**Introduction**

- Fabry Disease (FD) is an X-linked lysosomal storage disorder where deficiency of alpha-galactosidase A and accumulation of globotriaosylceramide (Gb3) within lysosomes results in slowly progressive organ damage and shorter life expectancy.
- FD is suspected based on the individual's clinical presentation, and can be diagnosed by an enzyme assay done on leukocytes to measure the level of alpha-galactosidase activity. An enzyme assay is not usually done on leukocytes to measure the level of alpha-galactosidase activity. An enzyme assay is not necessary for FD diagnosis, but may be suggestive of FD if excess lipid buildup is noted. Patients with FD and Fabry disease may have similar symptoms as well as increased risk of cardiovascular disease.
- Enzyme replacement therapy (ERT) has been shown to improve or stabilize many FD manifestations. Increasing evidence obtained during recent years points towards starting ERT promptly to obtain optimal effects, highlighting the importance of early identification of FD patients.
- Nevertheless, delayed diagnosis in the diagnosis of FD in males and females, respectively, illustrates the difficulty in providing early treatment for many patients.

**The Fabry Outcome Survey (FOS)**

- FOS was implemented to provide high quality real-world data from clinical practice, analyses and evidence to support improvements in the treatment and management of FD.
- Patients with Fabry disease entered into the database but not receiving enzyme replacement therapy will be followed in order to gain insights into the natural history of the condition.
- FOS collects long-term effectiveness and safety data on galactosidase alfa and captures patient demographics and disease- and treatment-related variables.
- Patients with confirmed FD diagnosis are eligible for enrollment. Participating sites require ethics committee and/or regulatory approval in accordance with applicable laws.
- The first assessments occur upon entry into FOS and follow-up assessments are performed according to treating clinicians' standard schedules of care.
- Data are routinely checked to ensure highest-quality reaching data completeness is challenging as a result of and pending the heterogeneity of the investigators.

**Table 1. Key Facts on Fabry Disease**

| Incidence | Estimated to be between 1 in 40,000 to 1 in 120,000 live births; some recent publications have shown an incidence around 1 in 500 in specific populations.
| Main Symptoms | Pain, Renal involvement, Cardiovascular manifestations, Dermatological manifestations (e.g. angiokeratomas), Ocular manifestations (e.g. cornea verticillata), Other manifestations.
| Number of patients | 200+ | 10-200 | 1 | 0-50 |
| Sites participated | 1 |

**Figure 1. FOS number of patients by country**

**Figure 2. FOS manuscripts in one decade and more in development**

**Patients with Fabry disease entered into the database but not receiving enzyme replacement therapy will be followed in order to gain insights into the natural history of the condition.**

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**Data are routinely checked to ensure highest-quality reaching data completeness is challenging as a result of and pending the heterogeneity of the investigators.**

**Table 2. Key Facts on FOS (as of March 2014)**

| Started | 2001 |
| Countries | 24 |
| Sites | 171 |
| Publications | 42 |

**Objectives of FOS**

- Collect and make available information about the long-term course of FD and the long-term effectiveness and safety of galactosidase alfa treatment.
- Describe the population of patients affected with FD.
- Collect long-term data on patients with FD.
- Provide high-quality real-world clinical data, analyses and evidence to support improvements in the management of FD.
- Understand the long-term effectiveness and safety of galactosidase alfa on the course of FD.

**Figure 3. FOS publication development process**

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**Figure 4. FOS had a significant geographic congress presence in 2012 and 2013**

**Disclosures**

- All authors are employees of Shire. Poster presented at The European Conference on Rare Diseases & Orphan Products (ECRD), Berlin, Germany, May 8-10, 2014.