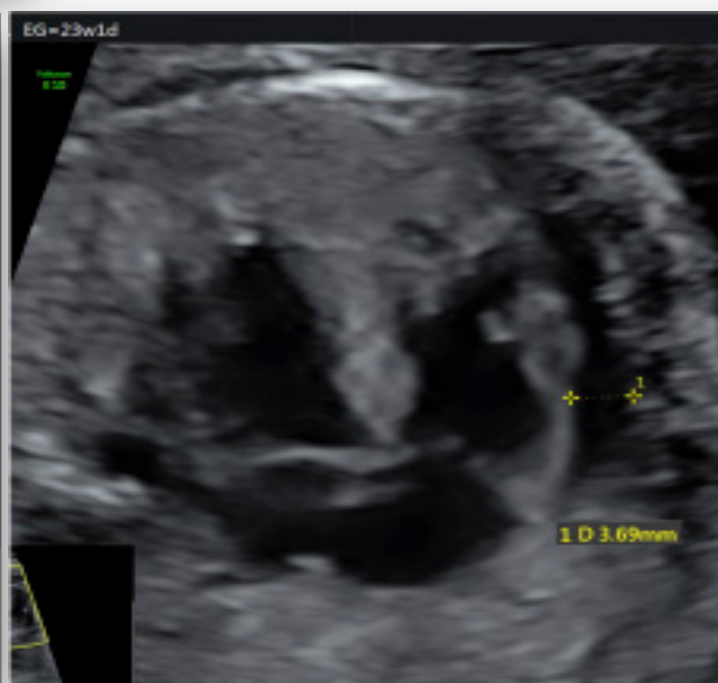
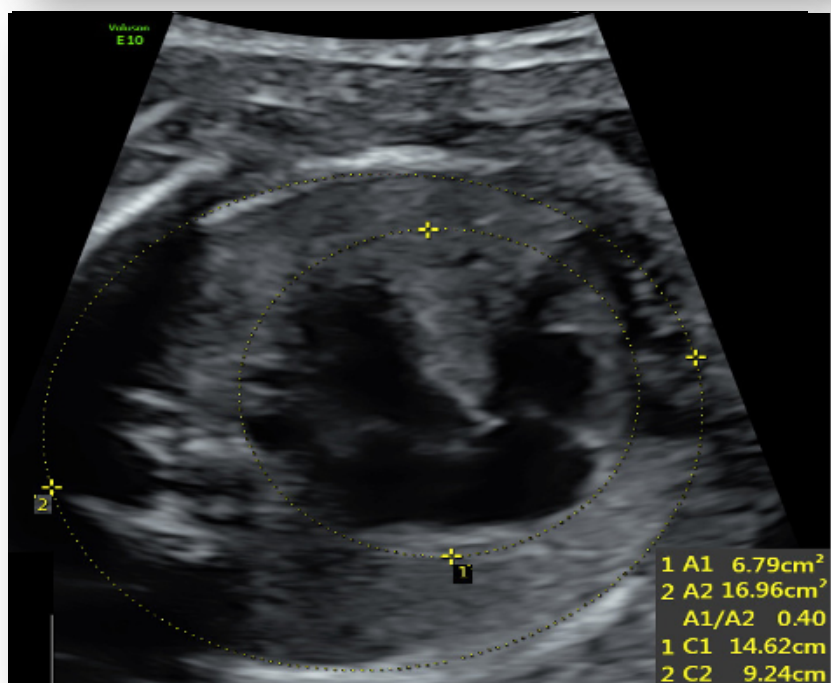
