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The title page

Everyday Life and Mastocytosis from a patient perspective – a qualitative study

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Abstract

Aim: To investigate and gain knowledge about patients' perspectives on everyday life with mastocytosis and how they experience, understand and manage symptoms and challenges.

Background: Indolent systemic mastocytosis (ISM) is a disease characterized by the accumulation and activation of mast cells. Symptoms are diverse and range from mild to severely debilitating or even fatal. It is considered rare but is underdiagnosed due to lack of awareness. Quantitative studies have shown that ISM can negatively impact quality of life. No qualitative studies have described everyday life with the disease.

Design: A qualitative interview study taking a phenomenological approach.

Methods: Seven qualitative, semi-structured interviews with adult patients with ISM. The analysis was inspired by Giorgi's phenomenological method. COREQ reporting guidelines were utilized.

Results: Three themes and five subthemes emerged from the analysis. 1. The everyday life with a rare disease, unknown to most people. Being perceived as a hypochondriac in the encounter with the health system. The diagnosis makes a difference. Expert knowledge is important. 2. Living with and handling the invisible and visible symptoms. The visible body. 3. Fearing an attack. Feeling safe and vulnerable at the same time.

Conclusion: Patients with ISM are severely affected in their everyday lives, especially in terms of their relationship with family and social network. Symptoms restrict and complicate activities and participation in social contexts and the fear of an anaphylactic attack is always present. The disease affects patients' self-perception and sexuality. The rarity of the disease and general low awareness seems to be of great importance in the encounter with the health care system, both before and after diagnosis, and there is a need for expert knowledge, support and care.

Relevance for clinical practice: The focus of counselling should not only be on the disease itself, but also on living life with the disease.

Keywords:

Nursing, systemic mastocytosis, chronic rare disease, counseling, patient experience, disease management, everyday life, patient perspectives, qualitative research, health personnel.

What does this paper contribute to the wider global clinical community?

- This study provides an insight into the influence of Indolent Systemic Mastocytosis in the everyday life of adult patients.
- When patients with Indolent Systemic Mastocytosis talk about their disease they do not only refer to a variety of custom symptoms, but to a complex of experiences, perceptions and emotions, arising in different situations and contexts in everyday life
- The health care system must be flipped away from the traditional ways of organizing and structuring consultations, and focus more on the patient's agenda, needs and experiences and not only the disease and treatment.

Introduction

Systemic mastocytosis (SM) is a chronic disease characterized by an accumulation, proliferation and activation of mast cells in several organs, typically skin, bone marrow, and gastrointestinal tract (Brockow & Metcalfe, 2010; Gulen, Hagglund, Dahlen, & Nilsson, 2015; Metcalfe, 2008). In a Danish registry study, the prevalence was estimated to be 10 out of 100,000, but the disease is likely underdiagnosed due to both lack of awareness among health professional, and the very heterogeneous symptomatology (Broesby-Olsen, Kristensen, et al., 2016; Cohen et al., 2014). Both children and adults can be diagnosed with mastocytosis. Mastocytosis in children usually resolves before puberty, whereas the disease is lifelong in adults. SM is divided into seven categories with differing severity and prognosis, of which indolent SM (ISM) is the most prevalent subcategory (Broesby-Olsen, Dybedal, et al., 2016; Valent et al., 2001). It carries a favorable prognosis with a life-expectancy that does not differ from the background population, as opposed to aggressive forms of SM. While the pathogenesis to some extent has been elucidated, and involves a somatic gain-of-function KIT mutation, leading to clonal expansion of mast cells, the basic cause of mastocytosis is unknown and, at present there is no known cure. (Broesby-Olsen, 2016).

A substantial proportion of patients with SM have experienced an anaphylactic reaction, which typically is elicited by insect stings (wasp/bee) or drugs, but which can also occur unprovoked or by a combination of factors, such as ingestion of specific food, alcohol, physical stimuli, etc. (Brockow, Jofer, Behrendt, & Ring, 2008; Gulen et al., 2015). At present, it is not possible to predict which patients are at risk of anaphylaxis, and therefore SM patients are advised to carry emergency medicine in the form of adrenaline auto-injectors, to be used in case of an acute life-threatening anaphylaxis (Brockow & Metcalfe, 2010). In addition to the very heterogeneous, chronic and fluctuating symptoms, the patients are thus also affected by the risk of anaphylaxis in their everyday lives.

