

# Just a late pre-term?

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# Context

Gestational diabetes on diet

Antenatal ultrasound : left kidney duplication + suspicion of micropenis

- NIPT XY
- AFT : no pathogenic variant detected

Emergency C-section for pre-eclampsia at 36w5/7

- Apgar 9-10-10

Weight 3,7kg (P96) – Size 51 cm (P90) – PC 34 cm (P72)

Normal physical examination apart from suspicion of micropenis vs buried penis and hydrocele

→ Maternity

# Maternity

- One episode of **greenish** vomiting
- Feeding difficulties and weight loss
- Clinical examination :
  - normal except from a mild jaundice + slight pre-tibial edema
  - Normal parameters (T° - sat – BP – bpm)



Admission in Kangaroo Unit at H48

- Blood test : Hb 18.2 g/dL, pl. 244 000, WBC 4880, neutro 4730, CRP 0.9, nl kidney fct, nl iono, TA nle

# At the Kangaroo Unit

## In the first week of life

### Respiratory/hemodynamic

- Borderlines saturations (85-92%) and intermittent saturation differential
- Normal BP

### Digestive

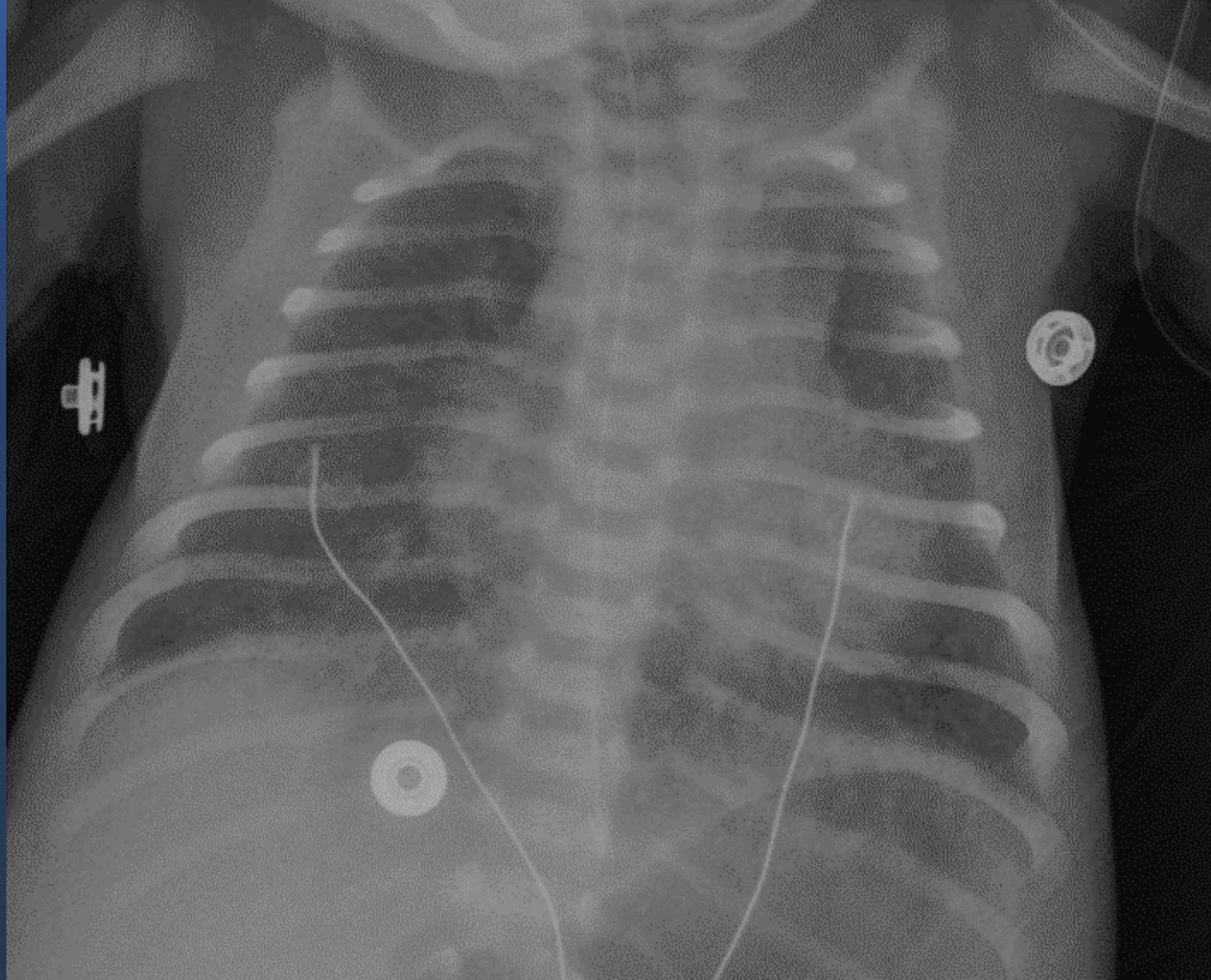
- Nasogastric feeding. Frequent regurgitation + gastric residue, not green anymore.
- Weight loss without feeding tube

