CASE 1： 21YR/M

- **C.C.: Paroxysmal palpitations**
- **since 7months ago， aggravated in the past month**
- **Accompanied by Proximal limb muscle atrophy with decreased muscle strength**
- **Family history（-），hypertension（-）**

Presented by Lili Wang, Yuhan Yi, Lei Zhao
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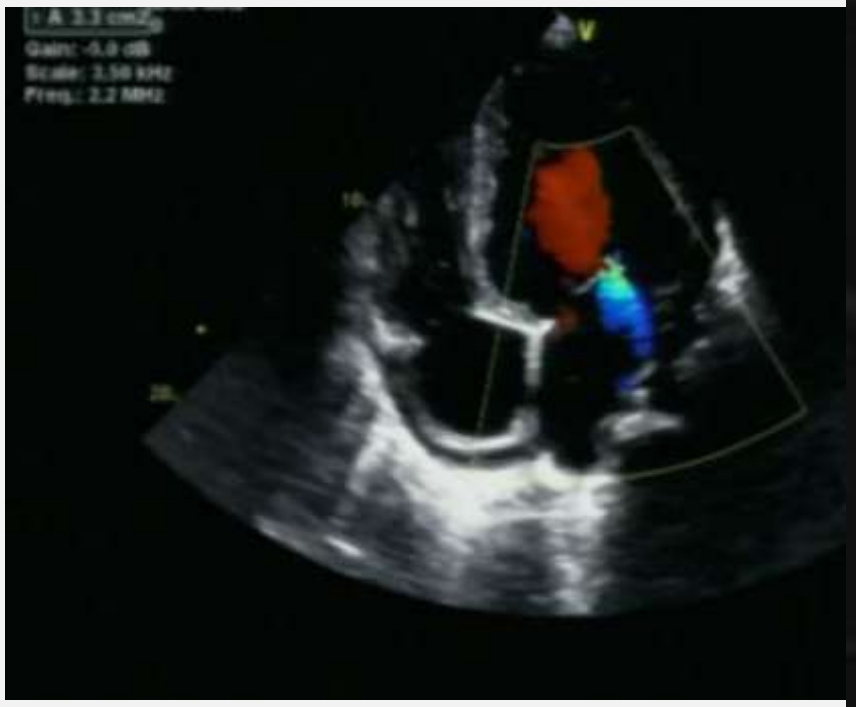
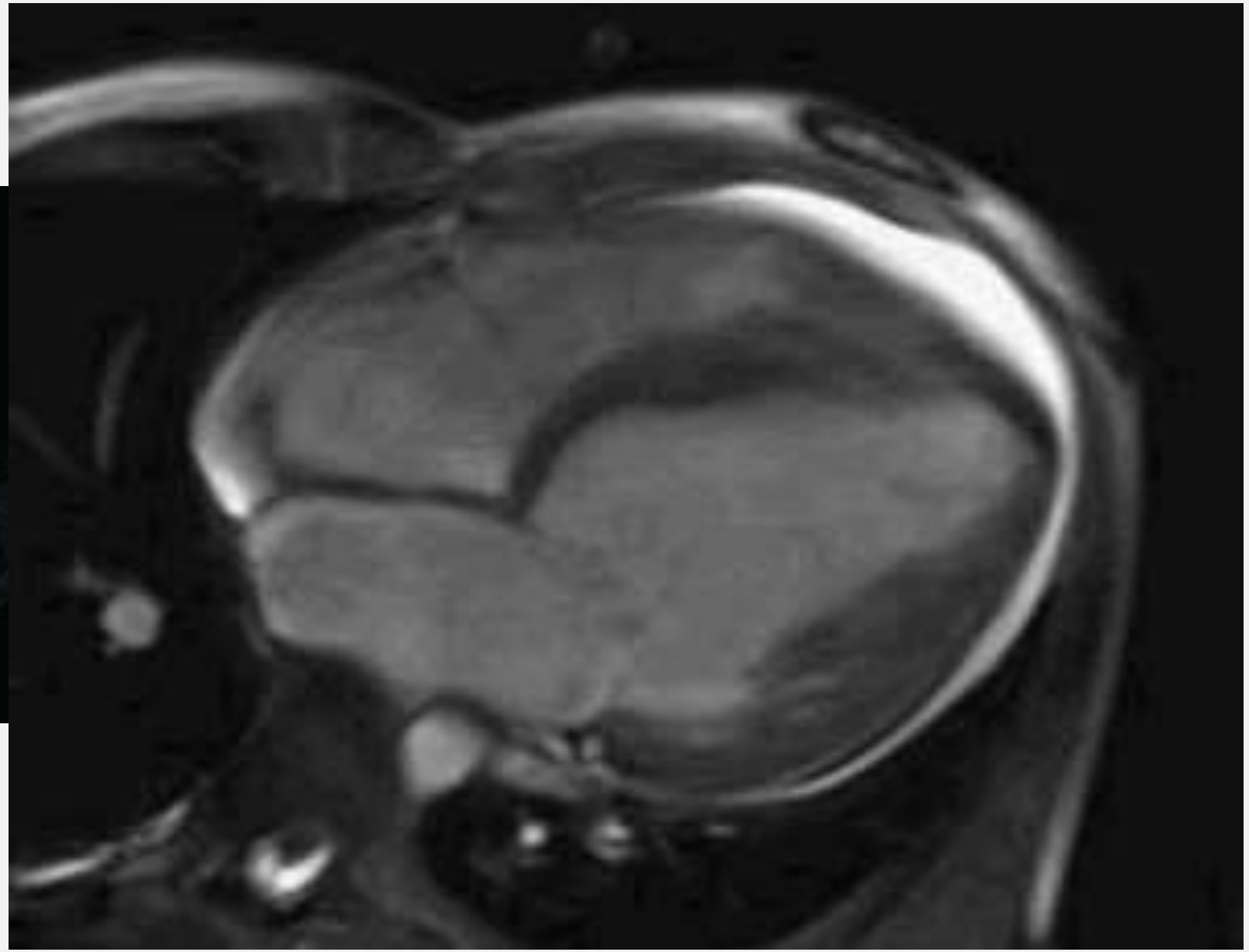
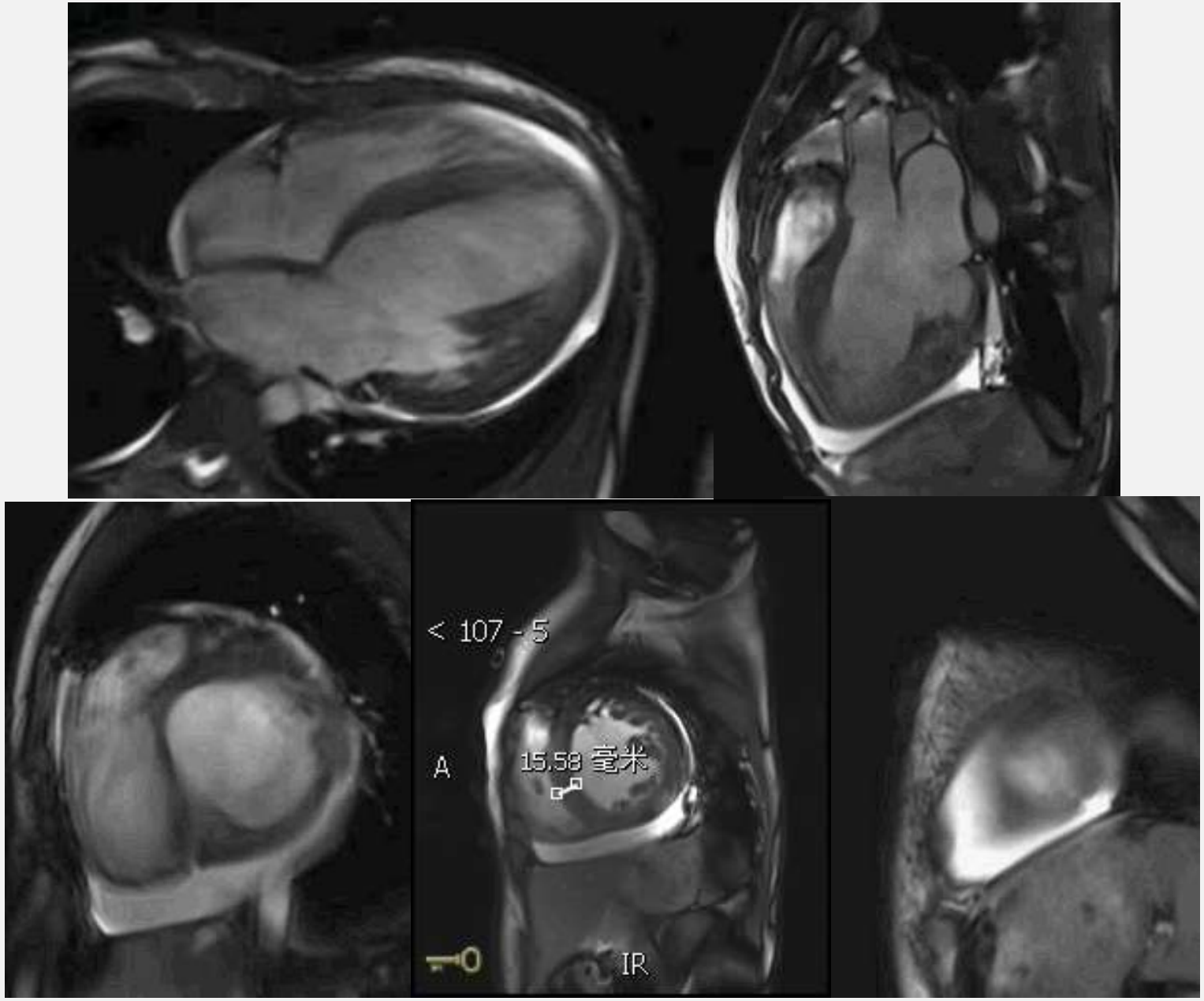
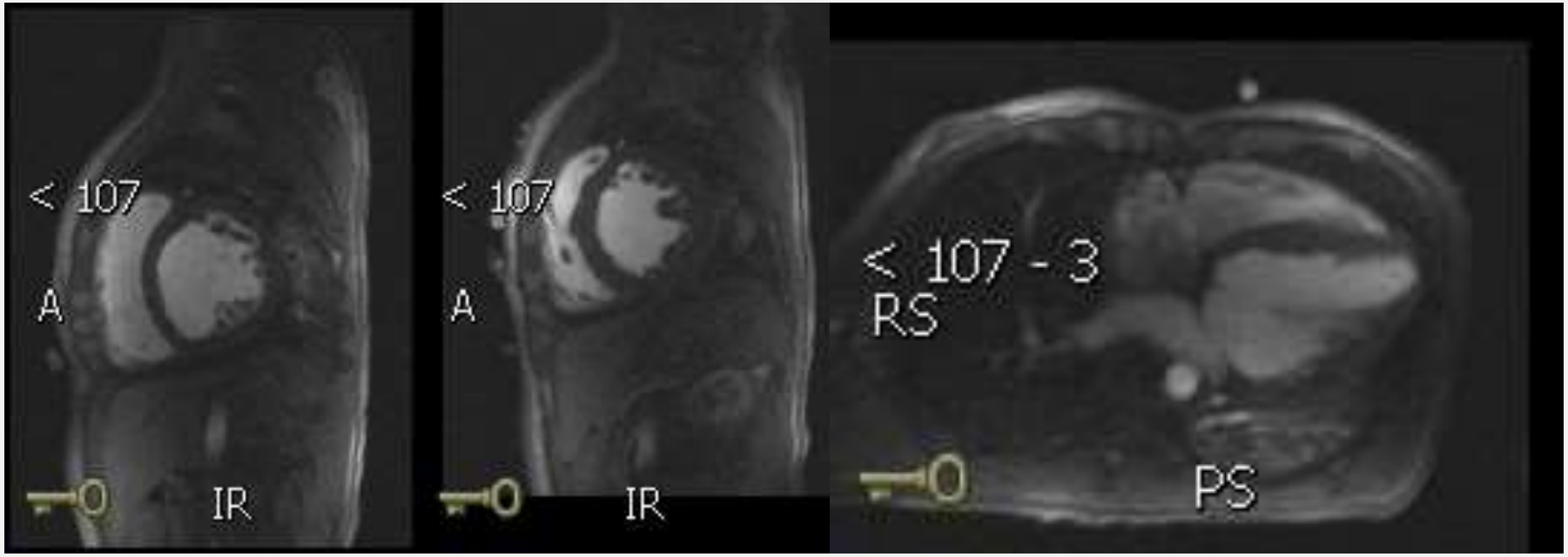

