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Mastocytosis

ABSTRACT

Mastocytosis is a rare and underdiagnosed disorder characterized by mast cell proliferation in the tissues and organs of the body. The gastrointestinal manifestations of the disease can be noted in approximately 70%–80% of those patients diagnosed with the disorder. Symptomatic manifestations of systemic mastocytosis can either be triggered spontaneously or be precipitated by a variety of situations, stimuli, and exposures. Common gastrointestinal complaints include abdominal pain, diarrhea, nausea, vomiting, and gastrointestinal reflux disease. Substantial numbers of mast cells have been noted in patients who have been diagnosed with gastritis, ulcerative colitis, and Crohn disease. Irreversible, with symptoms that run the gamut from the merely annoying to the severely life-threatening, mastocytosis is a disease that prevents an individual from leading a normal life. As the prevalence of gastrointestinal symptomatology in those patients who have been diagnosed with mastocytosis is so significant, it is an important and relevant disease of which gastroenterology nurses should be cognizant.

It is a day like any other, but not for me. I dress, but slowly, so as not to overexert my body. I am careful with my makeup and perfumes, because that could be the trigger that sends me into an anaphylactic attack. I drive to work instead of walking with my friends, because I am afraid to overtax my body with the short walk downtown from my home. Adjustments and sacrifices have become a way of life for me, and most of the time I don't even notice that I am making them anymore as they have become such an engrained portion of my daily lifestyle.

It is hot in the office and I hope the air conditioning can cool the room down quickly, before my body decides to object to an extreme in temperature it will not care for or tolerate. I leave work excitedly as I look forward to attending my sister's birthday party this evening. I arrive at the restaurant and take in the tinkling of silverware, the sparkle of lights, the laughing voices of happy people, the festive atmosphere, and am so happy to be here and a part of this special celebration tonight. I forget myself and become complacent while happily greeting family and friends, inhale too deeply as I think how good it is to be alive, and I am done. I have given my body the trigger it has been seeking and I have been trying to avoid all day. What is that trigger? I don't know and have no time to care. I start popping out in angry red welts and my airway begins to close. It is a day like any other, but not for me. I have mastocytosis, and must be forever vigilant and on guard against the demons of my own body.

A disease that is both chronic and rare can be very difficult for both the patient and their families to absorb into their psyches, more or less into their lives. Imagine a disease that all but the rarest of physicians will tell you that they do not feel comfortable treating. Imagine a disease that the majority of physicians will dismiss as anxiety, hypochondria, or both. Imagine a disease that is suddenly triggered with terrifying speed, usually at the most awkward of times and in the most embarrassing of social situations. Imagine a disease that has turned your body's innate protector, your immune system, from a friend into, at best, an adversary, and at worst, an enemy. This disease is mastocytosis.

Overview

Mastocytosis is a rare disorder characterized by mast cell proliferation and accumulation in one or more

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tissues and organs of the body, inclusive of the skin, bone marrow, liver, spleen, gastrointestinal tract, and lymph nodes. Recognized as a myeloproliferative disorder by the World Health Organization in 2008, the true incidence of mastocytosis is unknown, but current statistics mark it as a rare disease, with a prevalence of under 200,000 cases per year in the United States (Pardanani, 2013). This fact officially classifies it as an "orphan" disease, which equivocates to an estimated prevalence of approximately 1 case within a population of 10,000 people (Brockow, 2014).

A disease that can occur either in childhood or in adulthood, it manifests as one of two distinct subclass presentations—CM (cutaneous mastocytosis) or SM (systemic mastocytosis) (Valent, 2013). Although the etiology of mastocytosis remains unclear, it is considered to be predominately associated with the genetic mutation c-KIT D816V, which is thought to be the cause of the abnormal proliferation of mast cells in the tissues of the body (Tremblay, Carreau, Kremyanskaya, & Mascarenhas, 2015). Symptoms vary, as they are dependent upon disease subtype, and are related to either mast cell activation and/or organ infiltration by the mast cells.

