

5. Morrissey et al. Cardiac abnormalities in Anderson-Fabry disease and Fabry's cardiomyopathy. *Cardiovasc J Afr.* 2011;22:38–44;
6. Eng CM, Guffon N, Wilcox WR, et al. Safety and efficacy of recombinant human alpha-galactosidase A replacement therapy in Fabry's disease. *N Engl J Med.* 2001;345:9-16.
7. Elliott P, Baker R, Pasquale F, Quarta G, Ebrahim H, Mehta AB, et al. Prevalence of Anderson-Fabry disease in patients with hypertrophic cardiomyopathy: the European Anderson-Fabry Disease survey. *Heart* 2011;97(23):1957-60.
8. Hagege AA, Caudron E, Damy T, et al. Screening patients with hypertrophic cardiomyopathy for Fabry disease using a filter-paper test: the FOCUS study. *Heart.* 2011;97:131-136.
9. Terryn W, Deschoenmakere G, De Keyser J, Meersseman W, Van Biesen W, Wuyts B, et al. Prevalence of Fabry disease in a predominantly hypertensive population with left ventricular hypertrophy. *Int J Cardiol* 2013;167(6):2555-60.
10. Palecek T, Honzikova J, Poupetova H, Vlaskova H, Kuchynka P, Golan L, et al. Prevalence of Fabry disease in male patients with unexplained left ventricular hypertrophy in primary cardiology practice: prospective Fabry cardiomyopathy screening study (FACSS). *J Inherit Metab Dis* 2014;37(3):455-60.
11. Vieitez I, Souto-Rodriguez O, Fernandez-Mosquera L, et al. Fabry disease in the Spanish population: observational study with detection of 77 patients. *Orphanet J Rare Dis.* 2018 Apr 10;13(1):52.
12. Maron MS, Xin W, Sims KB, Butler R, Haas TS et al. Identification of Fabry Disease in a Tertiary Referral Cohort of Patients with Hypertrophic Cardiomyopathy. *Am J Med.* 2018 Feb;131(2):200.e1-200.e8.
13. Ommen SR, Nishimura RA, Edwards WD. Fabry disease: a mimic for obstructive hypertrophic cardiomyopathy? *Heart* 2003;89:929–30.
14. Recommendations for Cardiac Chamber Quantification by Echocardiography in Adults: An Update from the American Society of Echocardiography and the European Association of Cardiovascular Imaging, *Journal of the American Society of Echocardiography* January 2015
15. Echevarria L, Benistan K, Toussaint A, et al. X-chromosome inactivation in female patients with Fabry disease. *Clin Genet.* 2016;89:44-54.
16. Mehta et al. Natural course of Fabry disease: changing pattern of causes of death in FOS - Fabry Outcome Survey *J Med Genet.* 2009;46:548–52;
17. Meikle PJ, Hopwood JJ, Clague AE, Carey WF. Prevalence of lysosomal storage disorders. *J Am Med Assoc* 1999; 281:249–54.
18. Kampmann C, Perrin A, Beck M. Effectiveness of agalsidase alfa enzyme replacement in Fabry disease: cardiac outcomes after 10 years' treatment. *Orphanet J Rare Dis.* 2015 Sep 29;10:125. doi: 10.1186/s13023-015-0338-2.
19. Schiffman R, Kopp JB, Austin 3rd HA, Sabnis S, Moore DF, Weibel T, Balow JE, Brady RO. Enzyme replacement therapy in Fabry disease: a randomized controlled trial. *JAMA* 2001;285:2743–9.

Table 1 Clinical characteristics of study patients.

	HCM (n=80)
Male / Female, n (%)	53 (66%) / 27 (34%)
Age	41.5 ± 12.7
Ejection fraction (%)	60.7 ± 7.4
Interventricular septal wall thickness (mm)	18.2 ± 4.4
Posterior wall thickness (mm)	13.5 ± 2.1
LV end-diastolic diameter (mm)	47.4 ± 6.2
LV end-systolic diameter (mm)	27.8 ± 6.5
Left atrial diameter (mm)	36.7 ± 8.5
Systolic pulmonary artery pressure (mmHg)	34.5 ± 9.8
LV mass index (LVMI)(g/m ²)	171.05 ± 48.5

Data are shown as mean ± SD (range). HCM, hypertrophic cardiomyopathy; LV, left ventricular.

Table 2 Clinical characteristics and genetic results in patients with Fabry disease.

	Patient 1	Patient 2
Plasma a-Gal A activity ($\mu\text{mol/L/h}$) *	0.5 $\mu\text{mol/L/h}$	0.4 $\mu\text{mol/L/h}$
Lyso-Gb3 (ng/mL) #	7.8	5.10
Gender	Male	Male
Age (years)	49	38
Pattern of LVH	Concentric	Concentric
Interventricular wall thickness (mm)	18	20
Posterior wall thickness (mm)	18	20
LV end-diastolic diameter (mm)	52	54
LV end-systolic diameter (mm)	37	42
Ejection fraction (%)	60	44
LV mass index	241	278
Acroparesthesia	+	+
Angiokeratoma	-	-
Hypohydrosis	-	-
Corneal opacities	-	-
Proteinuria	+	+
Cerebral infarction	-	-
Mutations - Missense	p.R112C	p.R301Q
Nucleotide change	c.334C>T	c.902>A

*Reference value ≥ 3.3 $\mu\text{mol/L/hour}$. #Reference value $\leq 1.8\text{ng/mL}$.

