Case: 52yr / M

- C.C: Palpitations for three years
- Aggravated with syncope for over two months

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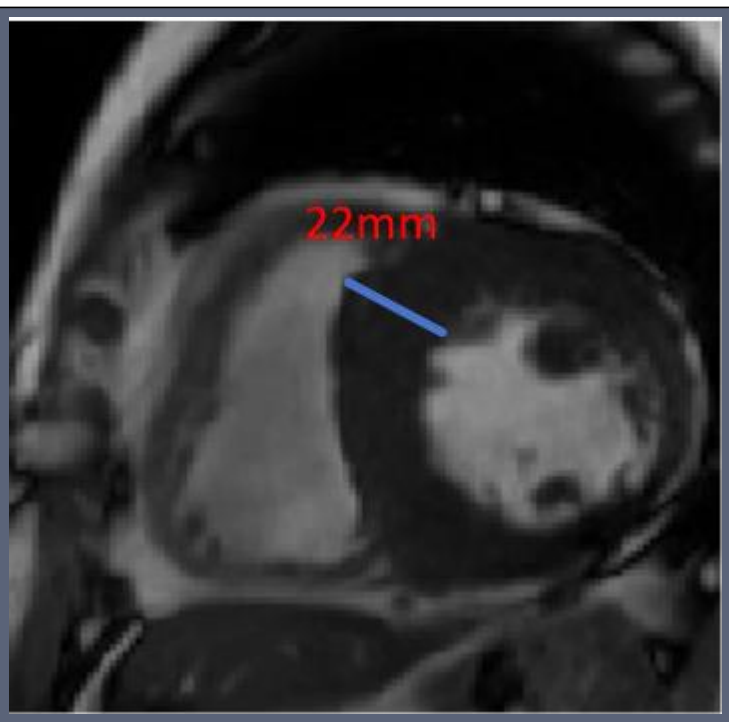
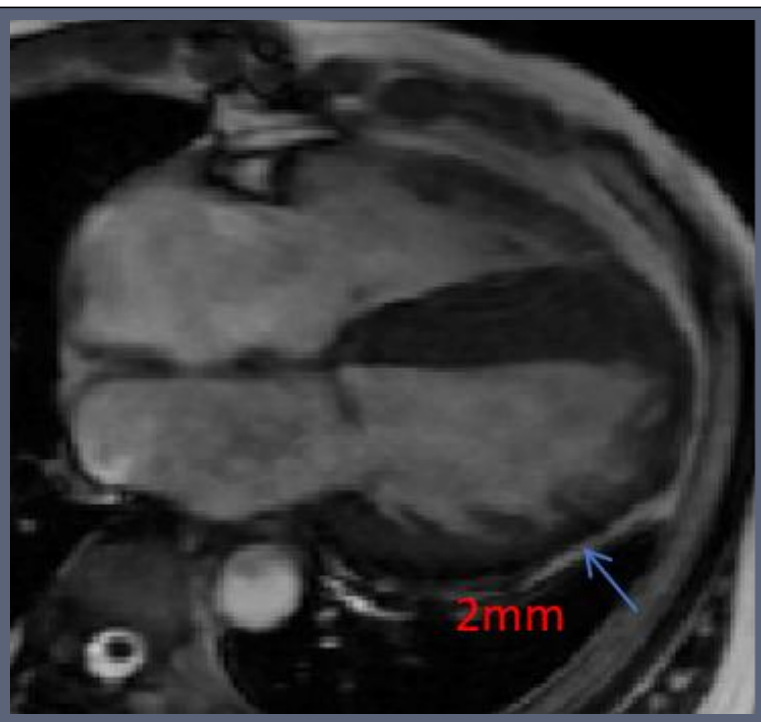
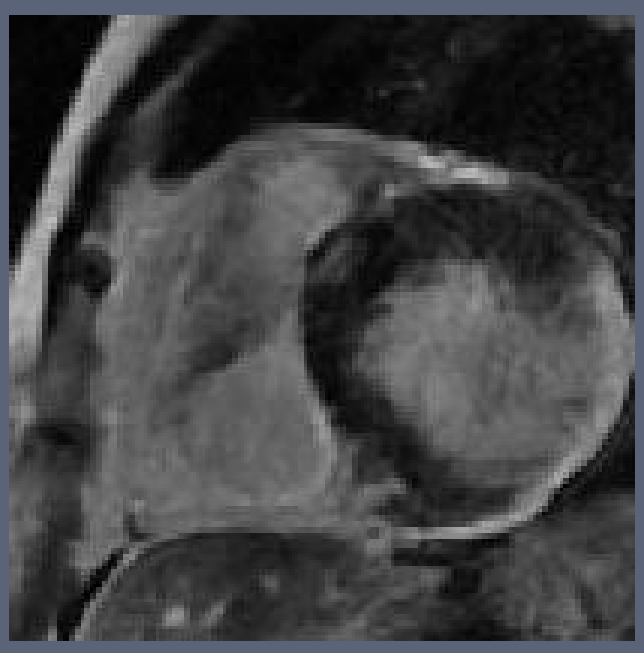
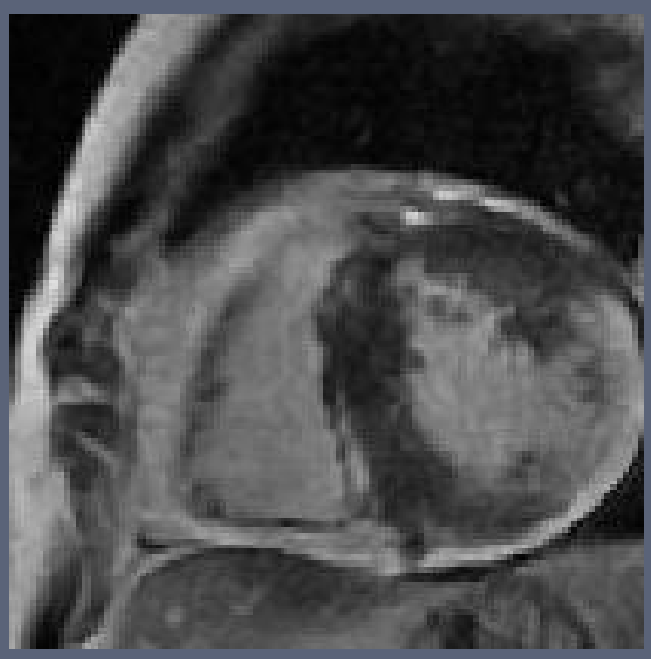
Cine CMR

