

TOUCHSTONE SM SURVEY

An overview of results from a survey
of people living with and treating
systemic mastocytosis (SM)



COMPASSION
in ACTION
PATIENT AFFAIRS



FOREWORD

Systemic mastocytosis (SM) is a rare condition where abnormal mast cells (a type of white blood cell) build up in the body. People living with SM may often experience severe and unpredictable symptoms, which can have a debilitating impact on their lives. It is estimated that approximately 32,000 people in the United States are living with SM.¹ In most people with SM, the condition is caused by a mutation, or a change, in a specific part of their DNA called the *KIT* gene (identified as *KIT D816V*).

The ***TouchStone SM Survey***, conducted by Blueprint Medicines in the United States during the summer of 2020, surveyed both patients and health care providers with the intent to better understand the holistic patient experience and the perceptions of SM disease burden. This report serves as a summary of those findings related to SM diagnosis, symptoms, disease impact, and management. Most importantly, it helps us understand the extent to which SM can impact various aspects of a person's life. It may not be occasional symptoms that flare and then subside; SM can affect a person's physical, mental, and emotional state. More progress is required to understand SM and its management in order to ease the burden on those who are suffering.

This survey study was intended to raise awareness of SM and to inform and empower patients living with the disease, as well as those treating them. We, along with countless health care providers and advocacy partners, are dedicated to driving this awareness and providing the support and collaboration needed to make a difference in the lives of patients living with SM. Join us on this journey to greater understanding and to driving advancements through innovation.

Ruben Mesa, MD & Mariana Castells, MD, PhD

TouchStone SM Survey Working Group Members

Reference: 1. Data on file. Blueprint Medicines Corporation, Cambridge, MA 2022.



**Cheri (caregiver) and
Mike (living with SM)**

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TOUCHSTONE SM SURVEY WORKING GROUP

The *TouchStone SM Survey* was conducted by Blueprint Medicines, a biopharmaceutical company, in collaboration with a working group of external SM experts from across the United States. These experts are listed below and include doctors, nurses, patients, and professionals from Blueprint Medicines who are interested specifically in SM research and advocacy.*

Ruben Mesa, MD¹

Mays Cancer Center at UT Health San Antonio; MD Anderson Cancer Center San Antonio, Texas

Erin Sullivan, PhD, MPH

Blueprint Medicines Corporation Cambridge, Massachusetts

David Dubinski²

Blueprint Medicines Corporation Cambridge, Massachusetts

Brittany Carroll

Blueprint Medicines Corporation Cambridge, Massachusetts

Valerie Slee, RN, BSN & Team

The Mast Cell Disease Society, Inc. Sterling, Massachusetts

Linda Bohannon, RN, BSN, MSM³

Cancer Support Community Washington, D.C.

Mariana Castells, MD, PhD

Brigham and Women's Hospital Boston, Massachusetts

The success of this work is due to the valuable participation of patients and health care providers. These groups gave their time to provide thoughtful responses to the survey questions. Their contributions will help the entire SM community better understand this difficult disease. The working group would like to thank all patients, caregivers, patient advocates, and health care providers who work daily to improve the lives of people with SM.

*External experts were compensated for their time as part of the TouchStone SM Working Group.

Please note the following updates since the time this survey was launched:

¹Now at Atrium Health Levine Cancer Institute - Charlotte, North Carolina

²No longer with Blueprint Medicines

³Now at Vital Strategic Insights - Indianapolis, Indiana

ABOUT SYSTEMIC MASTOCYTOSIS

What is SM?

Mastocytosis is a condition that occurs when too many abnormal mast cells build up in tissues or organs. Mast cells are an important part of the immune system and keep people safe from disease. When too many mast cells build up, the substances released can lead to inflammation or cause symptoms to make the body think there is an allergic reaction. This can sometimes result in organ damage.

Which parts of the body can be affected by SM?

A few parts of the body that may be affected:

- Skin
- Digestive system
- Brain
- Lungs
- Heart
- Muscles and bones

What are the common symptoms of SM?

Symptoms may change based on where mast cells build up. Common symptoms may be:

- Skin symptoms (like rash, redness, hives)
- Gut issues (diarrhea, abdominal pain)
- Anaphylaxis (severe allergic reaction)
- Brain fog or headaches
- Fatigue
- Bone pain
- Low blood pressure

What are some of the challenges of SM?

