WHAT IS BEHÇET’S DISEASE?

Behçet’s disease is a rare disorder that causes swelling in blood vessels, which can cause mouth, skin, and genital sores. Over time, this swelling and inflammation damage the blood vessels and put patients at high risk of complications, such as blood clots.

Behçet’s is likely an autoimmune condition, meaning it is caused by the immune system attacking the body’s own cells. There are several genetic mutations that doctors have linked to Behçet’s disease, but not everyone with these genetic markers develops it. Both genetic and environmental factors likely affect the development of Behçet’s disease.

Behçet’s disease can occur in anyone, but it is more common in people who live in the Mediterranean basin and Central and Eastern Asia.

There is no cure for Behçet’s disease, but medication can help manage symptoms.

COMMON SIGNS AND SYMPTOMS OF BEHÇET’S DISEASE

Behçet’s disease symptoms vary from patient to patient and often seem unrelated at first. Common symptoms include:

- Sores on the mouth, skin, or genitals. Mouth sores start as raised round bumps and develop into painful ulcers that look and feel similar to canker sores. Skin sores usually appear on the legs and can look like acne or tender red bumps. Genital sores usually appear on the scrotum or vulva and are usually round red bumps.
- Eye inflammation that causes redness, pain, and blurred vision.
- Swelling and pain in the knees, ankles, wrists, or elbows

Behçet’s disease can also cause the following complications:

- Blood clots in the veins and arteries that cause redness and pain in the arms or legs.
- Aneurysms (weakening and outpouching of blood vessel walls)
- Digestive problems like abdominal pain, bleeding, or diarrhea
- Headache, fever, and disorientation
LAB TESTS RELATED TO DIAGNOSING BEHÇET’S DISEASE

*Reference ranges are set by individual laboratories for their specific populations, so reference ranges might differ slightly.

There are no lab tests that can definitively diagnose Behçet’s disease, but they are used to rule out other conditions like lupus, Crohn’s disease, and rheumatoid arthritis. The lab team can also check for the genetic mutation related to Behçet’s disease, called HLA-B51. 30-60% of people with Behçet’s have this genetic mutation, but if you have the mutation, it doesn’t mean you will get Behçet’s disease. Many people with the genetic mutation do not develop Behçet’s.

**HLA-B51:** This is a blood test that checks for the genetic mutation that is associated with Behçet’s disease. If it is positive, the patient is at a higher risk of developing Behçet’s disease.

**ANTINUCLEAR ANTIBODY (ANA):** This test is used to help determine if a person might have an autoimmune disorder that affects many tissues and organs. It is important to note that a positive ANA test does not diagnose one particular disease, but it does indicate a potential autoimmune disorder. This test is important because it helps support the diagnosis of an autoimmune disease such as Lupus or Sjogren’s disease. A positive ANA test means that these particular autoantibodies are present in the patient’s blood.

**ALBUMIN:** This test measures the levels of albumin (a type of protein) in the blood. This test is important because very low levels or very high levels of albumin are a sign of a possible health condition. This test can be used to determine whether some symptoms are more likely related to an inflammatory bowel disease like Crohn’s or ulcerative colitis or a nutritional deficiency, as people who have nutritional deficiencies for dietary reasons or because they have issues with absorbing nutrition from the gut will have low levels of albumin in their blood. The typical reference ranges* are between 3.4 - 5.4 g/dL.

UNDER THE MICROSCOPE

*In this image, small blood vessels (green arrows) are surrounded by inflammatory cells (yellow arrow) which have been stained red to help the pathologist identify them. Some inflammatory cells can also be seen within the walls of the blood vessels.

QUESTIONS TO ASK YOUR DOCTOR

- Which treatment option(s) do you recommend?
- Why do you recommend that specific treatment option?
- What are some signs and symptoms of a flare-up of Behçet’s disease? How do I prevent a flare-up?
- Are there additional tests that could be used to understand my disease and prognosis better?
- What changes should I make to my lifestyle? Should I make any dietary changes?
- Are there support groups or other resources to help me navigate my condition?
ANTI-TISSUE TRANSGLUTAMINASE ANTIBODIES (ANTI-TTG): This test measures the level of antibodies to tissue transglutaminase that are present in the blood. This test is important because high levels of anti-tTG antibodies can be a sign of celiac disease, a cause of digestive problems and malabsorption of nutrients in the small bowel. The typical reference ranges* are

- Negative: 3U/mL
- Weak Positive: 4-10 U/mL
- Positive: 11 U/mL

COMPLETE BLOOD COUNT (CBC): This test helps determine your health status by looking at your overall blood count levels, including your red blood cells, white blood cells, and platelets. This test is important because it can indicate if you might have a condition or disease, such as an infection, anemia, inflammation, or cancer.