### Infectious

- Apyrexia

### Renal

- Suspicion of micropenis (endoc + genetic) : negative
- Kidney ultrasound : duplication of the left calicial system, moderately dilated



- **Echocardiography**

RV bigger than the LV. Flattened septum.

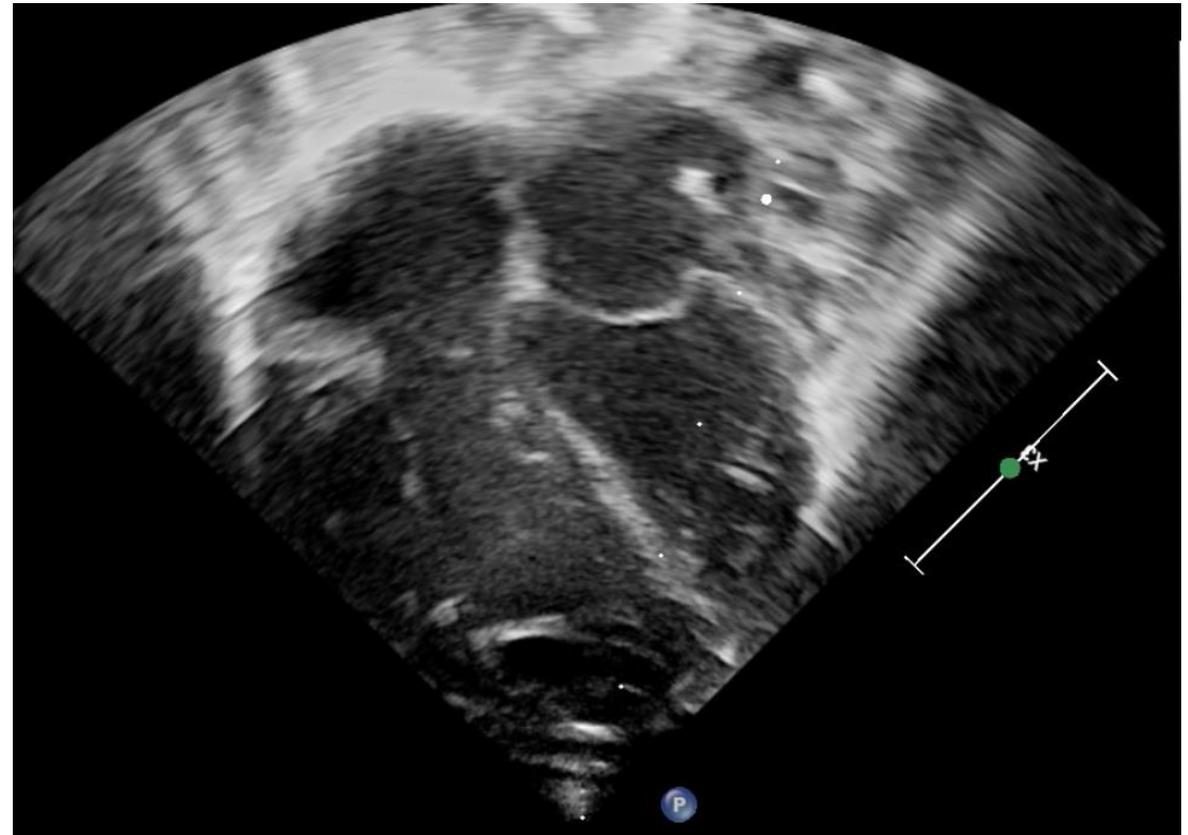
⇒ **Residual pulmonary hypertension,**

PFO, left aortic arch. Ductus arteriosus closed.