Mast cells, produced in the bone marrow, are the regulators of the immune system. A type of white blood cell, a mast cell is a type of granulocyte derived from the myeloid stem cells. Widely distributed in nearly every organ of the body (primarily in close proximity to the blood, lymphatic vessels, nerve endings, skin, and mucous membrane surfaces), they are particularly abundant in the skin, respiratory system, gastrointestinal, and genitourinary tracts. They develop from immature cells produced in the bone marrow and then migrate into the tissues, at which point they proceed to mature.

Mast cells produce chemicals that normally serve protective, inflammatory, and regulatory functions within the body. They can be activated by immune mechanisms such as immunoglobulin G (IgG) receptors, nerve growth receptors such as TrKA, and complement receptors such as C3aR and C5aR. They can also be activated by non-immune-precipitating mechanisms that include environmental and pharmaceutical triggers. Responsive to both allergic and inflammatory intrusions, they are intimately involved in both wound healing and the body's defense against allergens and pathogens, and thus play an important role in the defense of the body from disease (Amin, 2012). Their activity is induced by the release of multiple mediators, some of which are preformed and stored in granules, whereas others are synthesized and released in response to the inducing stimulus (Hodges et al., 2012). In the case of mastocytosis, these chemicals, or mediators, which commonly contain histamine, heparin, tryptase, and chymase, overproduce, thereby causing symptomatic reactions as they are secreted, including itching, sneezing congestion, swelling, and wheezing (Tete et al., 2012).

Symptoms of mastocytosis vary from individual to individual, and can either occur as "attacks" or manifest simply as fatigue or a feeling of ill-health. The rate of progression is individualized as well, both from person-to-person and episode-to-episode. Composed of a series of seemingly unrelated symptoms, each individual will present in a slightly different manner, with disparate triggers and divergent mechanisms of presentation.

Classifications and Stages

Cutaneous mastocytosis (CM) is a benign skin disorder noted predominantly in the pediatric population. Often resolving by the age of puberty, it affects little to none of the internal organs of the afflicted individual (Carter, Metcalfe, & Komarow, 2014).

Systemic mastocytosis (SM), on the other hand, is a chronic disease that is both persistent and aggressive, progressing into more advanced presentations of the disease state in a small percentage of cases. As the disease process develops, symptoms relative to one of its more distinctly specific subcategory presentations will begin to manifest. As these symptoms become more specific and predominant, the disease can be more definitively diagnosed and categorized, based upon the unique and distinct symptomatology of its presentation. Symptoms can run the gamut from skin reactions, such as itching and flushing, to gastrointestinal symptoms of vomiting and diarrhea, to muscle and joint pains, to alterations in mood (Horny, 2012). It can present neurologically with back pain, headaches, and syncopal episodes, or systemically just as extreme fatigue and weariness (Smith, Butterfield, Pardanani, Deluca, & Cutrer, 2011).

Clinical Progression

Mastocytosis can be divided into seven specific subcategories as per World Health Organization guidelines. These are cutaneous mastocytosis, indolent systemic mastocytosis (systemic mast cell disease), systemic mastocytosis with associated clonal hematologic non-mast cell lineage disease, aggressive systemic mastocytosis, mast cell leukemia, mast cell sarcoma, and extracutaneous mastocytoma (Akin & Valent, 2014).

Cutaneous mastocytosis presents most commonly in areas of the body where the skin has the most exposure to either pressure or rubbing. It manifests as small, brown spots that can be either flat or elevated, and which become surrounded by red, itching skin when scratched. This is known as Darier's sign (Theoharides, Valent, & Akin, 2015).

Indolent or slow-growing systemic mastocytosis is the most predominant form of this disease noted in the adult patient. Generally associated with a low mast cell burden, it commonly affects the gastrointestinal tract, and can present with an enlarged liver or spleen. In general, indolent forms of the disease, although they can progress in the severity of their symptom manifestations, will not progress to the more severe aggressive forms of the disease. Although mediator-related symptoms are quite common, the grade of bone marrow infiltration is low, usually remaining at less than 5% (Gulen, Hagglund, Dahlen, & Nilsson, 2016). Treatment is directed toward controlling the symptoms associated with mast cell activations (Pardanani, 2012).