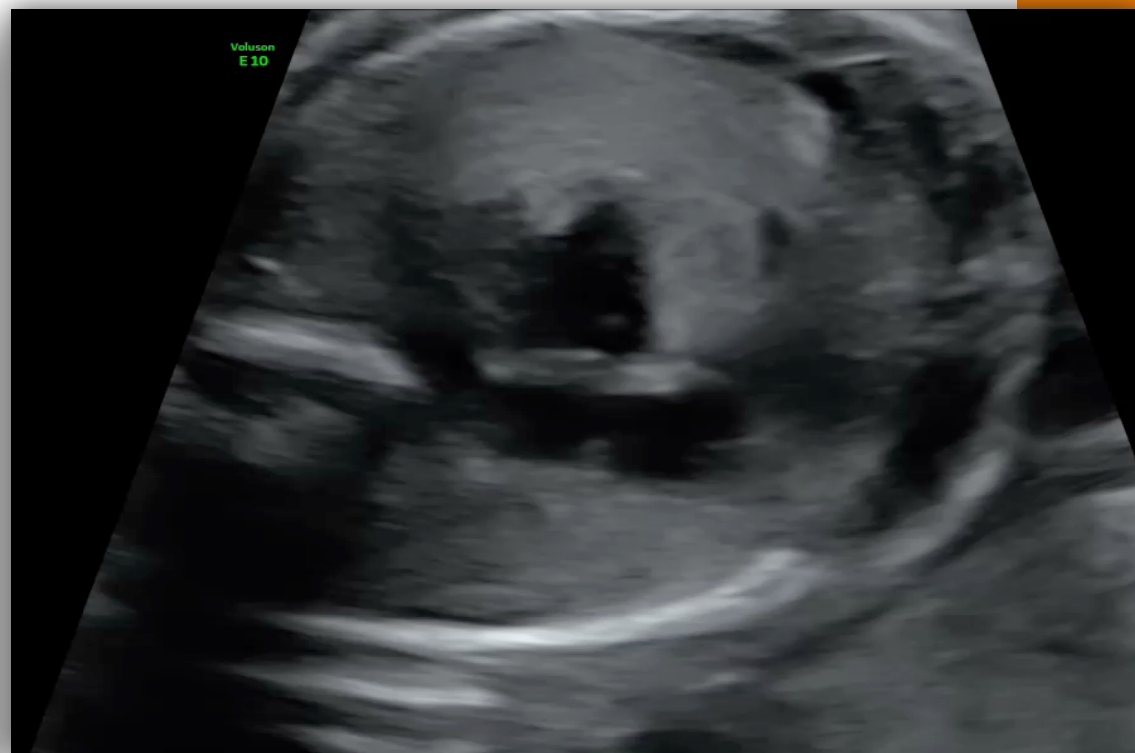
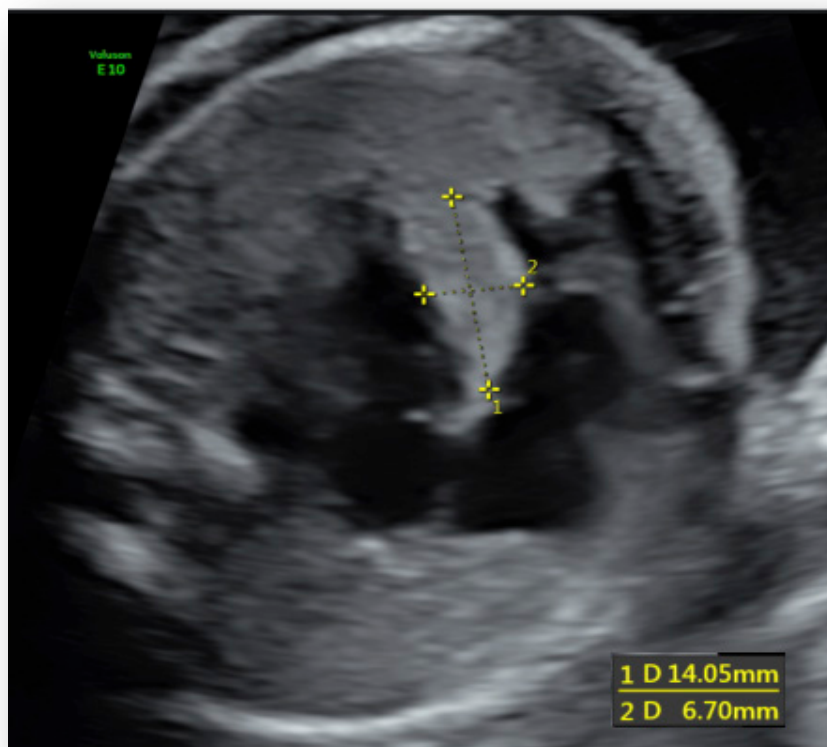