- **Blood tests : NI**

- **Respiratory microbiology (-)** (viral & bact)

**Treatment :** milk thickener



At D12

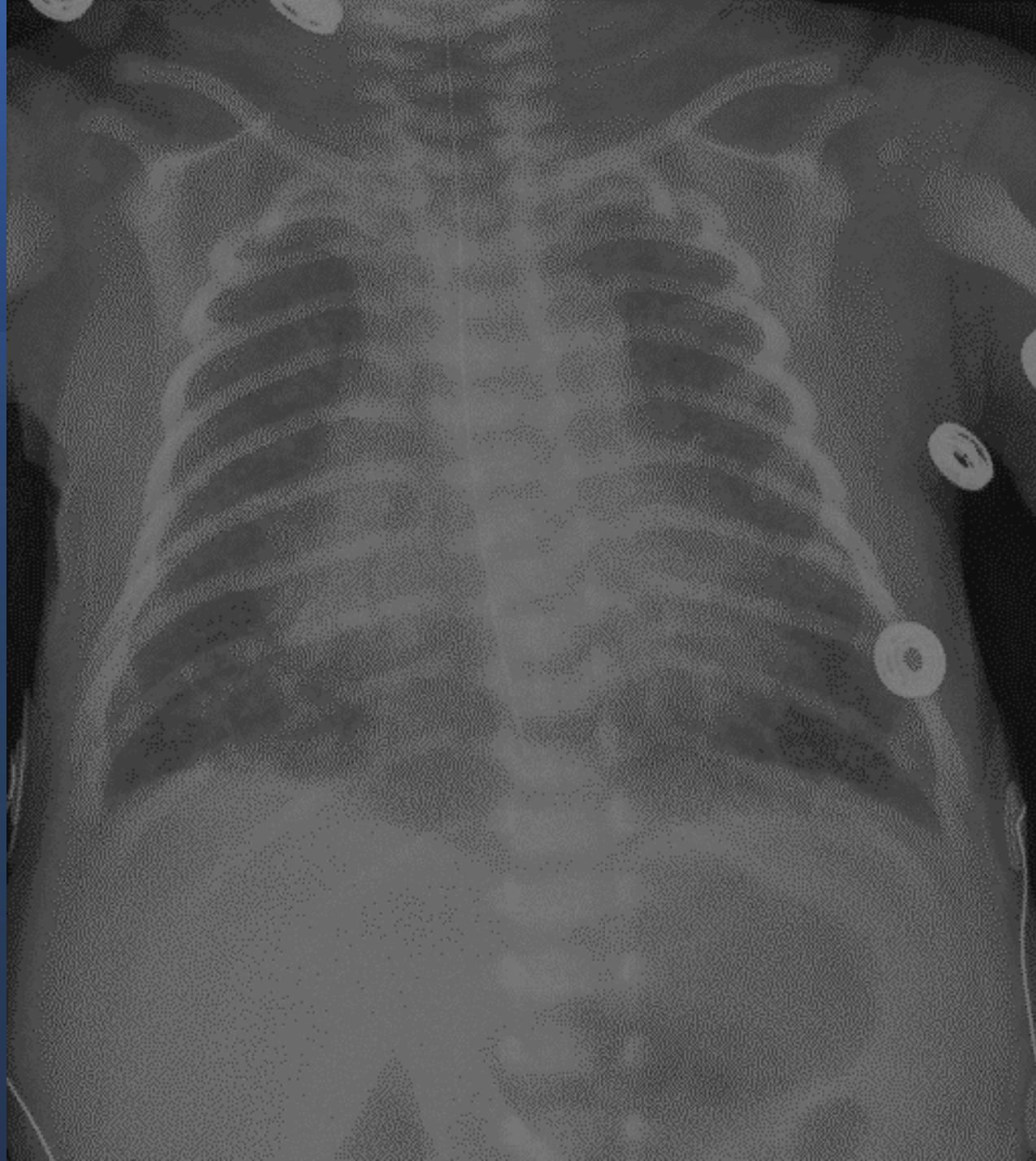
- Respiratory **deterioration** with intercostal retraction, polypnea and desaturation (80%). Normal breath sounds
- Systolic murmur 1/6, femoral pulse +/-, CRT <2 sec
- Mucous diarrhea - normal abdominal examination except for a hepatomegaly of 1 cm