A 3.3 cm2
Gain: -5.0 dB
Scale: 3.58 kHz
Freq.: 2.2 MHz



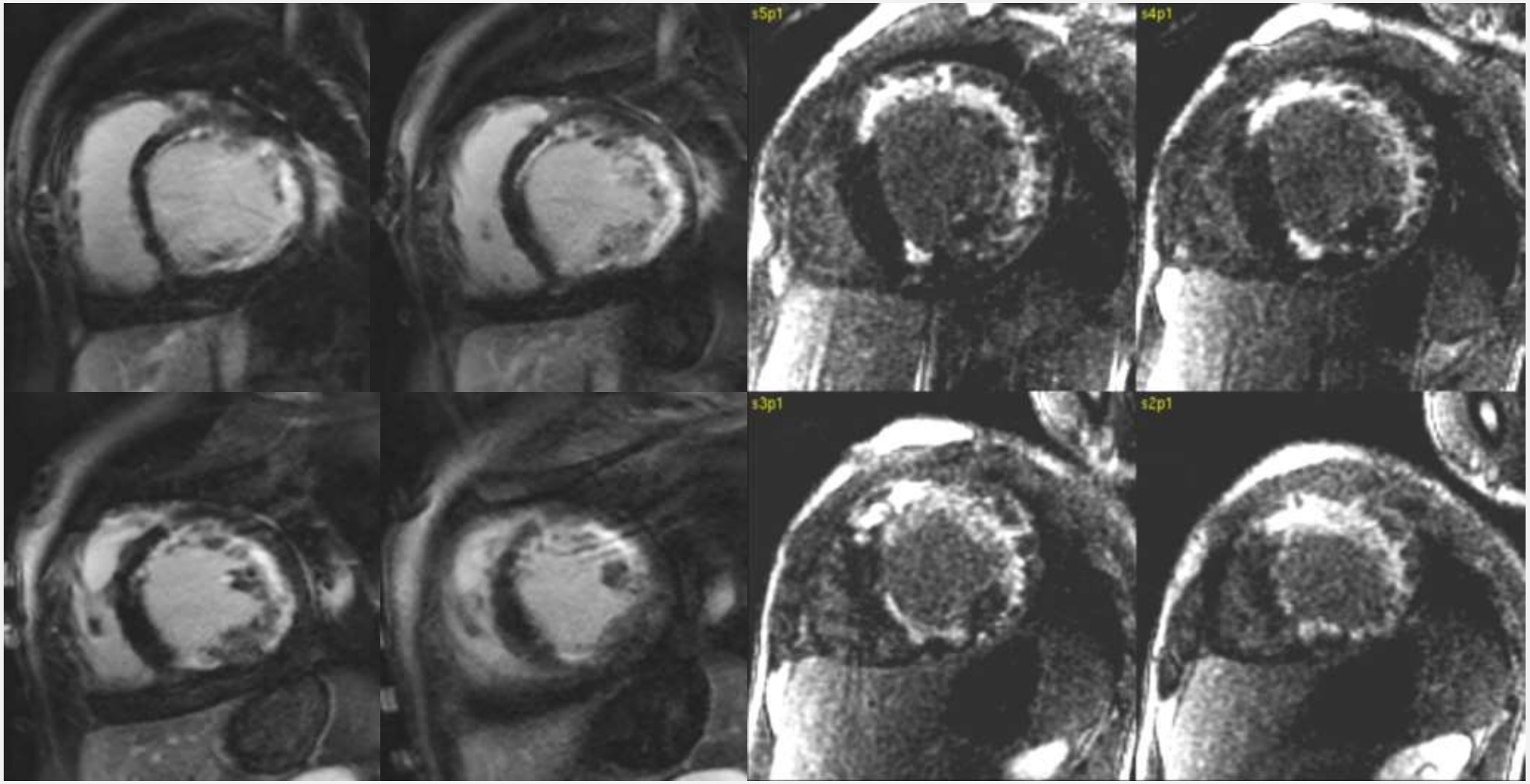
Cine-MRI



MRPI



LGE



Differential diagnosis?