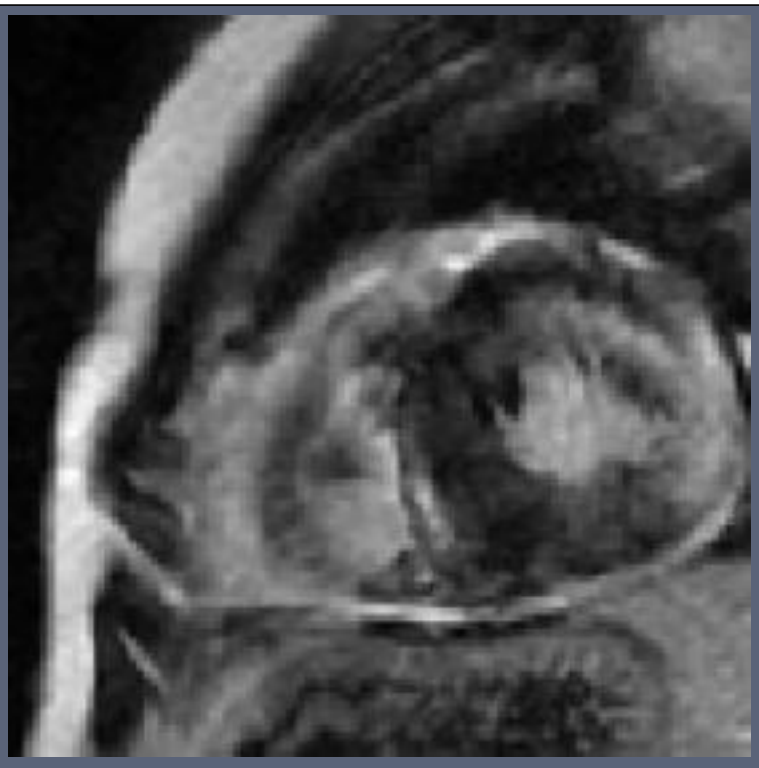


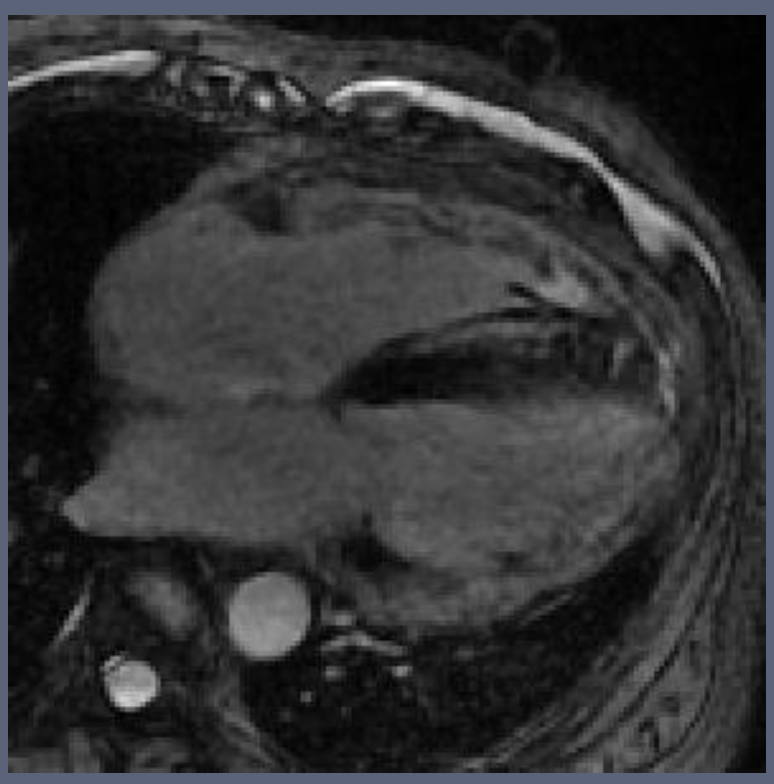


LGE







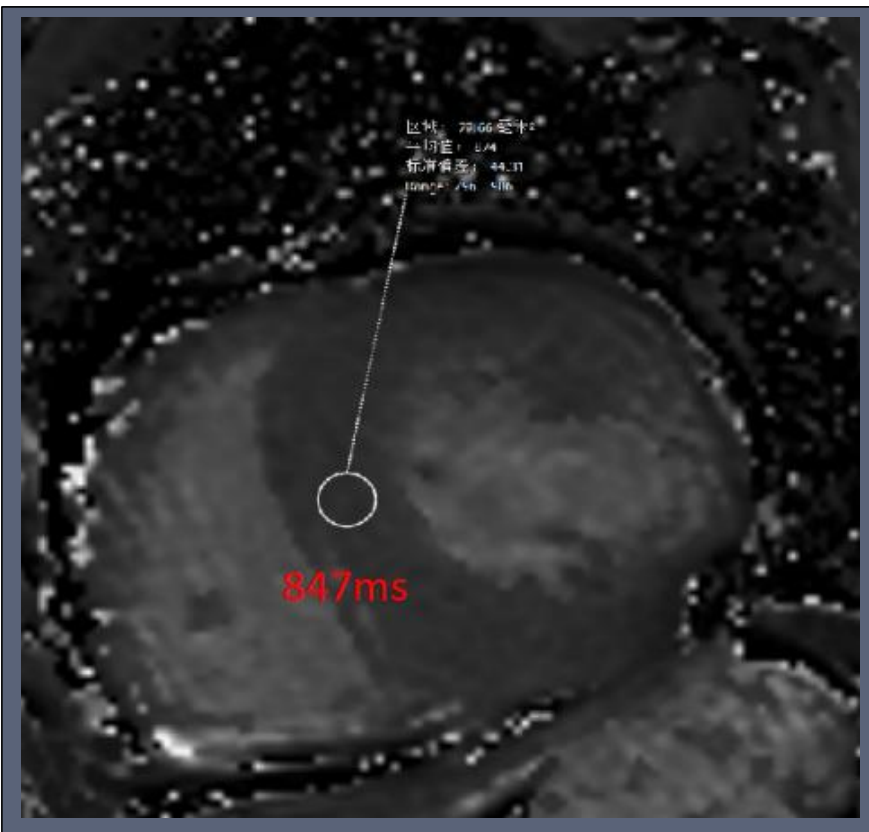


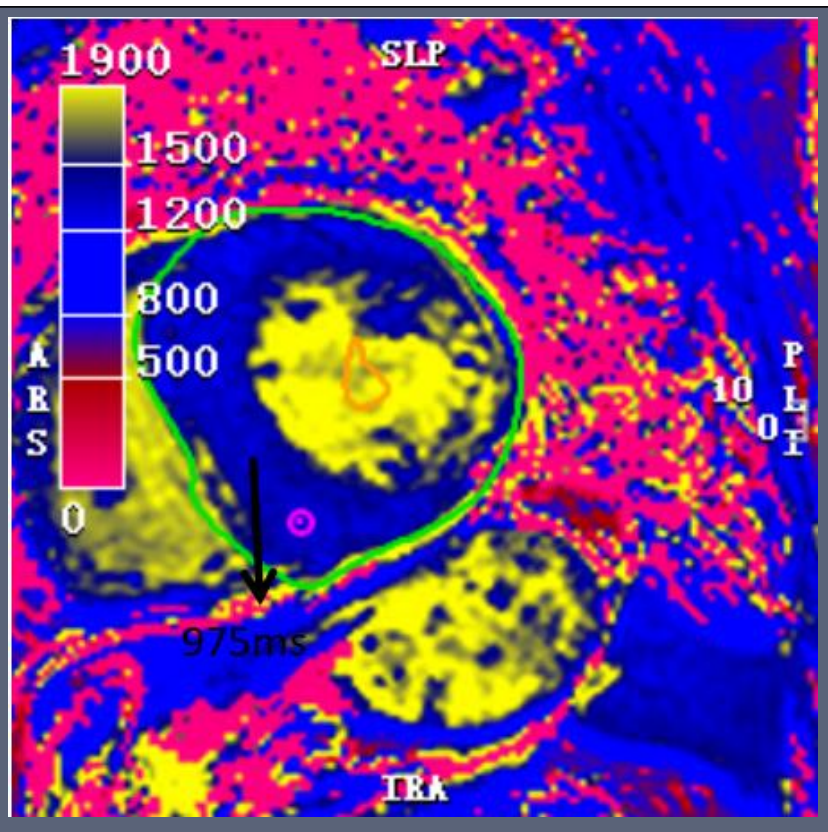
Differential Diagnosis?

- What do you have in your mind?

1. Hypertrophic cardiomyopathy
2. Hypertensive cardiomyopathy
3. Myocardial infarction
4. Cardiac amyloidosis

T1 mapping





Radiologic findings

- **MRI:**

1. Asymmetric left ventricular hypertrophy(maximal thickness at 22mm at the septum), thinnest in the inferior wall(2mm).

2. Transmural delayed enhancement is present in the thinned myocardium of the left ventricular inferolateral wall.