HIPERTRÒFIA MIOCÀRDICA NODULAR

SIGNE D'ESCLEROSI TUBEROSA?

Andrea Domènech - Resident 4t any
Hospital Universitari Mútua de Terrassa



Ecografia 2nT



UNIVERSITAT DE
BARCELONA

Estudi genètic

- Amniocentesi a les 21.2 SG
- QFPCR: 2cr 13, 2cr 18, 2cr21
- Cariotip: 46XX
- Arrays: arr (1-22,X)x2
- **Variant significat incert (VSI) gen TSC2:**

- ✓ QFPCR i cariotip
- ✓ CGH-arrays
- ✓ Mutacions
Esclerosi Tuberosa

Gen	Variant*	Zigositat	Herència	Classificació^
TSC2	NM_000548.4: c.539T>C	Heterozigosi	Autosòmica dominant	VSI
	NP_000539.2: p.(Leu180Pro)			

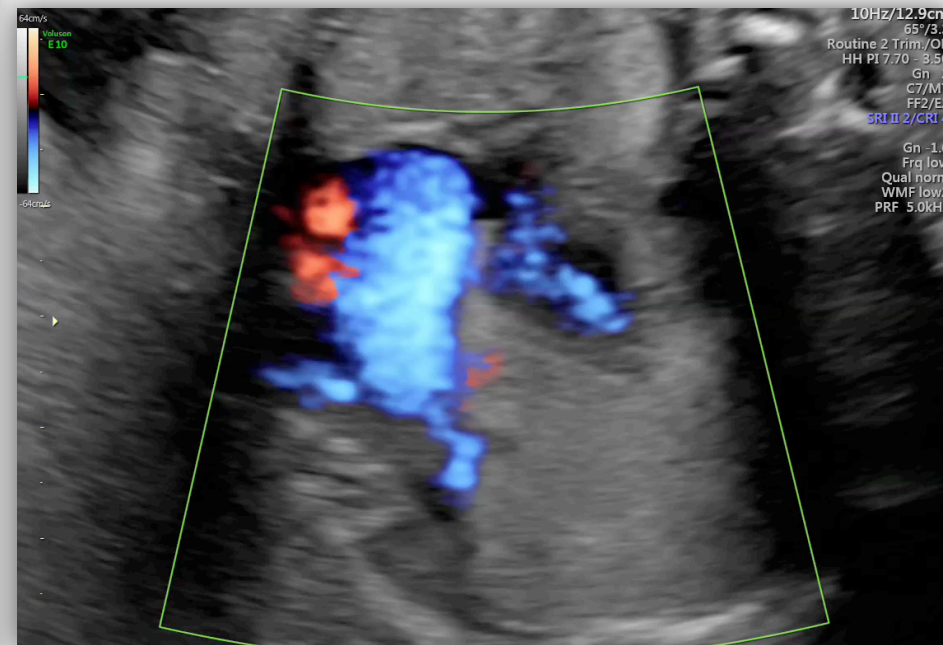
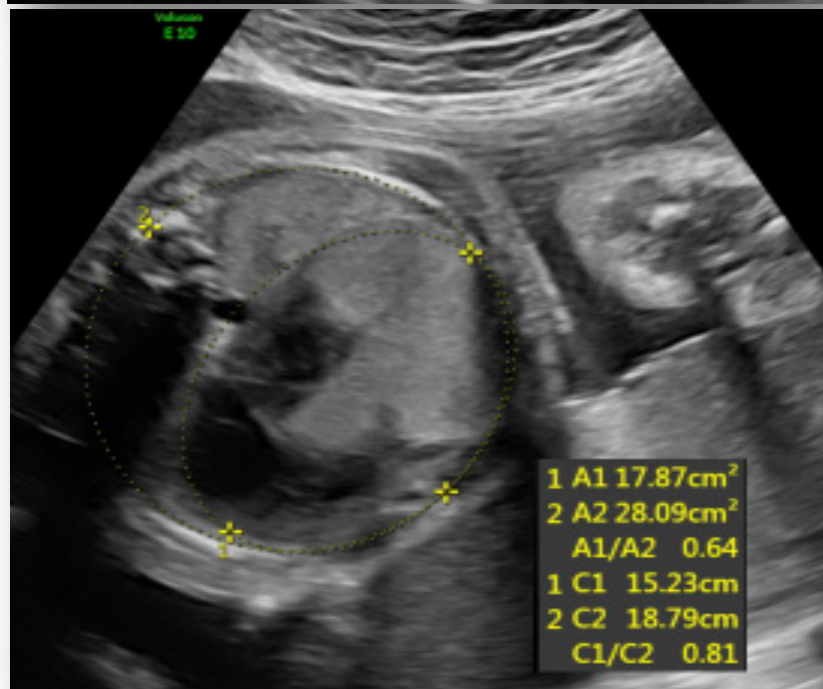
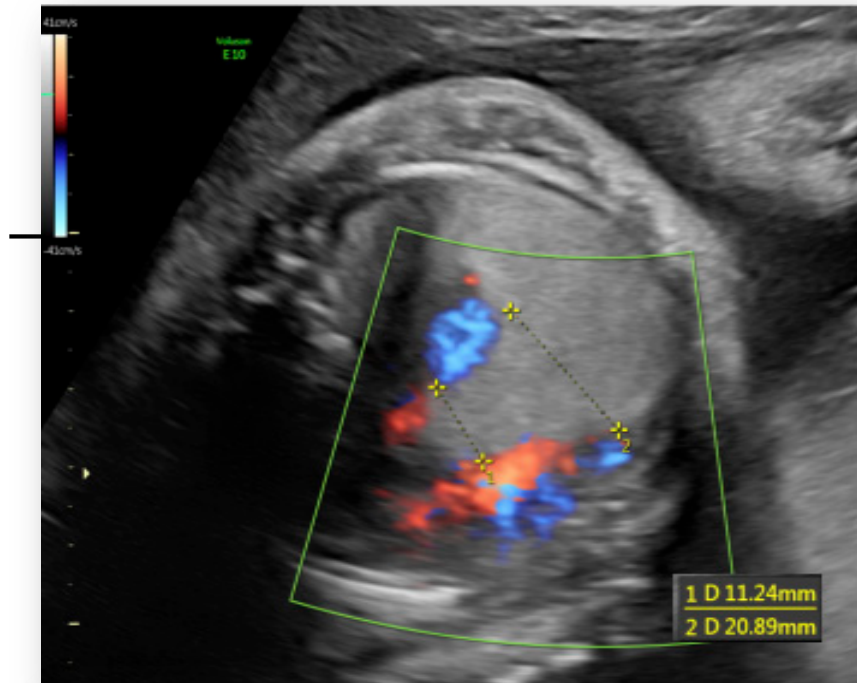