- The chronic nature of SM and the changing symptoms can take a toll on a person's mental and emotional well-being
- Symptoms can be unpredictable, which can significantly impact a person's daily life
- Individuals may need to make changes to their social activities and work responsibilities due to symptoms
- It may take a long time to be diagnosed with SM
- Many people with SM see multiple doctors and take multiple medications

SM IS RARE

It is estimated to occur in approximately 0.01% of the U.S. population (or 1 in 32,000 people).¹

In most people with SM, the condition is caused by a mutation, or a change, in a specific part of their DNA called the KIT gene.



ANAPHYLAXIS

One of the potentially severe symptoms of SM is anaphylaxis.

Anaphylaxis is a severe allergic reaction. It can occur very quickly after a person comes into contact with something they are allergic to, such as a food, plants, or insect bites or stings. It can be life-threatening if not treated quickly.

Reference: 1. Data on file. Blueprint Medicines Corporation, Cambridge, MA 2022.

TYPES OF SYSTEMIC MASTOCYTOSIS

	NON-ADVANCED		ADVANCED		
Type	ISM <i>(Indolent SM)</i>	SSM <i>(Smoldering SM)</i>	ASM <i>(Aggressive SM)</i>	SM-AHN <i>(SM with Associated Hematologic Neoplasm)</i>	MCL <i>(Mast Cell Leukemia)</i>
Prevalence	It is estimated that 1 in every 10,000 adults has SM. Of those that have SM, the majority have ISM. It is the most common type.	The prevalence of SSM is unknown.	It is estimated that 1 to 9 in every 1,000,000 people have ASM.	It is estimated that 1 to 9 in every 100,000 people have SM-AHN.	MCL is extremely rare.
Fast Facts	Many people with ISM experience fatigue, brain fog, and gut issues. People with ISM have a life expectancy similar to the general population.	Overall survival is lower than that of ISM. It is estimated that 9% of SSM cases will progress to ASM, SM-AHD, or MCL.	In ASM, there is more severe build-up of mast cells in different tissues. It is associated with progressive organ damage.	SM-AHN may indicate another blood disorder is present.	MCL affects the bone marrow and blood.

These SM subtypes were included in the TouchStone survey. In 2022, World Health Organization (WHO) published an updated classification criteria that further defined the subtypes. Please consult your doctor to learn more.

About 95% of SM cases are driven by activated *KIT* gene mutations.

ABOUT THE SURVEY

What was the purpose of the survey?

The purpose of the *TouchStone SM Survey* was to learn more about SM and how it affects those with the disease. The goal of learning this information was to help better understand how people are diagnosed with SM and what symptoms they experience.

Who took part in the survey?

There were 2 separate surveys. One was designed for people living with SM, and the other was for health care providers (HCPs) treating people living with SM. People living with SM had to be 18 years of age or older and have a self-reported diagnosis of SM to take part in the survey. They were invited to take part through the Mast Cell Connect patient registry.*

HCPs treating people with SM self-reported that they see at least 4 patients with SM and have been practicing for at least 3 years.

How many and what type of questions were in the survey?

The survey for people living with SM consisted of 100 questions and asked people to share information about:

- How they were diagnosed with SM and how long the diagnosis took
- What symptoms of SM they experience
- Past and current treatments for SM
- How often they see their HCPs, take medication, or go to the hospital
- How SM affects their daily life

The survey for HCPs treating people living with SM consisted of 51 questions and asked about their professional background, how they care for their patients with SM, and how they think SM affects their patients.

What did we learn from the survey?

Together, the 2 surveys have helped us learn about the differences between how HCPs and people living with SM think about SM. The surveys may also help us better understand how to help people living with SM and their HCPs better communicate to find tools and resources that might help ease the burden of SM.

*Mast Cell Connect was a volunteer online patient registry owned and managed by Blueprint Medicines, created to collect information from those with self-reported SM who signed up to share their experiences about the disease. The registry is now closed.

What were the limitations of the survey?