Mastocytosis with associated clonal hematologic non-mast cell lineage disease accounts for approximately one-third of all cases of systemic mastocytosis. Myeloproliferative and myelodysplastic disorders are commonly associated with this manifestation of the disease process. Successful treatment of this hematologic presentation of the disease improves the disease prognosis. However, the disease should be treated as two separate entities, with the hematologic disorder treated as a disparate disease entity from the systemic mastocytosis. As the number of mast cells in the tissues or organs of the body increases, the disease process progresses accordingly (Pardanani, 2013). Aggressive systemic mastocytosis presents with either the impairment or the loss of an organ function secondary to mast cell infiltration, most commonly targeting the liver, gut, bone, or bone marrow (Gulen et al., 2016). Mast cell leukemia is rare and associated with a fatal disease progression. An aggressive hematologic malignancy, its pathology is characterized by a level of circulating mast cells in the blood greater than 15% in tryptase-stained sections of the bone marrow. Mast cell sarcoma is a rare solitary tumor composed of abnormal mast cells, whereas extracutaneous mastocytosis is very rare and does not fit into any of the previously discussed criteria for either cutaneous mastocytosis or systemic mastocytosis (Pardanani, 2013).

Predominately targeting both men and women of the adult population equally, systemic mastocytosis runs the gamut from mild to life-threatening (Sokol et al., 2010). Seen more predominantly in the Caucasian population, the diagnosis of mastocytosis is based upon World Health Organization criteria, which are subdivided into major and minor categories. In order to be given the label of mastocytosis, a disease process must manifest with a minimum of one major and one minor criteria, or three minor criteria (Sanchez-Munoz et al., 2011). A major criterion would be the presence of more than 15 mast cells per high-power field in an extracutaneous organ. Examples of minor criteria are the presence of the C-KIT gene mutation, a serum tryptase level greater than 20 ng/mL, and spindle cells that comprise 25% or more of mast cell infiltrates (Tremblay et al., 2015).

Risk Factors

Mast cell degranulation can either be triggered spontaneously or be caused by a variety of situations, stimuli, and exposures. Triggers are multiple and varied, but thought to be related to an exposure to either a physical or emotional trigger that would then result in the degranulation of mast cells (Theoharides et al., 2015).

The most common, but not the only trigger, is food. Food, commonly an enjoyable and satisfying portion of a busy day, becomes a battleground, as it looms as a potential smoking gun for mast cell degranulation. It must be carefully selected and meticulously prepared, as the wrong spice hidden deep within a tasty dish can trigger a degranulation of mast cells. Certain foods present more of a threat than others, and include cheeses, various spices, and shellfish, as well as food preservatives and colorings (Vlieg-Boerstra, van der Heide, Oude, Kluin-Nelemans, & Dubois, 2005).

Other triggers include insect venom, drugs, and alcohol as a source of potential anaphylaxis (Bonadonna, Lombardo, & Zanotti, 2014). Emotional stress can initiate a "flare," as can environmental toxins such as perfumes and pesticides; bacterial, fungal, or viral infections; and physical stimuli or stressors such as heat, cold, sunlight, exercise, and fatigue (Escribano & Orfao, 2011).

In addition, mast cells can be coaxed to degranulate by physical exertion, as well as by sensitivity to certain categories of medication such as aspirin, antibiotics, and nonsteroidal anti-inflammatories. Mast cell mediators such as histamine, heparin, chemokines, cytokines, leukotrienes, and prostaglandin D2, can induce flushing, anaphylaxis, and generalized pruritus (Theoharides et al., 2015). In the longer term, the disease can predispose to osteoporosis, respiratory events, digestive disorders, depression, sleeping disorders, and interstitial cystitis. The majority of the symptoms, although not life-threatening, can significantly impair quality of life (Gulen et al., 2016).

Anesthesia presents a challenge as well, and the treatment algorithm for patients diagnosed with the disease is both intricate and lengthy (Pardanani, 2013). Beta-blockers are contraindicated for surgical patients as they have the potential of precipitating an anaphylactic episode (Dewachter, Castells, Hepner, & Mouton-Faivre, 2014).

