# Endocardial Fibroelastosis

**The Controversy Continues**

David A. Chiasson MD
DPLM – Division of Pathology
Hospital for Sick Children

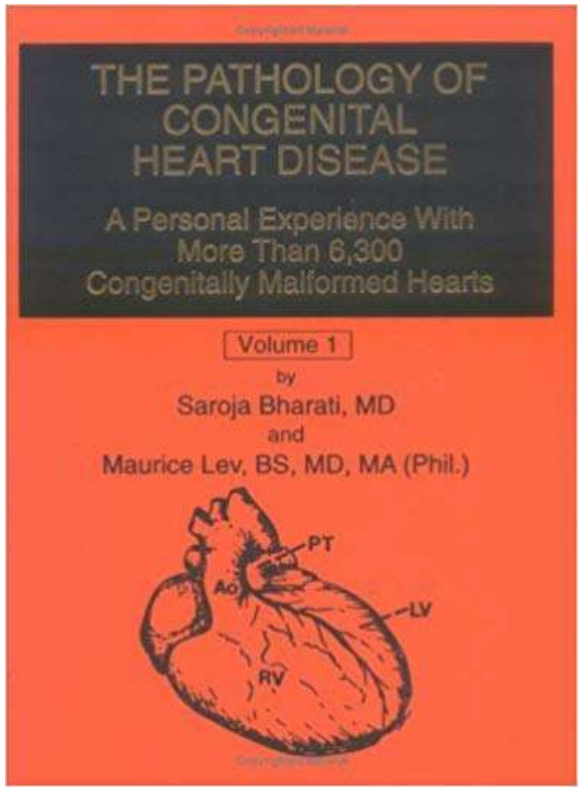
# Disclosure:

I am a pathologist

Maude Abbott

Jesse Edwards

Maurice Lev



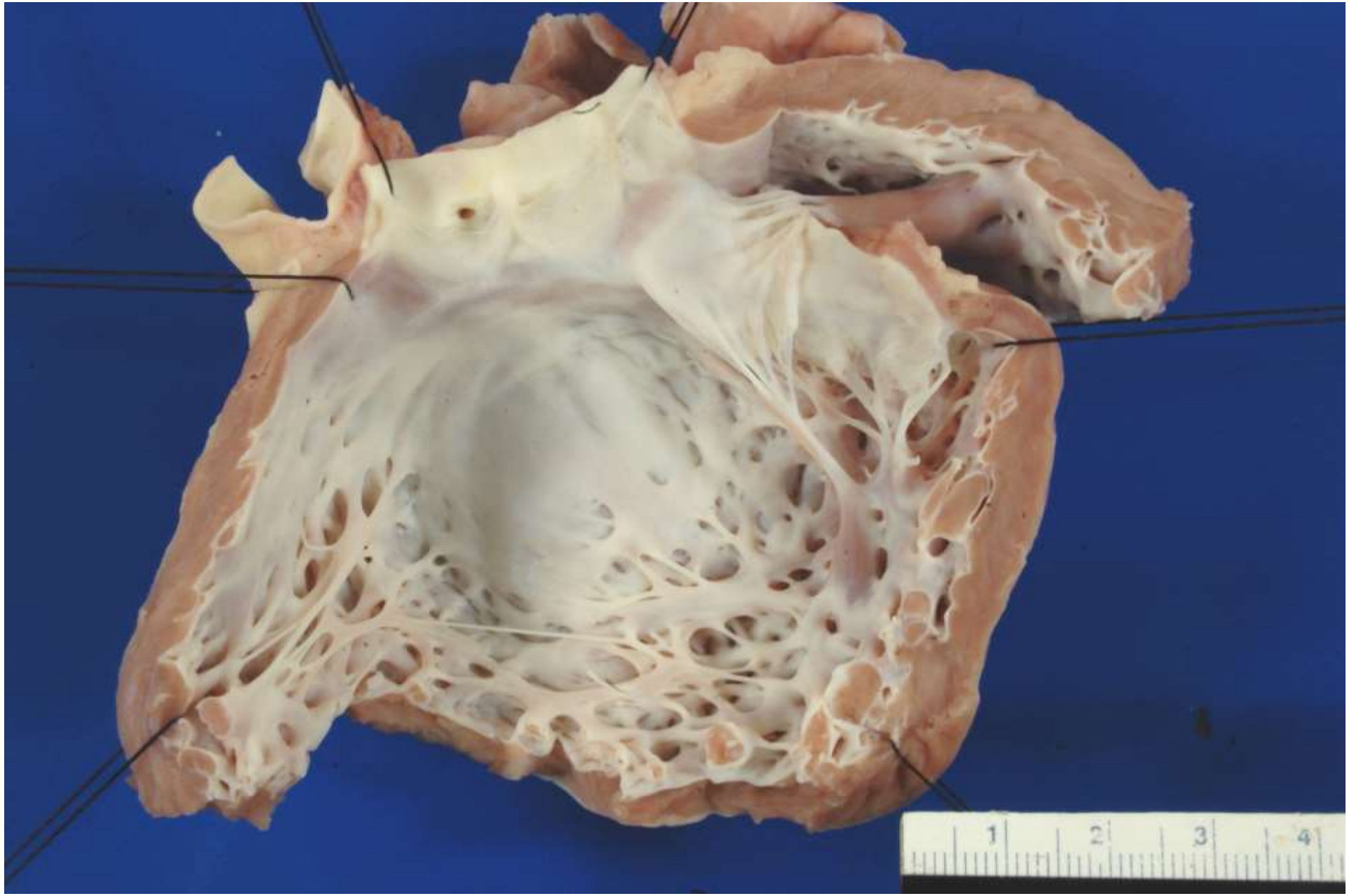
# The “Secondary” Pathology of CHD

- **General:**
  - Cardiomegaly
  - Chamber dilation
  - Wall hypertrophy
- **Epicardium**
  - Effusion
  - Inflammation
  - Fibrosis / adhesions
- **Myocardium**
  - Ischemic injury: acute / remote (replacement fibrosis)
  - Inflammation
  - Interstitial fibrosis
- **Endocardium**
  - Thrombosis: acute / organizing
  - Inflammation (endocarditis)
  - Fibrosis vs fibro-elastosis (EFE)

# Endocardial fibro-elastosis

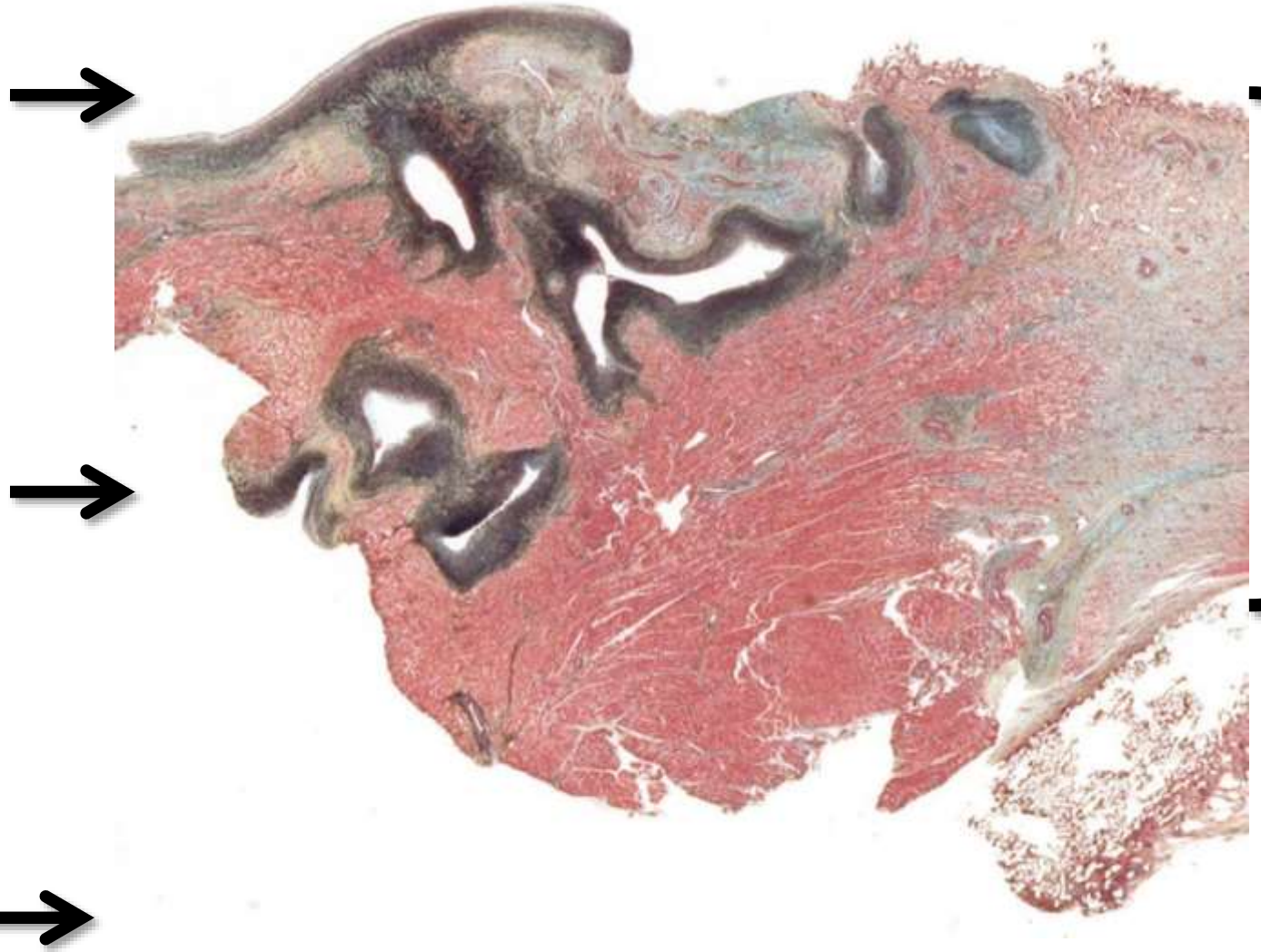
- **thickening of the endocardium by collagen and elastic fibers**
  - **does not = endocardial fibrosis**
- **“pediatric” entity**
- **localized / patchy**
  - **hemodynamic / “jet” lesions**
- **diffuse**
  - **Infantile DCM / 1° EFE**
  - **CHD: e.g. HLHS**

