

# DOCTOR DISCUSSION GUIDE FOR SYSTEMIC MASTOCYTOSIS (SM)



Not an actual patient or medical professional

Unexplained allergic reactions and recurring symptoms in your skin and gut could all be connected. If you suspect you have SM, use this guide to help you have an informative conversation with your doctor.

## THREE THINGS YOU MAY WANT TO COVER WITH YOUR DOCTOR



Your Symptoms



Your Triggers & Medical History



Tests & Next Steps

SM is a rare condition affecting approximately 32,000 patients in the U.S. (or 0.01% of the population)! **Because it can have so many different signs and symptoms, SM can be tricky to diagnose.**

**In approximately 95% of cases, SM has been associated with a genetic mutation in the gene called KIT D816V.** This causes the overproduction of abnormal mast cells (a type of white blood cell). If these cells build up and become overactive, they can cause symptoms in different parts of the body.

Only a doctor can make an SM diagnosis, and it's important to discuss all your symptoms, triggers, and medical history, so they can help determine if you should be evaluated. While awareness of SM is low, advocating for yourself and asking your doctor if you should be evaluated for SM could help you get the answers you need for an accurate diagnosis.

**My goal for this appointment is:**

# SYMPTOMS

Use this chart to identify symptoms you've experienced or rule out any you haven't.

**Airway symptoms** Nasal congestion, shortness of breath, wheezing, or throat swelling.

Severe     Recurring     Unpredictable     Unexplained     None

**Anaphylaxis** An extreme allergic reaction that may have resulted in fainting or dizziness, the use of an epinephrine autoinjector (e.g., EpiPen®), or a visit to the emergency room.

Severe     Recurring     Unpredictable     Unexplained     None

**Bone or muscle symptoms** Bone pain, muscle pain, bone loss, or weak and brittle bones that break easily.

Severe     Recurring     Unpredictable     Unexplained     None

**Gut symptoms** Abdominal pain, cramping, diarrhea, nausea, vomiting, bloating, heartburn, or acid reflux.

Severe     Recurring     Unpredictable     Unexplained     None

**Head or brain symptoms** Brain fog, difficulty concentrating, anxiety, depression, difficulty sleeping, or headaches.

Severe     Recurring     Unpredictable     Unexplained     None

**Heart symptoms** Low blood pressure, rapid heart rate, chest pain, dizziness, or fainting.

Severe     Recurring     Unpredictable     Unexplained     None

**Skin symptoms** Red or brown, spot-like rashes that become swollen or itchy, skin that swells up if it gets scratched, flushing that lasts more than a few minutes, or hives.

Severe     Recurring     Unpredictable     Unexplained     None

**Whole-body symptoms** Fatigue, weakness, weight loss, sweats, chills, or enlarged organs.

Severe     Recurring     Unpredictable     Unexplained     None

**Other symptoms:**

# TRIGGERS & MEDICAL HISTORY

Use this space to provide details that might help your doctor better understand your symptoms.

**The things that may trigger your symptoms, even if your triggers seem to change.**

For example: bee stings, certain food or drinks, stress (emotional, physical, environmental).

**Any medications, supplements, or over-the-counter products you are using.**

**What else are you doing to relieve your symptoms (beyond medications) and how well is it working?**

**Your medical history may contain past diagnoses that can help shed light on your current symptoms. SM is sometimes mistaken for: cutaneous mastocytosis, IBS, and others.**

You can request your medical records from your previous doctors and print them out or email them.

**How are your symptoms affecting your life?**

For example: are they causing you to lose sleep, avoid activities, stopping you from seeing family or friends, or making it difficult to work?



Not an actual patient  
Symptoms depicted are illustrative only

## Tip:

Loved ones can help you prepare for your appointments. They may notice things that you have forgotten or overlooked. Asking for input or bringing them to your appointment could be helpful to you and your doctor.

## TESTS & NEXT STEPS

If your doctor determines your symptoms may be associated with SM, there are some tests they might perform to evaluate you further. Below are some actions they may consider.

### BLOOD TESTS:

**Tryptase test** – This measures the amount of tryptase (an enzyme that is released by mast cells) in your blood.

- Your doctor may run the tryptase test more than once because levels can fluctuate. For more accurate results, your doctor may ask for a tryptase test after you have an allergic reaction.

**KIT D816V test** – The KIT D816V blood test can help aid in the evaluation of SM by identifying a genetic mutation that has been found in approximately 95% of patients with SM.

- There are different types of KIT tests available which vary in sensitivity, and high-sensitivity tests may be required to detect SM. Talk to your doctor for more information about KIT D816V testing options.

### BIOPSIES:

**Biopsy of skin lesions** – Your doctor or a dermatologist may order a biopsy to look at the number of mast cells in the deeper layers of your skin.

**Bone marrow biopsy** – This test looks at the number and shape of the mast cells to see if they are abnormal. Bone marrow is almost always affected by SM, so a bone marrow biopsy is a crucial diagnostic tool.

**Organ biopsy** – This test looks at the number of mast cells in other organs such as the liver, spleen, gastrointestinal tract, and lymph nodes.

### REMEMBER

Only a doctor can determine which tests may be appropriate for you. Blood tests may help aid in the diagnosis of SM and determine if you should receive other tests such as a bone marrow biopsy to confirm the SM diagnosis. If an SM diagnosis is confirmed, your doctor may perform additional tests to determine the SM subtype and develop a treatment plan.

### QUESTIONS FOR YOUR DOCTOR ABOUT NEXT STEPS:

- Do you recommend I see any other doctors or specialists regarding my symptoms?
- Who should I work with to schedule any follow-up test(s) or evaluation(s)?
- What do you recommend to help manage my symptoms while I wait for testing results?
- If SM isn't the cause of my symptoms, are there any other ways to help determine what is?
- If SM is diagnosed, can it progress?
- Are there any patient support or advocacy groups you recommend for support or additional information on SM?