Symptomatic manifestations of systemic mastocytosis run the gamut from mild to life-threatening. They can include anemia and coagulopathy, facial flushing, itching, abdominal pain, diarrhea, signs and symptoms

of gastrointestinal reflux disease, nausea, vomiting, gastric and duodenal ulcers, pruritus, flushing, headaches, lightheadedness, heart palpitations, bone pain, cognitive dysfunctions such as an inability to concentrate, and anaphylaxis. Multiple symptoms can occur at once. Life expectancy is decreased only in the more aggressive forms of mastocytosis, in which bone marrow infiltration of the mast cells is greater than 90%, or when the proliferation of mast cells in an organ results in the impairment of its function (Gulen et al., 2016; Siebenhaar, Akin, Bindslev-Jensen, Maurer, & Broesby-Olsen, 2014).

Recent studies have demonstrated that gastrointestinal manifestations of systemic mastocytosis are the second most common manifestation of the disease after pruritus, and can be noted in approximately 70%–80% of patients diagnosed with the disorder (Behdad & Owens, 2013). Common gastrointestinal symptoms are thought to be secondary to mast cell mediators in the gastrointestinal tract. They include abdominal pain, which occurs in approximately 50% of diagnosed patients; diarrhea, which occurs in approximately 40% of diagnosed patients; and nausea and vomiting. Systemic mastocytosis can involve any of the gastrointestinal organs because of the increased histamine production that occurs with the disease, thereby causing a variety of site-specific symptoms. These manifestations of the disease include esophagitis, gastroesophageal reflux, gastric ulcer disease, peptic ulcer disease, steatorrhea, and intestinal malabsorption (Behdad & Owens, 2013).

Despite the frequency of gastrointestinal symptoms, the disease remains underdiagnosed. Systemic mastocytosis can manifest anywhere along the gastrointestinal tract. As no treatment is curative, therapy is directed toward symptom management of anaphylaxis and its related symptoms of pruritus, flushing, and intestinal malabsorption.

The symptoms of systemic mastocytosis vary, and are dependent upon the specific site of the excess number of mast cells. Management primarily involves the avoidance of major triggers, such as alcohol and nonsteroidal anti-inflammatory medications. Other triggers can include aspirin, codeine, morphine, thiamine, quinine, opiates, gallamine, decamethonium, procaine, radiographic dyes, dextran, polymyxin B, scopolamine, and D-tubocurarine (Brokow, Jofer, Behrendt, & Ring, 2008). In addition, management also involves control of the symptoms related to mast cell mediator release, as well as the treatment of related comorbidities, as they arise (Escribano & Orfao, 2011). Therapies that prevent mast cell infiltration, such as fludarabine (Fludara and Oforta) and interferon-alfa, have only been used in the treatment of aggressive systemic mastocytosis. Also, only used for the treatment of the more aggressive form of the disease are newer treatment modalities that include tyrosine kinase inhibitors that target the c-KIT mutation (Siebenhaar et al., 2014). The only potentially curative treatment for aggressive systemic mastocytosis at this time is hematopoietic stem cell transplantation. Although this mode of treatment appears to be promising, it has not been studied sufficiently to date to be conclusively determined to be a viably curative treatment option at this time (Ustun et al., 2014).

Diagnosis

Physical examination might include signs of anemia. These would include pallor, hepatomegaly, splenomegaly, lymphadenopathy, and urticaria. Anemia, thrombocytopenia, leukocytosis, and, in some patients, eosinophilia, basophilia, thrombocytosis, and monocytosis might be present. A combination of anemia, thrombocytopenia, hypoalbuminemia, and excess bone marrow blasts greater than 5% signifies a poor treatment prognosis (Patnaik, Rindos, Kouides, Tefferi, & Pardanani, 2007). The blood level of tryptase might be elevated in the case of systemic mastocytosis (Gulen et al., 2016).

Diagnostic procedures include a bone marrow biopsy and aspiration, barium studies, and endoscopy for patients who are manifesting with gastrointestinal symptomatology; a liver biopsy in the setting of hepatomegaly; and a skin biopsy when the disease manifests cutaneously. The most common histological marker is the clustering of mast cells in the visceral organs. Although other tests can suggest the presence of mastocytosis, only a positive biopsy result (either of the skin or bone marrow) demonstrating an accumulation of mast cells can be determined to be a definitive diagnostic tool (Gulen et al., 2016).