The very diverse symptoms of ISM are caused by mast cell infiltration and the release of mast cell mediators, such as histamine; they range from mild to severely debilitating, and can be life-threatening. These include chronic fluctuating symptoms involving the skin and gastrointestinal tract, neuropsychiatric symptoms (depression, loss of concentration, moods, or fatigue), musculoskeletal symptoms (osteoporosis, bone pain, myoarthralgia) (Lieberman, 2014; Matito et al., 2014). Symptoms can vary from person to person, and acute episodes of potentially life-threatening anaphylaxis can occur unprovoked. Around three-quarters of ISM patients have skin involvement with pigmented skin lesions, traditionally termed urticaria pigmentosa, which may be severe and greatly affect the physical appearance of the patient.

A chronic illness leads to a new life situation for the patient and thus a new way of being in the world. One is forced to reassess one's life image in the environment of which one is a part and with the people with whom one engages (Paterson, 2003). ISM is a chronic disease with a variable and unpredictable course, and it affects patients in different ways and it affects everyday life (Jennings et al., 2014). Everyday life is where the individual's life is being lived; it is where repetitions and routines create the predictability and security that we as human beings need in the conduct of our lives (Dreier, 2008; Holzkamp, 1998).

In the existing literature, no qualitative studies and only a few quantitative studies have focused on patients with mastocytosis and their experiences of everyday life with the disease. In these quantitative studies, it has been demonstrated that quality of life in patients with mastocytosis is severely affected (Jennings et al., 2014; Nowak, Gibbs, & Amon, 2011; Sokol et al., 2010; van Anrooij, Kluin-Nelemans, Safy, Flokstra-de Blok, & Oude Elberink, 2016). A study from Jennings et al. (2014) using a web-based questionnaire in patients with mastocytosis, found that the majority of the patients felt that mastocytosis had an impact on them emotionally. The greatest negative impact was caused by the unpredictability of symptoms associated with the disease (Jennings et al., 2014).

Other studies (Hermine et al., 2008; Jennings et al., 2014; Nowak et al., 2011; van Anrooij et al., 2016) report patients' quality of life or symptoms in measurable, quantitative ways, but do not address living with the disease from an everyday perspective.

There is, overall, a lack of knowledge about patients' experiences of living with mastocytosis and how the disease influence everyday life.

The purpose of this study was therefore to investigate and to gain knowledge about patients' perspectives on everyday life with ISM, and how they experience, understand and manage the symptoms and challenges in everyday life.

Methods

We used a qualitative design with a phenomenological approach with focus on participants' opinions, understanding and experiences in relation to the phenomena that occur in actual situations in the life world, where people are living (Green & Thorogood, 2014; Richards, 2014). To generate data, individual qualitative semi-structured interviews were conducted (Holstein & Gubrium, 1995). COREQ guidelines (Tong, Sainsbury, & Craig, 2007) have been adhered to in the reporting of this study.

Participants

Participants were enrolled at [BLINDED] – a specialized, multidisciplinary centre, caring for more than 300 mastocytosis patients, spanning all disease categories. Patients were selected based on purposive criteria (Green & Thorogood, 2014; Richards, 2014). Eligible participants were over 18 years of age and had been diagnosed with ISM for at least one year, Danish-speaking, and had experienced at least one episode of anaphylaxis. Participants that were mentally ill, or had diagnoses other than ISM were not included. Participants were approached via face to face and no one refused

to participate. No relationship to the participants was established prior to the study. Seven patients were included (Table 1).

Qualitative interviews

In the interaction with the participants, we pursued the narratives that the participants found most relevant, from their own perspectives (Green & Thorogood, 2014; Holstein & Gubrium, 1995; Richards, 2014). An interview guide was thematically constructed based on the aim, research issues and existing literature (Green & Thorogood, 2014). Open-ended questions were used to elicit reflective, descriptive and detailed answers. Examples of interview questions: "Please, tell me how you experience everyday life with mastocytosis? Tell me about how you experience your course of treatment. Describe a situation in your everyday life".

Two participants chose to be interviewed in their own homes, and the remaining five interviews were performed at the hospital. Only the participant and the interviewer were present at the interview. The interviews were conducted by the first author who was a specialized female nurse, highly experienced in ISM. The interviews lasted between 45-65 minutes and were audio recorded. Subsequently, each interview was transcribed and analyzed, to identify themes, so that these could be further elaborated in the subsequent interview (Green & Thorogood, 2014; Richards, 2014). The data was collected from April to June 2016.

Data analysis

According to Giorgi (2008), the purpose of phenomenology is to gain knowledge of participants' experiences in a particular field, and to capture knowledge of how a phenomenon is experienced, understood or perceived for the individual, as it appears in the their life world (Giorgi, 2008).

Phenomenology thus differs from a positivist-based approach, where prior knowledge of the world is

based on objective beliefs (Green & Thorogood, 2014). In relation to the current study, a phenomenological approach included a focus on participants' awareness of the disease and on everyday life in their life world. According to Giorgi (1975), human consciousness is always aimed at something (Giorgi, 1975). The relationship between consciousness and a phenomenon, also called intentionality, will continuously affect living life and be given different meanings depending on who experiences the phenomenon (Giorgi, 2008). Thus, in the current study, we were interested in the individual patient's awareness or experience as it appeared in their specific life world.