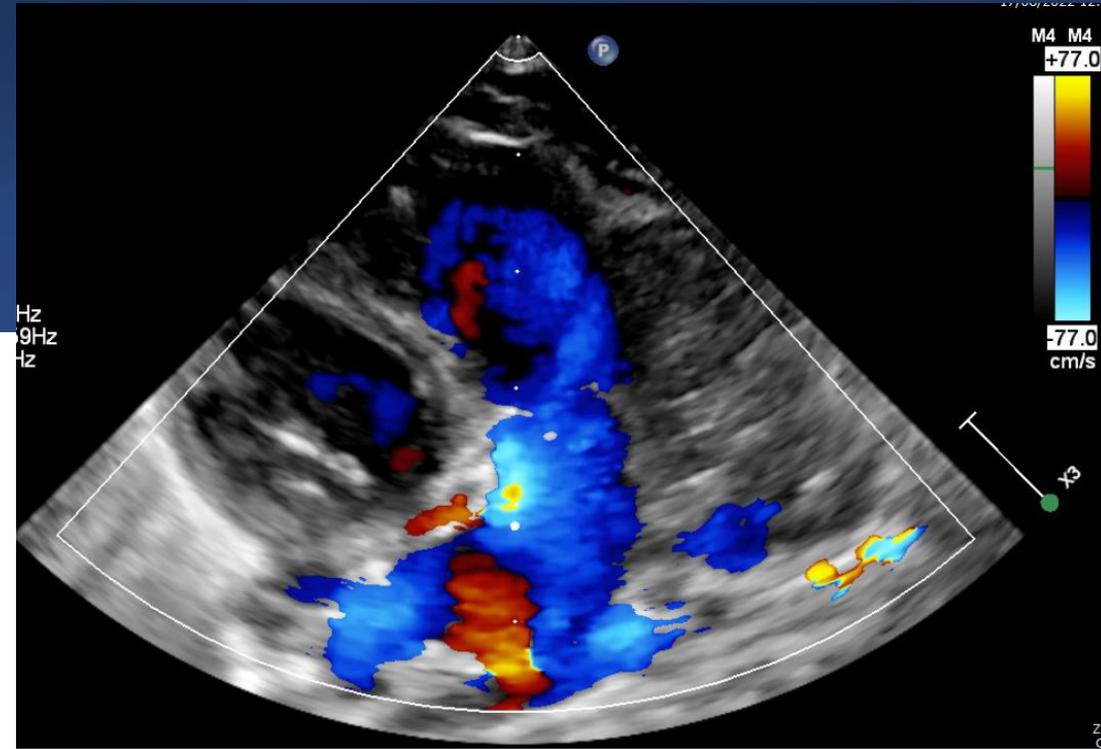
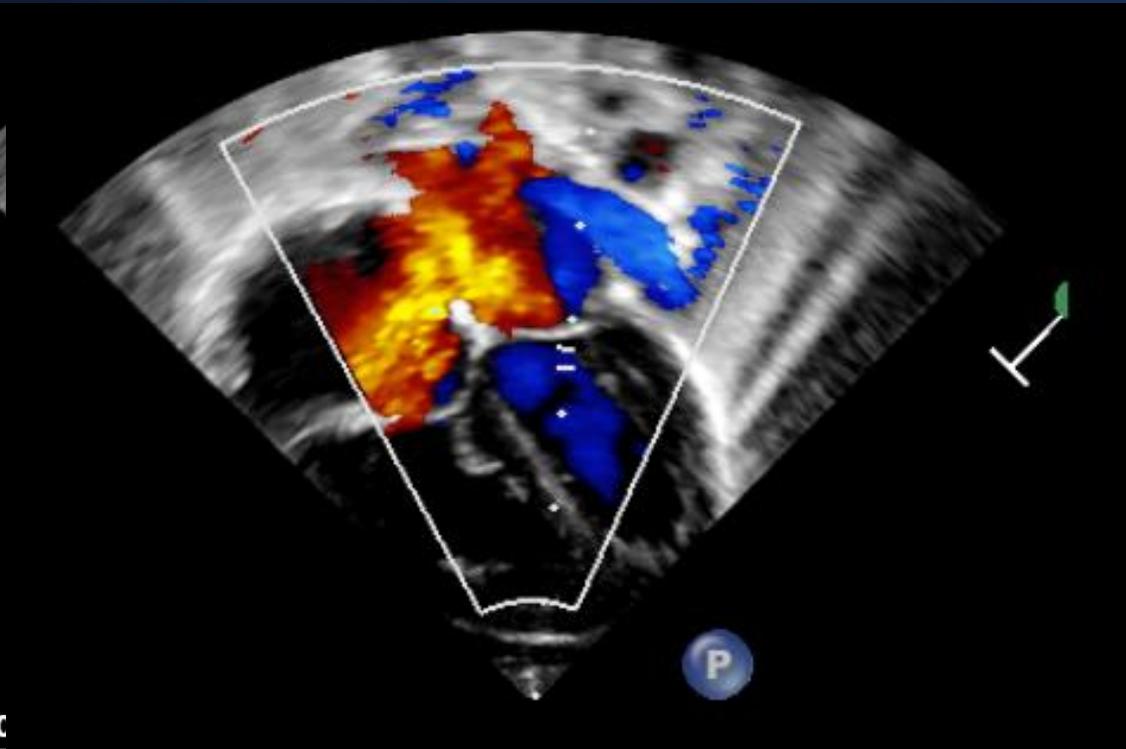


