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Demographic characterization of Brazilian patients enrolled in the Fabry Registry

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ABSTRACT. Fabry disease (FD) is an X-linked inborn error of metabolism caused by alpha-galactosidase A deficiency. The Fabry Registry is an ongoing observational database that compiles clinical data on patients with FD. We analyzed the Fabry Registry data of patients enrolled in Brazil to characterize the demographic and baseline clinical characteristics of this patient population. As of October 2010, 126 Brazilian patients were enrolled in the Registry (61 males, 65 females). The median age at onset of symptoms in males was 9.8 years, compared to 11.4 years in females. Males were diagnosed at a median age of 31.9 years and females at 27.1 years. The median time between the onset of first symptoms and diagnosis was 20.3 years in males and 14.3 years in females. Neurologic pain was the presenting symptom most frequently reported by both genders. Renal events were the most common clinical events reported in males, while cardiac events were the most common events in females. The results of

these analyses indicate that Brazilian patients were frequently not diagnosed with FD until many years after the onset of symptoms. Many Brazilian Fabry Registry patients report experiencing neurological pain, and many Brazilian women with FD exhibit substantial signs and symptoms. The prevalence of neurological pain as a presenting symptom among Brazilian Registry patients is consistent with previous reports from the overall Registry population. FD is treatable, and earlier diagnosis will allow for prompt initiation of appropriate treatment that may avert irreversible damage that could occur during the time from symptom onset to diagnosis.

Key words: Fabry disease; Alpha-galactosidase A deficiency; Fabry Registry; Brazil; Demographics

