



# A Singular Beginning, a Complex Journey

Gertjan Marissens



Universitair  
Ziekenhuis  
Brussel



## 000 A SINGULAR BEGINNING

An assisted start

- Maternal history
  - G1P0
  - No relevant medical history
- Conception
  - IVF-ICSI
  - Single Embryo Transfer

And then... the first ultrasound



Figure 1: Ultrasonic image of a DCDA pregnancy at 8 weeks gestation. (Source: Ville Y. et al., Hum Reprod Update. 2000;6(4):396-403. © Oxford University Press)

## 000 A COMPLEX JOURNEY

The fluid emerges

- **Routine follow-up:** normal
- **20-week ultrasound:** a second surprise — in twin 1



Figure 2: fetal US showing ascites



Figure 3: fetal US showing ascites



Figure 4: fetal US showing ascites

## 000 A COMPLEX JOURNEY

The abdomen that kept growing

- Ascites **progressed** gradually
- Extensive **diagnostic work-up**:
  - Amniocentesis
    - Genetics: Karyotype 46, XX; microarray normal
    - Infectious: Negative for CMV, enterovirus and parvovirus
  - an MRI was planned at 30 weeks
- **29+5 weeks** — rising concerns



Figure 5: Fetal US at 22+6 weeks

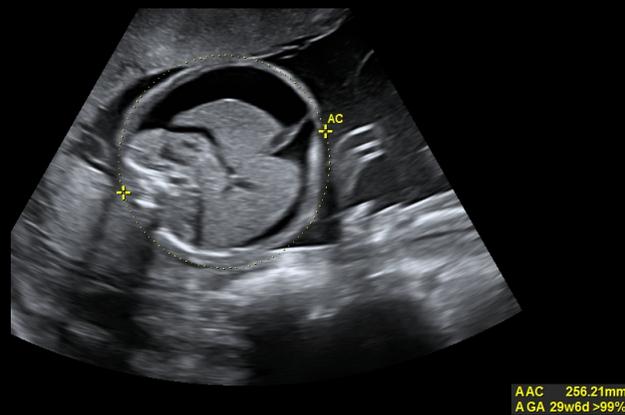


Figure 6: Fetal US at 23+5 weeks



Figure 7: Fetal US at 23+5 weeks



Figure 8: Fetal US at 29+5 weeks

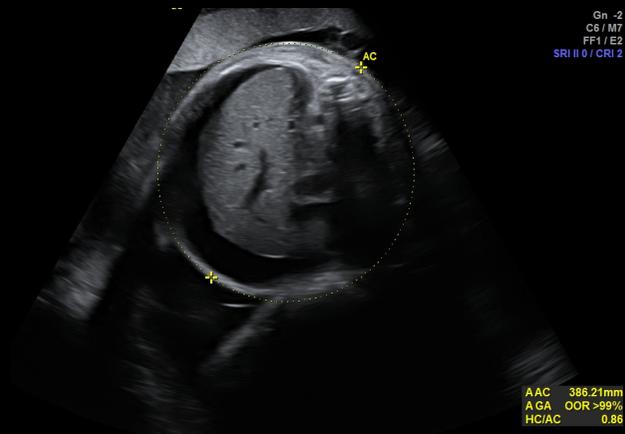


Figure 9: Fetal US of twin 1 at 31+5 weeks



Figure 10: Fetal US of twin 2 at 31+5 weeks

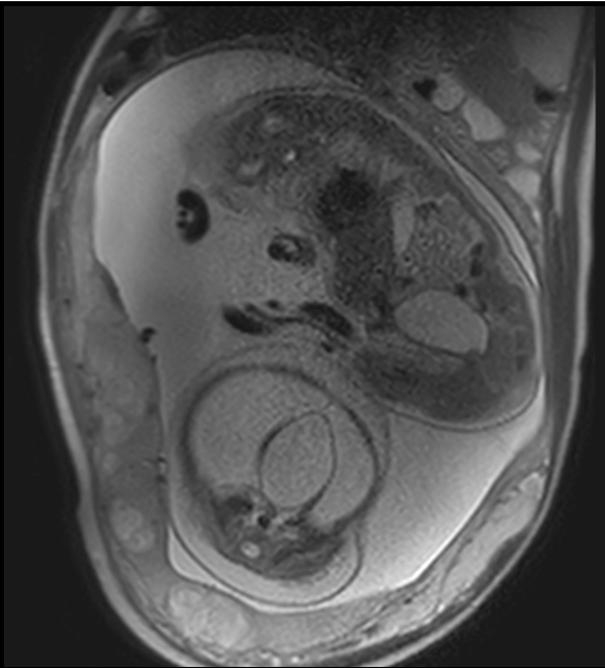


Figure 11: T2W image showing ascites

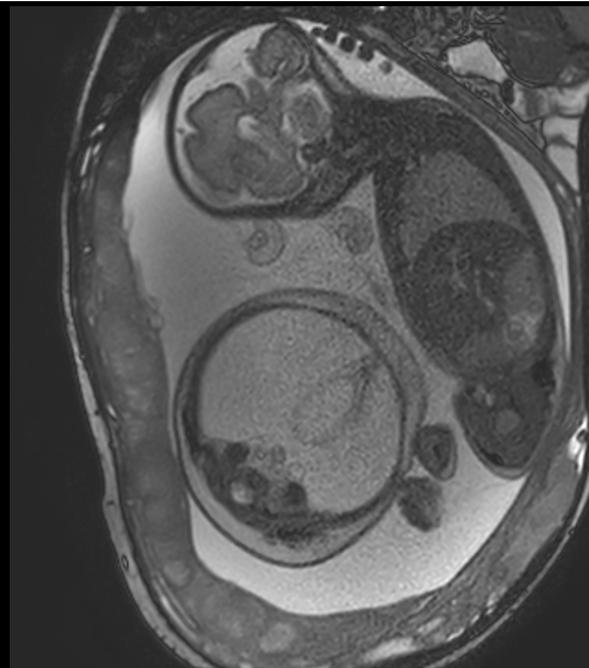


Figure 12: BTFE image showing ascites

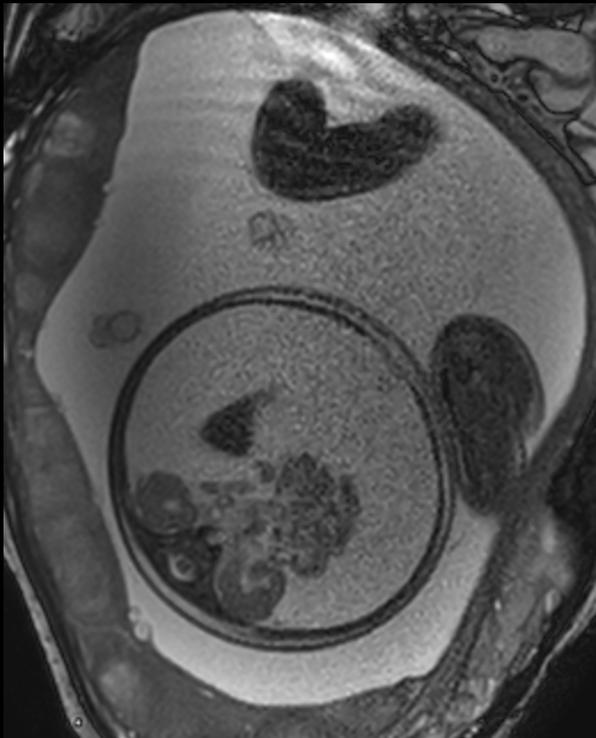


Figure 13: Axial BTFE image of twin 1 showing ascites and intestinal compression

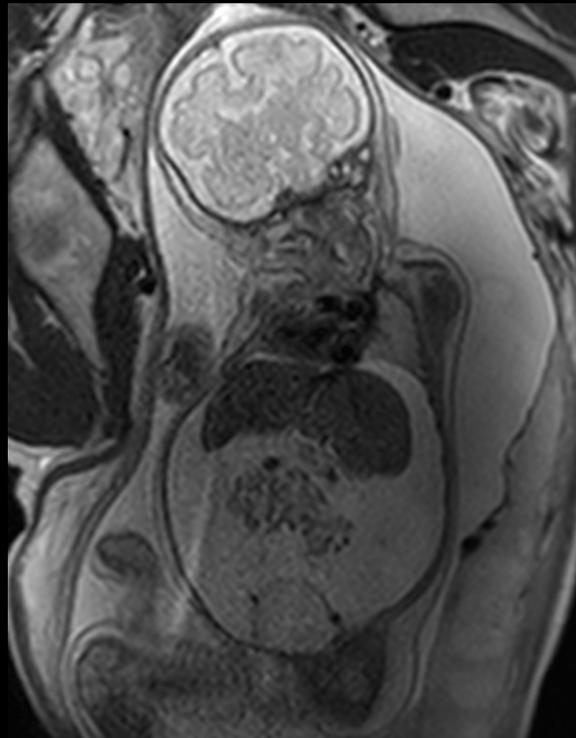


Figure 14: Coronal TW image of twin 1

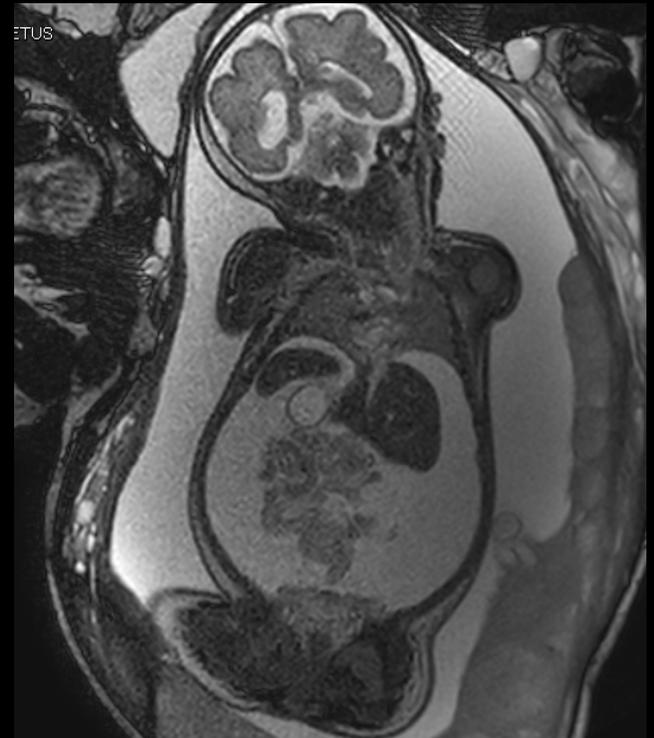


Figure 15: Coronal BTFE image of twin 1 showing intestinal and pulmonary compression

## 000 A COMPLEX JOURNEY

### A shift in balance

- **33+5 weeks** — emerging signs of fetal hydrops
  - **Urgent** caesarean section
    - Initially breathing, followed by **bradycardia and apnea**
    - **Difficult** ventilation
      - Paracentesis with drainage of 150 ml ascitic fluid
      - Persistent respiratory distress → intubation required

## 000 A COMPLEX JOURNEY

What's going on?

- What's your first impression of this case?
- Which investigations would you order?
- How would you manage this case?



## 000 A COMPLEX JOURNEY

### Initial findings

- **Clinical exam:**
  - Marked ascites and generalized edema
- **Laboratory results**
  - Ascitic fluid: ↑ cell count and ↑ triglycerides
  - Blood: Normal blood gas, no infection, mild anemia and leukopenia
- **Imaging**
  - Abdominal US: Ascites, left adrenal hematoma
  - Cardiac/ Lung US: Moderately reduced LV function
  - Cerebral US: Punctate white matter lesions