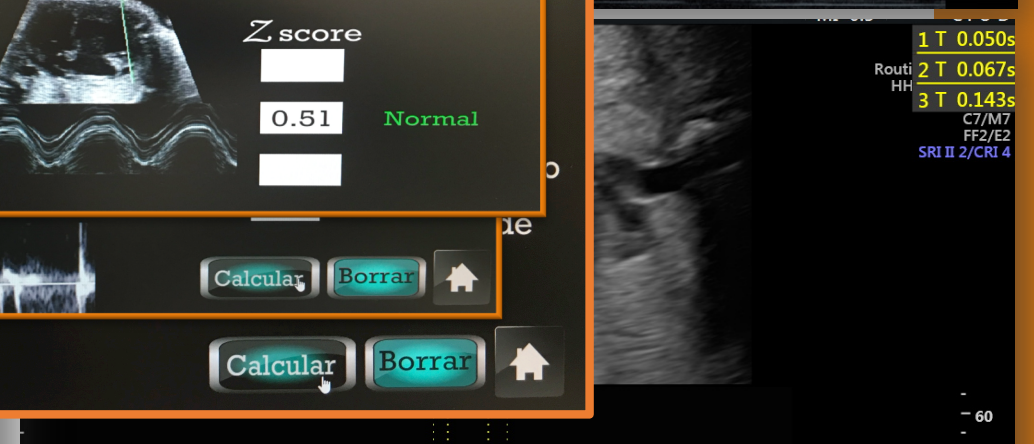
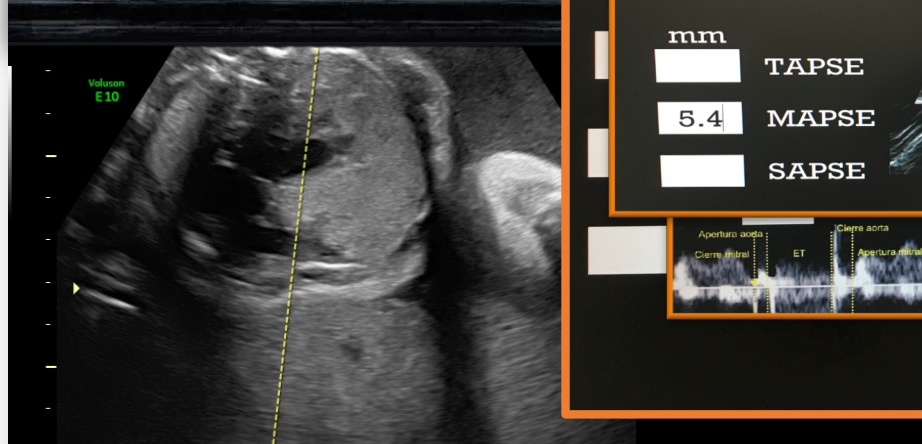
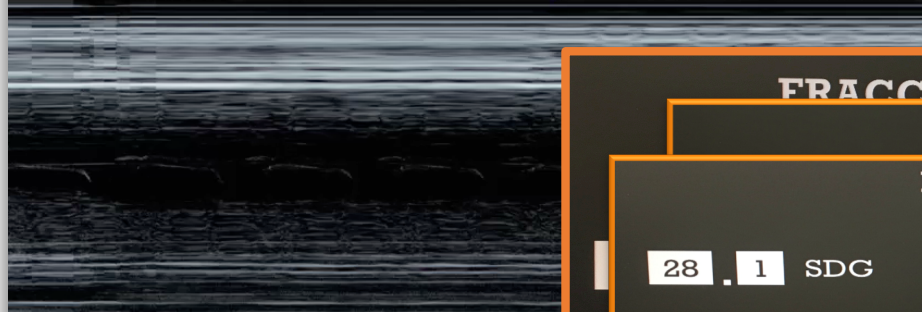
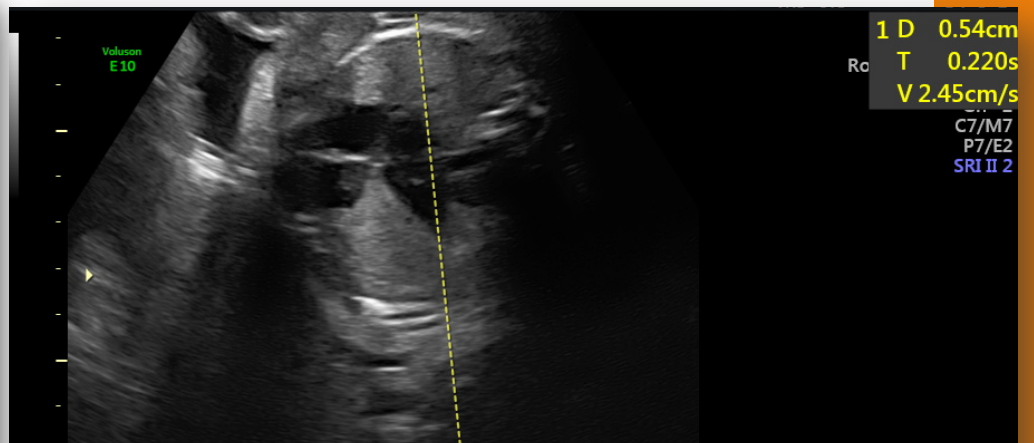
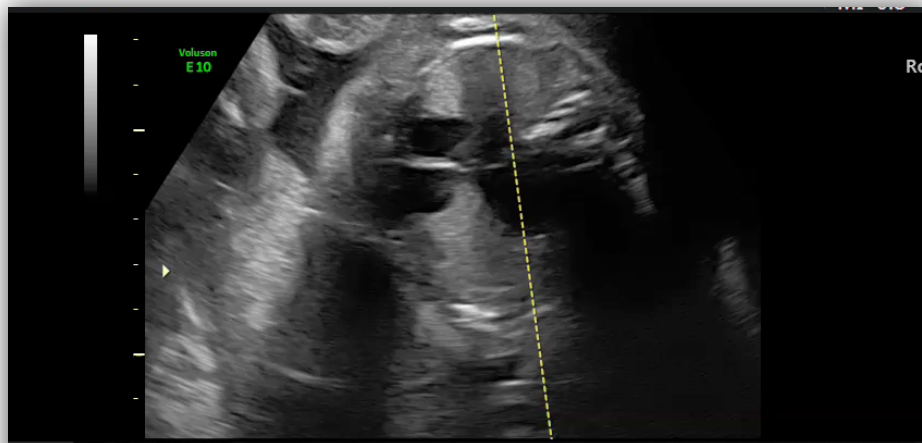
Estudi genètic dels pares

