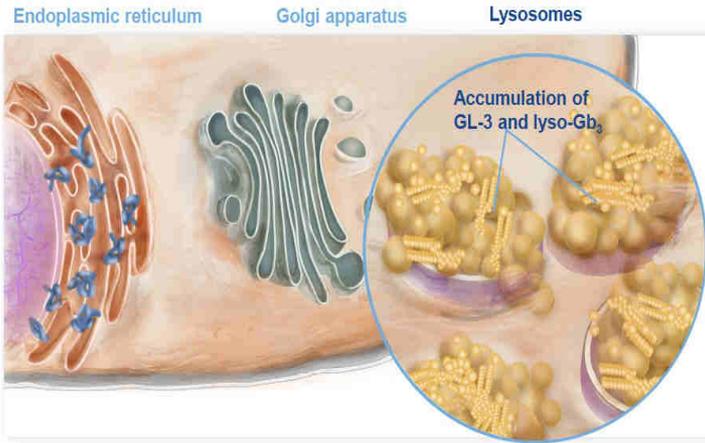




Visió del cardiòleg en la malaltia de Fabry

Jaume Pons Llinares

Cardiologia HUSE

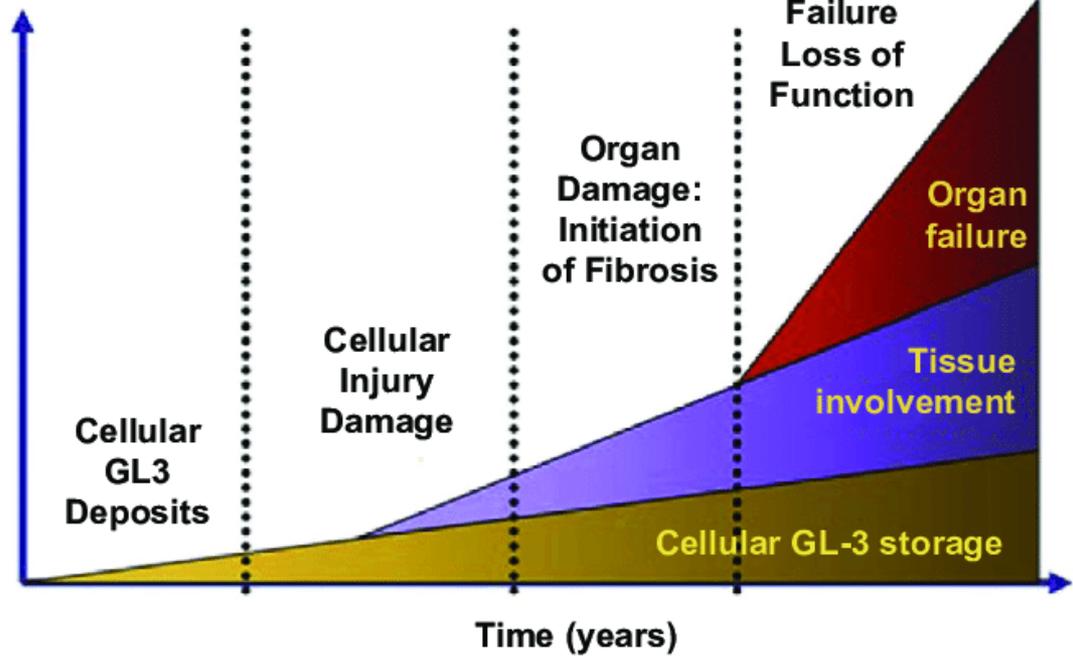


Stage 1
Burden
of Disease

Stage 2

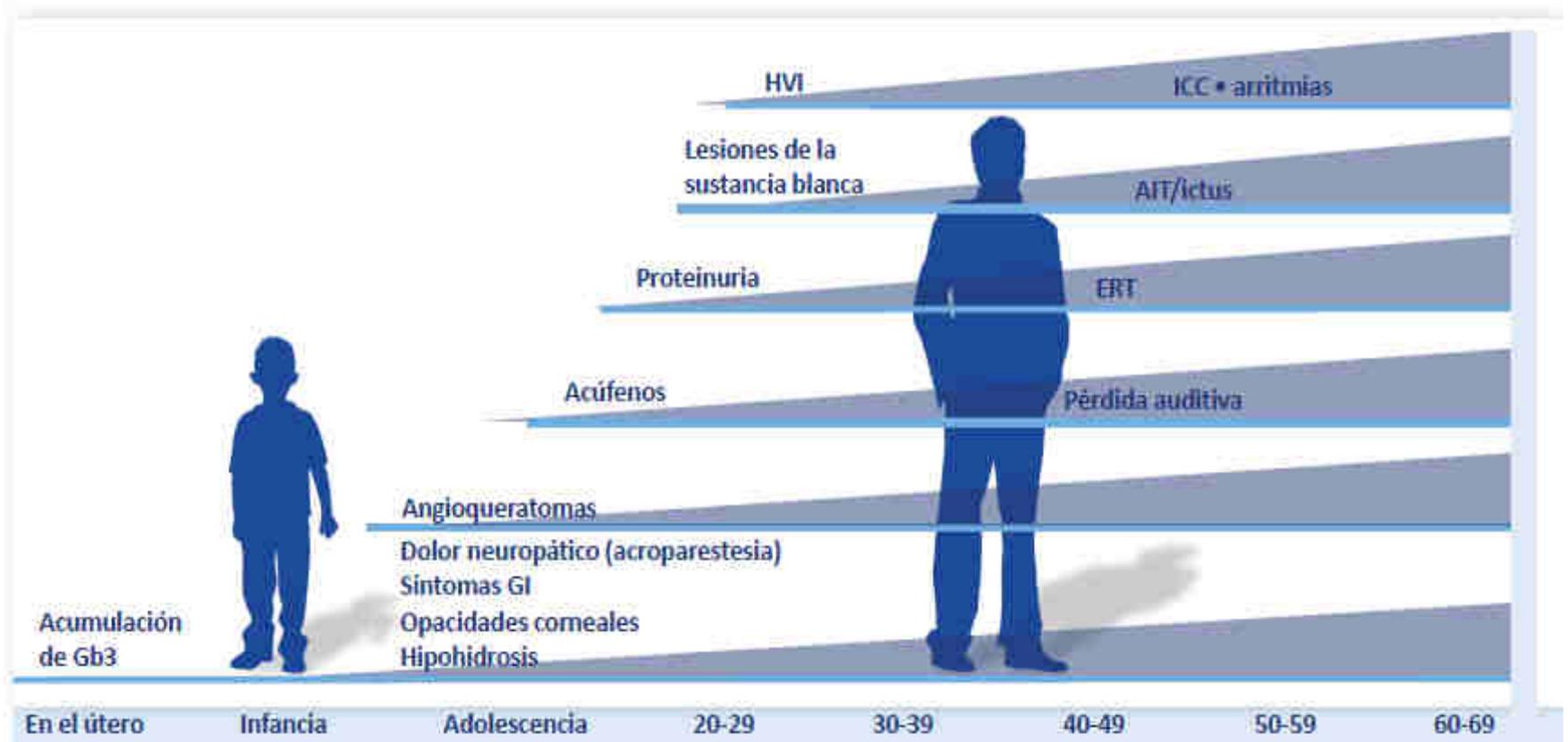
Stage 3

Stage 4



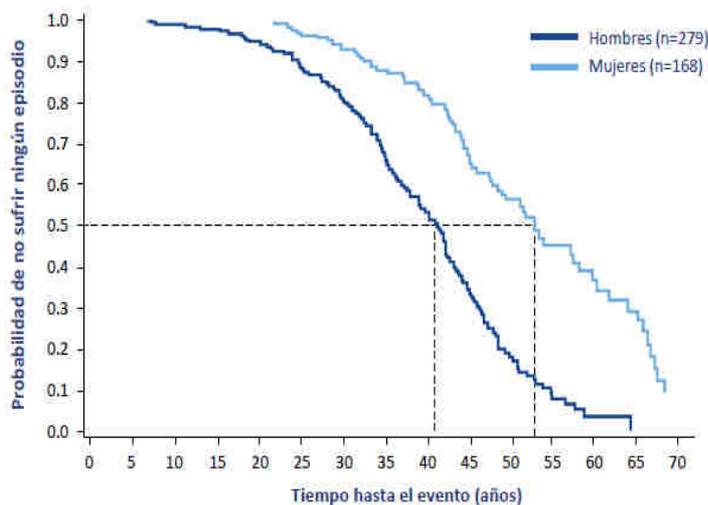
Time (years)