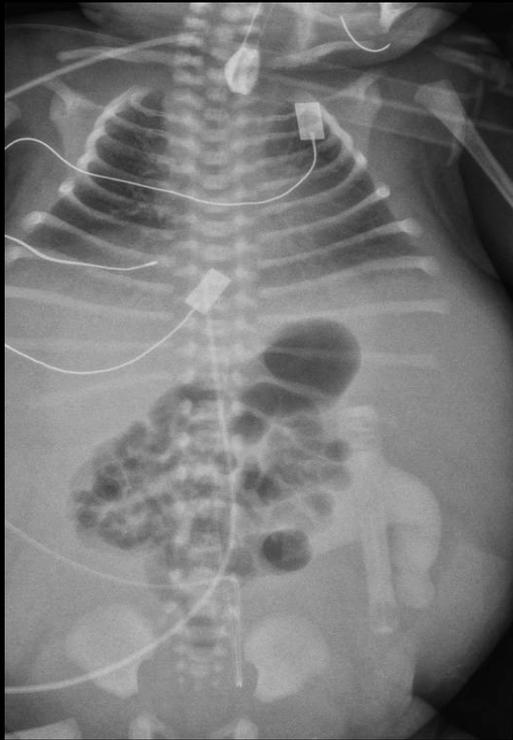


Figure 16: X-ray showing ascites

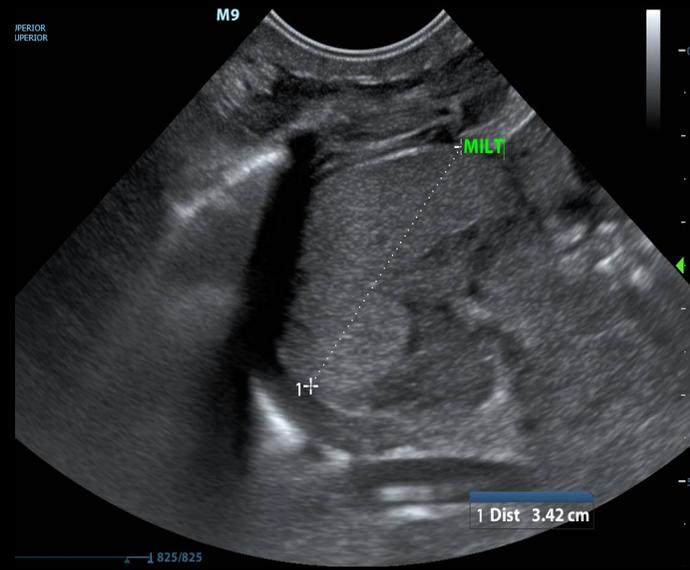


Figure 17: Ultrasound showing ascites around spleen

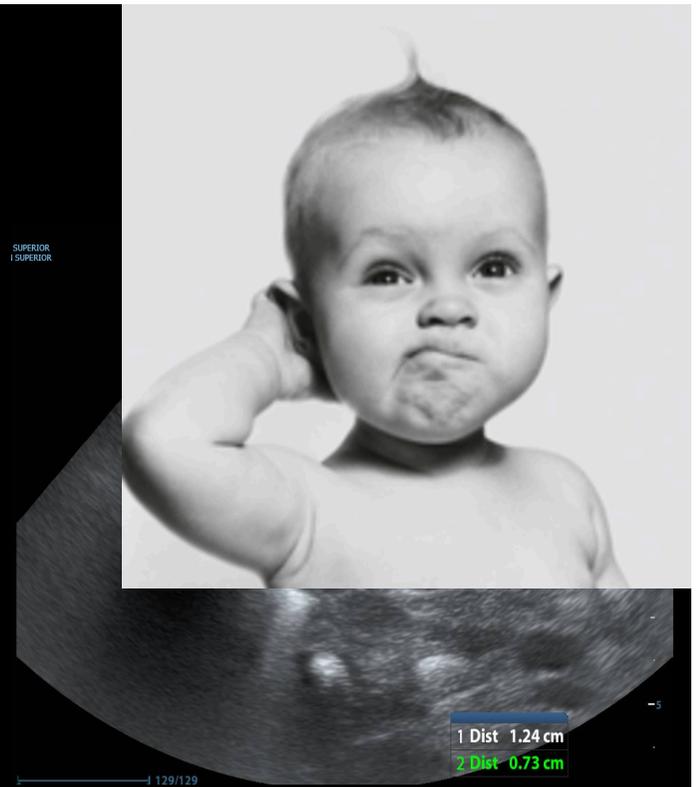


Figure 18: Ultrasound showing L adrenal hematoma



Figure 19: CUS showing punctate white matter lesions

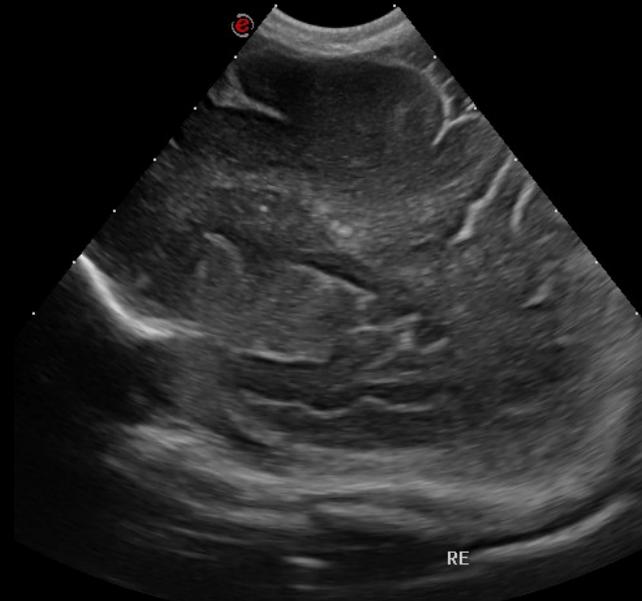


Figure 20: CUS showing punctate white matter lesions

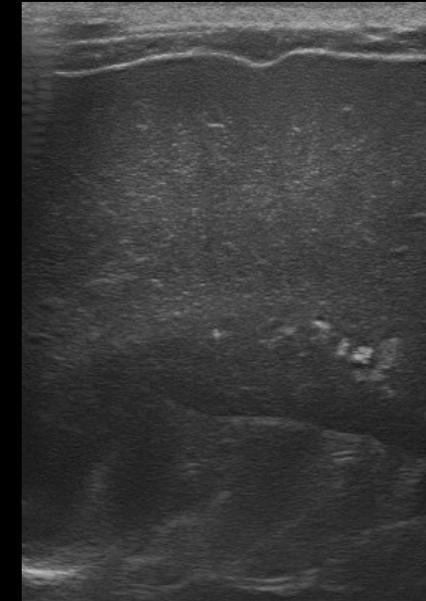


Figure 21: CUS showing punctate white matter lesions

## 000 A COMPLEX JOURNEY

How the case evolved

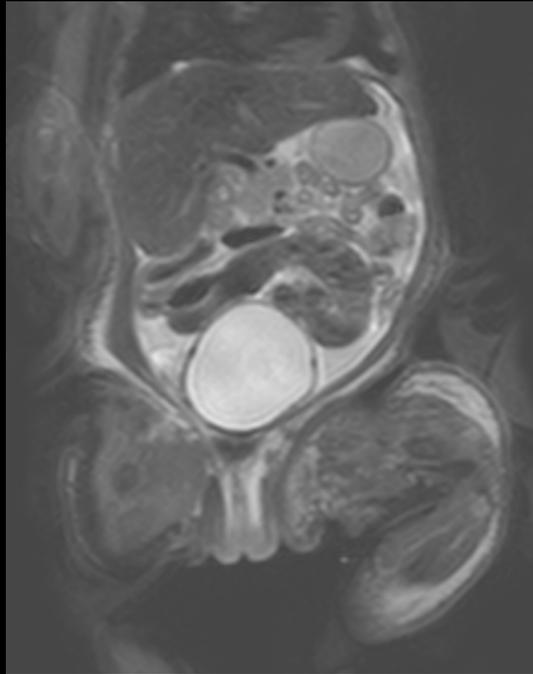


## 000 A COMPLEX JOURNEY

Identifying the underlying pathology



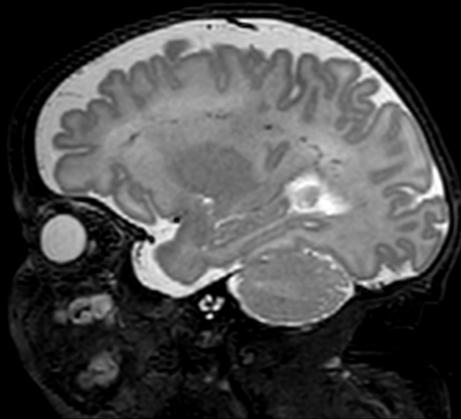
- Twin 1
  - Chylous ascites
  - Adrenal hematoma, white-matter lesions and LV dysfunction
- Further investigations
  - MRI Lymphangiography and Abdomen (Day 31):
    - Persistent ascites
    - Lymphedema of the left leg and abdominal wall
  - MRI Brain (Day 59):
    - Periventricular white-matter lesions
  - Genetics & metabolic screening



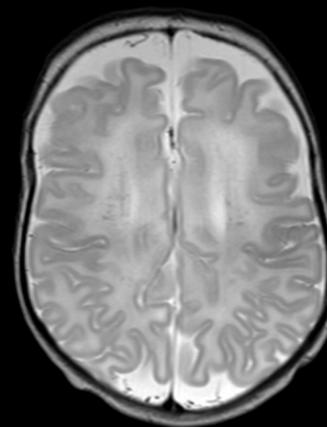
*Figure 22: T2 Cor FS showing ascites*



*Figure 23: T2 Cor showing ascites*



*Figure 24: T2W showing white matter lesions*



*Figure 25: T2W showing white matter lesions*

## 000 A COMPLEX JOURNEY

Beyond the neonatal period

- 5 months — Favorable evolution
- 8 months — Reticular rash
- 13 months — Soft, subcutaneous swelling
- 17 months — Asymmetric overgrowth