Treatment

Treatment of mastocytosis is dependent upon the disease classification and symptom presentation, the extent of the disease, and the person's overall health. Although there is no cure for the disease itself, several treatments in isolation or combination can be implemented to relieve the symptoms of a flare and as an attempt to prevent future occurrences. The basic and most primary treatment intervention, however, is the avoidance of any of the triggers that might cause a release of mast cell histamine.

Indolent mastocytosis may require no treatment at all. However, when intervention is indicated, the pharmacological choices are varied and directed toward specific problematic areas of the flare episode. Nonetheless, a majority of patients require at a minimum H1 and H2-blockers to reduce skin sensitivity and flushing, minimize reflux, vomiting, and diarrhea

and provide an initial intervention to the symptoms of anaphylaxis. Included in these groupings of medication are the H1-blockers, which include diphenhydramine (Benadryl), cetirizine (Zyrtec), loratadine (Claritin), fexofenadine (Allegra), and hydroxyzine (Atarax), as well as the H2-blockers, which include ranitidine (Zantac), famotidine (Pepcid), nizatidine (Axid), and cimetidine (Tagamet) (Pardanani, 2013). Nonsteroidal anti-inflammatory medications must be used with caution, as they have the potential to trigger the release of histamine and can thereby cause a severe allergic reaction (Carter et al., 2014)

If the first line of pharmaceutical interventions proves insufficient for symptom control, additional medication groups and categories can be introduced. The next stage of pharmacological intervention would include cromolyn sodium (Gastrocrom), a mast-cell-mediating medication that can decrease symptoms of pruritus, welts, and flushing, as well as the systemic symptoms of diarrhea, abdominal pain, bone pain, and disorders of cognitive function (Escribano & Orfao, 2011). Another mast-cell-stabilizing medication that could be used is ketotifen fumarate (Zaditen), an allergy medication. Medications also useful for the treatment of symptomatology as it appears are leukotriene inhibitors such as zafirlukast (Accolate), montelukast sodium (Singulair), and zileuton (Zyflo). Steroids are used for the control of malabsorption symptoms, ascites, and bone pain and for the prevention of anaphylaxis. Epinephrine is the medication of choice for the treatment of acute anaphylaxis. Although it does not block mast cells, it does neutralize some of the systemic reactions to its degranulation. Immunotherapy can be useful against the venom introduced by insect bites and stings. A medical alert bracelet should be in evidence at all times and emergency intervention protocols should be within easy reach (Carter et al., 2014).

Recent years have seen major advances in the understanding and recognition of this disease. More invasive treatment therapies for the treatment of the particularly aggressive and more recalcitrant cases include exposure to ultraviolet light for the relief of recalcitrant skin rashes, and laser therapy to treat skin manifestations of the disease. New treatment modalities also include interferon-alpha administration for the treatment of histologically confirmed bone marrow involvement, when prior therapy using less invasive measures has proven to be ineffective. Surgical excision of mastocytomas, electron beam radiation, and ultimately chemotherapy and stem cell transplants are also considered viable modalities of treatment (Gulen et al., 2016). In 2006, the Food and Drug Administration granted approval to treat aggressive mastocytosis that was not a result of the gene mutation D816V c-KIT with imatinib mesylate (Gleevec), a chemotherapeutic agent, the only TK1 medication that has received such a designation thus far (Siebenhaar et al., 2014). The ultimate goal of all of these interventions is to neutralize and eliminate the harmful mast cells (Tremblay et al., 2015).

As a result of the dearth of essentially effectual treatment options, clinical trials are commonly the treatment of choice for the more aggressive forms of the disease. Many are based upon evolving treatment modalities, relying upon volunteers to evaluate the efficacy of theoretical symptom management and the safety of treatment regimens. Investigational agents currently under evaluation include medications that target the KIT gene mutation, such as omalizumab (Xolair), masitinib (Masivet), ibrutinib (Imbruvica), SL-401, tamoxifen (Nolvadex and Soltamox), and cladribine (Leustatin), which suppress the activity of the immune system (Sokol, Ghazi, Kelly, et al., 2014; Tremblay et al., 2015).

When mastocytosis is limited to the skin, symptoms can improve or clear entirely. Alternatively, it is also possible it will progress into systemic mastocytosis. It is impossible to predict the course of the disease in any one individual patient. Systemic mastocytosis can progress slowly over a period of many years or can suddenly increase in its intensity. For the small percentage of patients who suffer disease progression, the course varies, and the prognosis becomes more guarded.

