THE MASTOCYTOSIS SOCIETY SURVEY ON MAST CELL DISORDERS:
Part 2- Clinical Experiences, Co-Morbidities, Diet, Families and Opinions

1Nancy Russell, Dr PH, 2Susan Jennings, PhD, 3Blair Jennings, BS, 4Valerie Slee, RN, BSN, 5Lisa Sterling, BS, 6Mariana Castells, MD, PhD, FAAAAI, 7Peter Valenf, MD, 8Cem Akin, MD, PhD, FAAAAI

1The Mastocytosis Society, Inc., USA; 2Mastocytosis Center, Department of Medicine, Division of Rheumatology, Immunology, and Allergy, Brigham and Women’s Hospital, Harvard Medical School, Boston, MA, USA; 3Department of Internal Medicine I, Division of Hematology & Hemostaseology, Medical University of Vienna, Vienna, Austria

ABSTRACT
Rationale: Mast cell diseases such as mastocytosis and mast cell activation syndromes involve abnormal proliferation or activation of these cells leading to many potentially debilitating symptoms. In order to determine the characteristics and experiences of people known or suspected to have a mast cell disorder, The Mastocytosis Society (TMS), a U.S. based patient advocacy, research and education organization, conducted a survey of patients.

Methods: This web-based survey was published through specialty clinics and the Society’s newsletter, Web site and online blogs. Both online and paper copies of the questionnaire were provided together with required statements of consent.

Results: The first set of results from this survey of 420 respondents has been previously published; the second set is now presented. These results include sources of diagnosis, clinical and laboratory tests performed, co-morbid conditions, dietary practices, possible familial occurrence of mast cell diseases, and perceptions concerning mast cell related medical care in the United States.

Conclusions: These patient survey results are provided to assist medical professionals in defining clinical approaches and research goals and to give patients with mast cell disorders the opportunity to review the experiences of similar patients.

BACKGROUND
Mature mast cells (MC) are found around blood vessels in all tissues, and also where the body interacts with its surroundings, well-positioned for quick reaction to environmental threats. MC disorders (MCD) include diseases involving abnormal proliferation and/or activation of these cells. Patients may have a primary MCD or non-clinical IgE mediated or non-IgE mediated MC activation including MC activation syndromes (MCAS). People with MCD may be at risk for anaphylaxis and chronic debilitating symptoms.

MC activation occurs by both IgE-dependent and independent mechanisms, causing MC to release mediators including histamine, tryptase, arachidonic acid metabolites, such as prostaglandins and leukotrienes, cytokines and chemokines, which initiate or exacerbate symptoms.

METHODS
Confidential cross-sectional survey questionnaire
Eligible participants were patients with MCD of all ages, or caregivers, living in or outside the US
Published through TMS (The Mastocytosis Society's) clinical notices of physicians working with TMS support groups, TMS website and online MCD-related blogs.
Posted online through TMS website link, April 15 - May 24, 2010.
Valid respondents defined as answering to least some questions beyond opening section, “Demographics and Diagnosis.”

Demographics
Respondents 530, Valid 420
Ages (of 416 subjects providing birth years) Average 44.8, Median 46, Range 1 - 80
Gender: Female 62.6%, Male 22.1%
Ethnicity: White 93.6% Native American, Hispanic or Other/mixed 6.0%
Residence (not collected due to confidentiality concerns) in U.S. in received by 84.3%

RESULTS:
Table I. “Who diagnosed your mast cell disorder?”

<table>
<thead>
<tr>
<th>Physician Type(s) as Source of Diagnosis</th>
<th>Total Respondents No. (% of 420)</th>
<th>Recalled Single Physician Type No. (% of 252)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Dermatologist</td>
<td>196 (46.7)</td>
<td>103 (40.9)</td>
</tr>
<tr>
<td>Allergist/immunologist</td>
<td>130 (31.0)</td>
<td>61 (24.2)</td>
</tr>
<tr>
<td>Hematologist/oncologist</td>
<td>111 (26.4)</td>
<td>38 (15.1)</td>
</tr>
<tr>
<td>Primary Care Physician</td>
<td>57 (13.6)</td>
<td>6 (2.4)</td>
</tr>
<tr>
<td>Gastroenterologist</td>
<td>49 (11.7)</td>
<td>18 (7.1)</td>
</tr>
<tr>
<td>Other*</td>
<td>26 (6.2)</td>
<td>9 (3.6)</td>
</tr>
<tr>
<td>Unspecified no./type (eg. &quot;doctor&quot;, hospital name)</td>
<td>21 (5.0)</td>
<td></td>
</tr>
</tbody>
</table>

*Other includes endocrinologist (13), internist-intern medicine specialist (5), pediatrician (4), hematologist (3), and others reported by 2 or less.