Limitations for the survey of people living with SM:

- An online survey may not allow for a truly diverse sample population. This means that the results we found may not be representative of the experience of all SM patients
- The survey was completed by people with self-reported SM, so there was no medical confirmation of an official diagnosis
- The survey was given during the time of the global COVID-19 pandemic, but people were asked to report on their health care experience the year prior to COVID-19. This may have changed the way people felt about their health. It may also have changed how often they visited their HCPs

Limitations for the survey of HCPs treating people living with SM:

- The 39% response rate to the HCP survey could be considered a limitation, but is consistent with HCP response rates documented in the literature
- Allergists, immunologists, hematologists, and oncologists play a crucial role in treating people with SM, but not including other specialties may affect the results of the survey
- The online survey method may have only attracted HCPs with electronic contact details
- HCP responses regarding patient experiences were not linked to a survey of their SM patients, which limits our ability to draw conclusions about whether HCP perceptions align with actual patient experiences

Those reading this summary should consider these limitations as they review the results.

ABOUT THE SURVEY PARTICIPANTS

As part of this survey, we wanted to understand the differences in how health care providers and people living with SM think about the effects of SM. To help meet this goal, we surveyed both **people with SM** and **health care providers who treat people with SM**.

56 PATIENTS

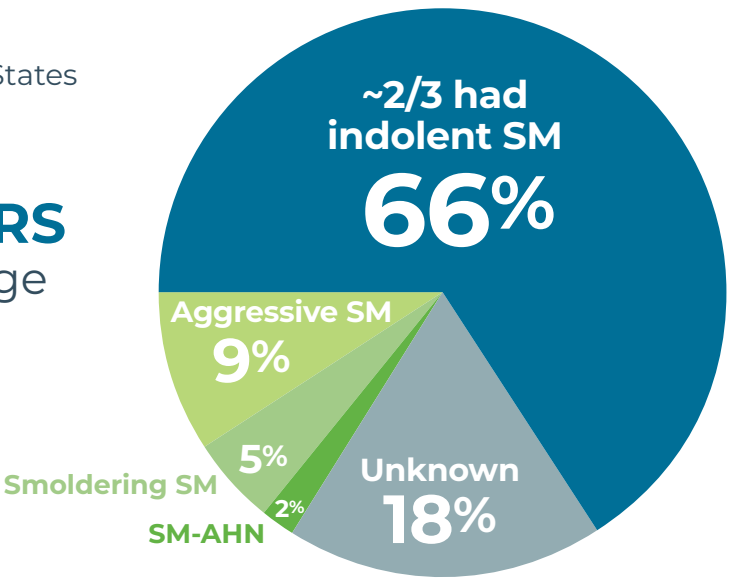
with self-reported SM living in the United States

89% female



48 YEARS median age

On average, individuals had been diagnosed for approximately **7 YEARS**



111 HCPs

treating patients with SM in the United States

57 Allergists/Immunologists



54 Hematologists/Oncologists



HCPs reported that they currently treat or were treating at least 4 patients with SM

JOURNEY FOR PATIENTS WITH SYSTEMIC MASTOCYTOSIS

Receiving an SM diagnosis can be a long journey for some people, and it may require many visits to many different types of doctors. We asked our survey participants to tell us about how long it took them to get a diagnosis and share details about that journey.



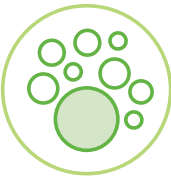
Time from first noticing symptoms to receiving an SM diagnosis took an **average of ~6 years**



However, HCPs reported that it took **~8.5 months to diagnose a patient with SM** after patients started experiencing symptoms



Under half (43%) of patients in this study received their diagnosis from an allergist/immunologist, and **59%** said this is the type of doctor that is primarily managing their SM care



HCPs estimated that **~47%** of their patients with SM had the **KIT D816V driver** mutation

“Indolent as a description of SM may be misunderstood in the thinking that symptoms are less severe; however, ISM patients reported negative symptoms that significantly impact their quality of life.”

Ruben Mesa, MD
TouchStone SM Survey Working Group Member

SYSTEMIC MASTOCYTOSIS SYMPTOMS

To fully understand the severity of symptoms for people with SM, we asked what the most bothersome symptoms were, how severe they were, and compared those answers to what health care providers thought their patients experienced.

