**Fabry Disease**
December 4, 2018

FDA held a Listening Session with Fabry Disease patients and caregivers on December 4th, 2018 to understand the symptoms of Fabry Disease that patients and caregivers consider the most important.

Patient Listening Sessions are intended to be a resource for the medical product Centers to expeditiously engage with patients or their advocates. Listening Sessions can either be FDA-requested (in cases where FDA has a specific set of questions to ask of a particular patient sub-population) or patient-led (when a patient community wants to share some perspectives with the FDA), and are generally focused on conditions of interest to staff across the medical product Centers.

*Discussions in FDA Rare Disease Listening Sessions are informal. All opinions, recommendations, and proposals are unofficial and nonbinding on FDA and all other participants. This report summarizes the input provided by patients and patient representatives at the meeting. To the extent possible, the terms used in this summary to describe specific manifestations of Fabry disease, health effects and impacts, and treatment experiences, reflect those of the participants. This report is not meant to be representative of the views and experiences of the entire Fabry disease patient population or any specific group of individuals or entities. There may be experiences that are not mentioned in this report.*

## Summary of topics discussed

1. **Impactful symptoms: The Fabry patients and caregivers described a variety of symptoms that significantly impact their daily life:**
    - Gastrointestinal (GI) symptoms were the most common symptoms mentioned by patients/caregivers. The GI symptoms experiences often include both constipation and diarrhea, as well as abdominal pain particularly in the lower abdomen. Other symptoms mentioned were cramping and bloating. The GI symptoms make it difficult for patients/caregivers to make plans, as well as eat at restaurants because of the quick onset of symptoms.
    - Cold and/or heat intolerance was another commonly mentioned symptom, which limits the activities patients/caregivers participate in during the summer and winter. Some participants indicated that they relocated their home to accommodate this symptom. It was often mentioned that they are unable to sweat. In some patients/caregivers, this symptom improved overtime.
    - Neuropathy, particularly in the extremities, was mentioned as a common symptom that impacts the patients/caregivers. Some mentioned that this symptom did not change overtime, and sometimes became worse with age.
    - Other symptoms that significantly impact patients/caregivers include tinnitus, hearing loss, fatigue, vision impairment/light sensitivity, headaches, and poor balance. This is not a complete list of the symptoms mentioned during the listening session.
    - Patients/caregivers indicated that they often experience depression, and this is a significant concern for the patient community.

2. **Effects of treatment: The Fabry patients and caregivers described a variety of symptom relief from treatments:**
    - It was frequently heard that enzyme replacement therapy (ERT) resulted in improved kidney function in Fabry patients.
    - Patients frequently reported that ERT help relieve fatigue. It was also frequently reported that patients experienced sweat, although sometimes this was minimal.

- The change in neuropathy and pain from ERT was inconsistent. Some patients experienced improvement, others had no change, and some had worsening pain.
- In general, GI symptoms did not change as a result of ERT. If there was an improvement in GI symptoms, it was often minimal.
- ERT infusions are often considered burdensome, however the patients/caregivers plan to continue receiving it.
- Some patients/caregivers indicated they experience allergic reactions to ERT, which might require them to have steroids and allergy medication when receiving treatment.

3. **Patient/caregiver concerns: The Fabry patients and caregivers described a number of concerns with their disease:**
    - Patients/caregivers indicated that they are worried that new treatments may not be developed. There is a concern that the development is stagnant because there are options available for Fabry patients. They hope that future treatments will be an improvement to what is currently available and will take into consideration that Fabry disease affects the entire body of the patient.
    - Patients/caregivers shared that they are concerned about the financial burden of treatment. It is especially worrisome to patients who are unable to work, do not have insurance, or are unable to receive disability benefits.
    - Patients/caregivers indicated that more education is needed about Fabry disease with other government agencies and healthcare providers.
    - While many patient/caregivers are excited about clinical trials and encourage others to enroll in them, there are some patients who cannot participate in trials due to the progression of their disease. Others indicated that they are concerned about the unknown associated with their disease progression.

**Partner organization**
National Organization for Rare Disorders

**FDA divisions represented**
- Office of the Commissioner, Patient Affairs Staff (organizer)
- Center for Drug Evaluation and Research (CDER), Division of Gastroenterology and Inborn Errors Products (DGIEP)

**Patients represented**
15 patients and caregivers

**Patient segments**
- Segment of classic male patients or their caregivers
- Segment of late onset patients or their caregivers
- Segment female patients or their caregivers

**Conflict of Interest**
- More than one participant indicated that they had previously been compensated by industry for sharing their experience of living with Fabry disease. These individuals are not receiving compensation for this listening session.