# Cardiac Explant - Dilated LV with +++ EFE




14 mo F S09-3596

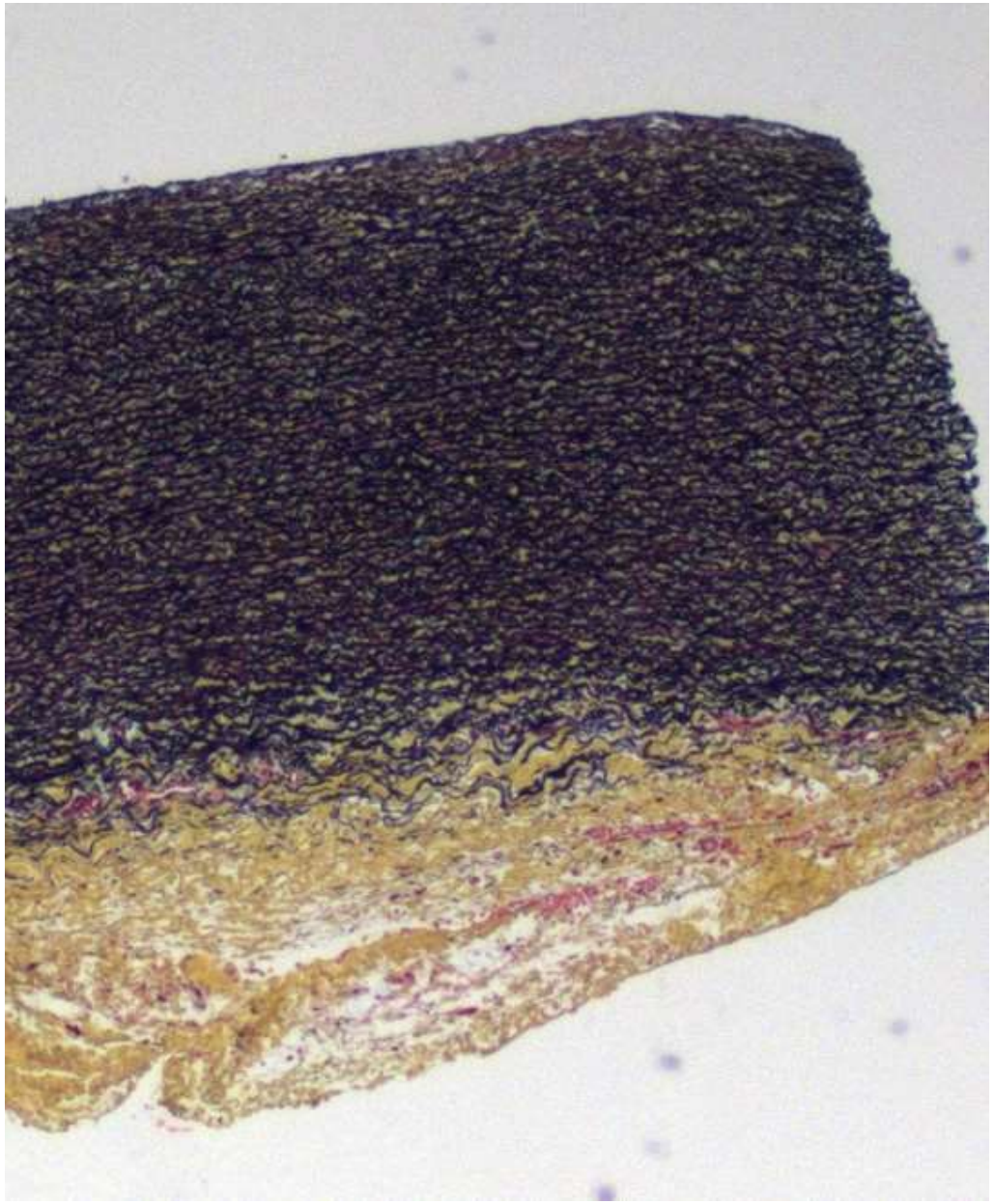
Whole mount section of LV wall from heart explant of 4 month old female DCM-CTx recipient (ELT stain)




S09-3930

**LV EFE**
**4 y with Aortic stenosis**

**SH19-0389**



**Elastic artery / Aorta**

# “Primary” EFE – Historical Perspective - I

- **1943 Weinberg - so-called fetal endocarditis**
- **1950 Prior – endocardial dysplasia**
  - **developmental disorder of mesenchymal tissue**
  - **to be classified with the congenital cardiac malformations**
- **1951 Hill - should belong to the collagen disease group.**
- **1953 Dennis - developmental defect**
  - **resulting from persistence and overgrowth of the primitive lining of the left bulbus cordis**
- **1955 Rosahn - disease is genetic in origin**

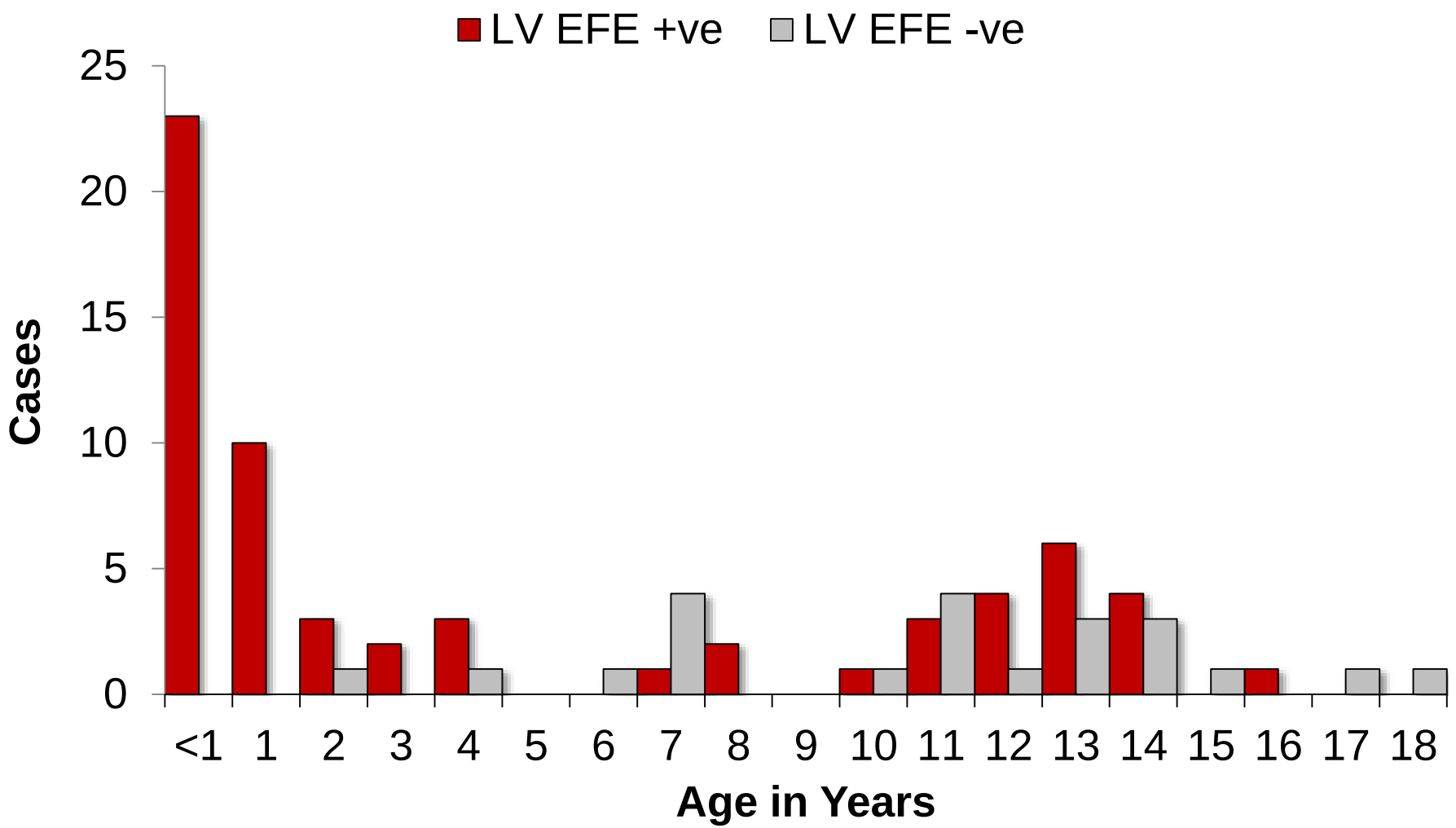
# “Primary” EFE – Historical Perspective – II

- 1956 Kelly: familial metabolic defect leading to myocardial weakness
  - endocardial changes are secondary.
- 1957 Black- Schaffer: mechanical explanation…
  - acquired in utero or in infancy
- 1961 Still: secondary to ↑ I-V pressure and dilatation
  - caused by some other cardiac anomaly
- 1962 Fisher: developmental defect probably of genetic origin
- 1960’s: association with + Mumps virus serology
- 1972- Hutchins: interstitial myocarditis of probable viral etiology
  - possible pathogenetic relationship
- 1973 Hunter - dominant autosomal trait rather than a recessive autosomal…….
- 1974 Schruyer: most probably of a viral etiology… a sequel to myocarditis or pancarditis.