9 out of 10 patients surveyed had experienced 10 or more SM-related symptoms throughout their lifetime

Patients (N=56) reported the following 4 symptoms as the “most bothersome”



Anaphylactic episodes
18%



Abdominal/stomach pain
16%

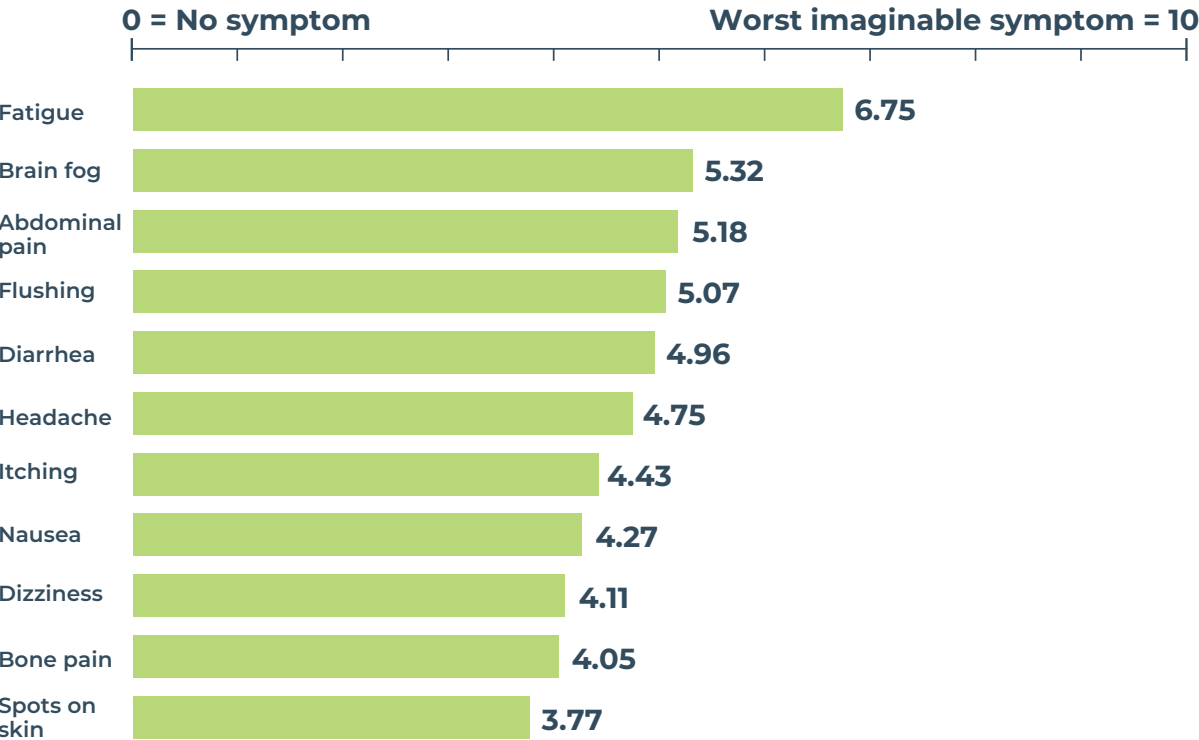


Diarrhea/loose stools
13%



Fatigue
11%

Patients scored the following symptoms on a scale of 0 to 10 over a 2-week period; the numbers below represent the average severity of ratings:



In the HCP survey (N=111), HCPs perceived that on average 75% of their ISM patients have no symptoms, mild symptoms, or moderate symptoms

ABOUT ANAPHYLAXIS

One symptom of SM is anaphylaxis, but people living with SM manage anaphylaxis in different ways. We wanted to understand how often people had to go to the emergency room vs how often they manage anaphylaxis on their own.

Over the past year, patients (N=56)...



30% reported going to the emergency room at least once for anaphylaxis



63% reported having anaphylaxis but treated it at home instead of going to the emergency room at least 1 time



88% reported keeping injectable epinephrine on hand for emergency use in case of an anaphylactic episode

Over 88% of patients were prescribed epinephrine, showing that health care providers are aware of the risk of anaphylaxis in people with SM.