1.5% a 4% Fabry en Hipertrofia VI inexplicada



Fabry Outcome Survey (FOS) (FOS)

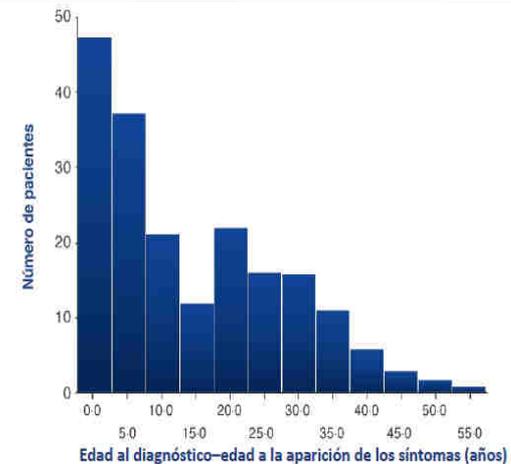
Estimación de Kaplan-Meier del tiempo hasta el primer episodio renal o cardíaco, ictus o muerte (N = 447)



Extraído de Schiffmann R et al. *Nephrol Dial Transplant.* 2009;24(7):2107.

50% varones sufre un episodio renal o cardíaco, ictus o muere en torno a los 41 años

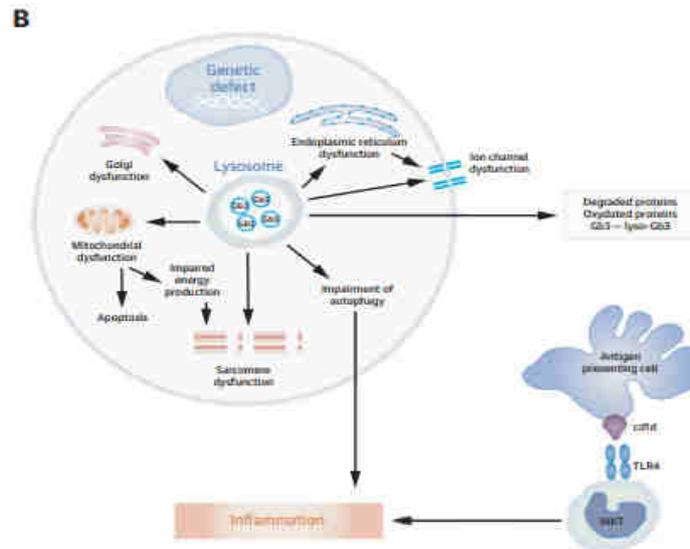
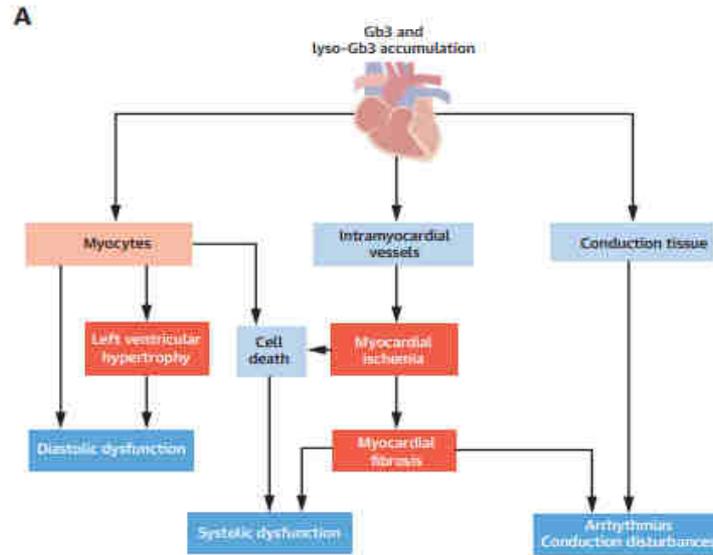
Retraso del diagnóstico desde la aparición de los síntomas en el registro Fabry Outcome Survey (n = 194)



Extraído de Mehta A et al. *Eur J Clin Invest.* 2004;34(3):238.

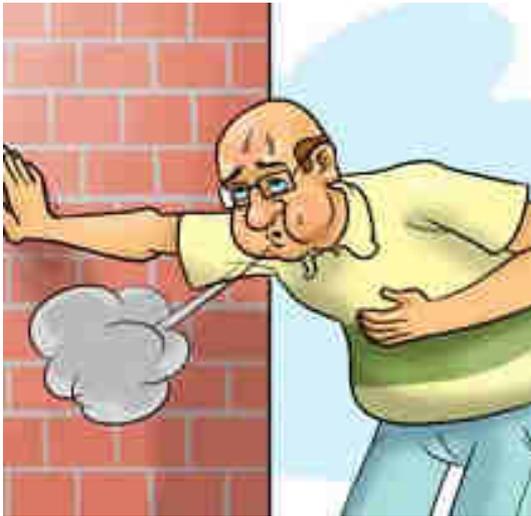
T medio entre síntomas y el diagnóstico fue de 13,7 y 16,3 años para hombres y mujeres.