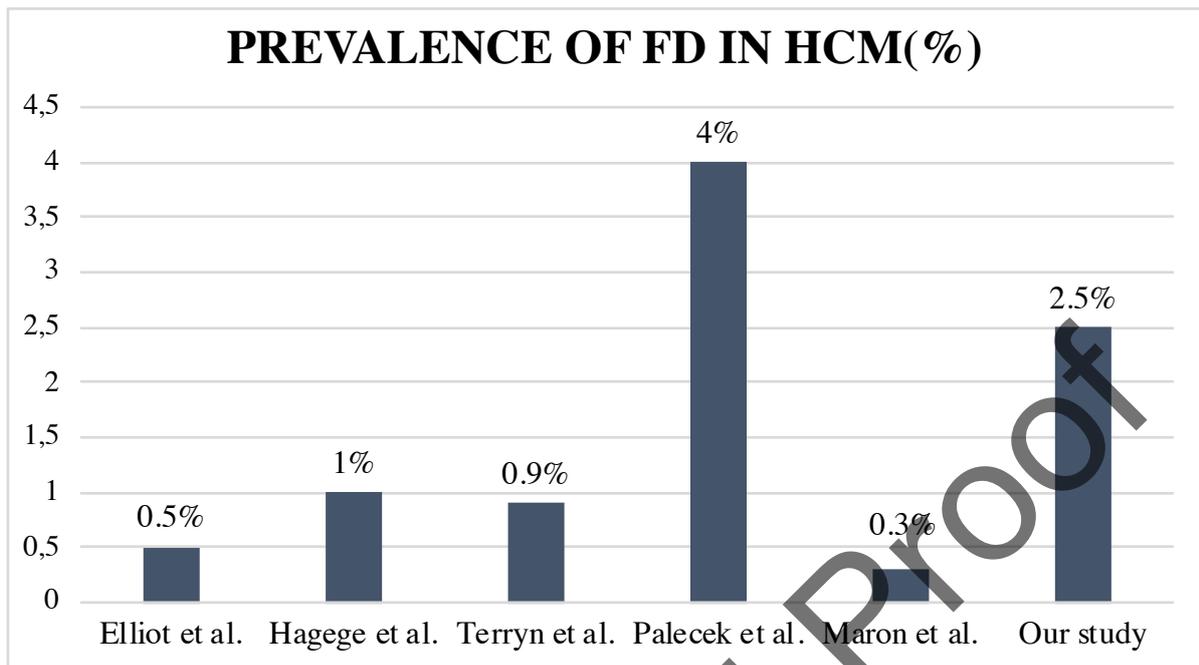


Figure 1. Prevalence of Fabry disease in various populations with hypertrophic cardiomyopathy patients

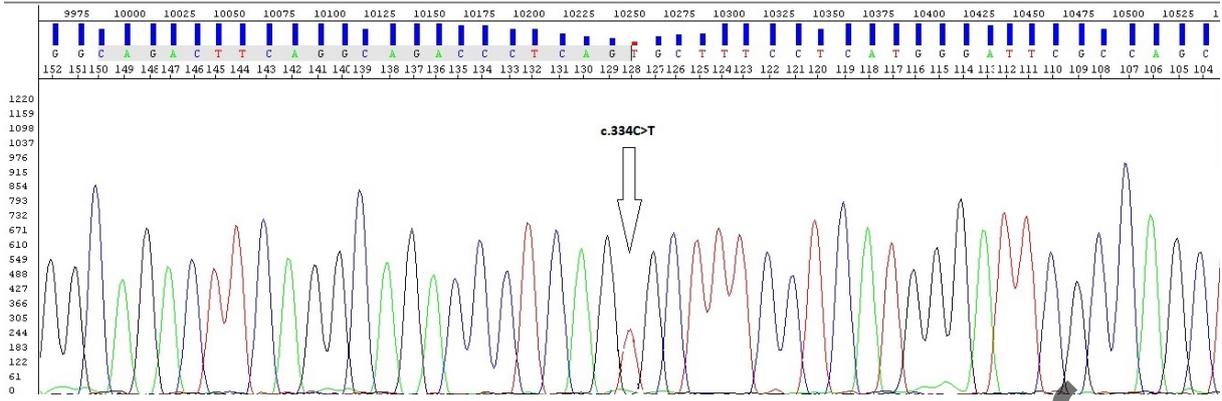


Figure 2.

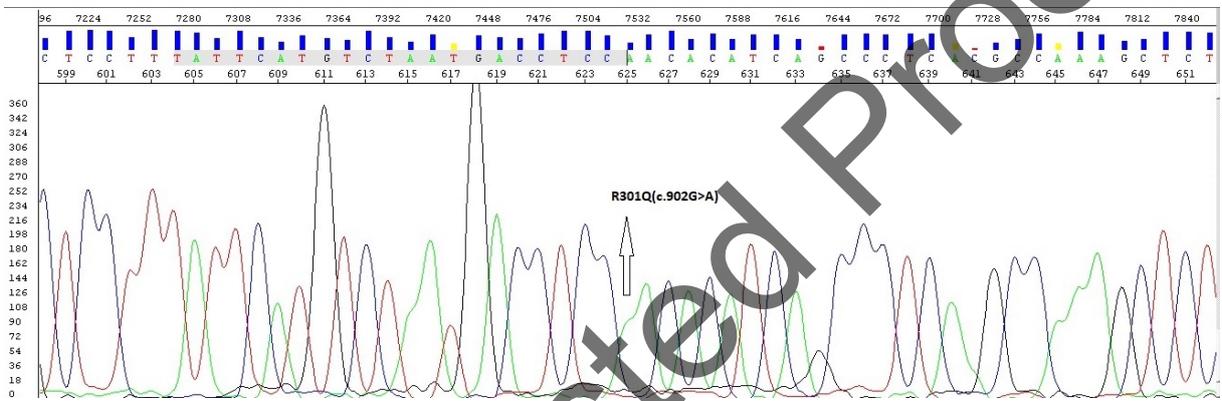


Figure 3.

Uncorrected Proof