The lifeworld is the world in which we live and take for granted (Giorgi, 1975). It is the actual everyday life reality that things appear as they really are, prior to explanations and independent of these. Therefore, in our study, we aimed to gain an understanding of how patients experience and understand mastocytosis and everyday life, in a search for responses as they appear and are perceived in the patient's life world – without the answers being guided by what is expected or assumed in advance (preconceptions). In the phenomenological approach, one's own preconception is put in brackets (Giorgi, 2008). The term epoché, which is an important prerequisite for phenomenological analysis (Giorgi, 2008), is applied to express the belief that it is necessary to focus on what comes from the participants' stories and experiences. It is essential that the researcher be aware of his/her own understanding, knowledge and experiences that relate to the phenomenon, to engage with the phenomenon openly and intuitively (Giorgi, 2008; Green & Thorogood, 2014). This is in order to be receptive to new and unexpected perspectives that patients bring forward in relation to what they perceive as relevant and meaningful in their everyday lives with mastocytosis.

The data analysis was inspired by Giorgi's phenomenological meaning condensation (Giorgi, 1975), which was developed on the basis of Husserl's descriptive phenomenology. Giorgi recommends the analysis be carried out in four steps. 1) Data collection, transcription and reading of data to get an overall understanding. 2) The identification of the natural meaning units. 3) Meaning units are transformed into themes. 4) Synthesis of the transformed meanings units into a general structure of the phenomena. The analysis process containing the four steps is described in more detail in Table 2.

The analysis process described above and in Table 2 involves meaning condensation. Meaning condensation means that the opinions expressed by participants in the interviews are reworded in shorter formulations where the essence of what is expressed are rephrased in fewer but more precise words (Giorgi, 1975; Green & Thorogood, 2014). An example of the analysis process from the identification of natural meaning units to the formation of a theme' form of theme is shown in Table 3. We validated the findings by discussing the interview transcriptions and the identified themes among the interdisciplinary research team.

Ethical considerations

Written informed consent was obtained from all participants. We provided participants with oral and written information about the study. Confidentiality was ensured, and recordings and transcribed material were stored in accordance with Danish data protection laws. According to [BLINDED] legislation, no formal approval from an ethics committee was required, as this was a qualitative interview study (DNVK, 2012).

Results

Based on the analyses, the following three themes and five subthemes emerged:

- Everyday life with a rare disease – unknown to most people
 - *Being perceived as a hypochondriac in the encounter with the health system*
 - *The diagnosis makes a difference*
 - *Expert knowledge is important*
- Living with and handling the invisible and visible symptoms
 - *The visible body*
- Fearing an attack

- *Feeling safe and vulnerable at the same time*

All themes related to conditions that affected how patients with mastocytosis experience, understand and manage with the disease in their everyday lives. These themes will be described in the following and supported by relevant quotes.

Everyday life with a rare disease – unknown to most people

Living with a rare disease that nobody knows about was described by the participants as having a great influence on their everyday lives, and in interaction with and meeting other people. The participants described many years of unfruitful clinical investigations of symptoms, and anaphylactic episodes. In the encounter with the health system, participants had experienced lack of knowledge, insight and understanding of their illness among health care professionals. Participants reported that those in their social networks also had difficulties in understanding the disease, since it was very rare and generally unknown, and therefore it was difficult for them to draw parallels to similar patient stories and experiences.

“... There is nobody who knows mastocytosis, my family, my colleagues, my doctor ... they don't know what it is ... it would be easier if it was diabetes or a broken arm, because everyone knows what that is ... they don't always understand it ... and it's difficult to explain” (Participant A).

For the participants, the rarity of ISM gave them the feeling of “loneliness with the disease”; this arose especially when they felt that friends, family or the health care professional did not really believe them, or didn't bother to listen to them tell about their symptoms, or did not try to understand how they felt. The experience of being alone with the symptoms and challenges related to the disease was described as a big burden.

Being perceived as a hypochondriac in the encounter with the health system

The participants reported a feeling of being perceived as hypochondriacs in their encounter with the health care system, before being finally diagnosed with ISM.

“Actually, I have always been told that it was my head that was wrong and there was nothing wrong with me ... I just had to pull myself together ...” (Participant F)

The participants expressed that they saw a barrier because their symptoms did not fit into any diagnosis that the health system had previously seen or recognized, and they were often sent home without further explanation or follow-up.