Fisiopatología



Clínica

Disnea



Palpitaciones

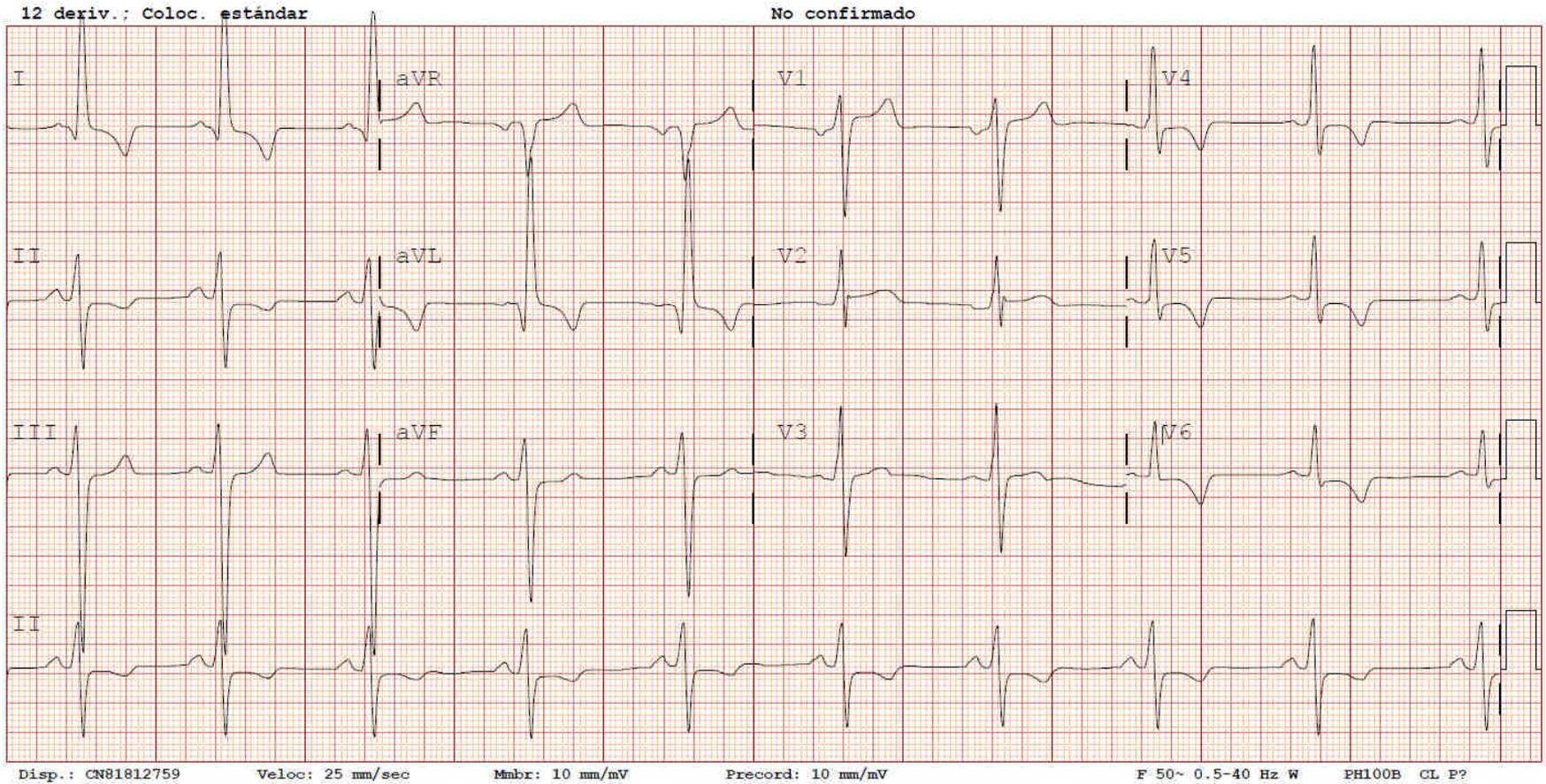


Angina

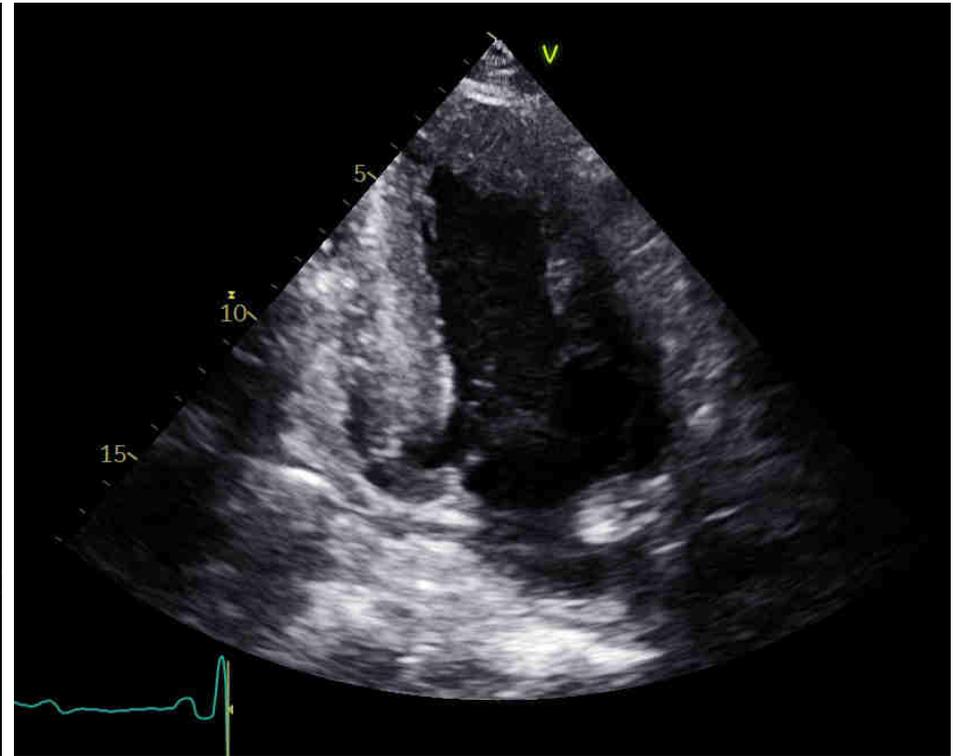
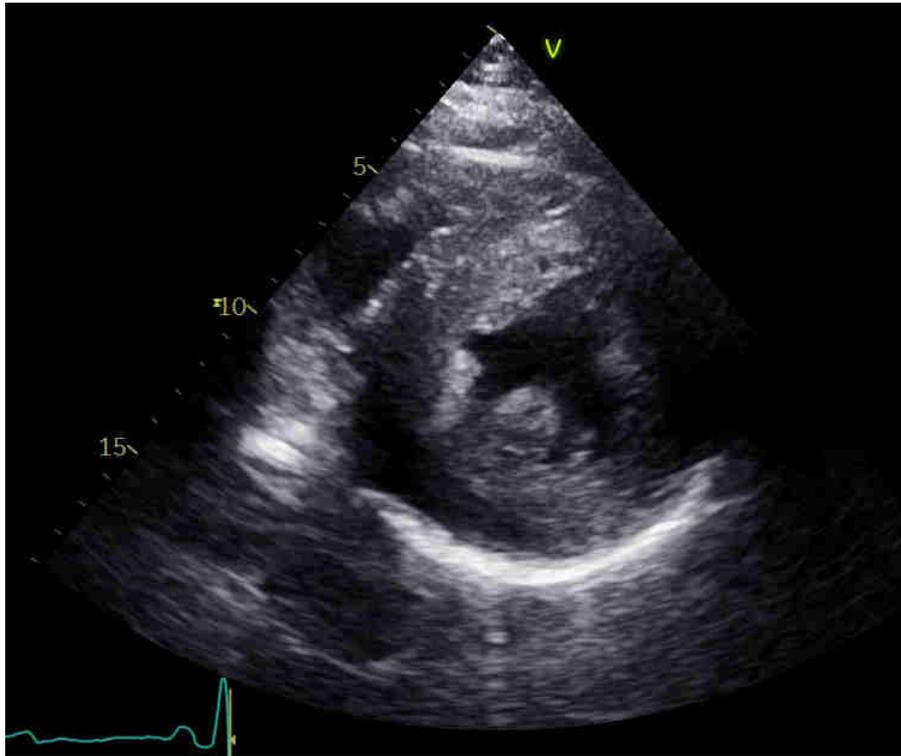