NEONATAL INTENSIVE CARE UNIT

Cpap +5 FiO<sub>2</sub> max 40%

**YOUR NEXT STEP?**





- **Echocardiography**

RV bigger than LV. Flattened septum.

ASD 2 vs large PFO

**PDA re-open with bidirectional shunt and preferentially L -> R**

**Residual pulmonary hypertension 2/3 systemic**

## Evolution

### At D15,

- Stop Cpap
- Feeding improvement - autonomy of 80%

### At D17, in the afternoon,

- vomiting and bloated belly
- Desaturations – O2 with nasal canula
- => Abdominal US : normal

D17, in the evening

## **Rapid worsening**

Peritoneal abdomen (pain, no transit,..)

Systemic signs (polypnea, retractions, tachycardia,..)

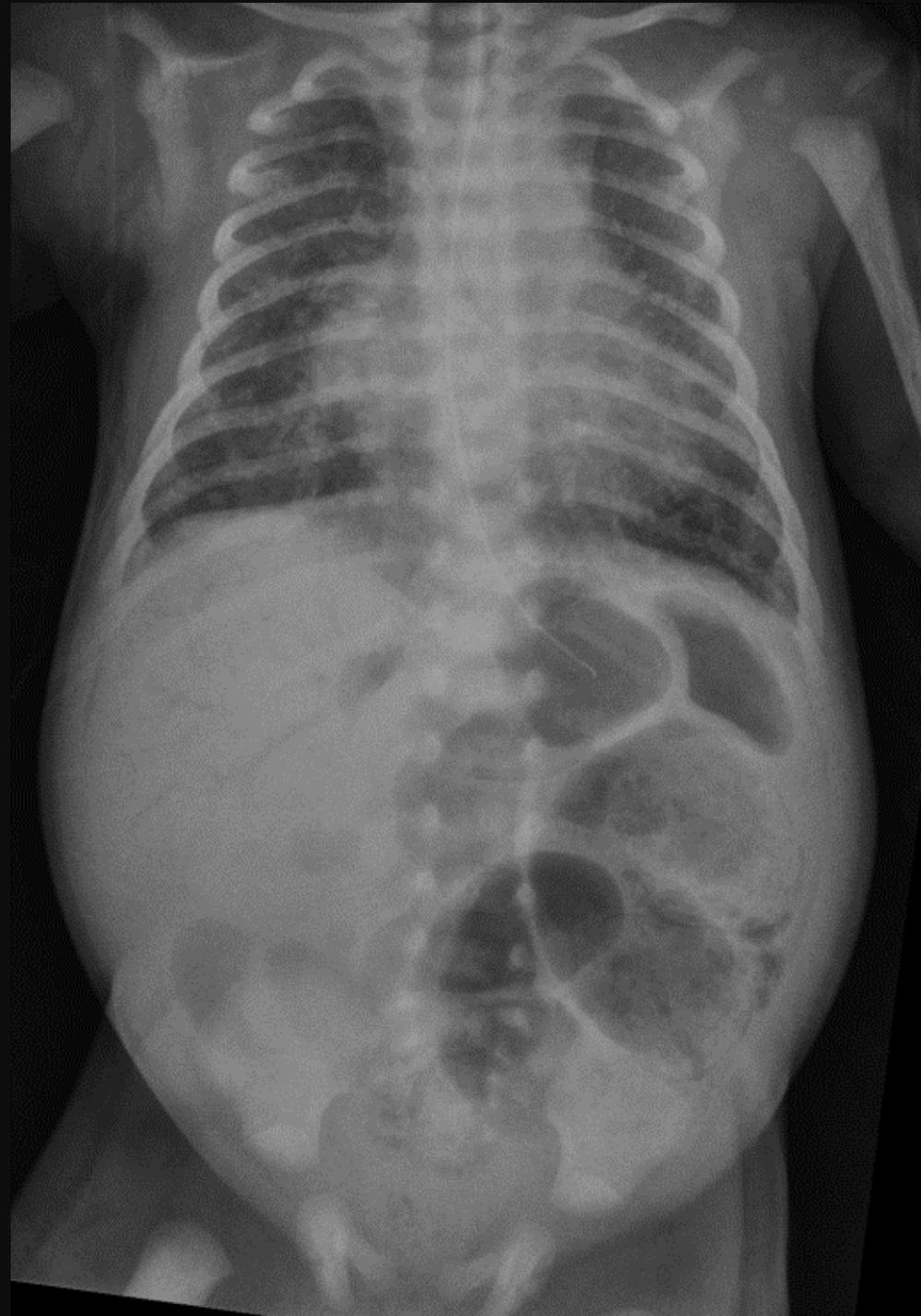
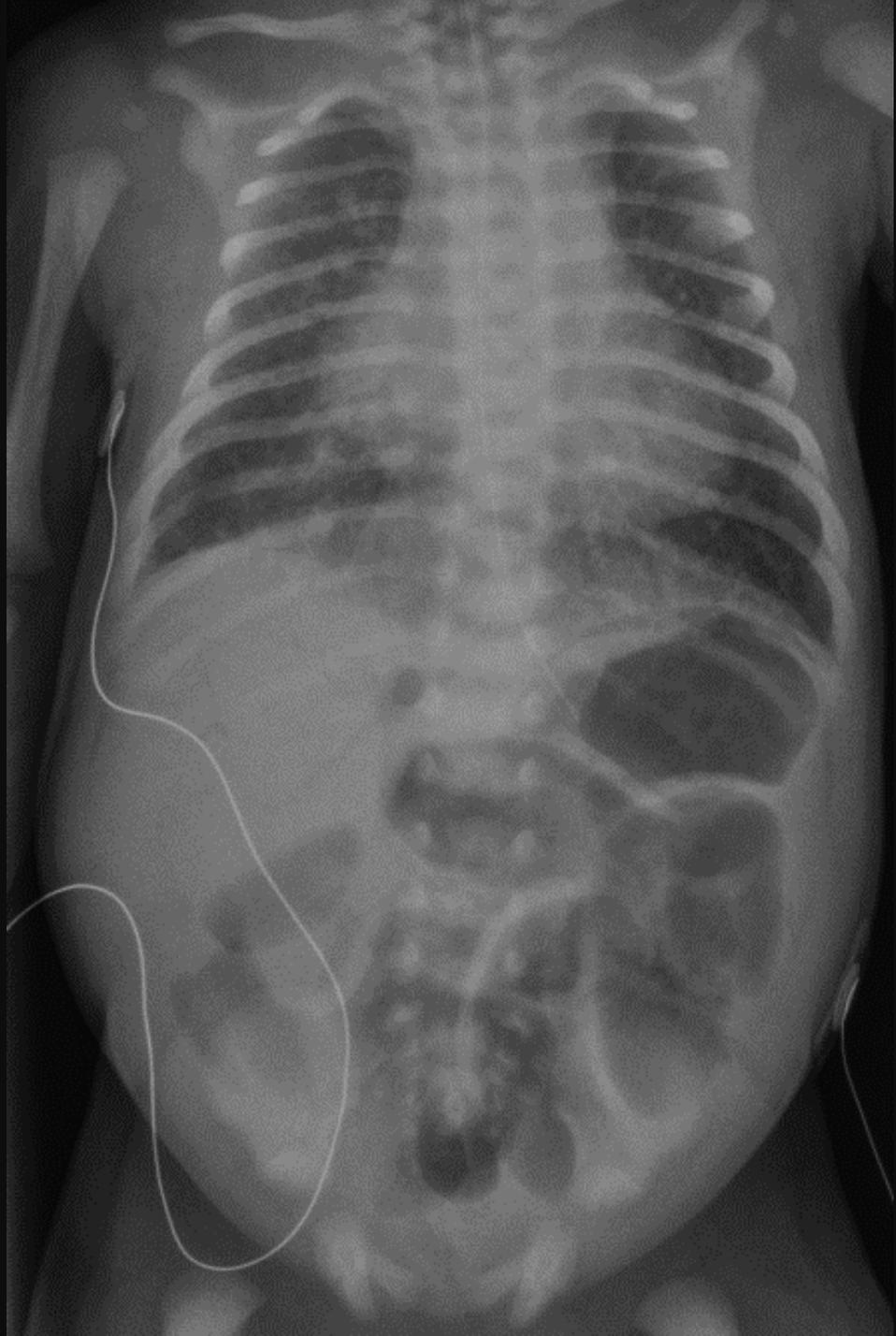
What do you do?