“... For many years I had pain in my stomach ... my skin itched and the spots got worse ... but when I went to the doctor, he just took a blood sample that was normal ... and said the spots were just hormone-related spots, and that I should enjoy life instead of inventing symptoms ... the doctor actually looked at me as hypochondriac ...” (Participant E).

Others described times when they had been hospitalized with an anaphylactic episode. In this context, they had been investigated intensively to rule out other diseases, but a diagnosis and a cause of their symptoms were not given. The experience of not always being believed was described as creating a lack confidence in the health care system.

The diagnosis makes a difference

The uncertainty about the body's reactions was described by the participants as being very frightening and made them feel scared and unsafe. Receiving a diagnosis was therefore described as a huge relief, because it subsequently gave them an explanation for their symptoms, while legitimizing their disease both in relation to themselves and the world around them. Being diagnosed was described as creating trust, meaning, order and security in an otherwise chaotic mass of symptoms, which were described as confusing and unmanageable in their everyday lives.

“... it was a relief to get a diagnosis and find out: well, I do actually have something ... it was not quite nonsense, all that I felt in my body ...” (Participant E).

The diagnosis also highlighted for the participants the seriousness of the disease, in that they got insight and knowledge about prognosis and treatment.

“... It was a relief to get a diagnosis ... it was not cancer, but at the same time it was also difficult because there's not really any treatment ... it's only symptomatic treatment ...” (Participant D).

Upon being diagnosed, the participants experienced that they were met with greater understanding by health professionals than ever before.

Expert knowledge is important

The encounter with the specialized center, where the health care professionals had expert knowledge, was described by the participants as very important. They felt that they were heard, and they were met with understanding and recognition of their symptoms. Participants described it as very positive and as giving a sense of security, to be linked up with a centre where there was expert knowledge and specialist skills in relation to their diagnosis.

“... It gives an enormous sense of security to know that there is a control of it here ... they (the staff) know what mastocytosis is and the right things are being done, so it has been very positive ...”(Participant B).

To be seen by an expert in mastocytosis allowed the participants to create a treatment structure, legitimize illness behavior and eliminate a certain amount of uncertainty. At the same time, they got specific advice and guidance on an individual basis. All this, they expressed, made them able to better understand, manage and live with the disease in everyday life.

Living with and handling the invisible and visible symptoms

The participants described that the unpredictability of the disease and the multiple visible and invisible symptoms had an influence on their everyday lives. The participants said that they often felt tired and in pain, and therefore had to limit their activities and social events, because of the disease.

Some of the participants expressed that they had to prioritize socializing because of their physical condition, and they could not participate on equal terms, compared to others. For example, they might have to leave early from parties and social events while their partner remained.

“... If we are at a party or something, then I'll go home earlier, and my husband stays, and he'll come home later ... it's actually my pain that causes it ... it fluctuates so 3-5 hours then I simply can't take it

anymore ... in the beginning people were enormously outraged by my behavior, but I have to take care of myself ...” (Participant F).

The women, in particular, reported that they could find themselves isolated and lonely, and unable to take part in the activities, experiences or communities that others participated in. Participants described a lack of energy to keep up with grandchildren, compared to other grandparents, and thus experienced not being able to live up to the ideal or expectations that they had of themselves or others had of them, and this affected their self-image negatively.

The participants described the experience of stigma and lack of understanding from their social networks in relation to why they did not do things like everyone else. Participants wanted to live a normal life, involving parties, alcohol and so on. They said that the invisible symptoms were difficult to understand for people around them. The participants said that, at times, they did not want to tell people about mastocytosis, and found other excuses and explanations. An excuse could be that they had to drive home, to avoid explaining why they did not drink alcohol. They explained that making excuses like these was important for their self-esteem and their interaction with others. Other invisible symptoms, such as memory problems, were described as disturbing and affected them in different situations.

“... Once in a while, you feel stupid when you can’t remember or you try to have a conversation with other people, friends, or colleagues and you can’t keep the thread of a conversation. It becomes quite mentally ... well, you get nervous about losing the thread ... It affects my self-esteem ...” (Participant E).

The participants said that they were trying to live life as others did, and they would not let the symptoms rule their everyday lives.

“... You learn to live with it as time goes on ... so, sometimes I go all in and then I know I am completely exhausted several days after ... but I then feel I am alive ...” (Participant G).

Participants said that they were constantly on the alert to minimize the potential risk of ending up in a severe situation that could trigger a reaction, and in the worst case, cause an anaphylactic

reaction. Therefore, the participants had regular habits and routines, which was adapted to the current situation in everyday life. Everyday life activities, such as cleaning and shopping, were planned, or their partner had to take over in order to avoid an exacerbation of symptoms.

Some participants had retired because of their many severe symptoms, which made it impossible for them to manage a job. Concerns about the future and the development of further symptoms were described by the participants.