Sincope

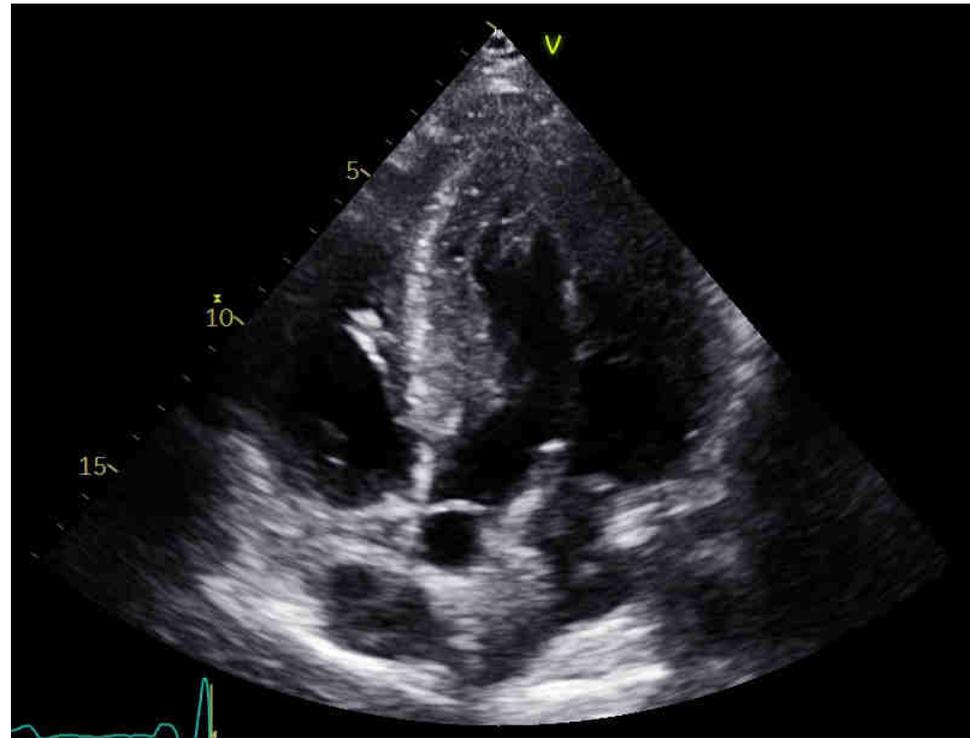
ELECTROCARDIOGRAMA



ECOCARDIOGRAMA



ECOCARDIOGRAMA

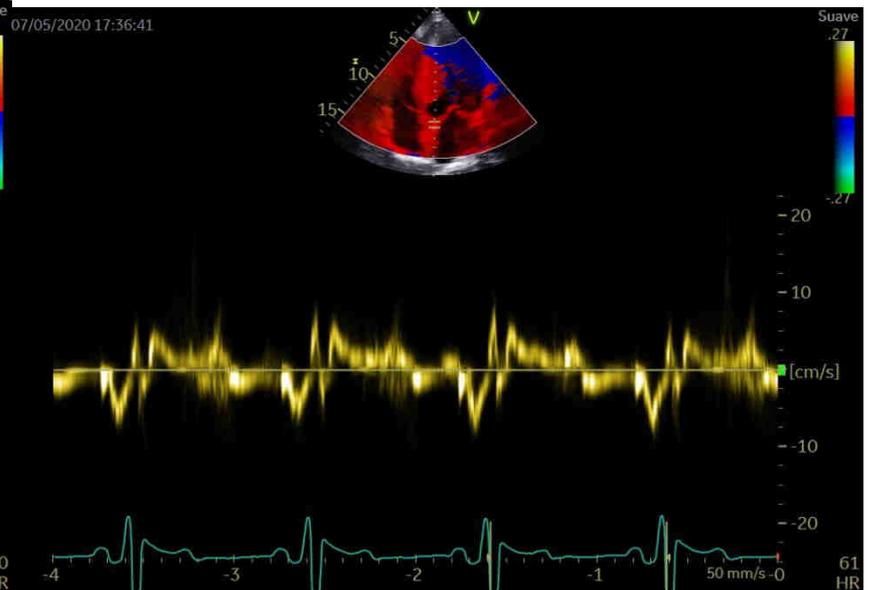


ECOCARDIOGRAMA

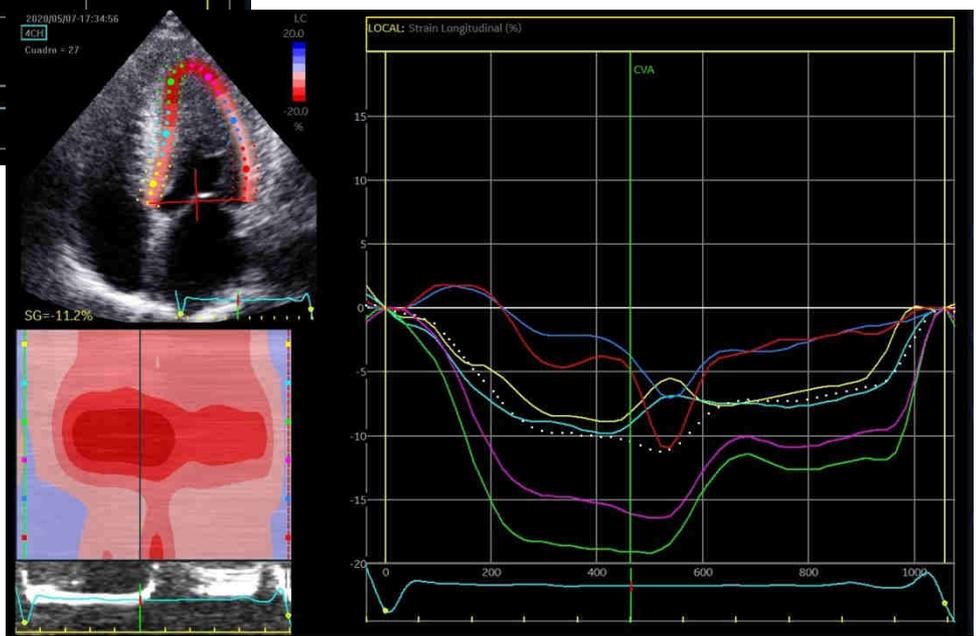
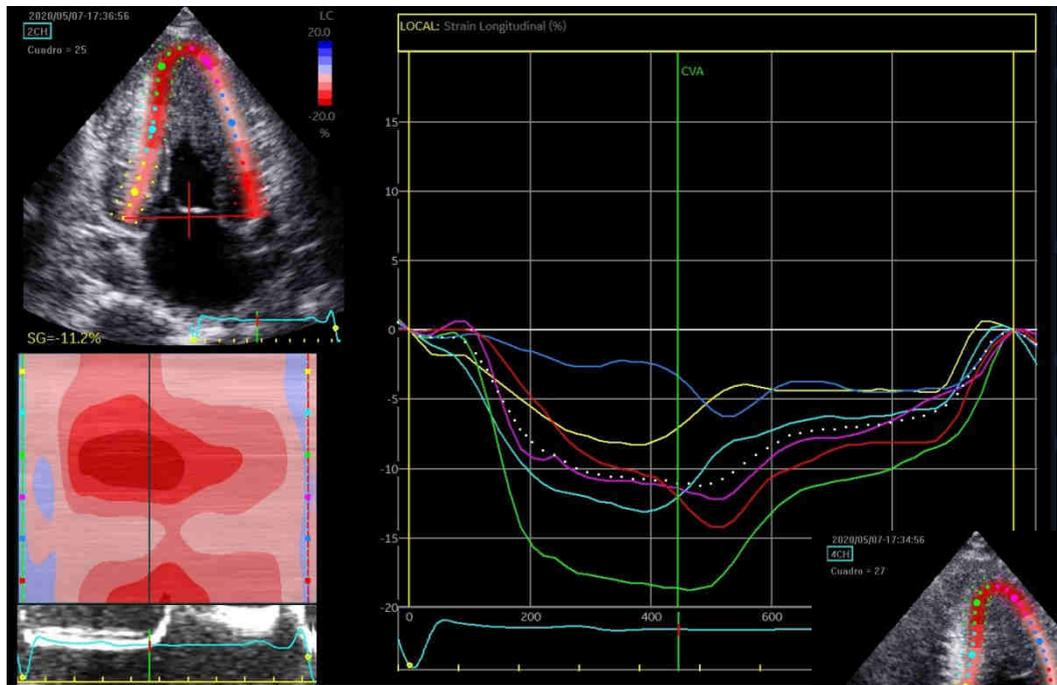


