

HARLEQUIN ICTHYOSIS

A Case Study

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UPDATE Congress 2009

PATIENT

- 21 year-old G2P1 patient with uncomplicated first pregnancy – NVD at term
- No known medical conditions
- No family history of any genetic or structural defects
- Uneventful pregnancy until 1 week before

INITIAL PRESENTATION

- 1ST visit at MAFU Steve Biko Academic Hospital at a gestation of 32w3 days
- Abnormal facial features noted at 3D scan done in private practice
- The patient had a dating scan early in the pregnancy and only presented to a gynaecologist at 29 weeks' gestation

INITIAL PRESENTATION

- Face
 - Normal eyes and orbita
 - Lip line intact
 - No nose visible
 - Large, protruding tongue
 - No visible auricles
 - 2 soft tissue solid masses protruding from the forehead
 - Abnormal profile

Lesaoana, Keneuwe

VP_117

MI: 1.2

12:58:10 PM 11/10/2008

FETAL MEDICINE - UNIV...



SIEMENS
CH4-1 / OB
2D
THI / 2.22 MHz
3 dB / DR 70
SC Off
Map K / RS 1
LMP
Age
EDC
EFW
EFW%

17fps 15cm

Fr521

Lesaoana, Keneuwe

VP_117

MI: 1.5

1:11:42 PM 11/10/2008

FETAL MEDICINE - UNIV...



SIEMENS
C7F2 / OB
2D
THI / 2.86 MHz
0 dB / DR 70
SC Off
Map K / RS 2
LMP
Age
EDC
EFW
EFW%

MI: 1.5



18fps

15cm

1.2



SIEMENS

CH4-1 / OB

2D

THI / 2.22 MHz

3 dB / DR 70

SC Off

Map K / RS 1

LMP

Age

EDC

EFW

EFW%

17fps

15cm

Fr542

INITIAL PRESENTATION

- Feet
 - Round, rocker-bottom shaped
 - 5 Small toes on each foot
- Hands
 - Clenched
 - Fingers not visible

11:48:16 AM 11/17/2008
FETAL MEDICINE - UNIV...

MI: 1.4



SIEMENS
CH4-1 / OB
2D
THI / 2.22 MHz
3 dB / DR 70
SC Off
Map K / RS 1
EFW
EFW%

17fps 15cm

Fr528

MI: 1.5



MI



18fps

15cm

Lesaoana, Keneuwe

VP_117

MI: 1.4

8:17:08 AM 12/08/2008

FETAL MEDICINE - UNIV...