# “Primary” EFE – Historical Perspective – III

- **Lurie 1988: EFE is not a disease**
- **Benson 1992*: …..suspicion that ……..primary type is secondary to ….some uncertain myocardial fault. (*Neonatal Heart D.)**
- **Aiello 1994 – a secondary phenomenon in dilated hearts**
- **Ni 1997 - sequela of a viral myocarditis (Mumps)**
- **Nield 2002 - occurs in the presence of autoantibody-mediated CHB**
- **Lurie 2010: not a disease but a reaction of the endocardium.**
  - **hope is for nosologic purity …… outworn but surviving concepts will be firmly rejected.**
- **Seki 2013 - clinically and pathologically different from DCM**
  - **should be recognized to promote understanding of the natural history and etiology**

# Age distribution of DCM-CTx explants according to presence or absence of LV EFE (n=85, 1 incomplete)

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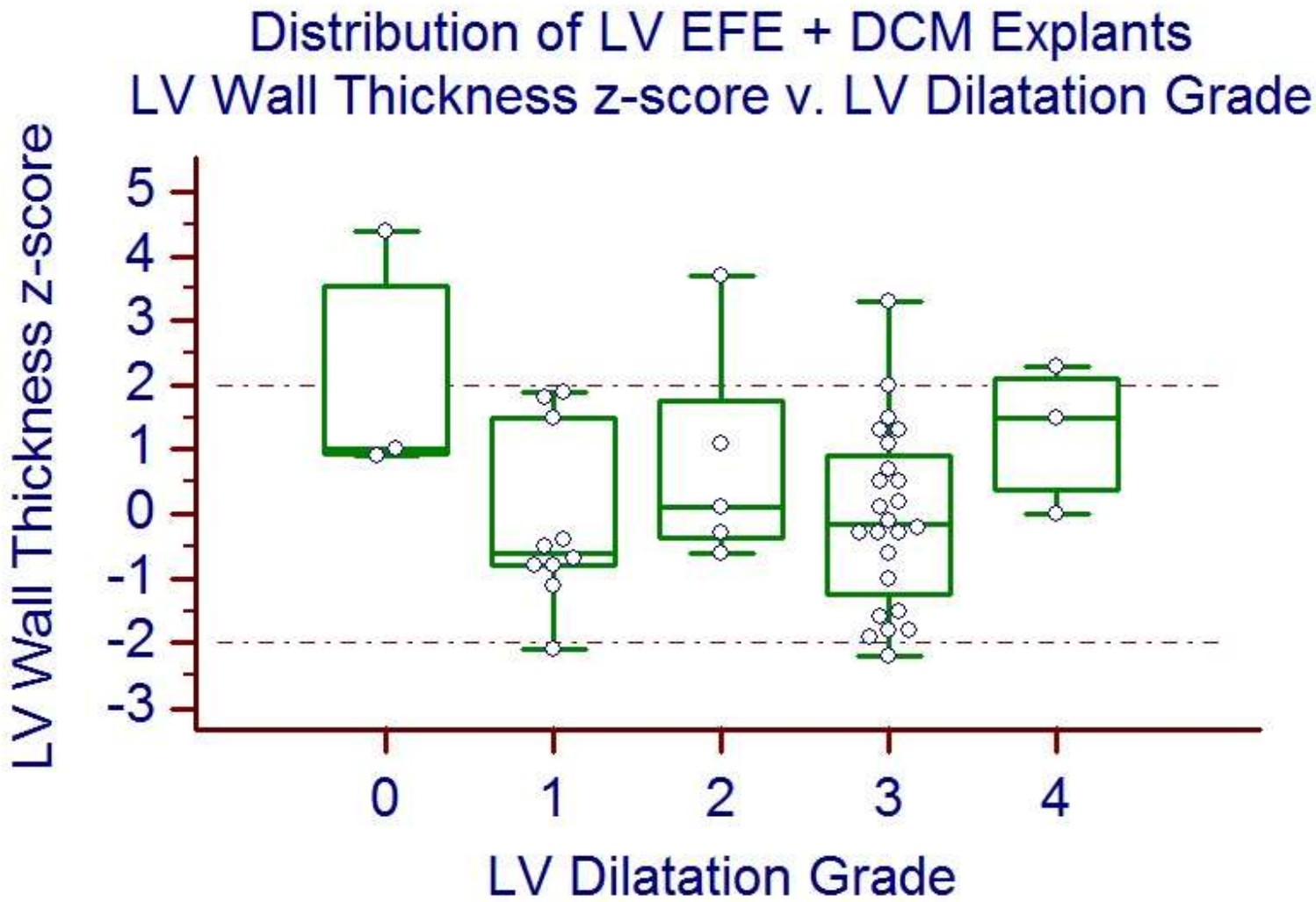
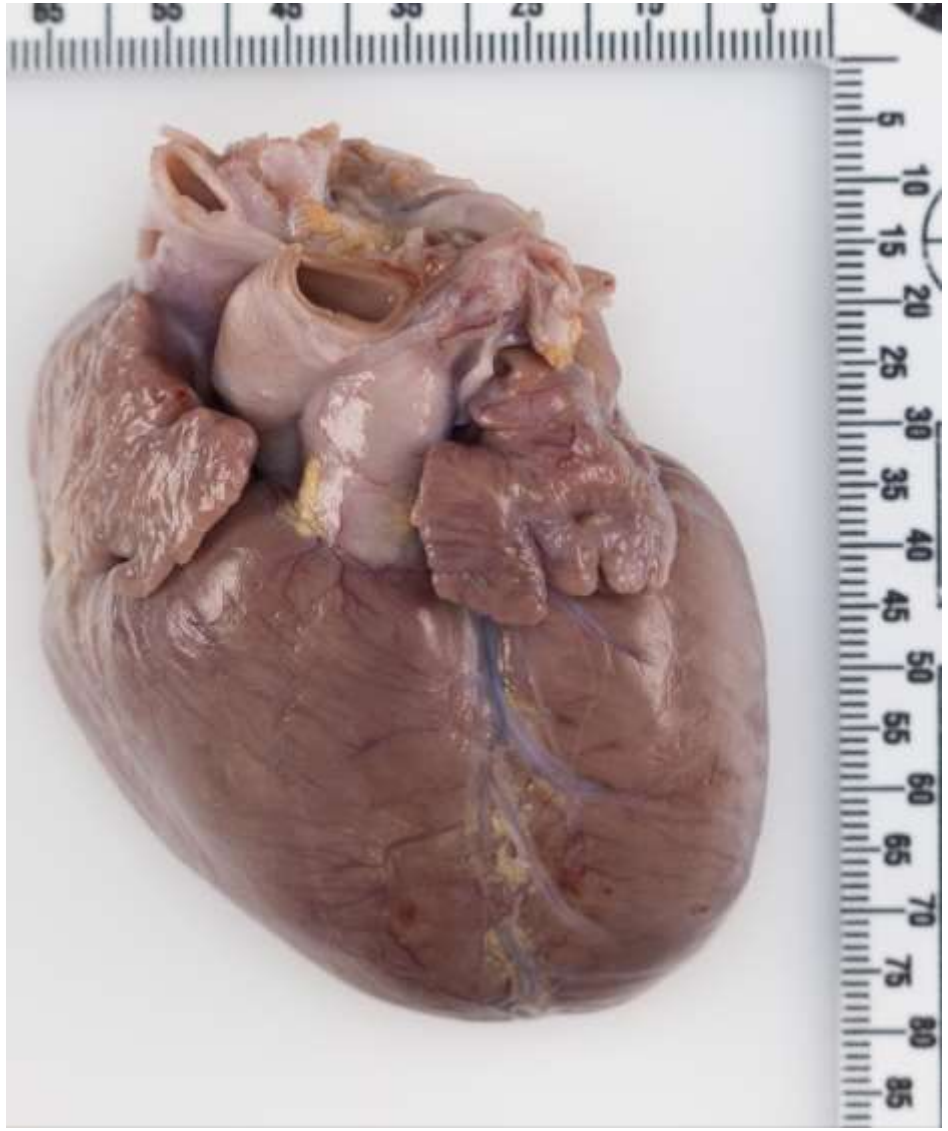
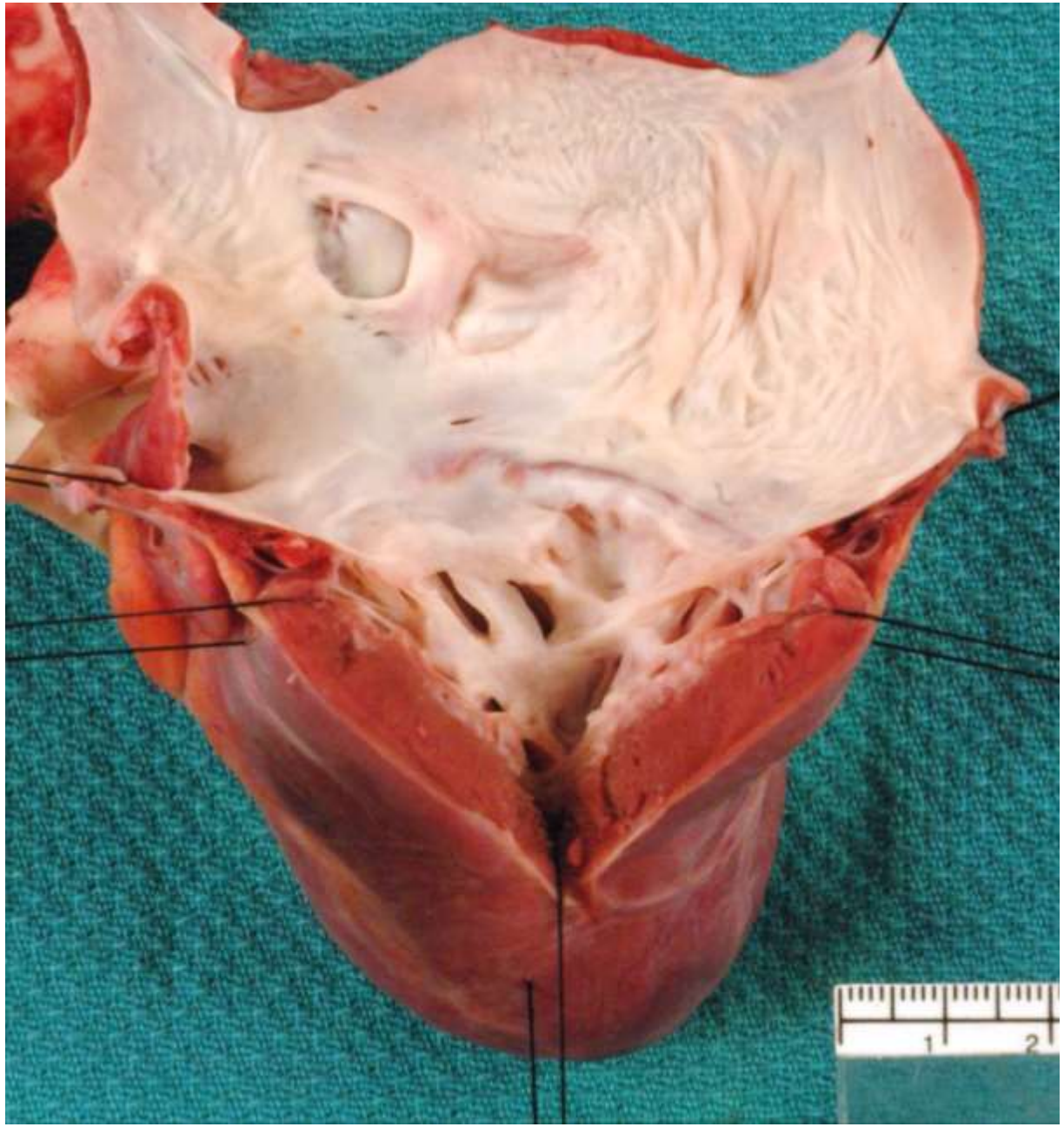