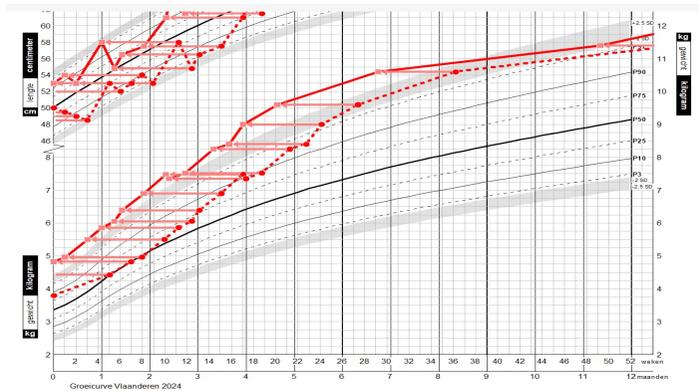


Figure 26: Growth curve (weight) in the first year of life

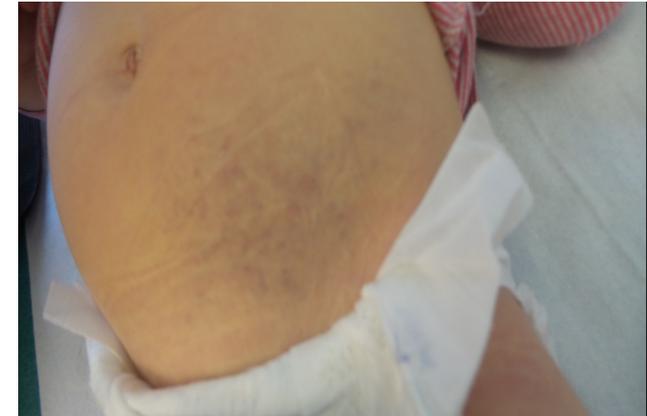
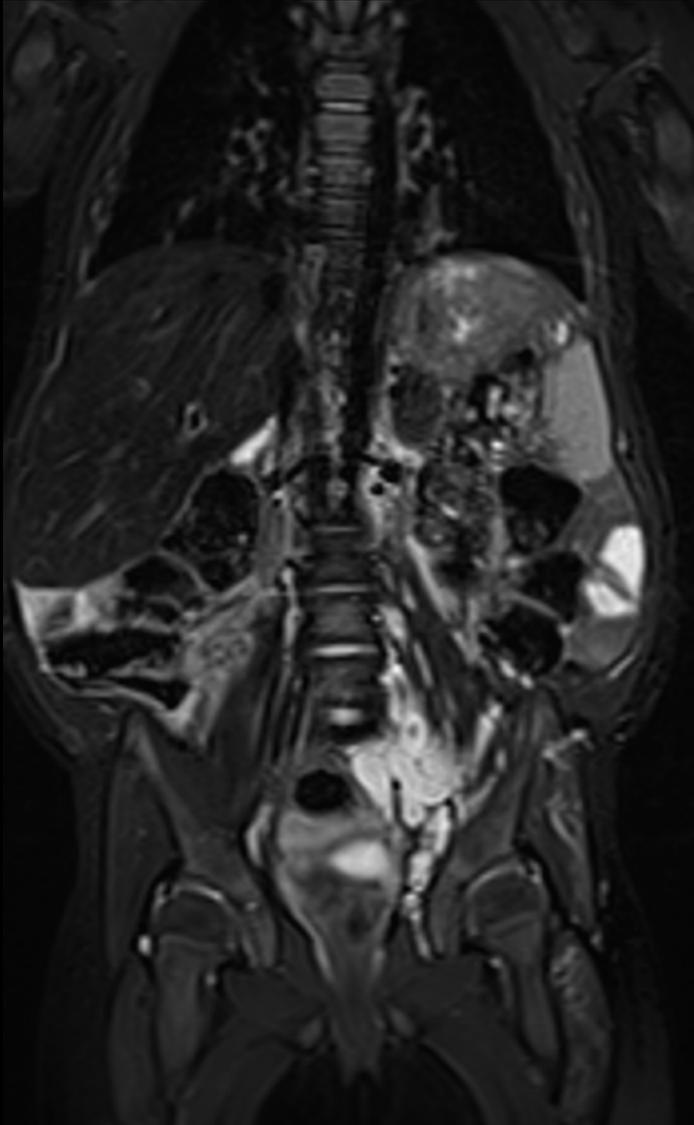