Diastole

Presiones de llenado VI

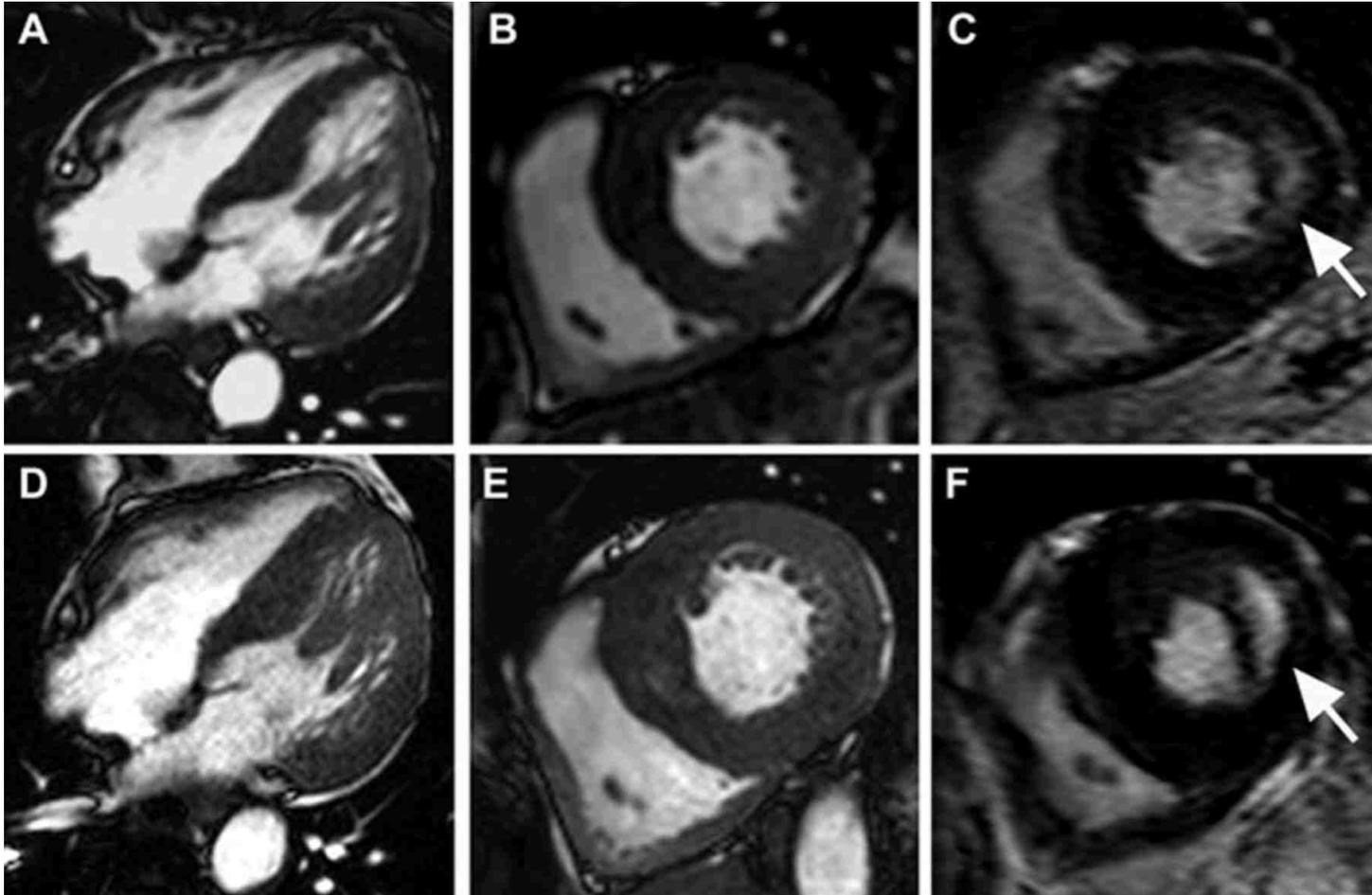


ECOCARDIOGRAMA



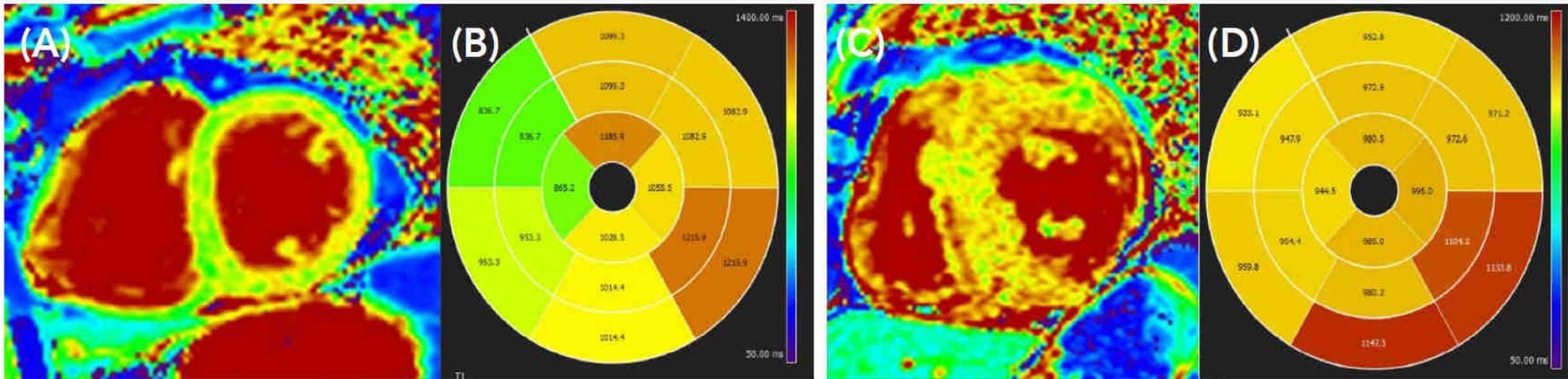
Strain Longitudinal

RNM CARDÍACA



RNM CARDÍACA

Imágenes T1 mapping en diferentes estadios de Enfermedad de Fabry



RNM CARDÍACA

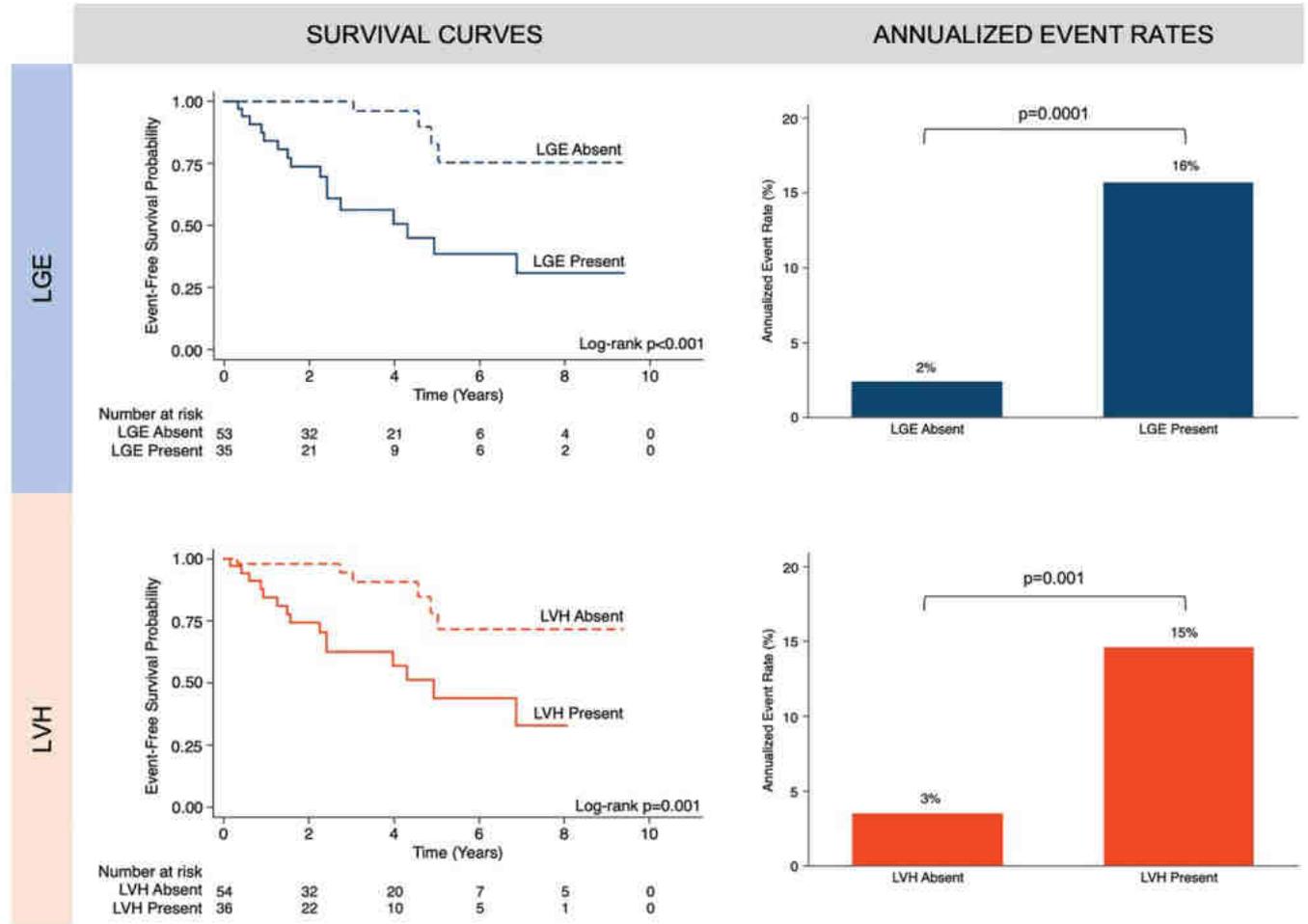
N=90