Distribution of LV EFE + DCM Explants
LV Wall Thickness z-score v. LV Dilatation Grade
LV Wall Thickness z-score
5
4
3
2
1
0
-1
-2
-3
0
1
2
3
4
LV Dilatation Grade

# SUID - DCM without EFE

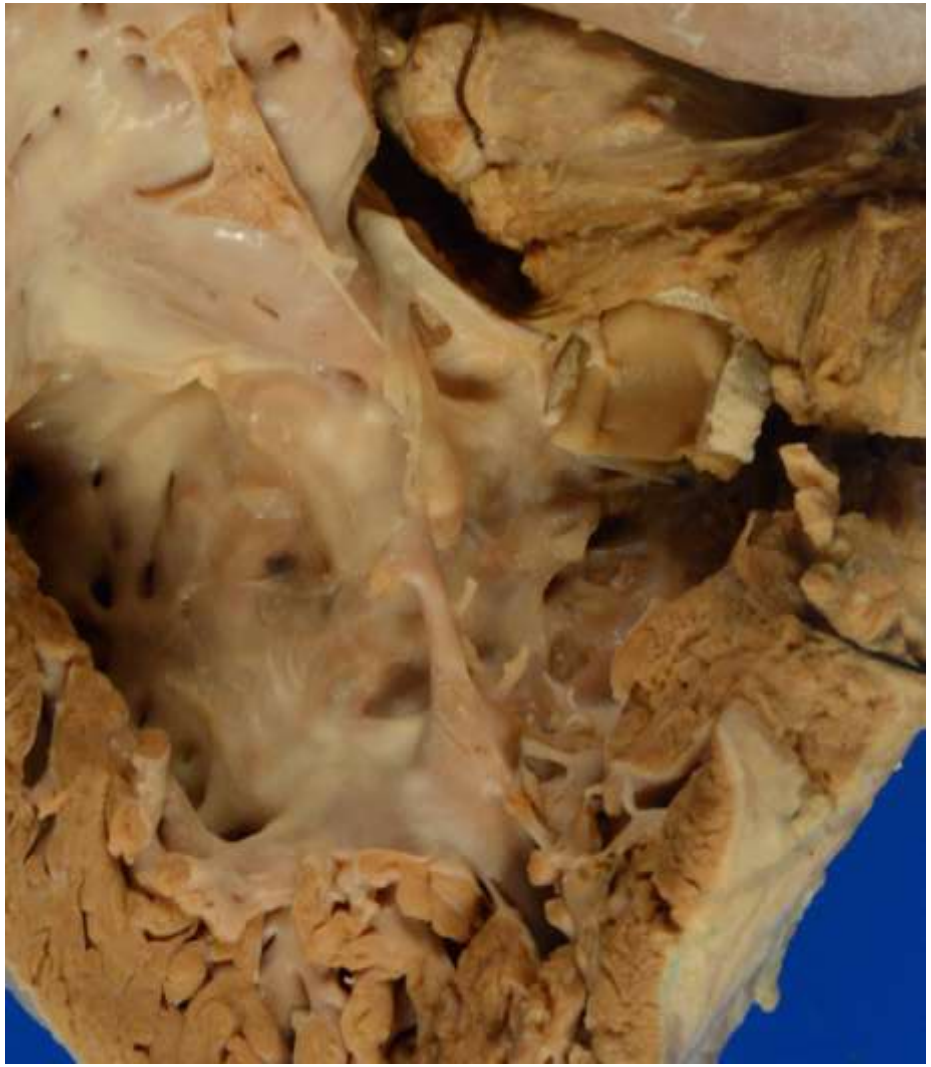


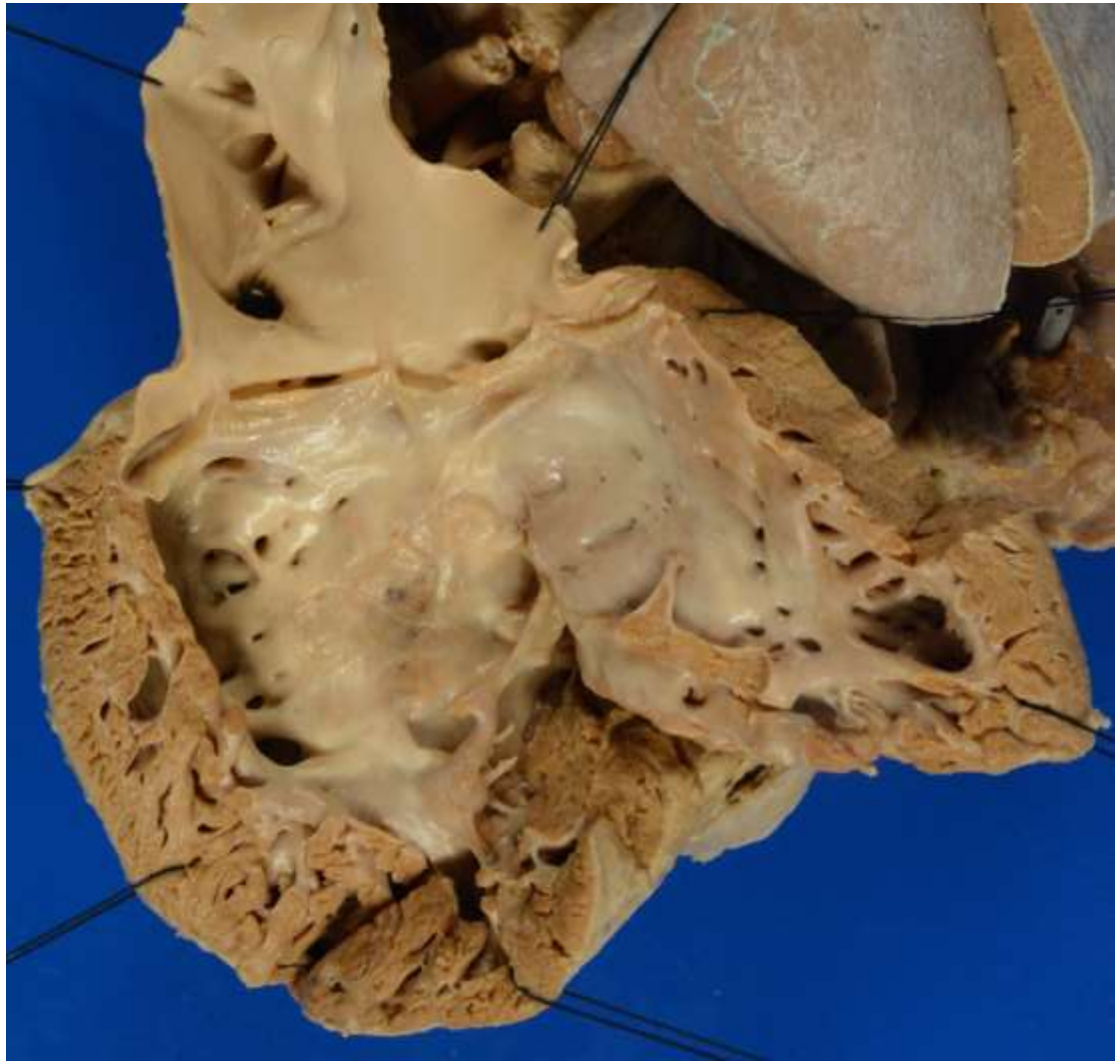
# EFE in Congenital Heart Disease

# LA Dilation with EFE
# Shone's Syndrome

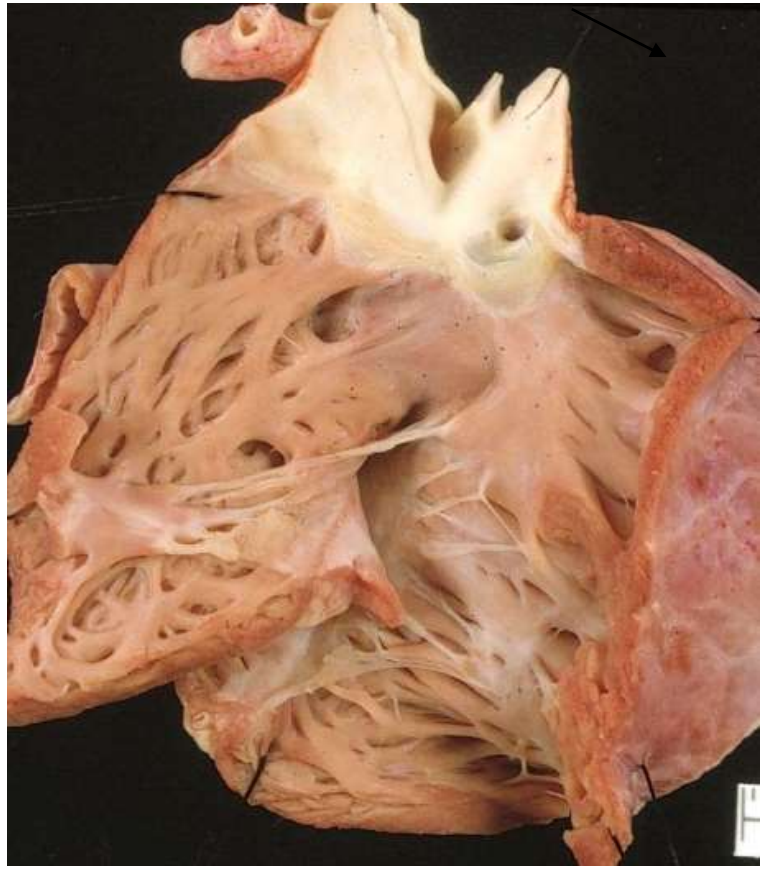


# EFE in cTGA - left sided ventricle



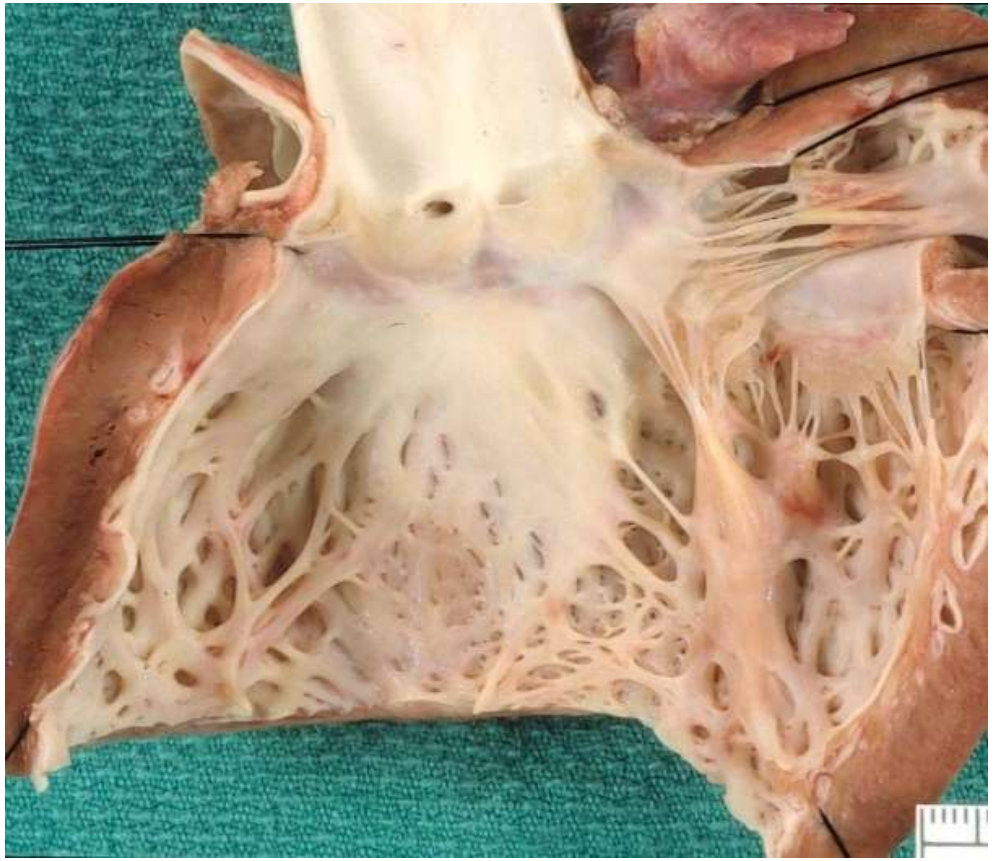


# Aberrant LMCA from MPA



-aberrant LMCA

arising from MPA



-single (right) coronary os

**- dilated LV with EFE**

**Case 1 (RFMA-1740)**

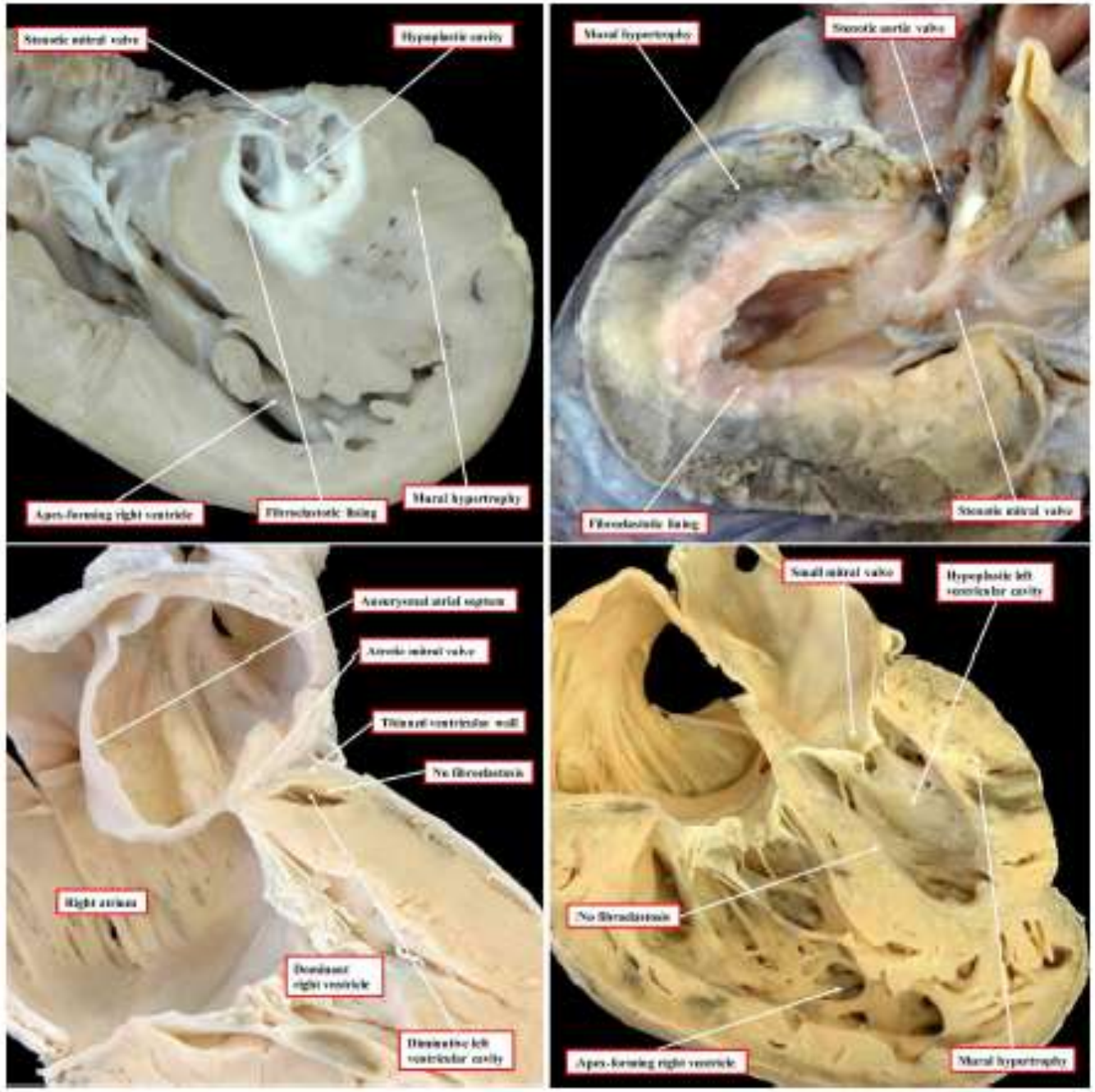



**Figure 1.** The images, all photographed by Diane E. Spicer and reproduced with her permission, show the phenotypic variants of hypoplastic left heart syndrome