The Heart of the Matter

Living with mastocytosis, as with any chronic disease, can be challenging and is most definitely life altering. It requires lifelong vigilance and offers no guarantees. An elusive disease to diagnosis, it is both a challenging and a frightening augmentation to the lives of those who experience it. The frustration begins with the diagnosis. Patients who suffer from a myriad of seemingly unrelated symptoms trudge from physician to physician hoping for some relief, an answer. The full picture is obscured beneath the pigeonhole of each physician's specialty area, and a complete and clear picture of an ailment composed of all its component parts is not a common occurrence. Most physicians do not recognize or understand mastocytosis. Many will fail to diagnose it when they do come across it, and the majority of those will not want to treat it even if they have. Medical breakthroughs into the diagnosis and treatment of the disease remain insufficient. Treatment is geared entirely toward symptom management rather than toward a disease-based therapy. It is possible that, for many of these patients, an accurate diagnosis will never be found, and they will attempt to live their lives treating isolated symptoms in a vacuum of misery, deprived not only of their quality of life but limiting performance of even the most basic of the activities of daily living as well.

In a study performed in 2011 evaluating the quality and impact of care on patients who were eventually diagnosed with mastocytosis, it was discovered that it took approximately 2 years for the majority of the subjects included in the study to receive a diagnosis. Furthermore, greater than half of those subjects had to consult with either three or more physicians before they received a definitive disease determination (Nowak, Gibbs, & Amon, 2011).

When the disease is finally diagnosed, the hard work has only just begun. Triggers of anaphylactic episodes are varied, but can then at least be analyzed, isolated, and hopefully avoided. However, increasing the stakes, a food or activity that might not have been a trigger on one day, may become a trigger on the next. An environment that might encompass multiple triggers may pose a higher risk of mast cell degranulation than exposure to a solitary trigger in isolation. Lives quickly become self-limited to the known and familiar, and even then there are no guarantees. Over time each individual must learn how to alter the manner in which they live their lives in order to slavishly conform to the demands of their unforgiving master—their disease.

Coping With Mastocytosis

Mastocytosis can present itself in a variety of ways and manifest with a myriad of symptoms, ranging from the inconvenient to the life-threatening. Typically, patients with the disease experience increasing limitations and greater suffering over their lifetimes. If they push against their symptoms, as our society encourages us all to do, the symptoms perversely just become more severe and a prolonged disease flare ensues. As symptoms follow no predictable pattern or path of progression, there is no way to predict when or how mast cells will be triggered to degranulate. In addition, current treatments do not remove or prevent all of the symptoms either of the mastocytosis itself or of the manifold secondary conditions that the disease predisposes patients with mastocytosis to contract.

Mastocytosis negatively impacts both the professional and personal lives of those with the disease. In the search for the avoidance of triggers, many people follow an extremely restrictive diet that consists of a small and limited number of foods that they have found they can safely consume. As physical activity can trigger a mast cell release, exercise of any kind on a regular basis, inclusive of walking, must be performed slowly and in small blocks of time. Many people with mastocytosis suffer from debilitating fatigue and weakness. Lack of concentration is common in a disease whose symptoms can vary from hour-to-hour and day-to-day. To live with mastocytosis is to live with the realization that there will be another event; just the time, the place, and the trigger remains the mystery.

Because medications alone cannot control the symptoms of mastocytosis, individuals with the disease must live with limitations to their lifestyles that are directed toward the prevention of symptoms and the avoidance of their known triggers. The problem is that a trigger can be anything that causes a mast cell to degranulate, and because this can include hormonal cycles; emotions, either positive or negative; strenuous activity; positive or negative stressors (either physical, emotional or environmental); a multitude of foods and beverages; alterations in temperature; and a multitude of medications; an individual who can function normally in our modern society is a biological impossibility.

Because it is largely unknown as to what either causes or triggers mastocytosis, and this can be anything at all that triggers the immune system in any particular individual on any particular day in any particular situation, it is important for each individual to evaluate their own triggers as soon as possible, thereby allowing them at least a modicum of control over their own destinies. However, as mastocytosis has no rhyme or reason to its presentation, there is no way to predict when or where symptoms will appear; just that, inevitably, they will appear.