Variant heretada del pare en herència **AD** amb penetrància **completa**, passant a ser variant **“probablement benigna”**

28 setmanes

- Augment tumoracions
- Index cardiotoràcic
- Insuficiència tricuspídea





FRACCIÓN DE EFUSIÓN
ÍNDICE DE TEI
MODO - M

28 . 1 SDG

mm

TAPSE

5.4 MAPSE

SAPSE

Z score

0.51 Normal

Apertura aorta

Cierre aorta

Cierre mitral

ET

Apertura mitral

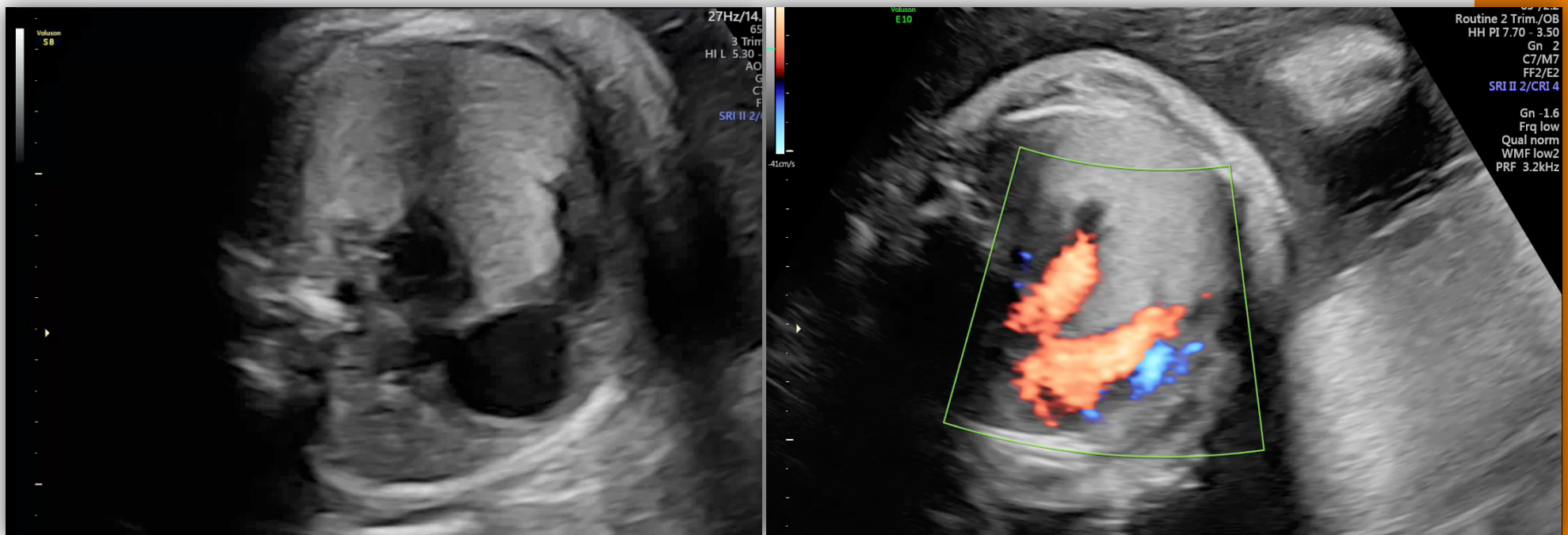
Calcular

Borrar

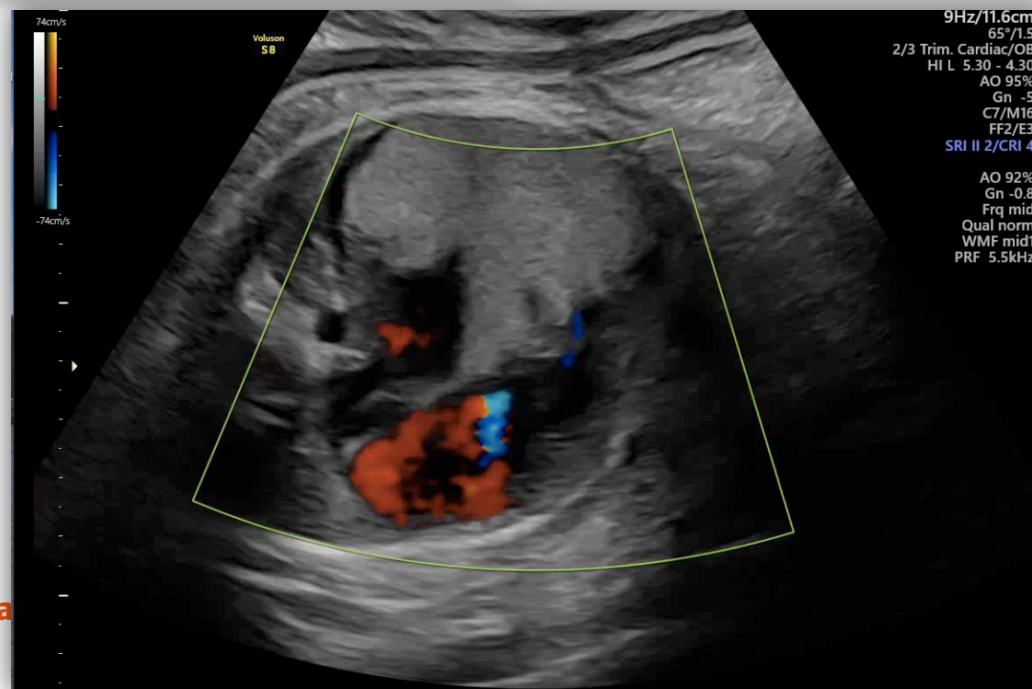
Calcular

Borrar

31 setmanes

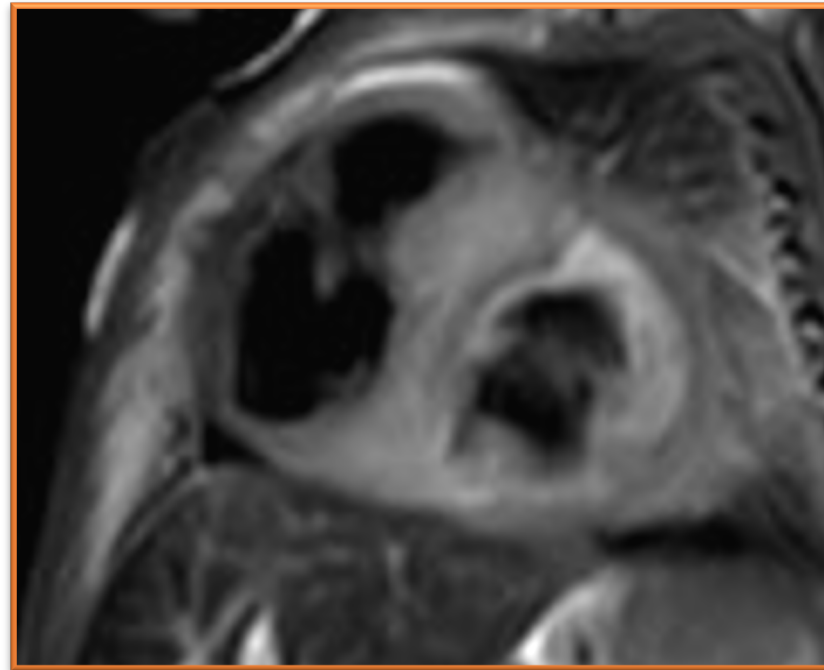
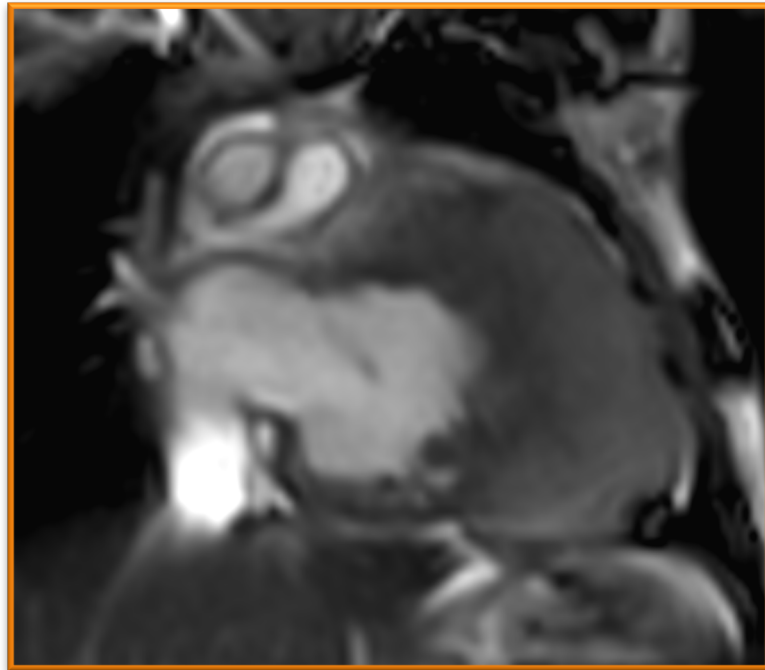