as seen in the clinical setting. The upper left panel shows the variant with mitral stenosis and aortic atresia. The heart in the upper right-hand panel has mitral and aortic stenosis. In the lower panels, to the left is seen the variant with mitral atresia, and to the right is the rarest variant with left ventricular hypoplasia with the small aortic and mitral valves, their size in keeping with that of the left ventricle although the aortic valve is not seen in the four-chamber section through the heart.

Journal of Cardiovascular Development and Disease

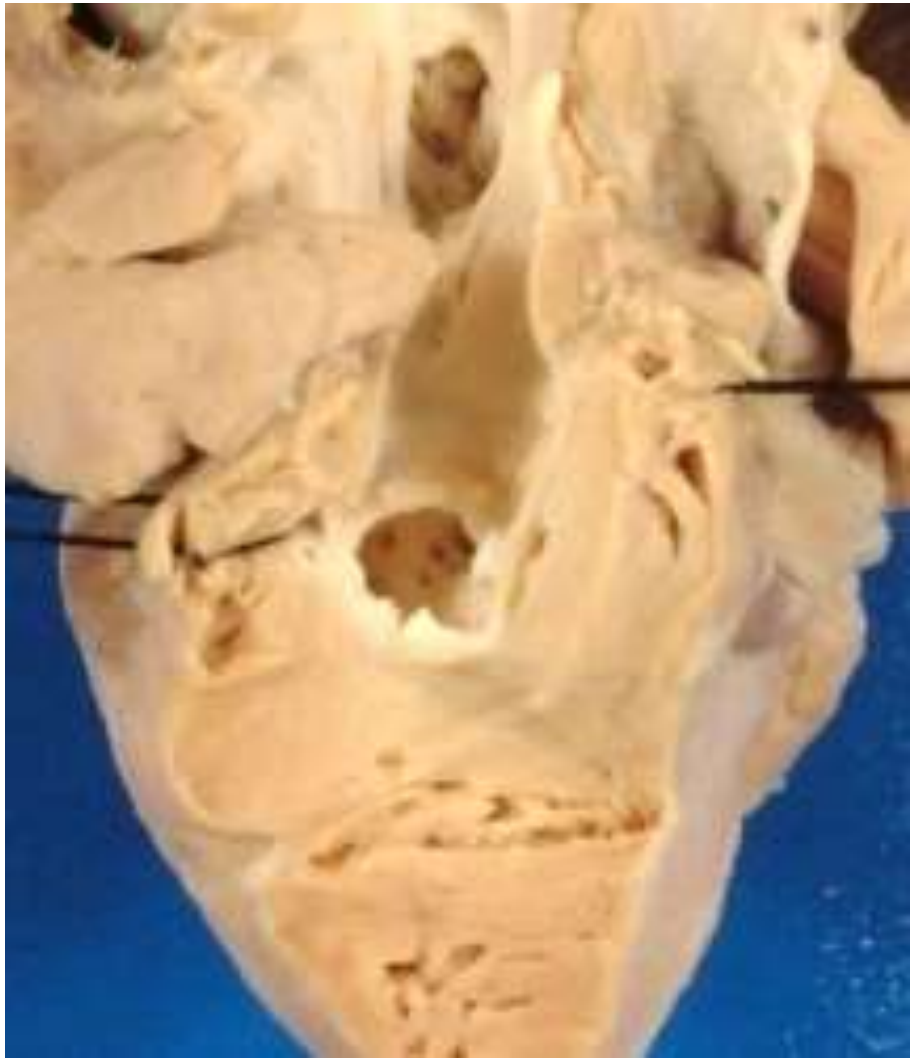
MDPI

*Review*

## Hypoplastic Left Heart Syndrome: A New Paradigm for an Old Disease?

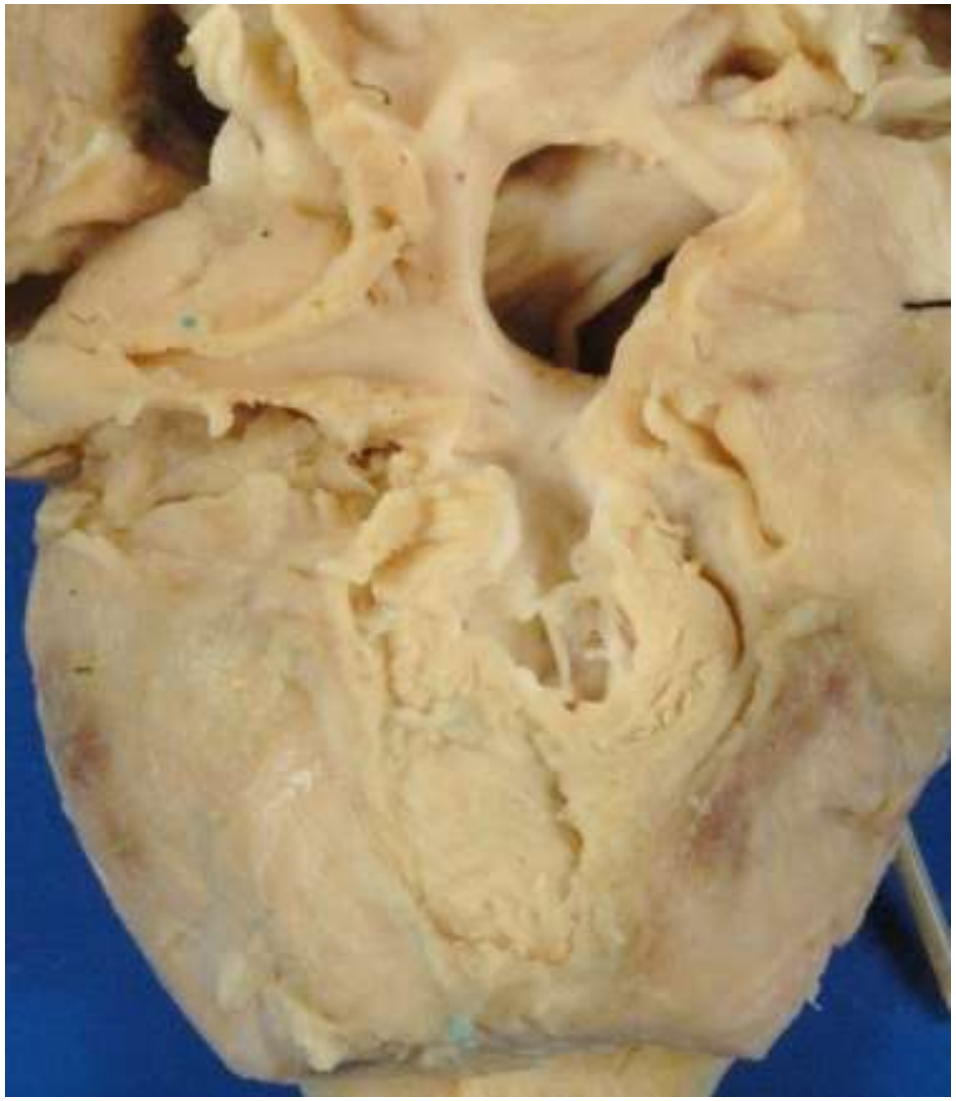
Paul Grossfeld [1,*], Shuyi Nie [2], Lizhu Lin [3], Lu Wang [1] and Robert H. Anderson [3]