The visible body

The visible symptoms of the skin were described by those participants who had skin lesions (urticaria pigmentosa) as very difficult and worrying, and as a great challenge in their everyday lives.

Symptoms such as itching, redness, swelling and heat that could arise suddenly were described by all of them as a big burden. Participants described the discomfort of their own body, that they felt less attractive and ugly, and that it was difficult for them to look at themselves naked. Their self-image became affected, and some described that it had a negative impact on their sexuality.

"... It doesn't matter to my husband ... but every time he touches my skin this image of my stomach pops up in my mind ... all the spots and there are brown bumps all over it ... So I think the skin is disgusting so I just can't ... just enjoy it ..." (Participant E).

Especially the female participants reported that skin symptoms constantly reminded them of the disease, and they deliberately chose makeup or clothing that covered the skin lesions, for example by using a scarf. The perception that people looked at or talked about them affected some of the participants, resulting in social isolation, anxiety, and mistrust of those in their surroundings.

"The skin means that I never go to the beach or the swimming pool, and that's because I do not feel comfortable ... to go to such places, when you have so many spots ... it's mental ... I feel people are looking." (Participant A).

The avoidance of social activities was also experienced by three participants. It was described as having an impact on their family, for example trips to the beach or beach holidays. For some

participants, the skin's appearance was not important, whereas symptoms such as itching and redness were expressed as being much more stressful.

Fearing an attack

The participants had experienced severe anaphylactic episodes that required emergency help and hospitalization. They described these as traumatizing experiences, both for themselves and their families. The feeling that they were close to death was expressed by the participants, as a feeling of severe anxiety:

"... I simply thought that I was going to die, it was the thought I had ... soon I would be lying in the coffin" (Participant C).

Participants said that previous attacks stood quite clearly in their memory.

"It just lies in my consciousness and affects me ... and does so in daily life. Suddenly, I feel an increased heart rate, and I haven't done anything ... then I think, now comes another attack ... and then I have to lie down, put my legs up, and then try to get a grip on the pulse again ..." (Participant A).

The participants expressed that their fear of having an anaphylactic attack could be reinforced when they were alone. Therefore, they very often allied themselves with neighbours, as their presence was perceived as making them feel safe. Others reported they would rather handle it on their own.

The fear of an attack happening in a public place, with nobody to provide competent assistance, created insecurity:

"... I have always been afraid that if it happened on the train or in a form of transport, where I do not know anyone, then it is better if it happens at work ... where there are colleagues who know what to do ..." (Participant D).

The experience of not being able to control an attack created fear and insecurity and affected both the participant and their family members in their everyday lives.

“... It comes all of a sudden ... it’s not under control ... I have tried several times, I have trained soccer girls, and then suddenly I have to go to the hospital, because I’m sick ... I am picked up by the ambulance. It’s a traumatic experience that way, and my daughter sees it every time, and my wife sees it too often ... it’s a great mental strain.” (Participant B).

These situations were described by the participants as having an impact on their relationships with partners, children or colleagues, who were extra concerned and aware of situations and symptoms. Participants explained how this resulted in preventive routines, such as not being the only adult present when bringing the children to soccer, or special agreements with colleagues who were able to assist, if needed. This extra attention was described as having an important impact on their relationships with others.

“... All of a sudden I become viewed as vulnerable – one to be taken care of ... but I’m a grown man ...” (Participant B).

Feeling safe and vulnerable at the same time

All the participants had been provided with an adrenaline pen, to be used as emergency medication in case of anaphylaxis. It created both security and fear for some participants. The sense of security was related to the ability to act in an uncontrolled situation, which could save their lives.

“It means I know I have a tool that can prolong my life in that 15-20 minutes it takes before the ambulance comes, you can be a little more, I will not say relaxed, but safe ...” (Participant G).

The adrenaline pen was described as making them feel safe because it could save their lives, but it was also stressful, because they never knew when they would need it. For some, it was an extra challenge always to remember to have it with them, and it could create a new cause for anxiety:

“So, if I have forgotten the adrenaline pen, no matter what happens, I’ll come back home, I do not go out without adrenaline. I’m simply ... it’s like getting claustrophobia ...” (Participant F).

The participants who had not tried to use the adrenaline pen yet described fear and uncertainty if they should experience an anaphylactic shock – the insecurity of not knowing what would happen,

not knowing how their bodies and those around them would react, did have an influence on their way of living and handling the disease.

Discussion

To our knowledge, this is one of the first qualitative studies to investigate patients' experiences with mastocytosis, and how it affects everyday life. The results show that living with a rare, chronic disease such as mastocytosis not only affects the physical body, but also has psychosocial influence on patients' everyday lives, and an affect on their family, work and social relationships.

