernous malformations are no more at risk for brain or spinal cord hemorrhage during pregnancy than at any other time. Women with cavernous malformation typically can have a vaginal delivery if they have not had a recent hemorrhage. However, these decisions should be thoroughly discussed with your physicians.

If your doctor suggests an MRI during your pregnancy, your MRI should not include gadolinium contrast.

Women who have epilepsy and are on antiepilepsy medications should discuss with their physicians their medications are safe for use during pregnancy. All treatment decisions should be made with your medical team, which may include a high-risk obstetrician.

If a cavernous malformation does hemorrhage during pregnancy, it can present extra treatment challenges. Surgery to remove a cavernous malformation may be an option during pregnancy in cases where waiting until after delivery is too risky.

NO. 14 ARE THERE SPECIAL ISSUES FOR *CHILDREN* WITH A CAVERNOUS MALFORMATION?

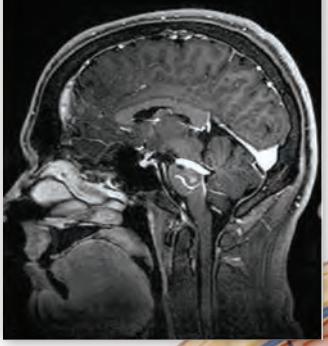
There is the potential for many special issues with children, depending on their age at diagnosis and their symptoms. Young children may not be able to tell you about symptoms, so it becomes important to schedule imaging regularly. It is also important to avoid overreacting so that children aren't exposed to unnecessary medical procedures, radiation from CT scans, MRI contrast medication, and sedation. It can be challenging to strike a balance. Trusting your instincts and having a knowledgeable medical team can help.

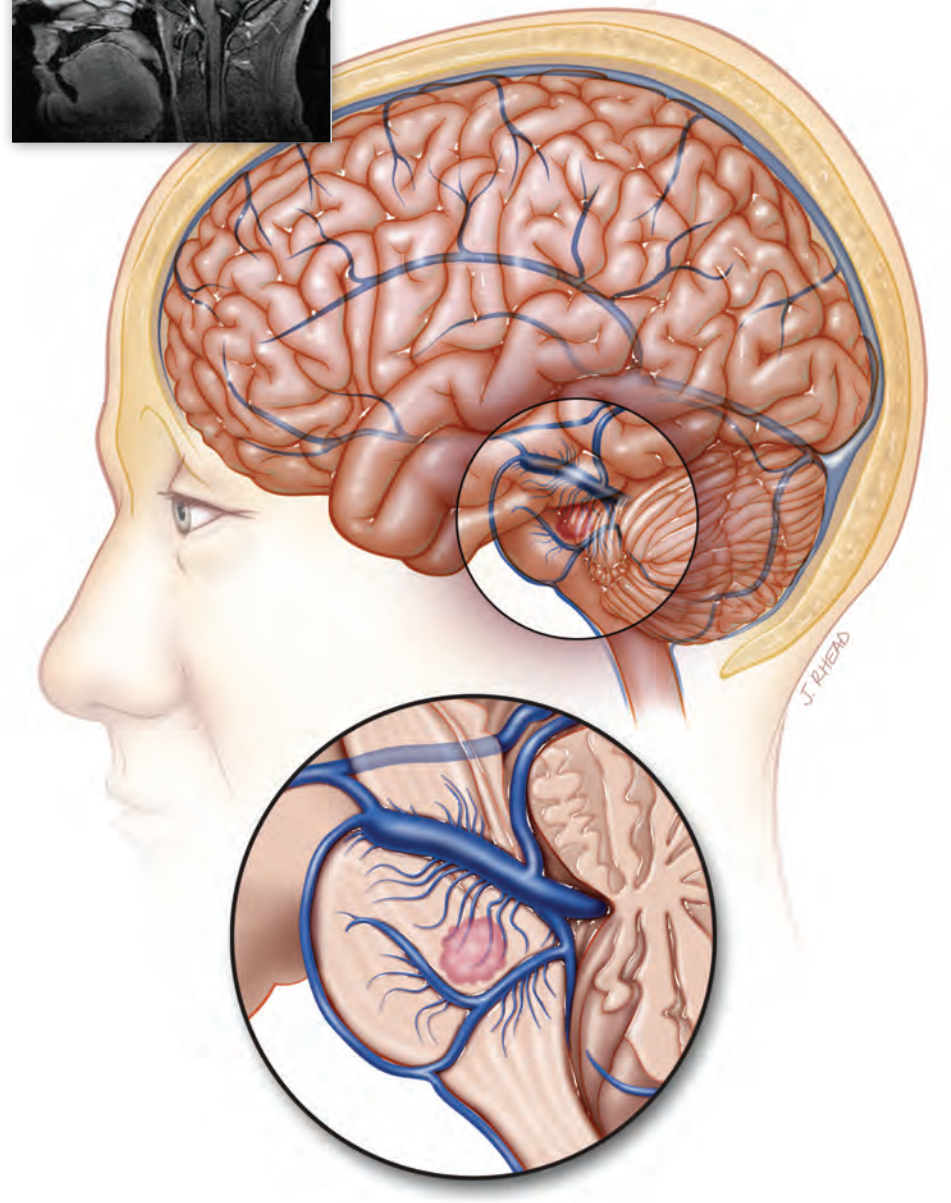
As a child grows older and parents become more experienced, managing this illness becomes easier.