# Hypoplastic left heart syndrome (HLHS)



**(RFMA-1731) AoV atresia, MV stenosis**

**Marked EFE**



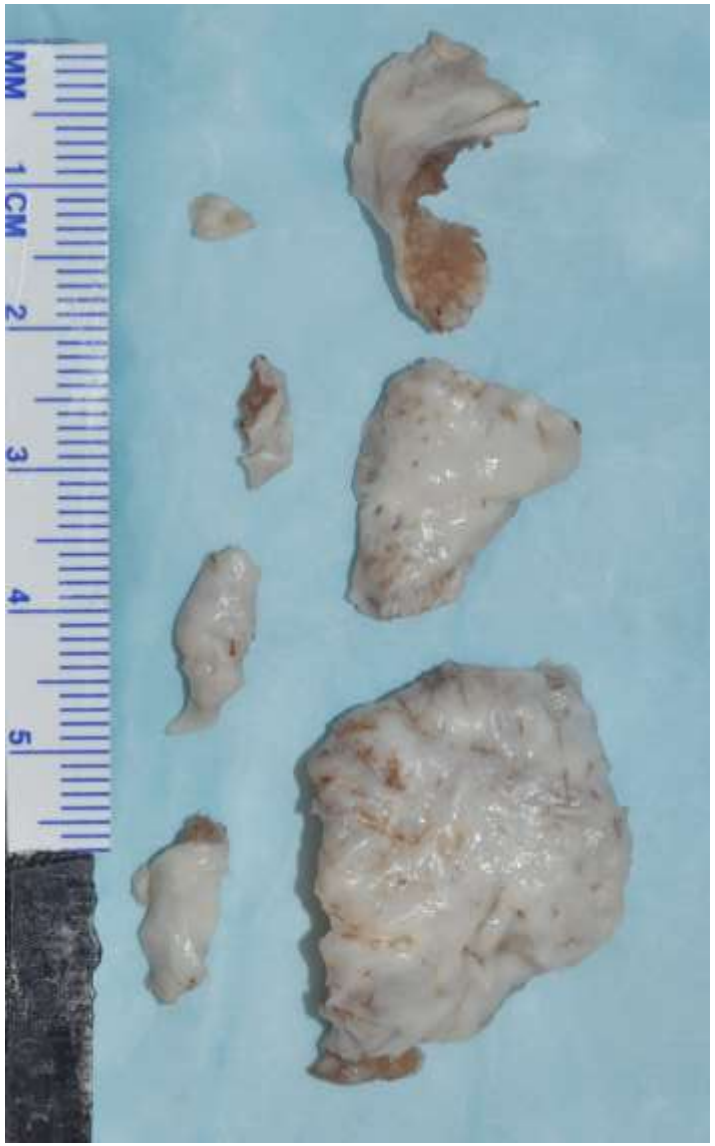
**(RFMA-1734) AoV atresia, MV stenosis**