TV

Bradicardia/MCP

ICC

Muerte CV



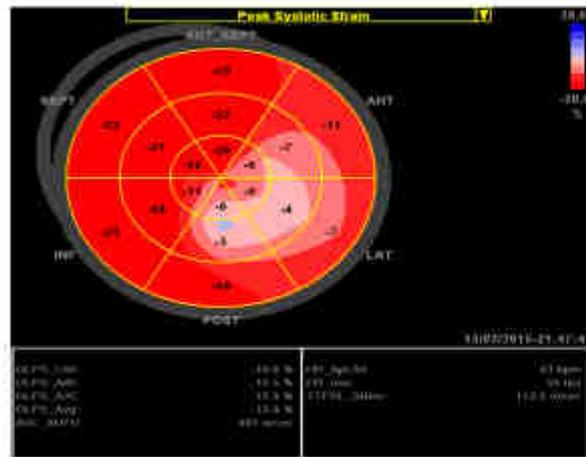
Intermediate Cardiomyopathy



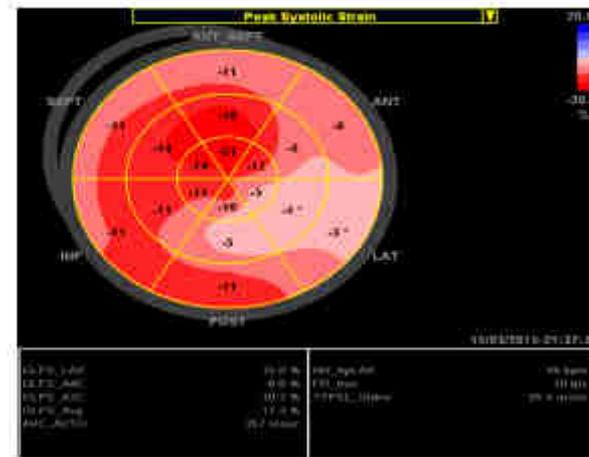
Advanced Cardiomyopathy



Early Cardiomyopathy:



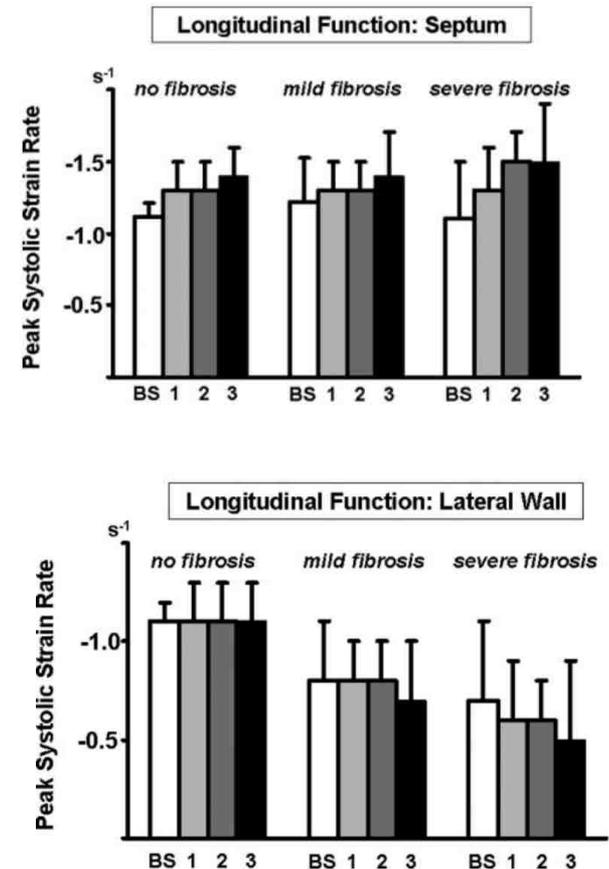
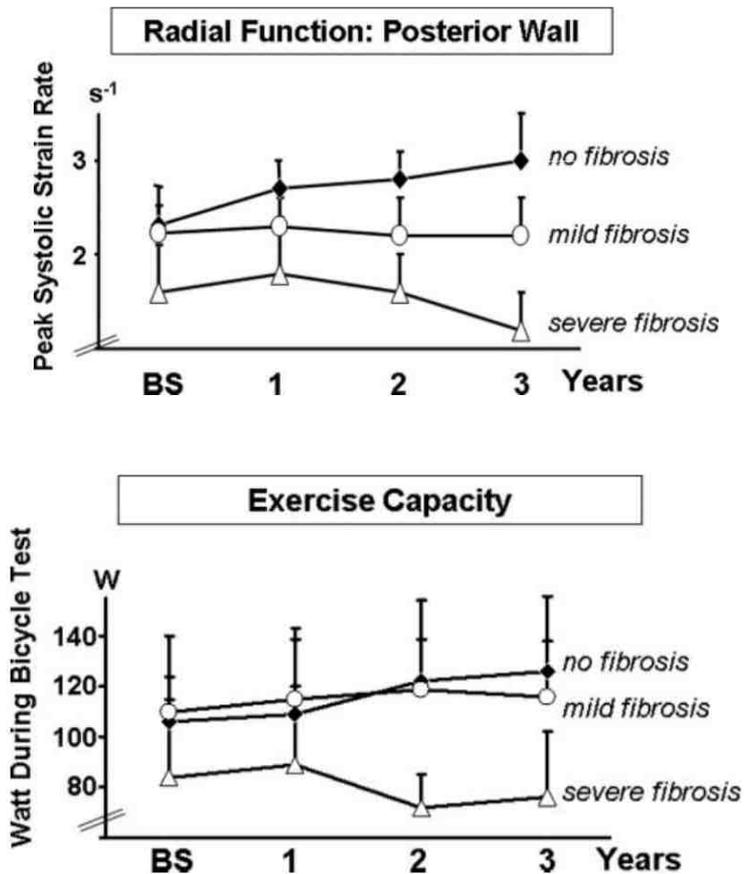
Advanced Cardiomyopathy:



Long-Term Effects of Enzyme Replacement Therapy on Fabry Cardiomyopathy

Evidence for a Better Outcome With Early Treatment

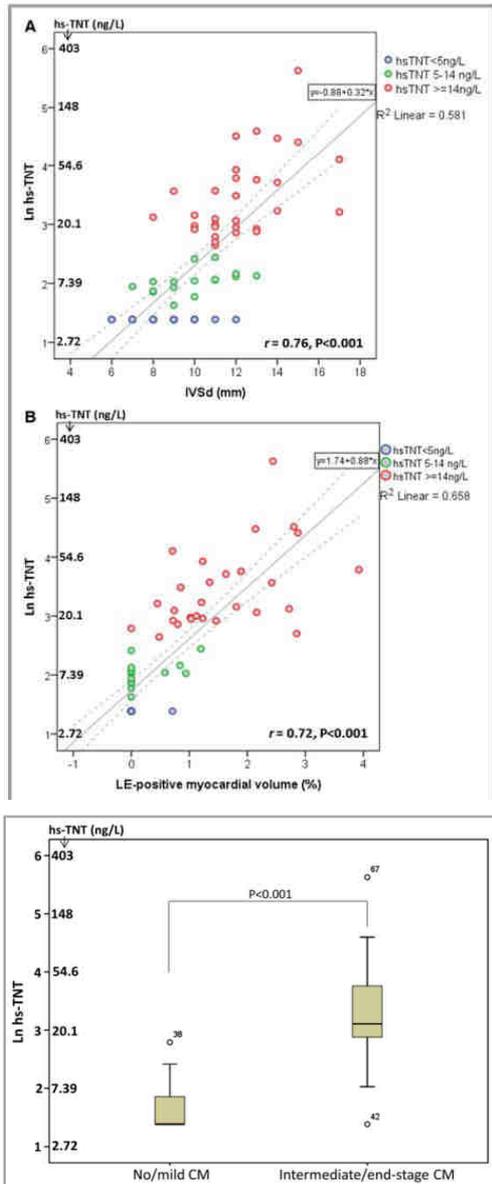
Frank Weidemann, MD*; Markus Niemann*; Frank Breunig, MD; Sebastian Herrmann;
 Meinrad Beer, MD; Stefan Störk, MD; Wolfram Voelker, MD; Georg Ertl, MD;
 Christoph Wanner, MD; Jörg Strotmann, MD



(Circulation. 2009;119:524-529.)

High-Sensitivity Troponin: A Clinical Blood Biomarker for Staging Cardiomyopathy in Fabry Disease

Nora Seydelmann, MD; Dan Liu, MD; Johannes Krämer, MD; Christiane Drechsler, MD; Kai Hu, MD; Peter Nordbeck, MD; Andreas Schneider, MD; Stefan Störk, MD, PhD; Bart Biinens, PhD; Georg Ertl, MD; Christoph Wanner, MD; Frank Weidemann, MD



J Am Heart Assoc. 2016;5:e002839

BIOMARCADORES

Role of Serum N-Terminal Pro-Brain Natriuretic Peptide Measurement in Diagnosis of Cardiac Involvement in Patients With Anderson-Fabry Disease

Caroline J. Coats, MBBS^{a,b}, Valentina Parisi, MD^a, Monica Ramos, MD^a, Kalaiarasi Janagarajan, MSc^a, Constantinos O'Mahony, MBBS^a, Anne Dawnay, PhD^b, Robin H. Lachmann, FRCP^c, Elaine Murphy, MBBCh^c, Atul Mehta, MD^d, Derralynn Hughes, DPhil^d, and Perry M. Elliott, MD^a

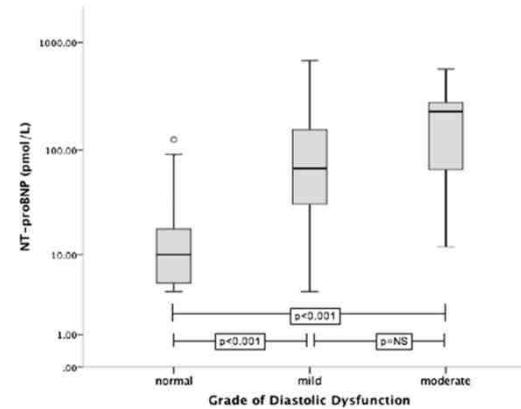
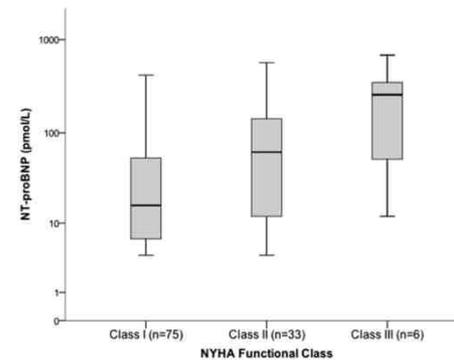


Figure 1. NT-proBNP increases with age in men and women (n = 114).