Telling your child about his or her diagnosis can be emotional and can require a period of adjustment for your child. Try to explain in simple, age-appropriate terms. Having a pediatric mental health professional ready to help can make this easier. A mental health professional can also help if a child with hemorrhage or surgery-related deficits struggles with peer relationships, restrictions on activities, or learning.

You will need to explain your child's illness many times to many professionals including school staff, friends and other caregivers. Good support from other parents in the Alliance to Cure community or your local special needs community can be helpful for you.

A neuropsychology evaluation can be helpful in determining strengths and weaknesses and what supports may benefit your child at school, home, and in the community.





DEVELOPMENTAL VENOUS ANOMALY

Also known as a venous angioma
or a venous malformation

PATIENT STORY: Vern

DVA with Cavernous Malformations

In 2004, Vern, then a 28-year-old father of two, hit his head while practicing jujitsu. From the subsequent CT scan, Vern and his wife Tiffany were surprised to learn that Vern had a more serious and long-term condition. Doctors diagnosed him with multiple cavernous malformations surrounding a large developmental venous anomaly (DVA), a dilated malformed vein, in his brainstem. While a DVA by itself typically does not cause symptoms or complications, Vern's DVA had set the stage for the development of more than one cavernous malformation.

It was not until two years later that Vern had his first hemorrhage. As a result, he closed his business as a mobile fleet service mechanic to focus on his recovery and his family.

"I couldn't drive," Vern explains. "My reaction time was just not near what it needed to be to operate a vehicle safely." Vern is driving again, but not on interstates or during times of heavy traffic. He also has been recovering from gait and vision deficits.

Vern has consulted numerous surgeons about removing the most problematic of his cavernous malformations. The DVA complicates surgery. Vern's wife Tiffany explains, "The angioma that keeps bleeding is located too close to his DVA to remove. The DVA looks almost like a hook, and the angioma is on the inside of that hook." Disturbing a large DVA like Vern's can cause a catastrophic stroke.

Tiffany continues, "Vern has learned that being busy and doing something productive each day makes a huge difference in his quality of life. Vern has always had a very active lifestyle, but since having his bleeds, he has learned how to do things safer and smarter. He follows what doctors have told him to do and not to do... well mostly. All in all, Vern's doing well."

NO. 15 HOW DO I *COPE*?

Receiving a diagnosis of cavernous malformation in the brain or spinal cord for you or a loved one can be difficult and upsetting. No matter your level of medical involvement, you will require a period of adjustment to a "new normal" which will include a time of grieving for what has changed. A counselor or other mental health professional can help if you experience emotional difficulty that interferes with your ability to get through your days. Close friends and family may have difficulty understanding, particularly if you don't appear ill. The Alliance to Cure Cavernous Malformation online community or our weekly virtual support groups can be helpful as you look for others who share your issues.

NO. 16 ARE THERE ANY LIFESTYLE IMPROVEMENTS THAT MAY REDUCE RISK?

In addition to working toward a cure, researchers are exploring whether there are actions you can take right now to reduce your risk. Here are suggestions:

VITAMIN D

Inflammation can drive cavernous malformation disease. Vitamin D is a natural anti-inflammatory and is beneficial to the immune system. Research has shown a correlation between a more aggressive course of cavernous malformation and low vitamin D levels. Your doctor can order a blood test to determine your serum levels of 25(OH) D. If your levels are low, your doctor can advise you on the D3 (cholecalciferol) dose you should take.