The study demonstrates that mastocytosis and the unpredictable symptoms have a significant influence on participants' routines and on how they live their everyday lives. The individual's conduct of everyday life and self-understanding (Holzkamp, 1998) is challenged by symptoms of sudden onset and exacerbation. Holzkamp (1998) describes the consequences of everyday life routines being interrupted by disease, symptoms and treatment. Holzkamp distinguishes between the everyday life, and the real life. The real life is characterized by events such as crises and disease, which differ from the daily routine (Holzkamp, 1998). From this perspective, the disease mastocytosis becomes a part of the real life, and the real life becomes stressful for the individual person, as it is characterized by unpredictability (Holzkamp, 1998).

In our study, participants experienced the unpredictability very stressful in their conduct of everyday life, because of the various symptoms; in particular, the risk of having an anaphylactic attack created disorder and chaos. Participants used different strategies, such as repetitions, routines and planning daily activities, to manage everyday life. Repetitions created security for the participants, and facilitated interpretation and understanding of a new everyday life as a whole and in specific contexts (Dreier, 2008; Holzkamp, 1998; Mørck, 2006). The participants used planning and routines to make life safer and more secure. These findings are consistent with other studies (Jennings et al., 2014; Nowak et al., 2011; van Anrooij et al., 2016), which show that the unpredictability of

mastocytosis similarly affected patients considerably, had an impact on their daily activities and social relationships, and created increased anxiety.

Furthermore, our study demonstrates that anxiety and negative impact related to the long diagnostic delay between the first onset of symptoms to a definitive diagnosis. Diagnostic delay is also described in the study by Nowak et al.(2011), which was based on a survey of mastocytosis patients and demonstrated a delay of diagnosis of up to 11 years (Nowak et al., 2011). The patients in the current study also experienced the long diagnostic delay as having an impact on everyday life, resulting in increased anxiety and uncertainty.

The current study showed that participants felt that they were perceived as hypochondriacs by the health care professionals and those in their social networks. This had a negative impact on their confidence in the health care system and created anxiety. The same challenges of having a rare disease were described, as a general problem for patients with rare diseases, in a European report on people with rare diseases, such as skin disease epidermolysis bullosa (EURORDIS, 2009; Lancet, 2009). In line with our findings, the report showed that patients experienced a challenge, by not being believed, being rejected, not being properly diagnosed or not receiving qualified advice and guidance (EURORDIS, 2009).

The current study illustrated that patients with mastocytosis had several years of unexplained and invisible symptoms behind them, which could not be explained or recognized by the existing biomedical explanatory model (Malterud, 2000). A qualitative study by Undeland and Malterud (2007), using focus group interviews with women diagnosed with fibromyalgia, underlined how important it was for patients to be diagnosed(Undeland & Malterud, 2007). This is similar with the experiences of the participants in this study where the diagnosis came as a relief; it helped patients to explain and legitimate their symptoms. Similar results were found in other qualitative studies in patients diagnosed with fibromyalgia, which showed that a diagnosis was important to patients,

particularly because it reduced anxiety and uncertainty associated with unexplained symptoms (Lempp, Hatch, Carville, & Choy, 2009; Sim & Madden, 2008).

Participants in our study experienced and explained that, after the diagnosis of mastocytosis had been made, they were taken seriously and they were believed. The diagnosis makes a difference in the encounter with the health system, as it affects the way patients are met, understood and treated by health care professionals (Brinkmann, 2016). People with a chronic rare disease need expert knowledge about their disease (EURORDIS, 2009) and, in relation to this, we found that participants experienced a lack of knowledge in general among health care professionals about mastocytosis, and that expertise was of great importance to them. The expert contributed to the establishment of specific routines and actions, which helped them to conduct their everyday lives (Holzkamp, 1998). Expert knowledge helped the patients to be more understood and recognized by others. Also, training in the use of the adrenaline pen made an important difference to most of the participants in our study. This is in line with the quantitative study by Nowak et al (2011). By using a retrospective questionnaire, they examined how patients with mastocytosis experienced the professional response to their disease. Here, expert knowledge and a connection to specialists were highlighted as important factors, in that these factors helped to eliminate a great deal of uncertainty that had filled their lives (Nowak et al., 2011).

The symptoms of mastocytosis led to a sense of stigma. Participant's perceived that they were stigmatized by those around them and felt social isolation, which was caused by their experience of visible and the invisible symptoms. This trend is also seen in other studies, in patients with psoriasis (Lakuta, Marcinkiewicz, Bergler-Czop, & Brzezinska-Wcislo, 2017), where the visible symptoms dominate, and by patients with fibromyalgia (Sim & Madden, 2008), where the invisible symptoms predominate. In comparison with psoriasis or fibromyalgi, patients with ISM have both invisible and visible symptoms, coupled with the risk of anaphylaxis (Hermine et al., 2008; Jennings et al., 2014; van Anrooij et al., 2016).