Jennifer, living with ISM

MANAGING SYSTEMIC MASTOCYTOSIS

Not only are the symptoms of SM bothersome to people living with it, but how SM is treated can also be a burden. People often have to take multiple medications, see many different types of doctors, and spend a lot of time at appointments.



61% of patients with SM reported taking **3 or more over-the-counter medications**



52% of patients with SM reported taking **3 or more prescription medications**

When asked to rank the treatment goals of their patients with SM, HCPs (N=111) said:

Patients with Advanced SM

- 1 Improved progression-free survival/overall survival
- 2 Better quality of life

Patients with ISM

- 1 Better quality of life
- 2 Improvement of symptoms

Patients (N=56) reported frequent visits to multiple specialists for management of their SM during a 1-year period

Primary Care Physician	Allergist/Immunologist	Gastro-enterologist	Dermatologist	Hematologist/Oncologist
80% Reported 1 or more visits	77% Reported 1 or more visits	56% Reported 1 or more visits	45% Reported 1 or more visits	48% Reported 1 or more visits
51% Reported 3 or more visits	45% Reported 3 or more visits	24% Reported 3 or more visits	7% Reported 3 or more visits	31% Reported 3 or more visits
24% Reported 6 or more visits	21% Reported 6 or more visits	11% Reported 6 or more visits	2% Reported 6 or more visits	11% Reported 6 or more visits
11% Reported 12 or more visits	13% Reported 12 or more visits	5% Reported 12 or more visits	0% Reported 12 or more visits	7% Reported 12 or more visits

Percentages have been rounded to the nearest whole number and may not add up to 100.

IMPACT ON QUALITY OF LIFE

Living with SM can impact people in all areas of their lives, including relationships, work, finances, and their overall quality of life. We asked questions to help us understand how much living with SM impacted their daily lives.



64% of patients (N=56) with SM said that they **avoid leaving their house** because of SM symptoms

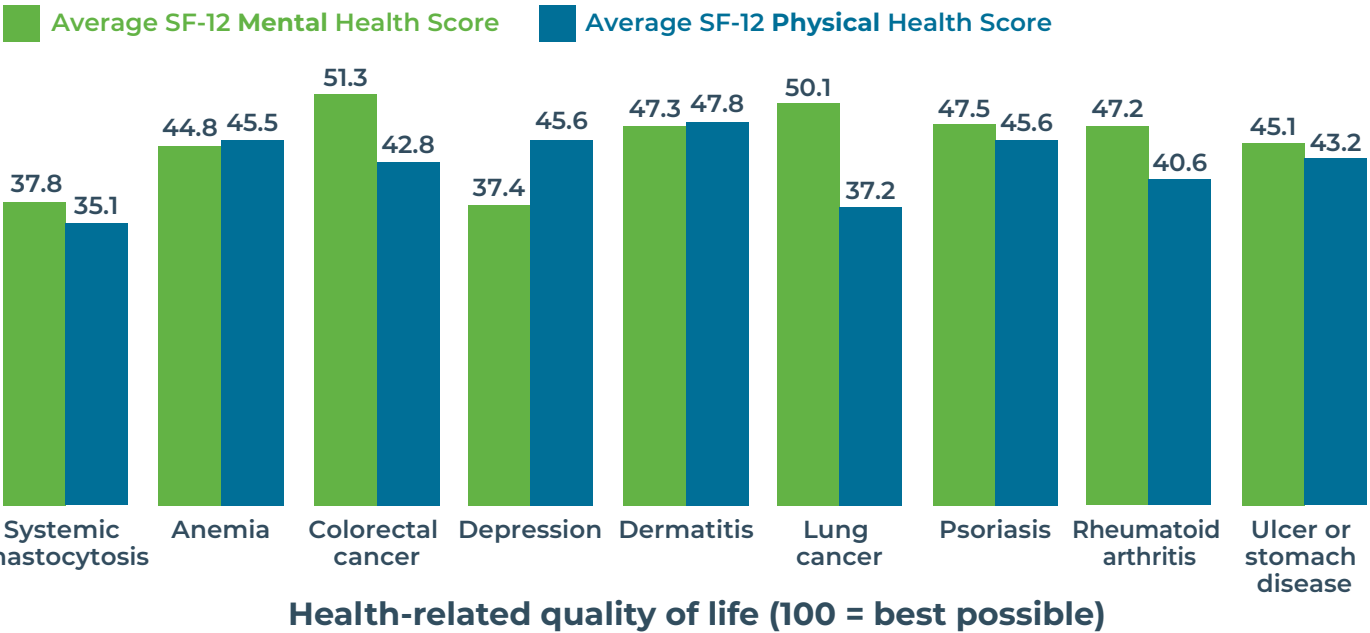


>50% of HCPs (N=111) said their patients with ISM felt quite a bit or a great deal **depressed or discouraged**