DIET

Fascinating research has linked the human microbiome to cavernous malformation disease. The human microbiome is a community of microbes (bacteria, viruses, parasites, and fungi) that live

in and on our bodies. Ideally, these microbes live in balance and play a supportive role in many processes in the body. Your gut microbiome requires a protective mucosal layer to prevent potentially harmful bacteria from entering your system and causing inflammation. As mentioned above, inflammation can drive cavernous malformation disease.

One of the best ways to protect your gut lining is by avoiding dietary emulsifiers and artificial preservatives. Dietary emulsifiers are food additives intended to keep ingredients combined. For example, oil and vinegar won't stay combined on their own. However, with the addition of a strong chemical emulsifier, they can be kept together. Sometimes emulsifiers are added to improve the mouth feel of a food or to increase their shelf life. Emulsifiers are added to many packaged foods and can be a challenge to avoid, particularly when it comes to convenience foods. Reducing processed packaged food is a good place to start.

INFLAMMATION

In addition to Vitamin D and diet, other actions can reduce inflammation. If you smoke or vape, stop. Your physician can help you develop a stop-smoking plan. Stay up to date on vaccinations, including annual flu and recommended COVID-19 vaccinations. Keep your cardiovascular system healthy by taking any cholesterol and blood pressure lowering medications prescribed by your doctor. Manage your stress levels. The next page offers suggestions for stress management.

WANT TO LEARN MORE?

Learn more about emulsifiers and gut health

NO. 16 ARE THERE ANY LIFESTYLE IMPROVEMENTS THAT MAY REDUCE RISK? *(Continued)*

STRESS MANAGEMENT

Research suggests that patients affected by cavernous malformation are more likely to experience anxiety. Living with this condition can be stressful! Here are some tips to manage that stress:

- Identify and accept your coping style. Some people reduce anxiety by researching all they can. For others, that same knowledge increases nervousness. Knowing where you fall on the spectrum can help you seek the right amount of information. People differ in their support needs, too. Do you feel better surrounded by friends and family, or do you need extra time to be alone with your thoughts and feelings? There is no right way to cope, but it helps to be aware of what helps you. Develop a circle of support. This can include the Alliance's online support groups if they are helpful to you as well as family, friends, and people from your faith community. You are not alone!
- Try meditation. There are countless apps that will help you learn to meditate. YouTube also has a wide variety of meditation, guided relaxation, and breathing technique videos available.
- Eat a balanced diet.
- Exercise (with your doctor's ok).
- Keep a gratitude journal.
- Talk to a therapist or spiritual advisor.
- Schedule pleasant activities into your day. This can help by both giving you something to look forward to and by boosting your mood when you engage in the activity.
- Hug someone you love.

NO. 17 ARE THERE OTHER WAYS I CAN BE SAFER?

- It's wise to have some of your medical records readily available. These are good to have with you when you travel and in case of emergency:
  - Either request a CD with your most recent MRI or download it from the patient portal.
  - A copy of your most recent radiology report
  - Any pertinent, recent medical records.
- Emergency contacts are an essential part of a safety plan:
  - Your emergency medical information should include the names & full contact information for at least two emergency contacts.
  - Emergency contacts should be very knowledgeable about cavernous malformation as well as your history, medications, treatment plan, and treating providers.
- Consider wearing a medical alert bracelet indicating you have a condition that can cause brain or spinal cord hemorrhage.
- Consider using a smartphone app to share information with medical personnel in an emergency.
  - Most cell phones offer apps that emergency personnel can access even when your phone is locked.
  - You can include information in more detail on the app and update it as things change.
  - You may need to change the permission settings on your phone to allow the app to show your information to emergency personnel when your phone is locked.

Visit our YouTube Channel for recordings of past webinars, videos from patient conferences, and so much more!

NO. 18 WHAT HOPE IS THERE FOR THE FUTURE?

Living with cavernous malformation can feel hopeless at times. However, treatments are on the horizon that promise hope for those suffering from this illness. Clinical trials of medicines to reduce symptoms and decrease the risk of hemorrhage have begun. Information about enrolling in these trials can be found on our website. Other medications and non-invasive treatments are under investigation in studies around the world. We believe there will be real treatments for cavernous malformation by 2025 and a cure by 2030!

VISIT OUR WEBSITE FOR THE MOST UP-TO-DATE INFORMATION ABOUT TREATMENTS IN DEVELOPMENT.

