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## **The Mastocytosis Patient Experience**

We are sharing information about the rare disorder mastocytosis and related mast cell disorders, which we have derived from a number of sources in Canada and internationally. Our intention is to educate by sharing mastocytosis patient experiences. These experiences are often beyond what has been researched and published in medical journals. This is intended for general information purposes only and of course does not constitute medical advice. PLEASE SEE YOUR PHYSICIAN for medical advice and treatment.

It is unknown what causes or triggers mastocytosis. However, prolonged stress, emotional or physical trauma, surgery of any kind, and even childbirth, have all been reported as contributing factors to the development or escalation of mastocytosis. The basis of mastocytosis is a malfunctioning yet important immune system cell called a mast cell. Anything which triggers the immune system, including surgery, will trigger mast cell degranulation. It is the chemicals released by mast cell degranulation which cause the severe and prolonged symptoms of mastocytosis. Mast cells are created in the bone marrow, and are found in every part and system of the human body, including the brain. Everyone has mast cells. In healthy people, mast cells function correctly. In mastocytosis patients, mast cells are out of control and/or behaving abnormally. Mastocytosis is classified as a neoplastic disorder (like cancer) and recently, also as a stem cell disorder. Patients experience mastocytosis as very similar to having an auto-immune disorder, because they suffer many of the same symptoms and limitations as other auto-immune disorder patients.

Mastocytosis Society Canada strives to educate our members and their physicians fully about mastocytosis so that they are better able to find helpful treatments to lessen suffering and hopefully slow down progression of the illness. There is presently no cure for mastocytosis.

Most physicians do not recognize or understand mastocytosis as it is highly unlikely that they were either trained in the subject or have personally encountered a patient with this rare disorder. Most of the physicians and specialists patients consult fail to recognize or diagnose mastocytosis, simply because they had never seen it before, nor had they heard of it. Additionally, the existing diagnostic tests have proven to be unreliable and inconsistent, evidenced by disparate results amongst the international mastocytosis patient base.

Internationally, there appears to be a consensus amongst mastocytosis patient groups, mast cell researchers, and physicians experienced in treating mastocytosis as to the following:-

1. Existing mastocytosis diagnostic tests need to be improved or completely new tests need to be

developed. We estimate that approximately half the mastocytosis patient base worldwide does not meet the current diagnostic parameters, however they do experience benefits from being treated for mastocytosis. They are being diagnosed based on their symptoms in an effort to help them find some relief. Many are being diagnosed clinically based solely on their symptoms. This approach is working, resulting in physicians acknowledging that medicine just does not know nearly enough about mastocytosis yet.

2. Patient experiences of symptoms and secondary conditions (arthritis, osteoporosis, pancreatic tumours, chronic fatigue, ehlers danlos, connective tissue diseases, POTS, fibromyalgia, raynauds, iritis/uveitis, vertigo, interstitial cystitis, autonomic nervous dysfunction, thyroid malfunction or cancer, cerebral aneurysms), are often beyond what has been medically outlined as within the mastocytosis definition. However, many mastocytosis patients suffer at least one of these complications. Some have/had more than one.
3. Medical research has barely scratched the surface in fully defining, diagnosing and treating mastocytosis. As a result, the stages or classifications of mastocytosis, and the resulting effects or symptoms of each stage, are misleading or incomplete. Medicine does not know this disorder or mast cells nearly well enough to provide answers to all that patients endure and suffer. Physicians experienced in treating mastocytosis patients are aware of this. Many mastocytosis patients experience some or all of the symptoms in the same severity. (*Reference: The Mastocytosis Research Institute in France <http://www.ncbi.nlm.nih.gov/pmc/articles/PMC2386235/>*). For example, Indolent Systemic Mastocytosis presents the same symptoms as Aggressive Systemic Mastocytosis, both in type and severity. Thus, many patients with Indolent Systemic Mastocytosis suffer continuously and live extremely restricted, largely isolated, quiet lives in order to best control their immediate environments. This is not a benign or minor diagnosis. Indolent Systemic Mastocytosis is in many cases life threatening and patients diagnosed with that stage experience continual escalation of current symptoms, and development of new symptoms they never used to have.
4. Current treatments do not remove or prevent all the symptoms. Patients with mastocytosis and related mast cell disorders suffer a wide range of life threatening and/or disabling and permanently damaging symptoms. This prevents many patients from living a normal life and basically existing in a state of catering to their disease to try to lessen their suffering. The most basic daily life activities induce suffering and internal damage over time for mastocytosis patients. For example, symptoms are triggered by simple activities such as daily bathing, inhaling airborne scents/chemical toxins, walking or any other exercise, eating, cognitive processing (concentration, learning and memory impairments), being in extreme temperatures (hot or cold in weather or water), talking for extended periods of time, engaging in social activities, stress or fatigue, and even for some, getting dressed every day. Friction and sensitivity to clothing, pressure, and coming in contact with man made materials, are enough to trigger symptoms for some patients.

Mastocytosis negatively impacts working and personal lives. Many mastocytosis patients have a restricted diet of very few foods they can safely eat, constant fatigue, episodes of vertigo and anaphylaxis, as well as inability to exercise on a regular basis (includes walking) due to fatigue and weakness and all the other symptoms. On a daily basis, many patients experience difficulty with tasks requiring concentration, learning, retention or anything relying on memory or information processing skills. Many people suffering with mastocytosis cannot commit to anything in terms of a schedule or regular routine because their symptoms vary in type and severity from day to day and often hour to hour. They are continuously symptomatic with disabling effects such as fatigue, sudden unexplained

episodes of drops in blood pressure (very common with mastocytosis), flushing, shaking and tremors, and poor concentration.

Most mastocytosis patients find that medications do not eradicate or prevent their symptoms. This disorder requires avoidance or minimization of exposure to triggers in addition to taking medications. Triggers can be anything which causes mast cells to degranulate. This includes hormonal activity, emotions (positive or negative), activities, positive/negative stress, foods and drinks, temperature, and exercise/exertion of any kind. It is only by controlling their exposure to triggers in addition to taking medications, that they can achieve some relief from the symptoms, but it is not total relief and this does not return them to functioning normally on a consistent and reliable basis. Their good days are very few and far between, rarely even lasting a full day. Most mastocytosis patients share this same experience.

Typically, mastocytosis patients experience increasing limitations and greater suffering over their lifetime. If they push against their symptoms, believing that "trying harder" or pushing to complete tasks (laundry, grocery shopping, visiting a friend, cleaning), the symptoms become more severe and prolonged and continue to ricochet setting off other symptoms for days, weeks or months on end. There is no predictability to mastocytosis symptoms. There is no way to make the symptoms behave consistently or to be able to garner better control over them. The medical research community has not yet fully figured out mastocytosis. In the meantime, patients are driving the medical research via their physicians reporting the escalating symptoms and secondary and tertiary complications suffered by their patients. Everyone wants to have the answers, patients most of all.

Mastocytosis patients often look normal or healthy to others, especially when they are enjoying one of their better days/moments when their symptoms are subdued or well controlled, as during these times they may be able to be more active. However, these stable periods do not endure and it is the fluctuating nature of the illness that causes much difficulty for the person with mastocytosis. Many mastocytosis patients are not seen regularly by people outside their immediate family or caregiver except for those rare occasions when they are feeling well. As a result, people fail to understand what mastocytosis is and the extent to which it prevents individuals from living a normal and active life.

Mastocytosis is irreversible and in most cases, the symptoms are extraordinarily difficult to manage.

By Mastocytosis Society Canada