Table II. Clinical Exams and Laboratory Tests reported by 389 respondents

<table>
<thead>
<tr>
<th>Exams/Tests</th>
<th>Total Respondents No. (%)</th>
<th>Frequency†</th>
<th>Routine</th>
</tr>
</thead>
<tbody>
<tr>
<td>History &amp; physical</td>
<td>323 (83.0)</td>
<td>183 (56.7)</td>
<td>113 (35.0)</td>
</tr>
<tr>
<td>Visual skin</td>
<td>321 (82.5)</td>
<td>185 (57.6)</td>
<td>101 (31.5)</td>
</tr>
<tr>
<td>Photographic skin</td>
<td>100 (25.7)</td>
<td>26 (26.0)</td>
<td>52 (52.0)</td>
</tr>
<tr>
<td>Diagnostic Tests</td>
<td>Complete Blood (CBC)</td>
<td>337 (86.6)</td>
<td>263 (78.0)</td>
</tr>
<tr>
<td>Serum chemistries</td>
<td>147 (37.8)</td>
<td>105 (71.4)</td>
<td>36 (24.5)</td>
</tr>
<tr>
<td>Serum ferritin</td>
<td>76 (19.5)</td>
<td>41 (53.9)</td>
<td>30 (39.5)</td>
</tr>
<tr>
<td>Skin biopsy</td>
<td>237 (60.9)</td>
<td>Not queried</td>
<td></td>
</tr>
<tr>
<td>Bone marrow biopsy</td>
<td>221 (56.8)</td>
<td>36 (16.3)</td>
<td>154 (69.7)</td>
</tr>
<tr>
<td>Diagnostic Markers</td>
<td>Serum tryptase</td>
<td>300 (77.1)</td>
<td>175 (58.3)</td>
</tr>
<tr>
<td>24hr urine histamine</td>
<td>198 (50.9)</td>
<td>51 (25.8)</td>
<td>128 (64.6)</td>
</tr>
<tr>
<td>24hr urine prostaglandins</td>
<td>84 (21.6)</td>
<td>29 (34.5)</td>
<td>44 (52.4)</td>
</tr>
<tr>
<td>C-kit - other genetic</td>
<td>123 (31.6)</td>
<td>Not queried</td>
<td></td>
</tr>
<tr>
<td>Total*</td>
<td>yes (test ever performed) plus with no answer, but selected or noted a frequency.</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Percent of 389 respondents.</td>
<td>84% of total for each test.</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Positive genetic tests (389 respondents)

C-KIT D816V mutation
Yes 47 (24.4%) No 61 (15.7%) Not sure 85 (21.9%) Not tested 191 (49.1%) Other genetic mutation Not tested 193 (45.2%)

Cells express CD2 and/or CD25
Yes 24 (6.2%) No 13 (3.3%) Not sure 185 (47.6%) Not tested 162 (41.6%)

Co-morbid conditions
Osteopenia or Osteoporosis 120 (31.4%) High blood pressure 88 (23.0%)
Hypercholesterolemia 82 (21.5%) Cancer 53 (13.9%)
Heart 10 (2.6%)

Family occurrence of MCD (376 respondents) 86 (22.9%) reported one or more relative(s) with either suspected or confirmed MCD.

Diet and nutrition 382 respondents (91.0%)

- Physician referred to dietitian 41 (10.7%)
- Physician recommended Low Histamine Diet 46 (12.0%)
- Physician recommended Low Histamine Diet 22 (5.8%)

Perceptions of 94 who followed a Low Histamine Diet
- Symptoms improved? Yes 48 (51.1%) No 17 (18.1%) Not sure or not answer 27 (28.7%)
- Nutrition adequate? Yes 48 (51.1%) (same 48 as above) No 32 (34.5%)
- Not sure 14 (14.9%)

Medical care for MCD in U.S. 317 respondents (84.3%)
- Number of centers sufficient 51 (16.1%)
- Being treated by MCD specialist 124 (39.1%)
- Physician said could not treat them 118 (37.2%)
- Comfortable if local & MCD specialist 263 (83.0%)
- Well informed by physician 208 (65.6%)

SUMMARY AND CONCLUSIONS
Survey respondents described a wide variety of diagnostic physician types and tests, but many were unsure what was performed or its purpose.
- Up to 30% of respondents had co-morbid condition(s).
- Dietary approaches have been tried with success for some, but others believed that their symptoms had not improved and that their nutrition was not adequate.
- A fifth of respondents indicated relative(s) with a MCD.
- Medical care for MCD in the U.S. is limited, but most patients felt they were well informed and would adapt to their physician collaborating with a MCD specialist.

Dr Lisa Sterling, BS, 2Dr Nancy Russell, Dr PH. 3Dr Susan Jennings, PhD. 4Dr Blair Jennings, BS. 5Dr Valerie Slee, RN, BSN. 6Dr Lisa Sterling, BS. 7Dr Mariana Castells, MD, PhD. 8Dr Peter Valenf, MD. 9Dr Cem Akin, MD, PhD. 10Dr Nancy Russell, Dr PH. 11Dr Susan Jennings, PhD. 12Dr Blair Jennings, BS. 13Dr Valerie Slee, RN, BSN. 14Dr Lisa Sterling, BS. 15Dr Mariana Castells, MD, PhD. 16Dr Peter Valenf, MD. 17Dr Cem Akin, MD, PhD. 18Dr Nancy Russell, Dr PH. 19Dr Susan Jennings, PhD. 20Dr Blair Jennings, BS.