Our study showed that the visible symptoms of the skin, and thus the body's appearance, was of great significance to the participants in their encounter with other people in everyday life, and in relation to their partners and sexuality. In line to our findings perceptions of the body and the consequences for self-esteem, close relationships and sexuality are frequent in chronic skin diseases (Hermine et al., 2008; Jennings et al., 2014; Lakuta et al., 2017; Magin, Heading, Adams, & Pond, 2010), such as psoriasis and atopic eczema. The results in our study indicated a linkage between the appearance of the skin, body image and, especially, the female participants' self-esteem. These findings agree well with other studies showing patients with skin disorders, such as burn victims (Price, 2009) or psoriasis, experience that their body image and self-esteem are greatly affected (Lakuta et al., 2017; Magin et al., 2010). The diverse symptoms and unpredictability of mastocytosis challenge everyday life in several different ways and in different contexts (Hermine et al., 2008; Jennings et al., 2014; van Anrooij et al., 2016). This appeared in the current study, in which participants experienced not having control and never knowing what was going to happen. The fear of having an attack had consequences across different social contexts.

Strengths and limitations of the study

In the current study, no gender perspective was taken into account, as there was an over-representation of women. This is, however, consistent with the reported incidence of mastocytosis, where there is a slight female predominance (Cohen et al., 2014).

The interviewer is a specialized nurse, highly experienced in ISM; her preconceptions might have influenced the interpretation of the data, which was addressed by having close dialogues with her supervisor and other occupational professionals (Green & Thorogood, 2014). The choice of method is assessed as relevant to elicit patients' experiences and perspectives on living with mastocytosis (Green & Thorogood, 2014).

Our study only included patients who had experienced anaphylaxis, which probably influenced the

results. It is a limitation of our study that patients with no previous episode of anaphylaxis were not included. We included only the more severely affected ISM patients.

Data saturation is reached when there is enough information to replicate the study, when no new information can be attained, and when further coding is no longer feasible (Fusch & Ness, 2015).

We carried out seven in-depth interviews with participants with mastocytosis. A small number of participants may be enough to provide a sufficient amount of data, because it is not the number of participants that determines whether or not there is enough data to reach data saturation (Kvale, 1999). According to Fusch et al. (2015) if saturation is considered as a matter of degree, there will always be the potential for the “new” to emerge. They suggest that saturation should be more concerned with reaching the point where further data collection becomes ‘counter-productive’, and where the ‘new’ does not necessarily add anything to the overall story (Fusch & Ness, 2015).

Conclusion

Patients with mastocytosis are severely affected in their daily lives because of the conditions of the disease. In particular, patients’ social lives and relationships with family and those in their social networks are significantly affected. The unpredictability of symptoms limits and complicates activities and participation in social activities. The fear of having an anaphylactic attack is always present. The disease affects patients’ self-esteem and sexuality. The encounter with the health system and those in their environments both before and after diagnosis was of great importance to the patients, which underlines that there is a crucial need to teach health care professionals about the disease.

Relevance to clinical practice

It is important that the patient's experiences from an everyday life perspective are taken into account in counselling and when tailoring treatment to the individual patient's needs. An interaction between the everyday perspective and the biomedical approach, which focuses primarily on medical treatment, is needed in clinical practice in order to help the patient in the best possible way. There is a need for health professionals to think holistically and involve the patient on the basis of his or her needs and wishes.

The results of this study show that when patients with ISM talk about their disease they refer not only to a variety of custom symptoms, but to a complex collection of experiences, perceptions and emotions, that arise in different situations and contexts in everyday life.

The focus of counselling should not only be on the specific organic disease manifestations, but also on living life with the disease. By a detailed description of patients' experiences, perceptions and management in different contexts and situations, results from the study could help health care providers who care for mastocytosis patients to have a better understanding of the disease and the burden on patients.

Often, the information, guidance and counselling of the patient and family is based on an agenda that is set by the health system; the agenda may be very different to the context and everyday life of the patient. A future perspective of our study is the discussion about whether our health system should be flipped away from the traditional ways of organizing and structuring consultations, to focus instead on what is important for the individual patient, where the starting point is the patient's agenda, need and experiences.