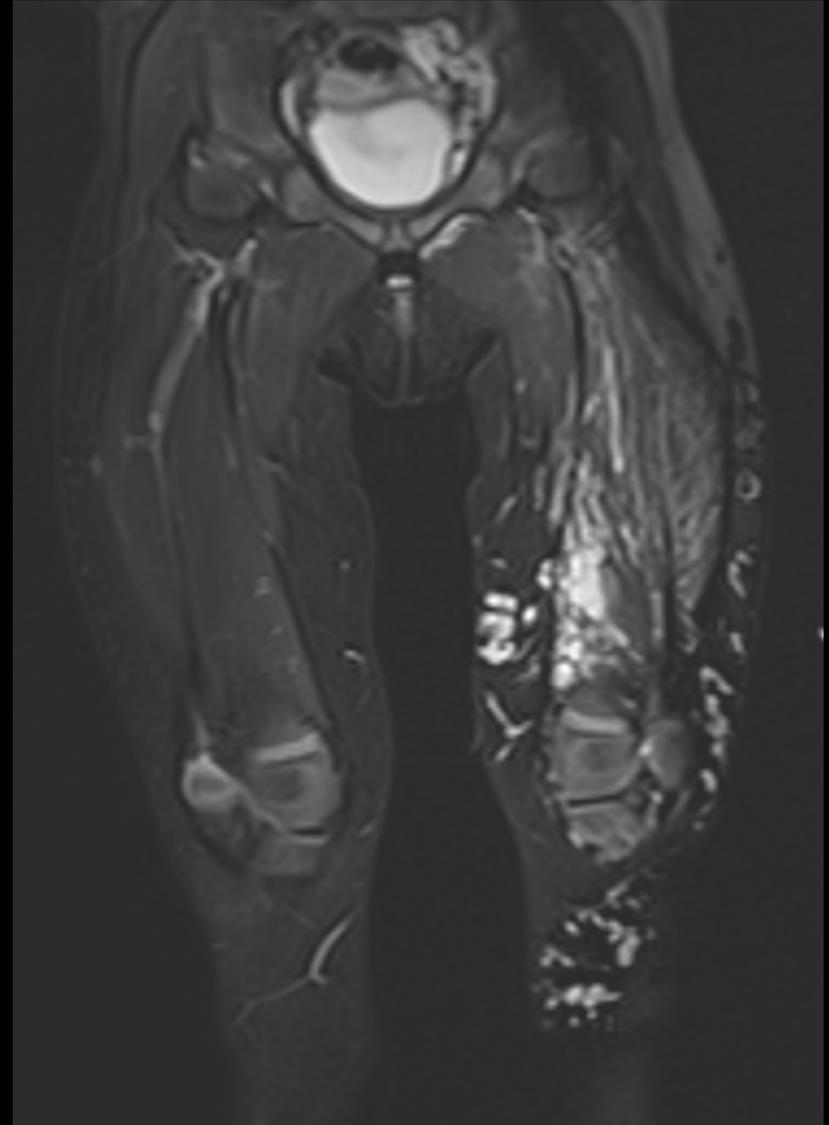
Figure 27: reticular rash as observed at 13 months



Figure 28: reticular rash at 13 months



*Figure 29: vascular malformation in left pelvis at 17 months*



*Figure 30: vascular malformation in left thigh*

## 000 A COMPLEX JOURNEY

### Establishing the final diagnosis

- Loss-of-function mutation in the *GLMN* gene
  - Essential for normal vascular development
  - Causes glomuvenous malformations (GVMs)
    - (Sub)cutaneous venous malformations
    - Typically present at birth, slowly enlarge during childhood
  - Inheritance
    - Autosomal dominant
    - Incomplete penetrance
    - Variable expressivity



Take home messages

## 000 TAKE HOME MESSAGES

### Key Clinical Insights from This Case

- The importance of follow-up
- (Epi)genetic differences may occur in monozygotic twins
- Neonatal chylous ascites
  - Rare and challenging condition
  - Key management principles
    - Decompression
    - Reducing chyle production

# 000 ANY QUESTIONS?