## **Workup**

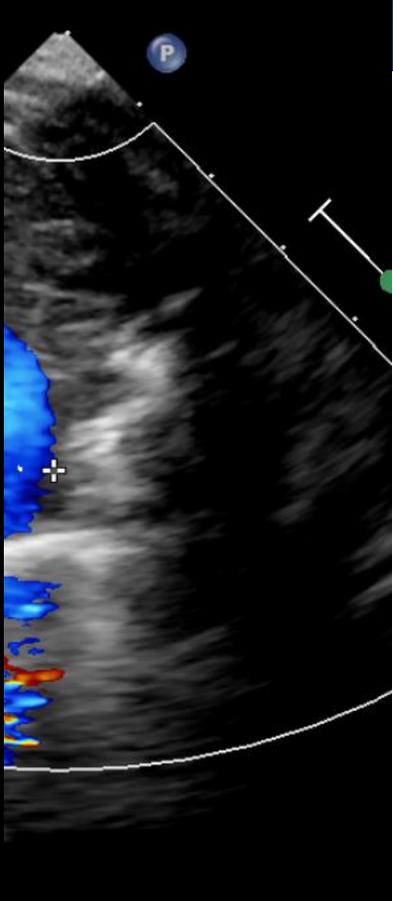
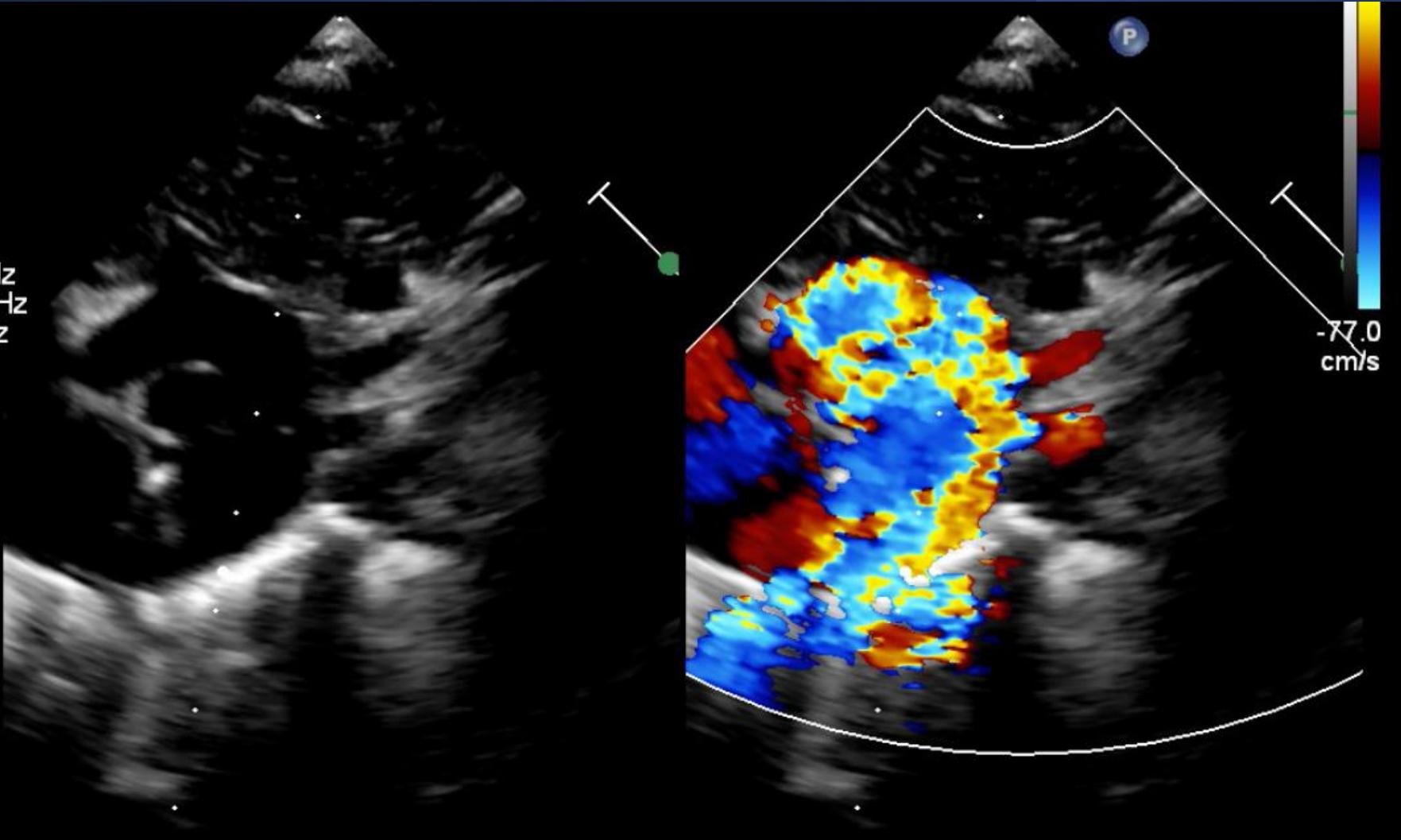
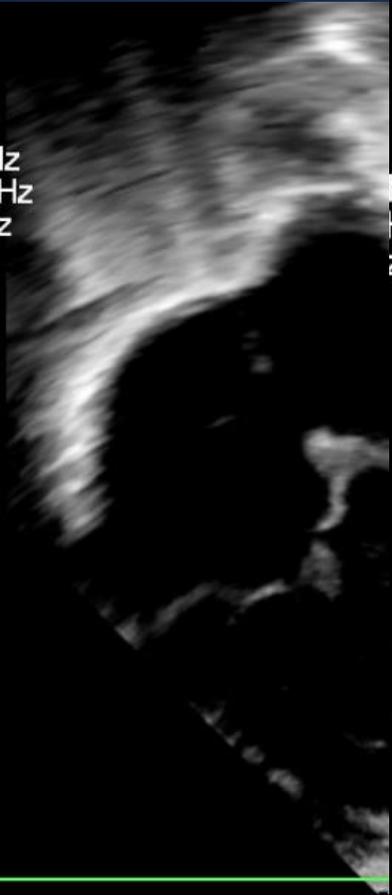
Repeat radiology

