Fabry disease is caused by mutations in the GLA gene encoding lysosomal enzyme α-galactosidase A (α-GalA). Absent or deficient α-GalA activity results in progressive accumulation of globotriaosylceramide and related glycosphingolipids within lysosomes in a variety of cell types throughout the body. The spectrum of clinical presentations of Fabry disease is wide. At the severe end of the spectrum, the classic Fabry phenotype comprises a multisystemic presentation with skin, neurologic, gastrointestinal, renal, cardiac, and cerebrovascular symptoms.1

The p.N215S (p.Asn215Ser) genotype has been labelled as a late-onset cardiac variant although the classic Fabry phenotype comprises a multisystemic presentation with skin, neurologic, gastrointestinal, renal, cardiac and cerebrovascular symptoms.1

In males, median IVST was >12 mm in the 25-34 years age group (median 13.0 mm, range 9.0-15.0 mm, abnormal in 60% of males in this group) (Fig. 1). Median LVPWT was abnormal (>12 mm) in the 35-44 years age group (median 13.0 mm, range 9.0-16.4 mm, 56% abnormal) (Fig. 2). Median IVST and LVPWT values in males increased in later decades (Figs. 1 and 2).

In females, median IVST and LVPWT values remained within the normal range for all age categories, except for IVST in patients aged 65-74 years (median 12.3 mm, range 10.0-14.0 mm, 50% abnormal) (Figs. 1 and 2). Abnormal LVPWT and/or IVST values (>12 mm) were found before the age of 65 in 12% of the females, possibly due to skewed X-chromosome inactivation.2

Agsidasiase beta (Fabrazyme®), Sanofi Genzyme, is indicated for use in patients with Fabry disease.3

The global Fabry Registry (NCT00196742; sponsored by Sanofi Genzyme) was initiated in 2001 as part of an effort to help healthcare professionals involved in the treatment or diagnosis of Fabry disease better understand the disease and its management and to help create Fabry disease treatment monitoring guidelines. Patient participation in the Fabry Registry is voluntary. Each independent site is responsible for obtaining a patient’s informed consent to submit his/her health information to the Registry and to use and disclose this information in subsequent aggregate analyses, such as journal articles, annual reports, education materials, and public health reports.

D.P.G.: Travel grants and honoraria for lectures on Fabry disease from Sanofi Genzyme, Shire HGT and Amicus Therapeutics; E.B.: Speaker honoraria from Sanofi Genzyme and Shire HGT; F.C.: Honoraria for lectures and research grants from Sanofi Genzyme; J.K.: Sanofi Genzyme employee; D.A.L.: Member of the Fabry Registry Board. Consults with FibroGen; D.F.U.: Consultant for Sanofi Genzyme; S.S.: Research funds and travel support from Sanofi Genzyme; R.K.: Consultant for Sanofi Genzyme; M.H.: Sanofi Genzyme employee; D.J.: Speaker honoraria and travel support from Sanofi Genzyme; S.G.: Consultant for Sanofi Genzyme; A.J.: Speaker honoraria for lectures from Sanofi Genzyme; R.J.: Consultant for Sanofi Genzyme; F.B.: Speaker honoraria from Sanofi Genzyme and FibroGen; J.A.: Consultant for Sanofi Genzyme; L.M.: Research fund and travel support from Sanofi Genzyme; Shire HGT; A.L.: Consultant for Sanofi Genzyme; E.B.: Speaker honoraria from Shire HGT and FibroGen; J.O.: Consultant for Sanofi Genzyme; E.B.: Member of the Fabry Registry and has received honoraria from Sanofi Genzyme, Shire HGT and Amicus Therapeutics; A.G.: Member of the Fabry Registry and has received research grants from Sanofi Genzyme, Shire HGT and Amicus Therapeutics. These activities have been monitored and found to be in compliance with the conflict of interest policies at Emory University School of Medicine; S.T.: Member of the Fabry Registry and has received research grants and honoraria from Sanofi Genzyme, Shire HGT and Amicus Therapeutics; S.G.: Consultant for Sanofi Genzyme, Shire HGT and Amicus Therapeutics; A.J.: Member of the Fabry Registry Board and has received advisory board fees and honoraria for lectures on Fabry disease from Sanofi Genzyme, Shire HGT and Amicus Therapeutics.

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The authors are responsible for all content of this poster.