Making the disease even more insidious, patients who have mastocytosis appear normal and healthy to others on their good days, when their disease is subdued and in a lull. Thus, a public manifestation of disease symptoms can be surprising and unnerving to startled friends and coworkers, and embarrassing to the patient in anaphylaxis, compounding the medical emergency. This causes many people who have the disease, whether diagnosed or not, to limit their social interactions to a small, safe group of family and friends, and keep their social interactions close to home where the added stressors of possible community judgment and discrimination will no longer be an issue (Jennings et al., 2014).

Irreversible, with symptoms that are at best difficult to manage and at worst life-threatening, mastocytosis is a disease that prevents an individual from leading an active and normal life. It is important for sufferers from this disease to maintain a strong support network, especially as it relates to both their triggers and the interventions required in the event of a flare episode. Communication is key, with both the patient's personal and professional support team.

People with mastocytosis suffer from a vast array of physically debilitating and emotionally draining symptoms every day. It prevents many of them from leading a normal life. It requires special diets, predetermined routines, and alterations in both their professional and personal lives that cater to the unique challenges it presents. The survivors are those who do not let their disease define them, but gain strength from the journey

and incorporate those lessons into who they are and what they want to become.

Mastocytosis and the Gastroenterology Nurse

By this point you might be asking yourself, what should this mean to me? Is this relevant to my practice as a gastroenterology nurse? The answer is a resounding yes. And here are the reasons why. Mast cells play an important role in the regulation of the visceral sensitivity of the gastrointestinal tract, linking them to such functional disorders as noncardiac chest pain, nonulcer abdominal pain, and irritable bowel syndrome (Ramsay, Stephen, & Doman, 2010). Increased numbers of mast cells have been found in the cecum, terminal ileum, and jejunum of patients with irritable bowel syndrome (Reggiani et al., 2015). Substantial numbers of mast cells have been noted in patients who have been diagnosed with gastritis, ulcerative colitis, and Crohn disease (Reggiani et al., 2015). And, although mast cells are prevalent in all of the body's tissues, they have a particular affinity for the gastrointestinal tract, a factor that has not been fully appreciated until recently (Chrysakopoulos et al., 2014). Gastrointestinal symptoms can present as peptic ulcer disease, steatorrhea, and malabsorption.

Although the precise role mast cells play in the gastrointestinal tract in their various manifestations remains significantly obscure, it nonetheless, with its large surface area, offers a sizable canvas upon which mast cells can exert a great deal of influence, thus affecting the secretion, absorption, and motility of the gastrointestinal tract. Moreover, proliferation of mast cells in response to perceived triggers such as gluten and celiac sprue can alter the function of the gastrointestinal tract enough to cause a symptomatic reaction, as it is one of the primary sites that allergic reactions to ingested food products occur (Kumar, Verma, Das, & Dwivedi, 2012).

Common gastrointestinal symptoms manifested in the vast majority of those patients who have been diagnosed with the disease are abdominal pain, diarrhea, nausea, and vomiting. In addition, there is evidence for the involvement of mast cells in the inflammation of the bowel wall, potentially causing motility disorders, gastroparesis, and postoperative ileus (Sokol et al., 2010).

Conclusion

The prevalence of gastrointestinal symptomatology noted in those patients who are diagnosed with mastocytosis is significant. The plight of those who remain undiagnosed is sorrowful. Let us never forget that as nurses, we are advocates for our patients. The next time you see a patient whose symptoms you cannot quite explain, think of mastocytosis. You might just save a life. ✪

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RESOURCES

The Mastocytosis Society, Inc
PO Box 129, Hastings, NE 68902
http://www.tmsforacure.org

Genetic and Rare Diseases (GARD) Information Center
PO Box 8126, Gaithersburg, MD 20898
http://rarediseases.info.nih.gov/GARD

Mastokids
PO Box 2706, Bluffton, SC 29910
http://www.mastokids.org

Cancer.Net (American Society of Clinical Oncology)
2318 Mill Road, Ste 800, Alexandria, VA 22314
http://www.cancer.net

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