37 setmanes



Part i període neonatal

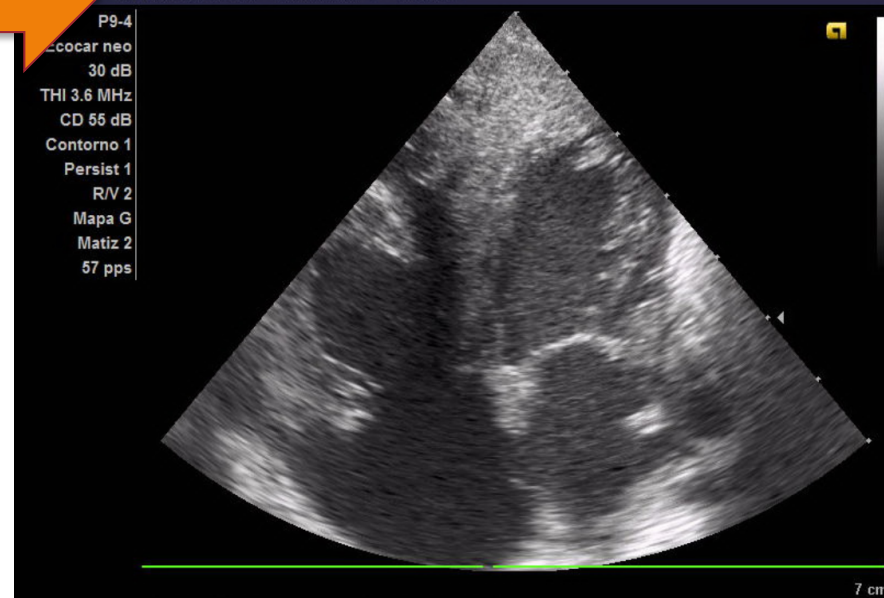
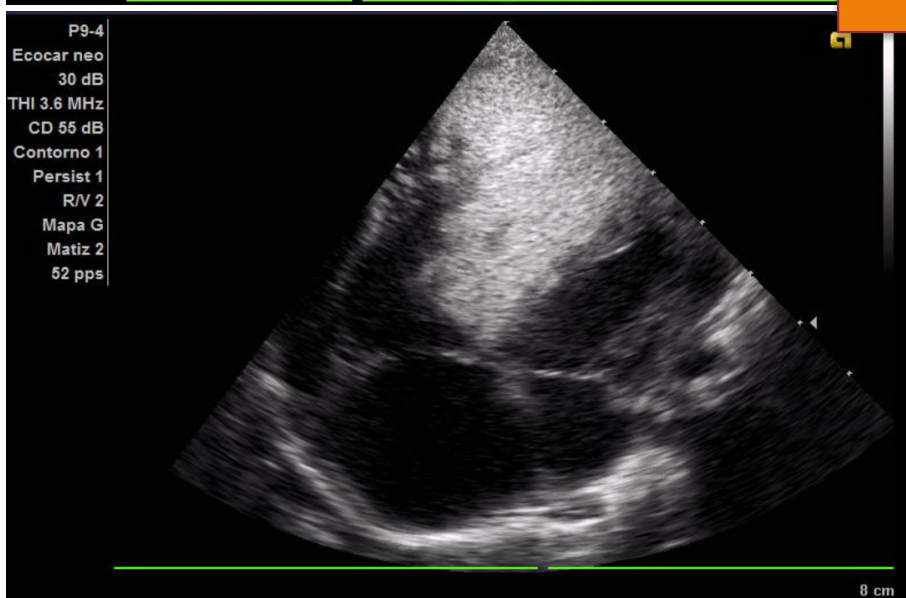
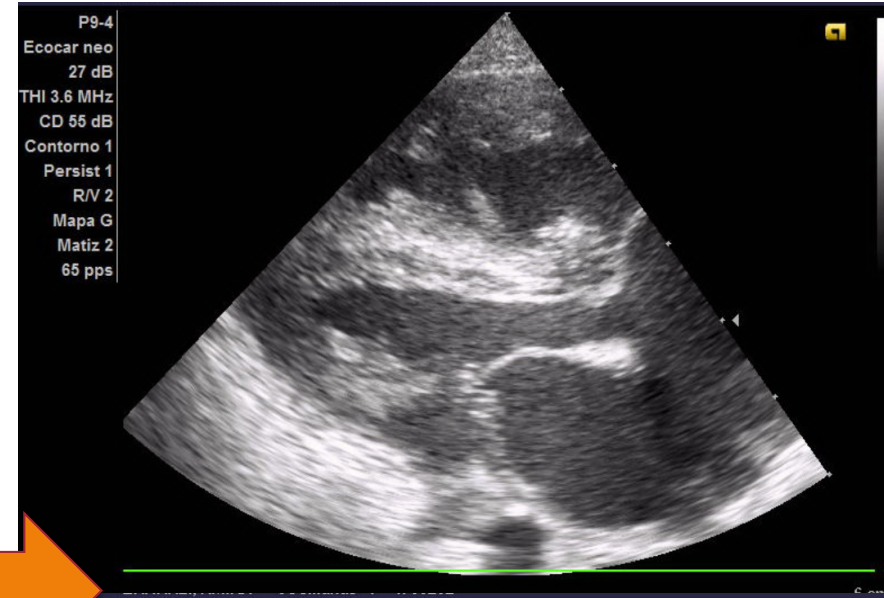
- Part distòcic amb fòrceps a les 39.4 SG
- APGAR 7/9, 3580g, saturació Hb 78-85%