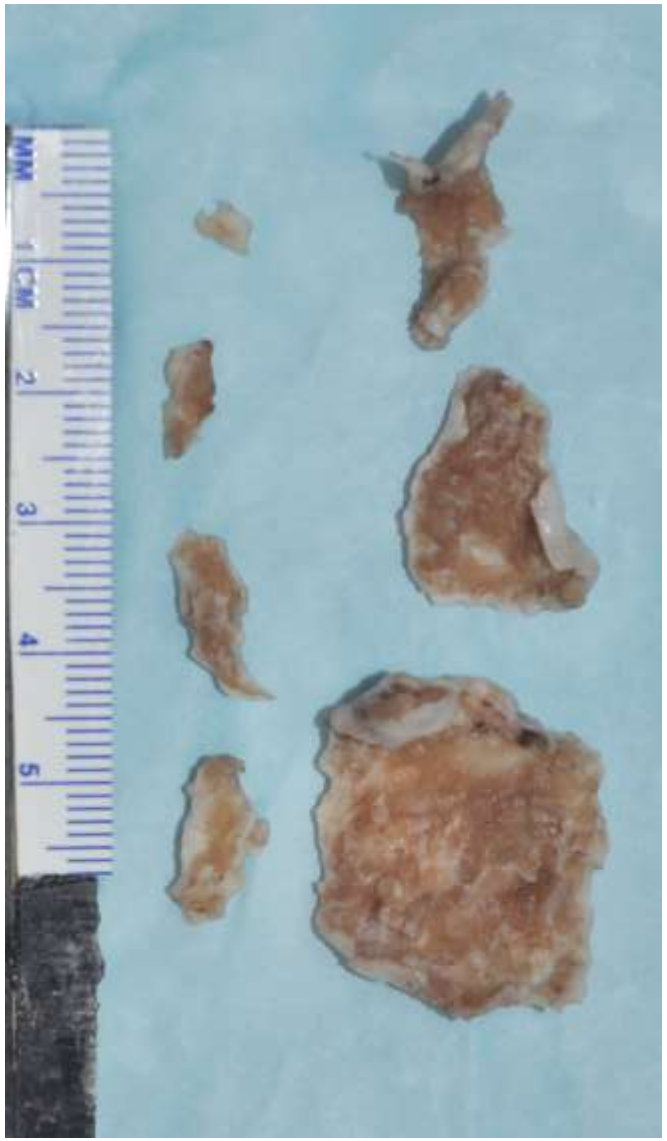
**No significant EFE**

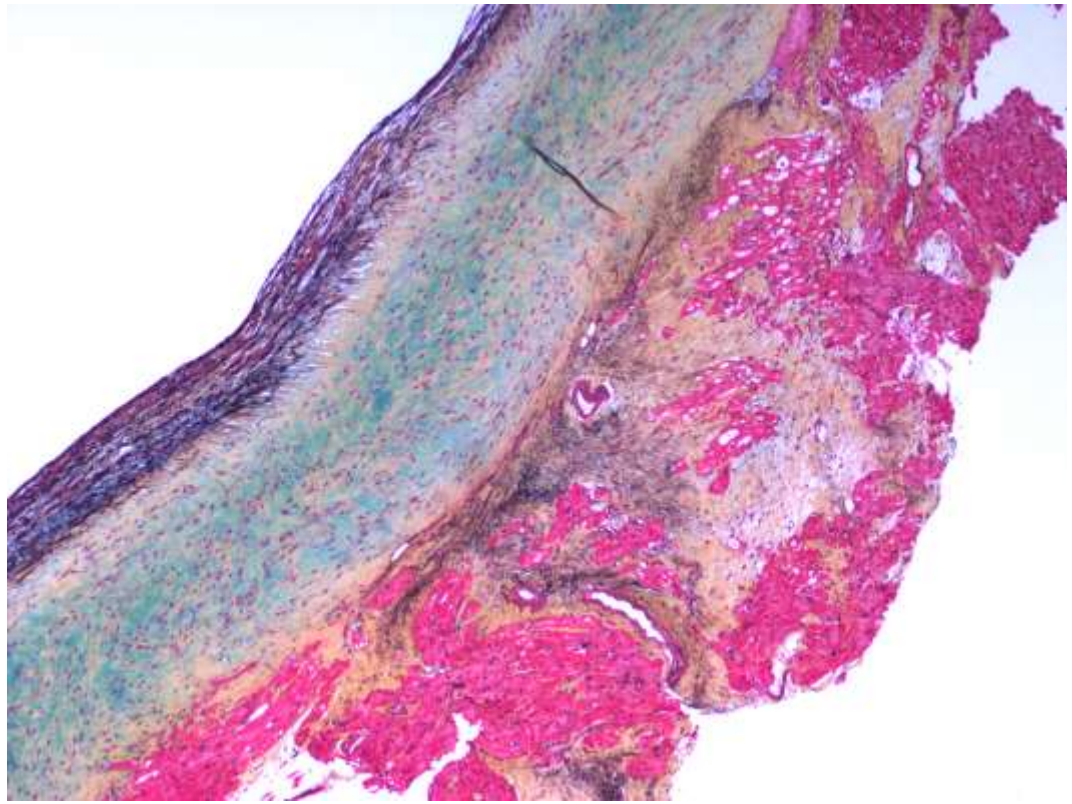
# EFE – Surgical Pathology

# Aortic stenosis with EFE

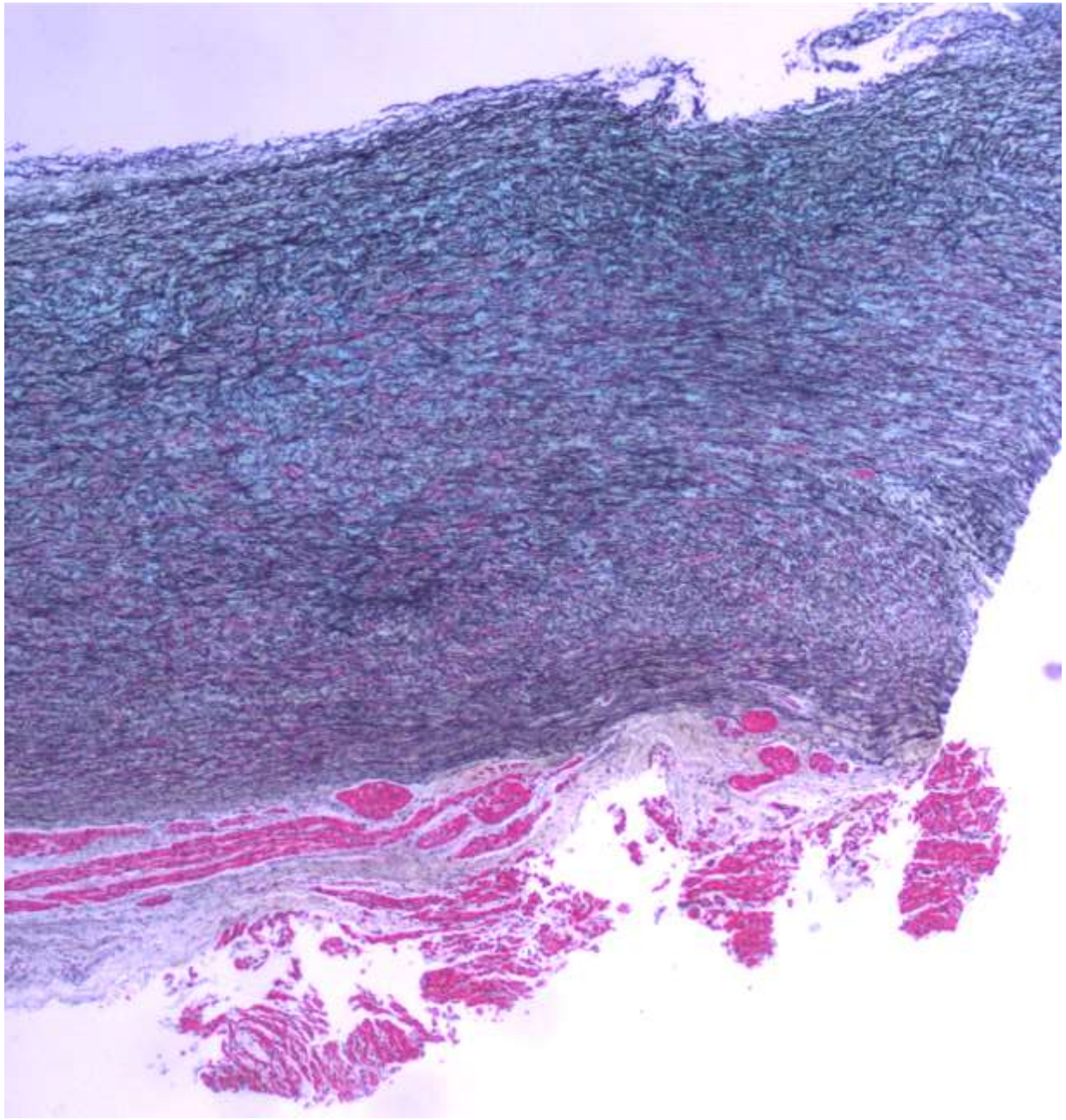
## Endomyocardial resection @ age 12 months



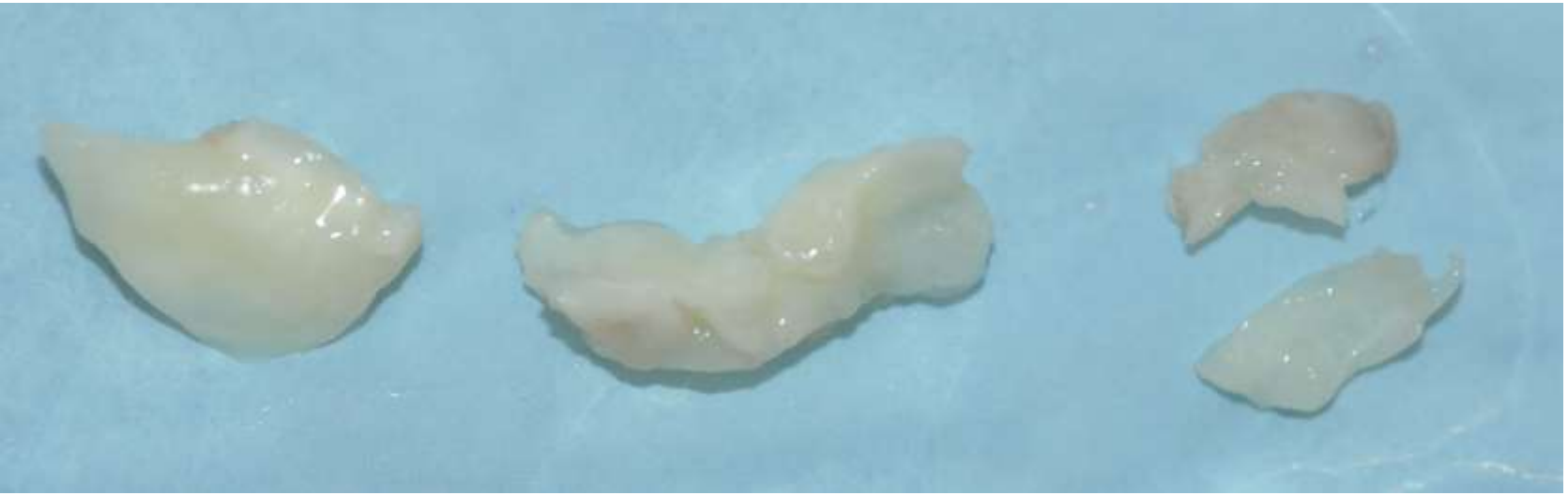



S16-595



# HLHS (MS/AS) with EFE

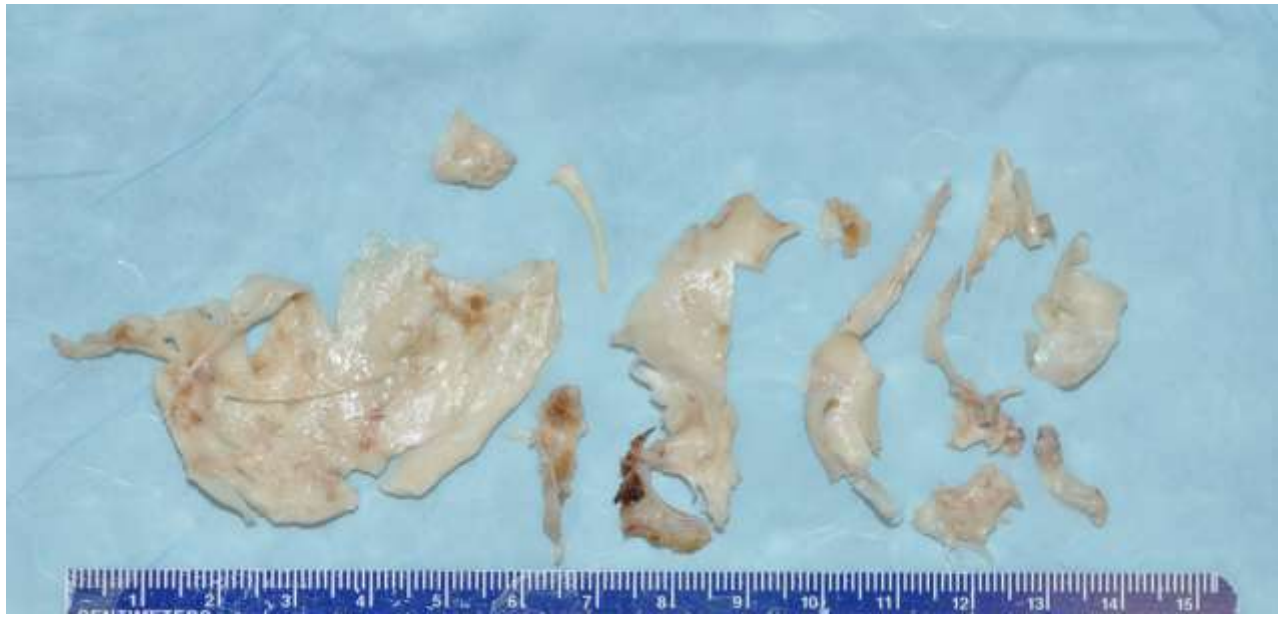
**Endocardial Resection @ age 5 months**



# Age 4 y - Aortic Valve Resection

**SH19-0389**

# EFE stripping in 4 yr old with HLHS: MS / AS



# Conclusion

- The widely recognized cardiac abnormality most commonly termed endocardial fibroelastosis (EFE) ……remains today just as much an enigma and mystery as at any time previously.
- …..To recapitulate, the abnormality here described is poorly understood and may prove to be a heterogeneous group of conditions.
- This aspect of the problem should receive much more investigative attention, since we do need insight into what the condition is.

Folger GR Jr. Endocardial fibroelastosis: a continuing and unsolved dilemma. Clin Pediatr (Phila). 1971 May;18(5):246-247.