Quality of Life in Comparison to Other Diseases

SM symptoms can significantly affect health-related quality of life. To better understand how SM affected people, the survey included 12 questions from a questionnaire called the SF-12. The SF-12 asked about two main areas of well-being: physical and mental health. Results were then compared between people with SM to patients with other diseases, including several types of cancer. Higher scores reflect a better quality of life.*

On average, **people with SM reported lower or comparable physical health and mental health scores** when compared to people with other serious diseases.



*On the Short-Form 12-item health survey (SF-12), scores range from 0 (worst possible health) to 100 (best possible health). Scores from patients with other serious diseases were collected by other researchers in a separate study, where the study design and its participants may not have been similar to the SM survey participants. Comparing the quality of life between people with SM and those with other conditions may not be accurate or fair due to the unique characteristics of each condition, including disease duration, life expectancy, and other factors.

WORK STATUS AND PRODUCTIVITY

Patients (N=56) reported SM symptoms had significant impact on their ability to work and perform usual activities*



54%
Reduced hours at work



27%
Voluntarily quit job



37%
Had gone on medical disability



16%
Had been terminated



27%
Reported no impact on work by SM

*Patients could select multiple answer choices.

More than 50% of HCPs surveyed (N=111) perceived that ISM symptoms limit their patients in the following areas:

- School/university/work
- Sports/physical activity
- Sleep
- Sexual activity
- Leisure time
- Relationships
- Ability to care for children

Percentages have been rounded to the nearest whole number and may not add up to 100.

LOOKING FORWARD

Findings from the **TouchStone SM Survey** show an overall negative impact of SM on people’s lives.

Despite reporting use of multiple over-the-counter and prescription medications and frequent doctor visits to manage their SM in this survey, people reported impaired physical functioning and mental health, decreased work performance and productivity, and difficulty completing daily activities. Many also reported having multiple episodes of anaphylaxis, including frequent visits to the emergency department. The survey results demonstrate that there are needs in this population of people that are not yet met, and future studies evaluating new treatments for SM should measure the impact on symptom burden and quality of life.

In addition to the results about quality of life for people with SM, we also found that there are several areas where the community needs to learn even more, such as:

- **Ways to improve communication between HCPs and their patients**
- **How the experiences of people with SM compare with one another (the survey group told us that other patients are one of the most important sources of information about SM!)**
- **How people with SM are feeling so others can offer understanding and support**

We hope that the results from this survey will help patients, caregivers, HCPs, and researchers better understand what it is like to live with SM. People who care for others with SM can read the survey report to learn how their loved ones may be feeling and to identify areas where they may be able to offer understanding and support. HCPs can read to find an honest assessment of their patients’ feelings about their disease and their health care. This may prompt them to ask new questions or start different discussions when they meet with their patients who have SM.

Researchers can use this survey as a foundation to ask additional questions:

What is important to the patient (ie, treatment goals)? Does this differ by the type of SM a person has or by their age, race, gender, or other characteristic? How can we help people better avoid anaphylaxis? How much do people with SM pay out-of-pocket for their care?

The working group for this survey would like to thank everyone involved in the survey, especially the people who are living with SM who so thoughtfully shared their experiences.

For more information on the TouchStone SM Survey, please scan the codes below to visit two papers published in the medical journal *Cancer: An International Interdisciplinary Journal of the American Cancer Society*.



Patient-reported Outcomes Among Patients with Systemic Mastocytosis in Routine Clinical Practice: Results of the TouchStone SM Patient Survey



Perceptions of Patient Disease Burden and Management Approaches in Systemic Mastocytosis: Results of the TouchStone Healthcare Provider Survey