To learn more about **RESEARCH** going on around the world.

To learn about minimally and non-invasive **TECHNIQUES** under investigation.

NO. 19

WHAT DOES THE *ALLIANCE TO CURE* OFFER PATIENTS AND FAMILIES?

Our mission is to inform, support, and mobilize those affected by cavernous malformation and drive research for better treatments and a cure.

PATIENT INFORMATION Alliance to Cure Cavernous Malformation provides extensive information on our website and in our newsletter. We announce new research findings as they are published. We host patient conferences and webinars with presentations by disease experts.

PATIENT SUPPORT Alliance to Cure Cavernous Malformation offers patient support through private Facebook groups and weekly virtual support group meetings. At our patient conferences, we offer time for attendees to share their stories.

GENETIC TESTING Alliance to Cure Cavernous Malformation offers free genetic testing to individuals with multiple cavernous malformations who are not able to obtain coverage through their insurance.

OPPORTUNITIES TO PARTICIPATE IN RESEARCH Alliance to Cure Cavernous Malformation has a patient registry where you can sign up to be notified about research studies, including clinical drug trials that may be recruiting for patients. We also collaborate in multiple projects that are exploring the impact of the illness on patients' quality of life.

DRIVE RESEARCH In addition to these patient and family activities, Alliance to Cure Cavernous Malformation sponsors an annual International Scientific Meeting, which brings together researchers from around the world. We offer grants to researchers and provide consultations to assist with their clinical drug trial planning.

NO. 20 WHAT SHOULD I ASK MY *DOCTOR*?

If you don't already know the answers to these questions, you may want to ask them at your next appointment and write the answers here.

WHAT *SIZE* IS THE CAVERNOUS MALFORMATION?

HOW *MANY* CAVERNOUS MALFORMATIONS DO I HAVE?

WHAT IS THE *EXACT LOCATION* OF THE CAVERNOUS MALFORMATION?

WHAT *FUNCTIONS* DOES THIS AREA OF THE BRAIN CONTROL?

DOES THERE APPEAR TO BE A DEVELOPMENTAL VENOUS ANOMALY NEAR THE CAVERNOUS MALFORMATION?

DOES IT APPEAR TO HAVE *BLED PREVIOUSLY*?

IN YOUR OPINION, WHAT ARE THE CONDITIONS UNDER WHICH YOU WOULD RECOMMEND *SURGERY* TO REMOVE THIS CAVERNOUS MALFORMATION?

IF I HAVE *ANOTHER BLEED*, WHAT SYMPTOMS WOULD YOU EXPECT?

WHAT SYMPTOMS WOULD WARRANT A CALL TO YOU OR A TRIP TO THE ER?

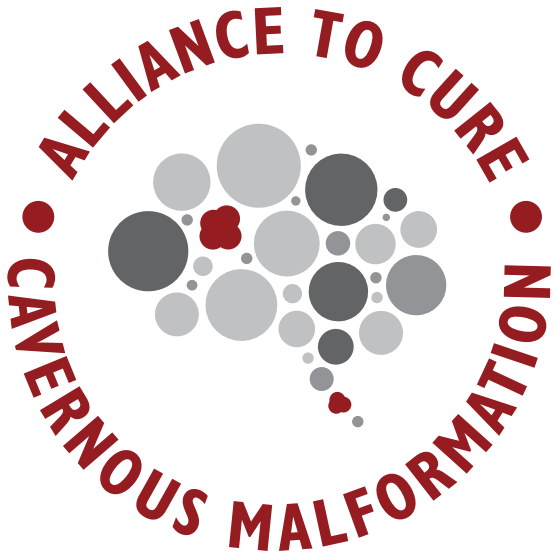
HOW OFTEN WILL I HAVE *FOLLOW UP TESTS* (AND WHICH ONES)?

HOW OFTEN WILL I *FOLLOW UP WITH YOU*?

WANT TO LEARN MORE?

Our website has additional questions and resources for your reference if your doctor recommends surgery.

GOING FORWARD



A cavernous malformation diagnosis may seem overwhelming. **ALLIANCE TO CURE CAVERNOUS MALFORMATION** is an excellent resource for you and your family members to meet others and learn more. Even though the diagnosis is rare, our online patient and family communities are large and growing. We are committed to helping each other. Please visit us on our website at AllianceToCure.org or on Facebook.

NOTES

NOTES

ALLIANCE TO CURE CAVERNOUS MALFORMATION

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