COMPREHENSIVE METABOLIC PANEL (CMP): This panel includes the components of a Basic Metabolic Panel (BMP), which measures the current status of several components of your blood, including glucose, electrolytes, markers of kidney function, and fluid balance, with additional measures related to liver activity and function. This panel is important because your body tightly regulates your physiology and these values can quickly indicate when there is a problem.

C-REACTION PROTEIN (CRP): This test measures the level of C-reactive protein in the blood. This is important because high levels of CRP are a sign of increased inflammation in the body. The typical reference range* is less than 10 mg/L.

CYTOMEGALOVIRUS TEST (CMV): This test measures the presence of CMV antibodies in the blood. This test is important because a CMV infection can worsen symptoms for patients with inflammatory disorders of the bowel like Behçet's disease.

ERYTHROCYTE SEDIMENTATION RATE (ESR): This test measures how quickly erythrocytes (red blood cells) separate from other components of blood (serum or plasma) and sink to the bottom of a test tube. This test is important because a fast fall rate indicates increased inflammation in the body. The typical reference range* for men is between 0-22 mm/hr and 0-29 mm/hr for women.

FERRITIN: This test measures the levels of ferritin, a protein that stores iron, in the blood. This test is important because very low and very high levels of ferritin in the blood can be signs of inflammatory conditions. The typical reference ranges* for adult males are between 20-250 ng/mL and 10-120 ng/mL for adult females.

LUPUS-SPECIFIC ENZYME-LINKED IMMUNOSORBENT ASSAYS (ELISA): These tests measure the presence of specific antibodies associated with lupus. ELISA can be used to test for the anti-double-stranded DNA antibody (anti-dsDNA) or anti-Smith Antibody (anti-SM). Anti-dsDNA is important because it is found in about 65 - 85% of people with lupus and less than 1% of healthy individuals. The typical reference range* for anti-dsDNA in adults is less than 10 IU/mL. Anti-Sm is also important because it is found in 9% to 49% of patients with lupus and rarely in healthy individuals. The typical reference range* for anti-Sm in adults is less than 7 U/mL.

SERUM IRON/SERUM FE: This test measures the level of iron in the blood. This test is important because low or high iron levels are a sign of its deficiency in the body and can help diagnose this condition. The typical reference range* is between 60-170 mcg/dL.

TOTAL IRON BINDING CAPACITY (TIBC): This test measures the body’s ability to attach iron and move it around the body. This test is important because low levels of TIBC can be a sign of inflammation in the body. The typical reference range* is between 240- 450 mcg/dL.

VITAMIN D TEST (25-HYDROXYVITAMIN D TEST): This test measures the amount of 25-hydroxyvitamin D in the blood. This test is important because it is used to screen for fat-soluble vitamin deficiencies which can occur in patients with diseases that can cause inflammation in the gastrointestinal tract like Behçet’s disease. The typical reference range* is between 20 and 40 ng/mL.
While she didn’t have a name for her condition, Jasmin endured symptoms her entire life. She spent a lot of time getting shuffled to different specialists who inevitably couldn’t give her answers. After several doctors and dozens of negative laboratory results, she began to wonder if her symptoms were all in her head. Jasmin majored in Clinical Laboratory Science at Winston-Salem State University, and once she learned about different aspects of laboratory testing, she suggested some tests for her doctors to order. Working together, they made a diagnosis of Behçet’s disease.

Jasmin received her diagnosis the day before her 25th birthday. While she felt relieved to finally have a name for all her symptoms, she was also scared. She learned to live with Behçet’s disease one day at a time, even if some days she embraces it and others she’s angry at the world. Over time she’s discovered that having a positive outlook, a solid support group, and advocating for others helps her achieve her dreams.

To learn more about other lab tests, go to www.ascp.org/patients.