

FIBRO-ADIPOSE VASCULAR ANOMALY

WHAT IS FAVA?

- FAVA is a rare vascular anomaly
- FAVA most often develops in muscles in extremities of the body — arms, hands, legs, feet — but is also seen in other areas, (ex: chest, gluteal, paraspinal)
- FAVA falls under the PROS (PIK3CA-Related Overgrowth Spectrum) umbrella
- FAVA lesions are not malignant; FAVA does not increase your risk of having cancer

FAVA lesions are made up of adipose tissue (fat) and fibrous tissue (which can be like scar tissue). This abnormal tissue is caused by a change in the somatic (body) genes that occurs **only** in the area where the FAVA is located. This genetic change causes abnormalities of tissue growth that manifest as a FAVA lesion.

A FAVA lesion does not destroy the healthy tissue surrounding it. However, because of the abnormal growth of tissue, FAVA may infiltrate the muscle, blood vessels, tendons, and nerves of the area.

WHAT ARE COMMON SYMPTOMS?

- Pain
- Enlargement of the limb or other area of the body at the site of the FAVA
- Stiffness
- Decreased range of motion
- Impairment of function
- Lesion is firm to the touch
- Abnormal skin — sometimes a bluish tint or large, visible veins

Pain, often severe, could be dull or sharp, localized or general, persistent or intermittent, tingling or burning, radiating, or the feeling of pressure. Stiffness and decreased range of motion are due to contracture — tightening and shortening — of the tissues in the affected muscle.



WHEN SHOULD I CONSIDER FAVA AS A POSSIBLE DIAGNOSIS?

- You have been diagnosed with another type of vascular anomaly
- You are experiencing one or more symptoms associated with FAVA
- You have been treated with several rounds of sclerotherapy with no improvement
- A lesion does not shrink, fade, or disappear over time

FAVA is a complicated diagnosis that is often challenging to properly identify. Its features, upon physical examination and imaging, overlap with those of other vascular anomalies.

Techniques used to diagnose FAVA include medical history and examination, MRI, ultrasound, venography, and biopsy.

An interdisciplinary team of medical specialists is often the best option for addressing the complexities involved in properly diagnosing and assessing care.

WILL MY FAVA GET WORSE?

- FAVA may get worse over time
- Its growth patterns vary and are not fully understood
- It is currently believed FAVA does not spread within the body beyond the affected muscle(s)

While individuals are born with FAVA, symptoms arise at different times for different people, most often between childhood and young adulthood. Progression of symptoms is particularly significant during puberty and may worsen in women during pregnancy.

Despite a lack of scientific investigation, the best current explanation of FAVA growth and correlating pain is that, as fibro-fatty tissue surrounds more nerves, pain, and often swelling, increase. If a FAVA has a significant venous component, pain may be caused by swelling and inflammation of the vein (phlebitis/phlebectasia).

TESTING FOR GENETIC MUTATIONS?

Testing is possible, but only via biopsy of the tissue at the site of the FAVA.

Because FAVA is most commonly caused by a mosaic PIK3CA mutation, and the mutation is not found in every cell of the body, genetic testing can be extremely challenging and produce inaccurate results.

The percentage of mutated cells in a tested tissue sample could be as low as 1%, requiring an ultra deep sequencing technique with high coverage in order to produce a more specific result.



HOW DID I GET FAVA?

- FAVA is congenital — it occurs from birth
- For some people, FAVA involves a mutation of the PIK3CA gene
- For others, FAVA possibly involves a mutation of the TIE2/TEK gene
- Both genetic mutations are somatic — they cannot be passed down to your children

Due to a phenomenon known as mosaicism, a mutation of the PIK3CA or TIE2/TEK gene occurs at random during fetal development, as the cells divide inside the embryo. Because of this, only some cells in a person's body are affected by the mutation. FAVA only develops in body parts containing the altered cells.

Because the mutations that cause FAVA occur randomly, it has no greater prevalence in any particular population.

HOW MANY PEOPLE HAVE FAVA?

- The total number of people currently living with FAVA is unknown
- Some with FAVA may have received an incorrect diagnosis of another condition
- FAVA is a rare disease

In the United States, a disease is considered rare if it affects fewer than 200,000 people; in the European Union, if it affects fewer than 1 in 2,000 people.

FAVA was first documented as a unique vascular anomaly in 2014, and thus diagnoses have only occurred since then. Prior to that, those with FAVA were diagnosed with a variety of vascular malformations and tumors. FAVA is still often misdiagnosed today.

JOIN PROJECT FAVA

To join, learn more about FAVA, or make a tax-deductible gift, go to www.projectfava.org.

Project FAVA is a patient advocacy organization devoted to furthering research and making a positive impact in the lives of those with fibro-adipose vascular anomalies.

We encourage you to join Project FAVA to stay informed of the latest news and breakthroughs in FAVA care and to learn about clinical trials, surveys, and other research studies seeking participants.

As a not-for-profit organization, Project FAVA relies on and greatly appreciates contributions from friends, corporations, and foundations. We are a qualified 501(c)(3) tax-exempt organization, and all donations are tax deductible to the fullest extent allowable by law.

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All medical information contained in this document has been reviewed and approved by members of Project FAVA's Medical Advisory Board.

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