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Table 1: Summary of the participants in the study

Participant	Sex/ Age	Married	Job	Year with diagnosis	Symptoms
A	Woman, 47 years	Yes	Yes	2 years	Severe skin symptoms, gastrointestinal discomfort, fatigue, concentration difficulties, headaches, pain in bone / joint, sadness, anxiety, many cases of anaphylaxis
B	Male, 43 years	Yes	Yes	2 years	No skin symptoms, beginning osteoporosis, gastrointestinal discomfort, many cases of anaphylaxis
C	Women, 65 years	Yes	Retired because of mastocytosis	6 years	Skin symptoms, gastrointestinal discomfort, fatigue, headache, pain in the body, many cases of anaphylaxis
D	Male, 45 years	Yes	Yes	4 years	Severe skin symptoms, onset of osteoporosis, gastrointestinal discomfort, mood swings, multiple cases of anaphylaxis
E	Woman, 47 years	Yes	Yes	7 years	Severe skin symptoms, pain, nausea, gastrointestinal discomfort, fatigue, memory problems, mood swings, anxiety, two cases of anaphylaxis
F	Woman, 49 years	Yes	Retired because of mastocytosis	6 years	Severe skin symptoms, chronic pain of the bones / joints, fatigue, gastrointestinal discomfort, difficulty concentrating, memory problems, anxiety, one case of anaphylaxis
G	Woman, 53 years	Yes	No, unemployed	13 years	Severe skin symptoms, pain in bones, nausea, abdominal pain, fatigue, memory problems, anxiety, two cases of anaphylaxis

Table 2: Description of the four steps in the analytical method, inspired by Giorgi

<p>Step 1</p> <p>Data collection, transcription and reading of data to get an overall understanding</p>	<p>The transcribed interview material was read, to achieve an overall understanding of the entire transcript. in order to derive a global sense of the whole (Giorgi, 1975). This was done with the greatest possible openness to data where preconceptions were put aside and without taking the specific aim of the study into account.</p>
<p>Step 2</p> <p>The identification of the natural meaning units</p>	<p>Natural meaning units were identified as expressed in the participants' own words (Giorgi, 1975). The written material was read again in order to create natural sense units of the participants' experiences related to the investigational phenomenon: in this case, the disease mastocytosis and everyday life. According to Giorgi (Giorgi, 2008), it was important that the theme only emphasized the importance of each unit as it first seemed, and did not try to relate it to the aim of the study or try to interpret what it meant. These sense units were separate entities which together emerged as a meaningful whole, just as it appears. No further analysis was carried out at this time.</p>
<p>Step 3</p> <p>Meaning units are transformed into themes</p>	<p>The material was re-read and meaning units were transformed into themes that expressed the essence of the phenomenon related to the aim of the study (Giorgi, 1975). There was a focus on both the natural meaning units and the central themes and the purpose of the study was also in focus: how patients with mastocytosis experience, understand and manage the symptoms and changes in their everyday lives that occur in relation to mastocytosis. The theme that dominated the individual natural meaning unit was reformulated as simply as possible, so that meaningful units were themed from the interview participant's point of view, as it was understood and interpreted (Giorgi, 1975). Meanings units were redefined in a more comprehensive research and academic language. Each meaning unit was studied systematically for what it showed about the studied phenomenon in a specific context for the specific person.</p> <p>The transformed meaning units for each interview participant were linked, so that a specific situation structure was obtained that was a more consistent and descriptive text. The specific situation structures were important because they provided the basis for the general state</p>

<p>Step 4</p> <p>Synthesis of the transformed meanings unit, into a general structure of the phenomena</p>	<p>structure in force in general, and described the essence of the phenomena. Looked at what was common, i.e. universal, and brought it forward to the essence of phenomena. Sub-themes and topics were identified in each interview and across the seven interviews (Giorgi, 1975). The general descriptions of the themes are shown in the Results section.</p>
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Table 3: Illustration of an example of the analysis process, from the sense units to themes

Natural meaning units arise from the interview	Categories	Sub-themes	Final themes
<p>Participant E: "... I have always been sick ... I've always had stomach problems and rashes and scratched my skin. It scratched for years without there being anyone who knew why, and I felt then like a person who invents diseases, a hypochondriac ... The doctor says you are healthy, thus, it may well be I have stomach problems, but they cannot prove anything, so I am healthy ..."</p>	<p>Hypochondriac - they cannot prove she's sick</p>	<p>Being perceived as a hypochondriac in the encounter with the health system</p>	<p>Everyday life with a rare disease – unknown to most people</p>
<p>Participant F: "I've actually always been told that it was my head that was wrong, there was nothing wrong with me, I just had to pull myself together, so it was so nice to get a diagnosis and find out, that I actually was ill it was not just nonsense, everything that I felt in my body ..."</p>	<p>Nice to get a diagnosis – she actually was ill</p>	<p>The diagnosis makes a difference</p>	
<p>Participant C: "... both family, friends, there's no one who knows mastocytosis, they know not what it is, and it is a little annoying when you come to your own doctor and say that you have mastocytosis, and he doesn't know what it is at all ..."</p>	<p>Nobody knows mastocytosis</p>	<p>Expert knowlegde is important</p>	