- What do you have in your mind?

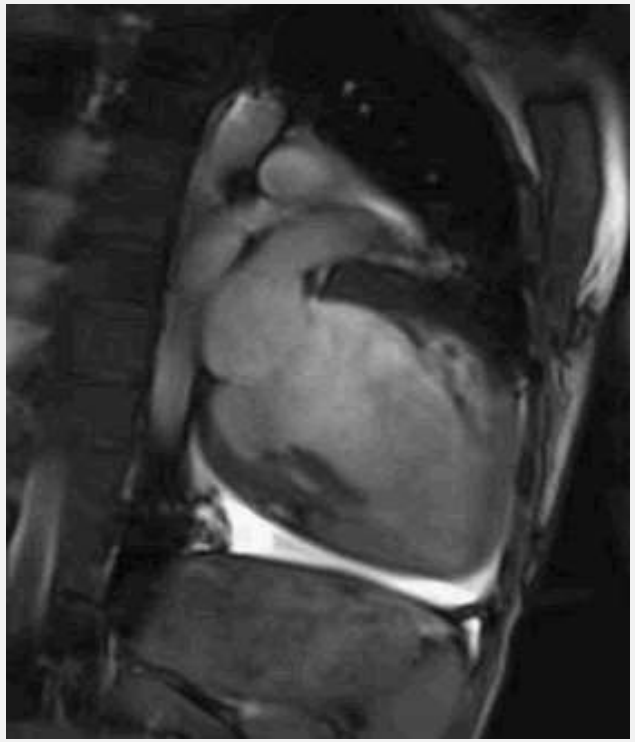
1、hypertrophic cardiomyopathy

2、Friedreich ataxia

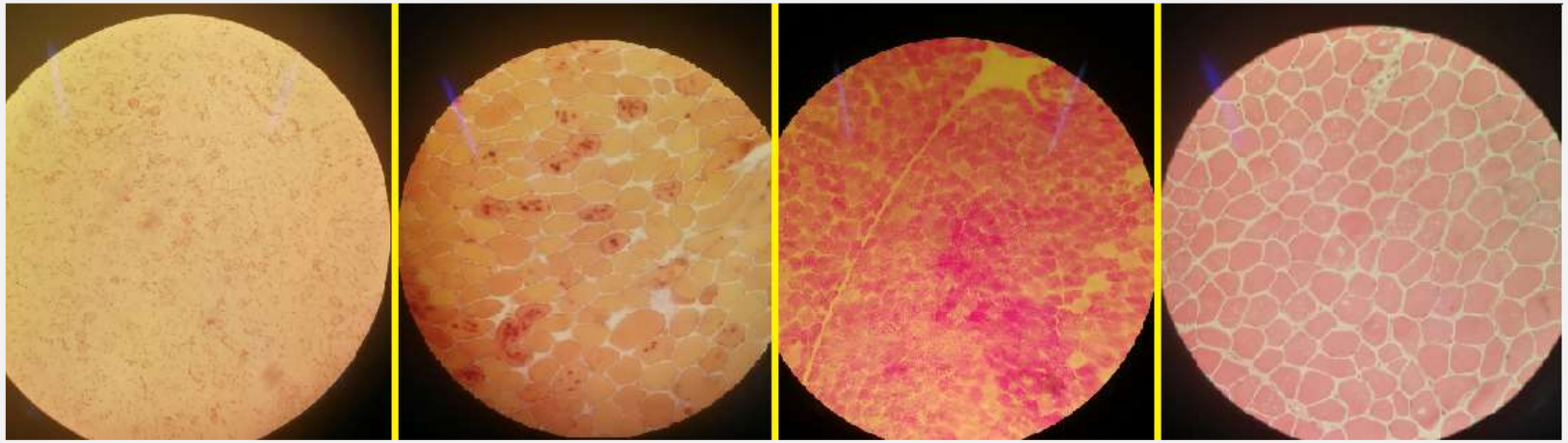
3、Cardiac Amyloidosis

4、Fabry disease

5、……

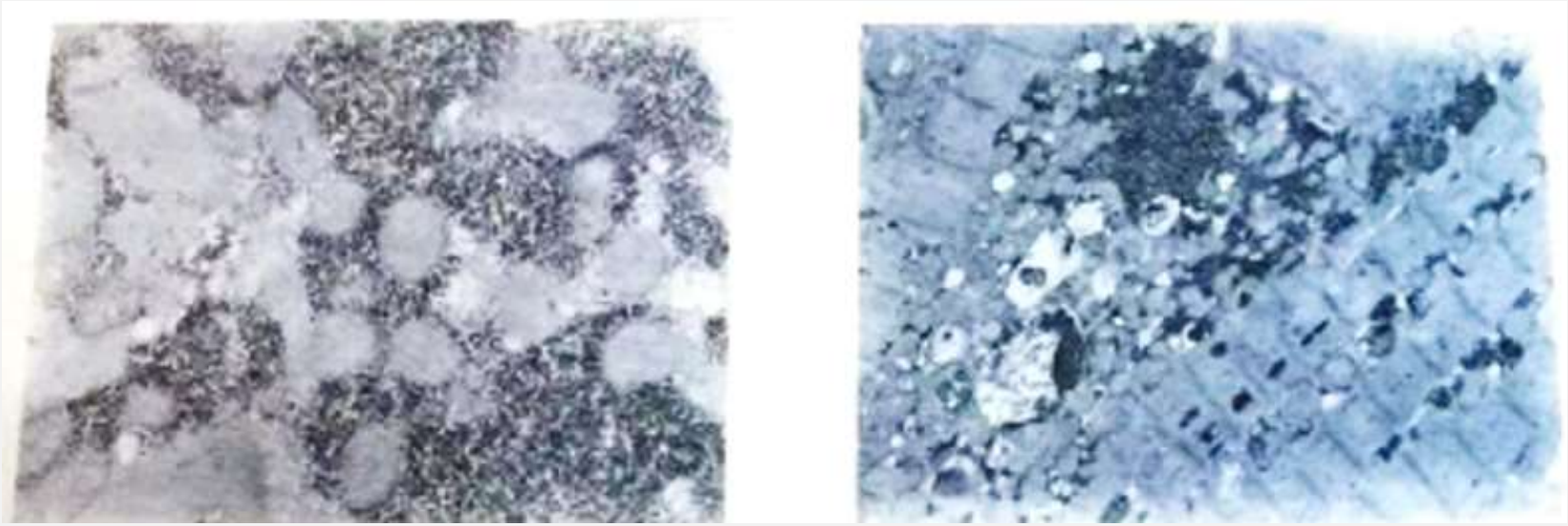


Muscle biopsy



•H&E staining： small and slightly larger vacuoles within some muscle fibers.
•NSE staining： deeply stained granular material within some muscle fibers.
•Desmin staining： a significant deposition of desmin-positive material within relatively numerous muscle fibers

Myocardial biopsy



- a substantial quantity of high electron-density free glycogen granules are diffusely distributed within the interstitial space of the myofibrils with some vesicles and myelin-like structures.

Genetic Testing

基因	突变信息	HGMD信息	基因疾病信息				NGTS1900068701未知	人群携带频率	clinvar信息	功能预测1	
LAMP2	c.973delC chrX-119575705 p.L325Wfs*21 frameshift	DM Danon disease 973del	Danon病	XL	99	有较多大片段重复缺失报道，恶性突变84.1%，错义突变6.8%，启动子1.1%	Hom 1.00/42(0.98)	0 0	----	ExACEAS 突变频率 新发/排除	0/0 0/0

Diagnosis

Danon Disease

- Coronary CTA：no significant abnormalities
- CMR：
  - a mild hypertrophy in the left ventricle and interventricular septum
  - Hypoperfusion of the subendocardium
  - Heterogeneous strong enhancement after Gd

Danon Disease

- a severe X-linked autophagic vacuolar cardioskeletal myopathy
- loss of function variants in the LAMP2 gene
- heart failure , extracardiac neurological, skeletal and ophthalmologic manifestations
- Sex variations:
  - Males: more severe; concentric LVH
  - Females: relatively mild; variable expressions
- Requires systematic management and treatment

Hong KN et al. *Journal of the American College of Cardiology*. 2023;82(16):1628-1647.