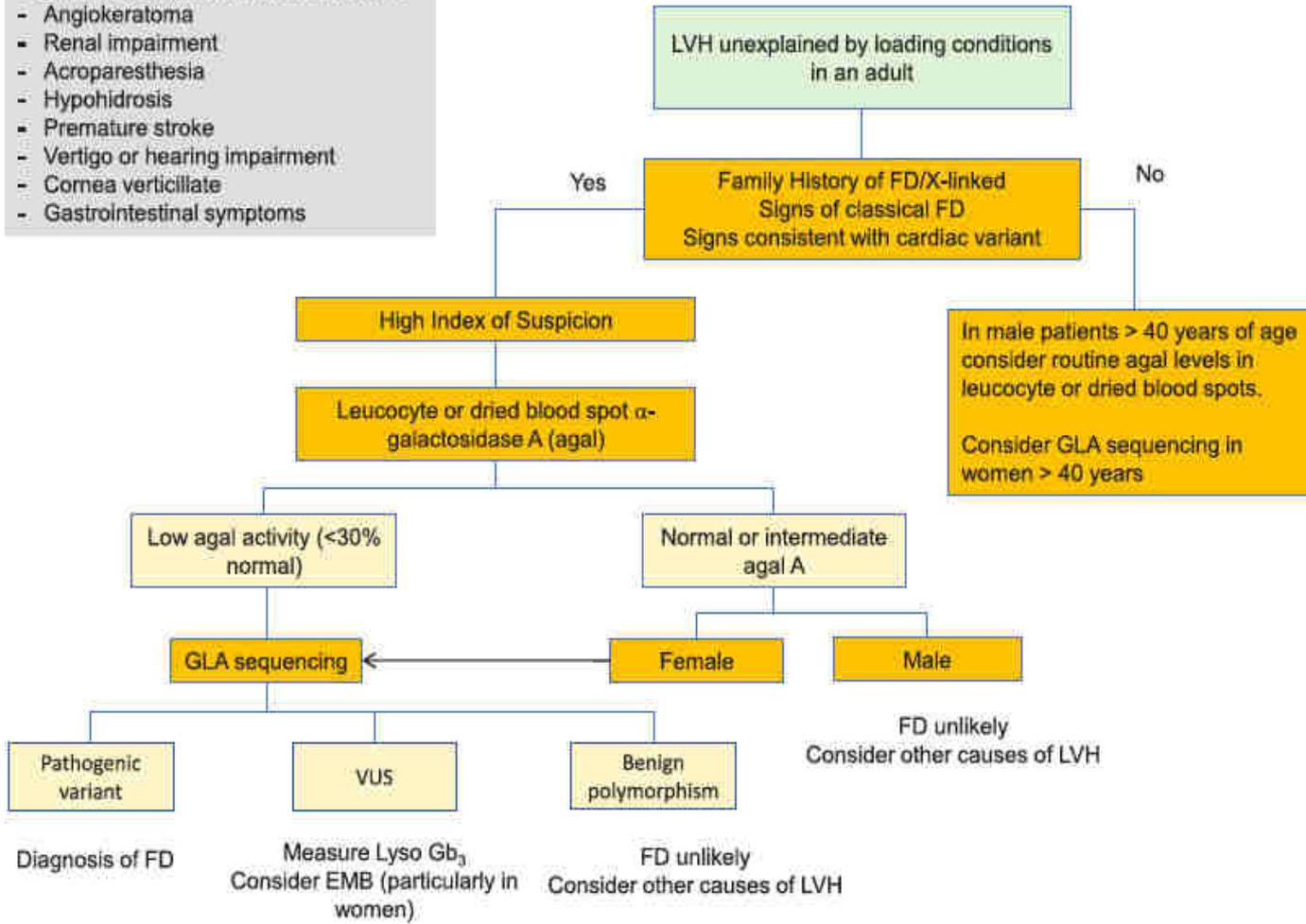


Am j Cardiol 2013;111:111-117

DIAGNÓSTICO

Signs and symptoms of classical FD:

- Angiokeratoma
- Renal impairment
- Acroparesthesia
- Hypohidrosis
- Premature stroke
- Vertigo or hearing impairment
- Cornea verticillate
- Gastrointestinal symptoms



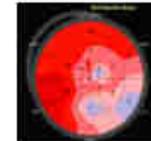
Cardiac Signs of FD



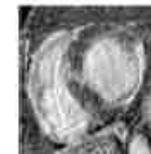
Short PR
Conduction disease



Concentric LVH
RVH
Papillary muscle hypertrophy



Reduced GLS in posterior wall



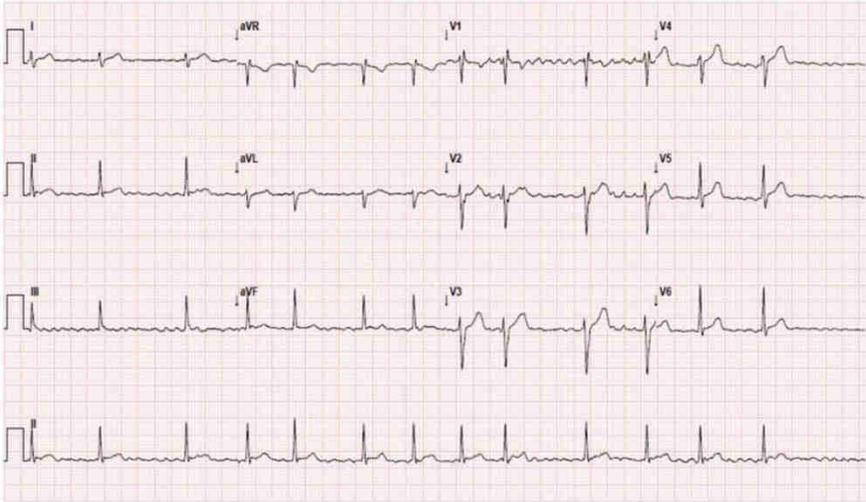
Posterior wall scar
Low T1

Fibrilación auricular

Intentar control del ritmo

ANTICOAGULAR !!! ACOD

Betabloqueantes

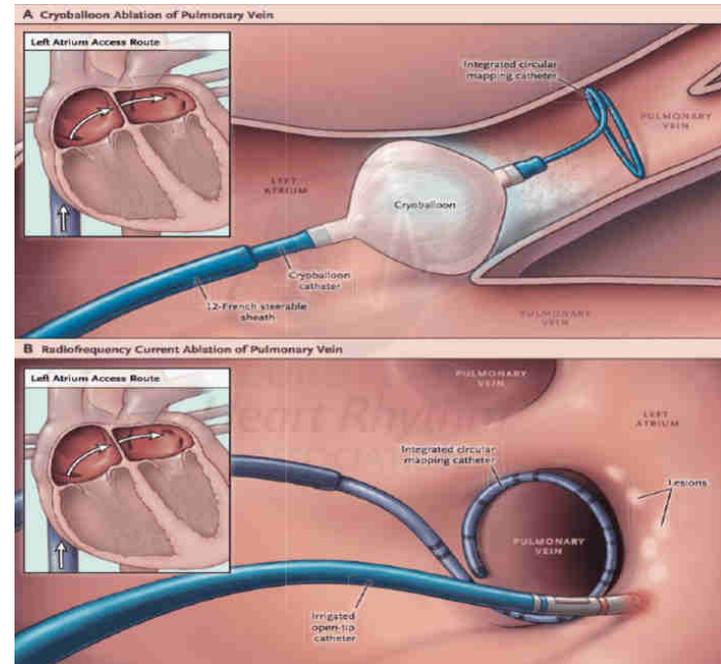


~~Flecainida~~

~~Propafenona~~

~~Amiodarona~~

~~Dronedaron~~



Insuficiencia Cardíaca

- 25% en registros
- Diagnóstico diferencial y específico de la disnea
- Tratamiento específico
 - HeFReF
 - IECA/ARA No evidencia ARNI
 - Bbloc/Ivabradina con precaución
 - Antialdosterónico con precaución
 - ¿ISGLT2?
 - HeFPeF
 - Antialdosterónico con precaución
 - ¿ISGLT2?
- Estimulación biventricular

CENTRAL ILLUSTRATION Proposed Evolution of Cardiac Involvement in Fabry Disease