3. T1 mapping shows low native T1 values of the myocardium(847±22ms, normal reference value 1100±20ms).

enzyme activity determination and genetic testing

送检项目： α-半乳糖苷酶酶活力检测

临床诊断： 法布里病

检 验 结 果

项目名称	检测结果	单位	参考区间
α-半乳糖苷酶	0.9↓	nmol/g/min	24.5~63.6
β-半乳糖苷酶	231.5	nmol/g/min	144.7~350.4

提示：α-半乳糖苷酶酶活力检测结果偏低，提示法布里病（Fabry 病），需结合临床症状进行诊断。

主诉：换肾，室速
既病史：-
已有检测结果：-
家族史：-
临床印象：法布里病
重点关注基因：GLA

检测结论

检测到可能与临床表型相关的变异，暂无明确结论。

基因检测结果

该样本在Fabry病、Fabry病，心脏变异型相关基因GLA存在一处半合子变异。
该样本在肾病综合征20型相关基因TBC1D8B存在一处半合子变异。

基因	变异位点 (GRCh37/hg19)	合子型	正常人群携带率	转录版本 基因亚区	家系验证	ACMG 变异评级	疾病信息
GLA	c.37G>C chrX-100662855 p.A13P	半合子 0/25 1.00	-	NM_000169.2 exon1	-	VUS	1、Fabry病(XL) 2、Fabry病，心脏变异型(XL)
TBC1D8B	c.1031G>T chrX-106069463 p.R344L	半合子 0/26 1.00	-	NM_017752.2 exon6	-	VUS	1、肾病综合征20型(XL)

Anderson-Fabry Disease

- A rare X-linked inherited lysosomal storage disorder.
- Involvement of multiple systems throughout the body, primarily categorized into classic and late-onset phenotypes.
- Cardiovascular involvement usually manifests as left ventricular hypertrophy, myocardial fibrosis, heart failure, and arrhythmias.
- Genetic analysis and pathological biopsy are the gold standards for diagnosing FD.
- CMR has typical features such as LGE and low native T1 values.