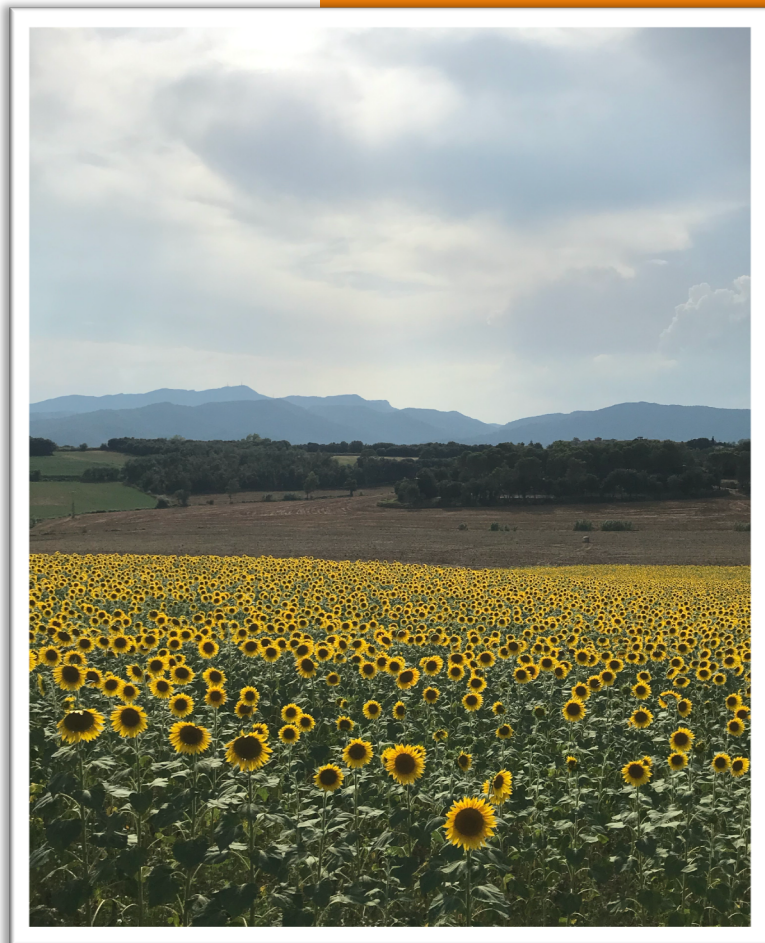


- Biòpsia miocàrdica (6 dies): **no concloent.**



Evolució amb Everolimus





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