Systemic Mastocytosis Patient Experience from Mast Cell Connect, the First Patient-Reported Registry for Mastocytosis

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BACKGROUND

- Systemic mastocytosis (SM) is a rare, underrecognized hematologic disorder characterized by excess mast cells in the bone marrow and other organs.1,2
- Patients with SM often present with symptoms that are not effectively addressed by current treatments.3,4
- Little is known about the patients’ journey, their perspective on the burden of disease, and real-world treatment approaches for SM.
- Systematically characterizing the natural history of SM and its impact on patients will facilitate the development of new therapies.

THE MAST CELL CONNECT REGISTRY

- Mast Cell Connect (MCC, www.mastcellconnect.org), the first patient-reported registry for mastocytosis, began enrolling on Dec 1, 2015.

Key study objectives:

- To facilitate the development of new therapies by improving collective understanding of the natural history of mastocytosis and its impact on patients, as well as increasing participation in clinical trials.
- To provide evidence to support the need for enhanced care and the development of new treatments for patients with SM.
- To increase awareness among healthcare providers about SM.

METHODS

- The Mastocytosis Society, expert physicians, and media coverage enabled rapid participant enrollment.
- Participants enrolled on a secure online portal and completed a 25-item survey providing demographic, disease, and treatment information. This survey was developed specifically for Mast Cell Connect and incorporated standard questions used in rare disease patient registries, questions from the EORTC QLC-C30 quality of life questionnaire, and questions specific to patients with mastocytosis.
- Site-agnostic enrollment enables broader participation and collection of real-world data, particularly in the US, where many patients are seen in the community setting.
- Inclusion criteria were a diagnosis of SM or cutaneous mastocytosis (CM).
- Participants reporting diagnoses of both SM and CM were categorized as SM participants.
- Diagnoses were self-reported; however, participants were asked to provide medical records to support their diagnosis.
- The study is IRB-approved and informed consent was required to join.

RESULTS

- Participants (n=208) joined Mast Cell Connect at time of data cut-off (September 30, 2016). 20 participants had joined Mast Cell Connect.
- The majority of participants were female (74%) and reported a diagnosis of SM (63%) or both SM and CM (37%).
- SM participants reported concomitant CM, seen in 37% of indolent SM (ISM), 11% of SM subtype unknown, and 1% of advanced SM (AdvSM).
- All SM participants had at least one symptom attributed to patients with mastocytosis.

Table 1. Participant demographics

<table>
<thead>
<tr>
<th>All (n=208)</th>
<th>SM (n=137)</th>
<th>CM (n=47)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Age, median (range)</td>
<td>46 (7–77)</td>
<td>52 (7–77)</td>
</tr>
<tr>
<td>Gender (%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Male</td>
<td>54 (26)</td>
<td>54 (27)</td>
</tr>
<tr>
<td>Female</td>
<td>154 (74)</td>
<td>133 (76)</td>
</tr>
<tr>
<td>Race, n (%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>White</td>
<td>189 (91)</td>
<td>130 (95)</td>
</tr>
<tr>
<td>Country, n (%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>US</td>
<td>187 (90)</td>
<td>123 (90)</td>
</tr>
<tr>
<td>Canada</td>
<td>8 (4)</td>
<td>6 (5)</td>
</tr>
</tbody>
</table>

**Table 2. Time from symptom onset to diagnosis of SM**

- **Participants (n=150) were asked to report the time from symptom onset to diagnosis.**
- **SM subtype unknown: 29%**
- **Diagnoses were made most frequently by specialists in dermatology (39%), hematology (25%), and general practitioners (24%).**
- **Participants reported a diagnostic odyssey of 8 years from symptom onset to diagnosis** (Figure 2; Table 2).
- **Fatigue/tiredness** was the most common symptom (affecting ≥50% of all SM participants) reported to be most problematic (X: Figure 3).
- **The use of imatinib (7%) was higher than expected, as it is not indicated for patients with SM.**
- **Most SM participants, across all subtypes, are currently using symptom-treating medications.**
- **Despite frequent use of symptom-treating medications, many patients suffer from uncontrolled symptoms and limitations on daily activities.**
- **The most common symptoms (affecting ≥50% of all SM participants) were fatigue (71%), difficulty concentrating (60%), pain (other than abdominal: 54%, abdominal: 3%), and difficulty sleeping (35%).**
- **Several gastrointestinal symptoms, difficulty concentrating, difficulty sleeping, anxiety, and depression tended to be reported more frequently in AdvSM participants.**

**Table 3. Frequency of participants experiencing moderately to severely interfered with activities of daily living**

- **SM impacts daily living**: Across all subtypes of SM, 35% of participants reported that SM impacts ‘quite a bit’ or ‘very much’ on their work or other daily activities and family life (Figure 3).
- **There was a trend for greater impact to be reported more often by AdvSM participants.**

**Table 4. Frequency of participants currently using each medication (%)**

- **Anti-inflamators**: 35% of SM participants reported being on anti-inflamator medication.

**CONCLUSIONS AND NEXT STEPS**

- **Ongoing enrollment and data collection to date: 208 participants joined Mast Cell Connect, suggesting the mastocytosis community is highly motivated to participate in research.**
- **Participants reported a diagnostic odyssey of 8 years from symptom onset to diagnosis, indicating that SM is largely underrecognized.**
- **Despite frequent use of symptom-treating medications, many patients suffer from uncontrolled symptoms and limitations on daily activities.**
- **Newer therapies are needed to improve quality of life and outcomes.**
- **Continued collaboration among researchers, patient advocates, and industry is needed to advance care and the development of new treatments for patients with SM.**

**For more information, please visit www.mastcellconnect.org or contact us at patients@blueprintmedicines.com**

**ACKNOWLEDGMENTS**

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**Disclosures**


**REFERENCES**


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**Table 2. Time from symptom onset to diagnosis of SM**

**Table 3. Frequency of participants experiencing moderately to severely interfered with activities of daily living**

**Table 4. Frequency of participants currently using each medication (%)**

**Figure 1. Frequency of SM subtypes**

**Figure 2. Age at symptom onset and diagnosis of SM**

**Figure 3. SM impacts daily living**

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