Biology

What else?



Ec



- ASD 2 vs PFO, shunt L->R, LPA 15-18mm,
- Normal abdominal aortic doppler
- High suspicion of **aorto-pulmonary window of 7-8 mm**

# Evolution

## D19

Transfer to the intensive care unit of the CUSL  
(possible surgery & CHD)

Hemodynamic deterioration with suspected intestinal perforation

- Exploratory laparotomy
  - NEC with pancolitis
  - Intestinal **malrotation**
  - **Congenital short bowel** (+-60 cm)

+ Repeat  
physical  
examination

Aorto-pulmonary window

Intestinal malrotation + congenital short bowel

Bilateral hydronephrosis

White matter hyperechogenicity

Bilateral mydriasis



Suspicion of « Multisystemic Smooth Muscle  
Dysfunction Syndrome » (MSMD)

# MSMD (<1/100 000 cases in the world)

## **Symptoms :**

- PDA with pulmonary hypertension ; other CHD
- Vasculopathy
- Cerebrovascular disease Moyamoya-like
- Intestinal malrotation and intestinal hypoperistalsis
- Hypotonic bladder
- Congenital mid-dilated pupils non-reactive to light

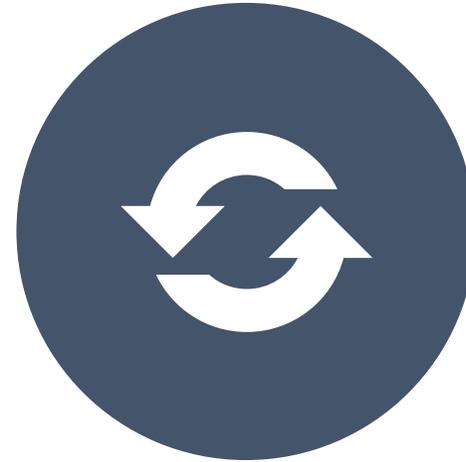
**Complications :** high risk of aortic dissection, thrombosis, stroke, pulmonary complication

**Diagnosis :** Genetic confirmation by the presence of recurrent mutations (Arg179) in the ACTA2 gene

# TAKE HOME MESSAGE



**PHYSICAL EXAMINATION**  
**(EYE IS NOT EASY)**



**REPEAT, REPEAT, REPEAT**

We only find something out if we're looking for it