SIEMENS
CH4-1 / OB
2D
THI / 2.22 MHz
3 dB / DR 70
SC Off
Map K / RS 1

17fps 15cm

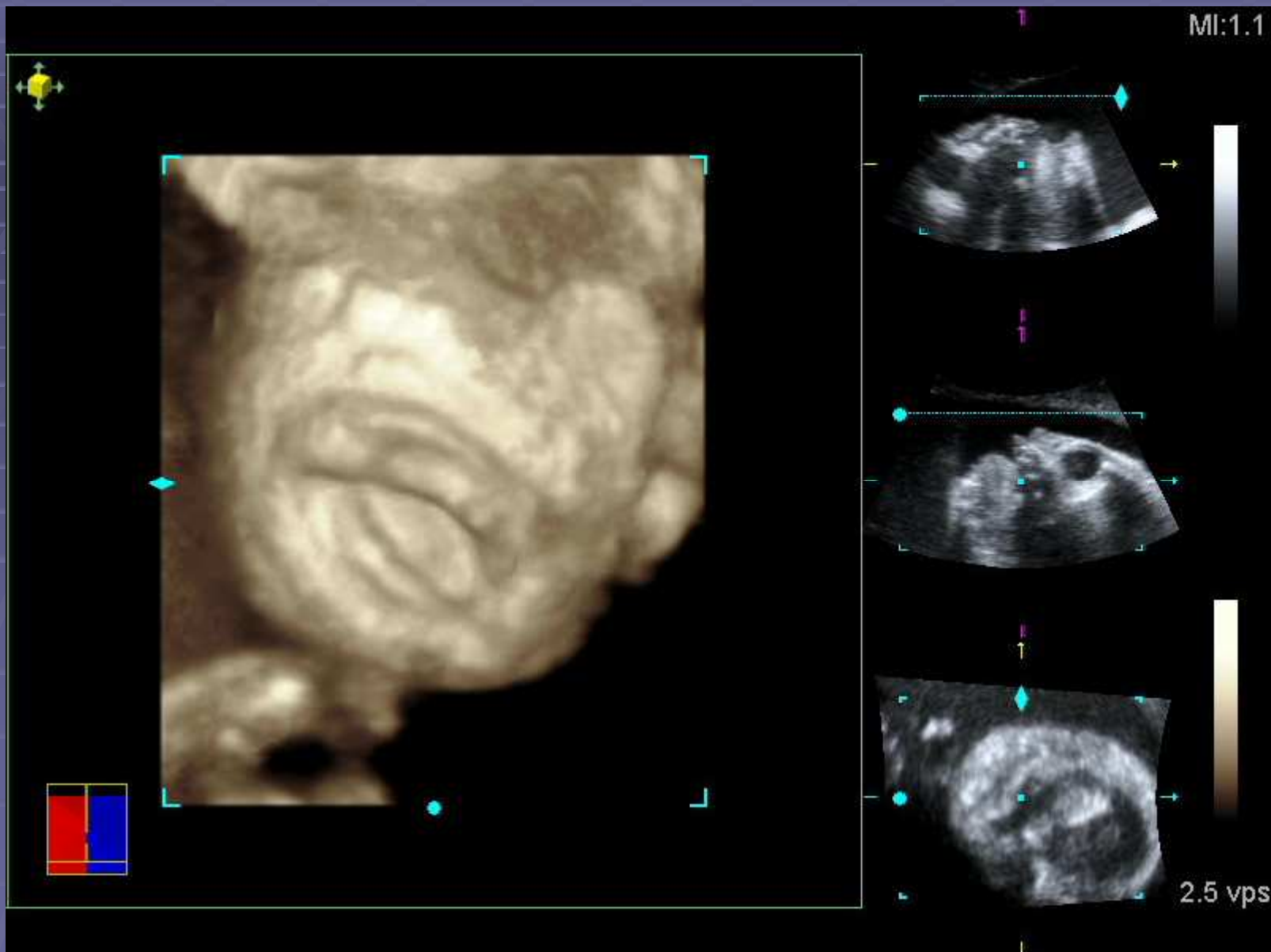
Fr541

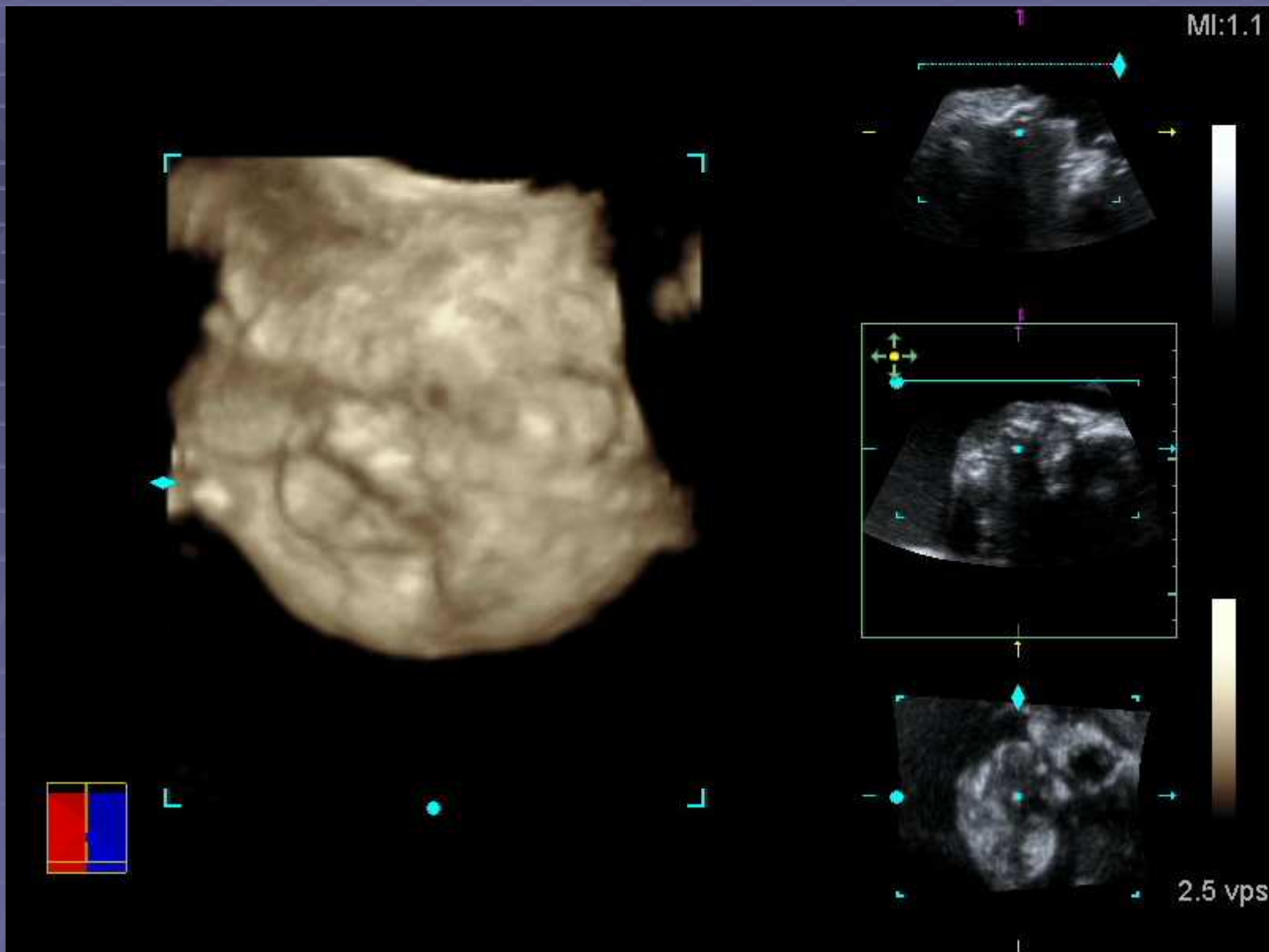
INITIAL PRESENTATION

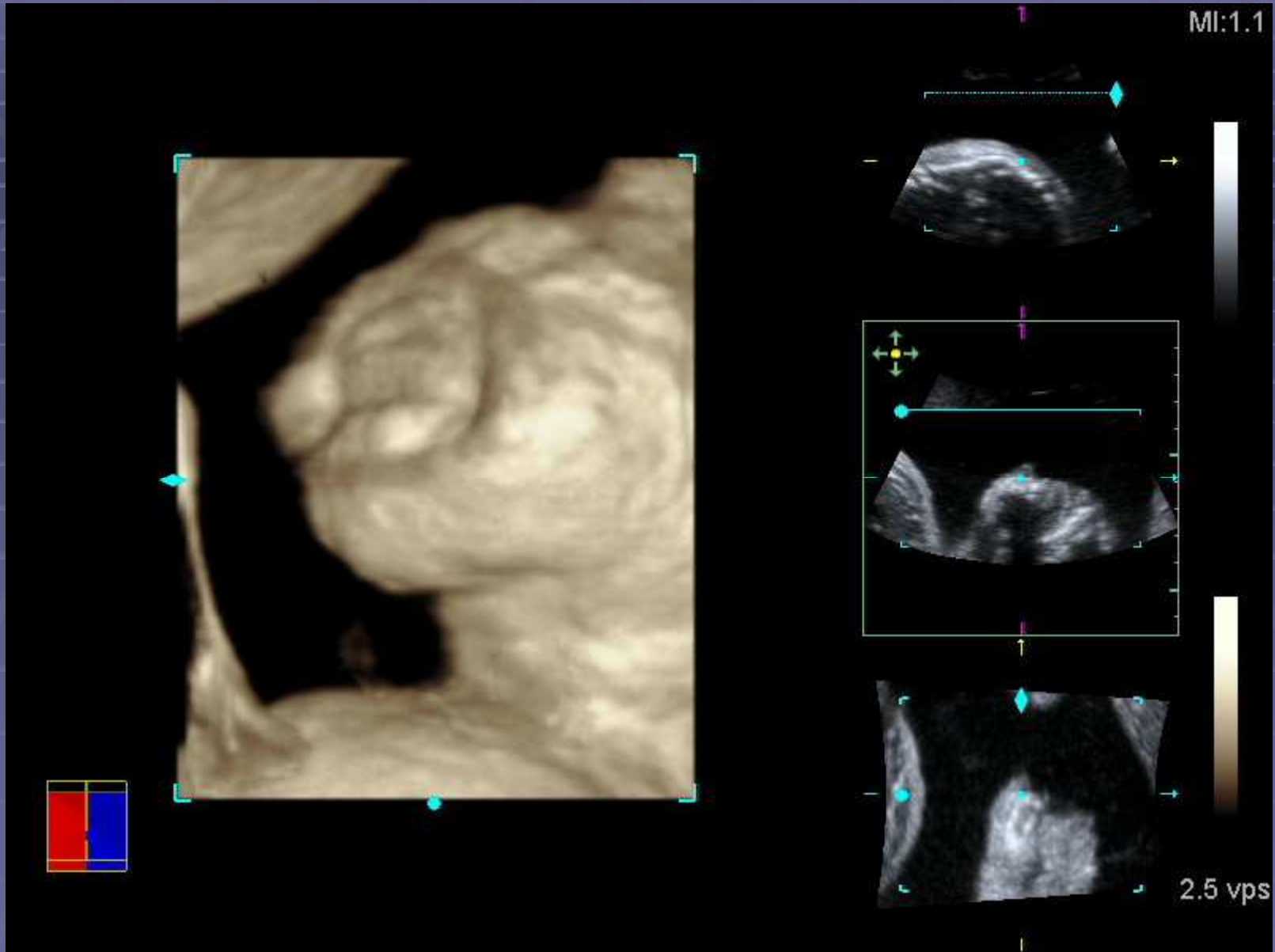
- Head, brain, heart and great vessels, abdominal wall, GIT, kidneys and bladder appeared normal

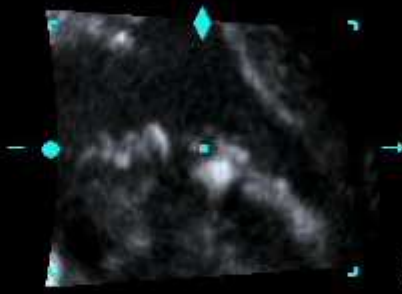
2nd Visit

- Cordocentesis done
 - Normal karyotype
- Hands better visualised
- 3D scan yielded better views of the face and extremities

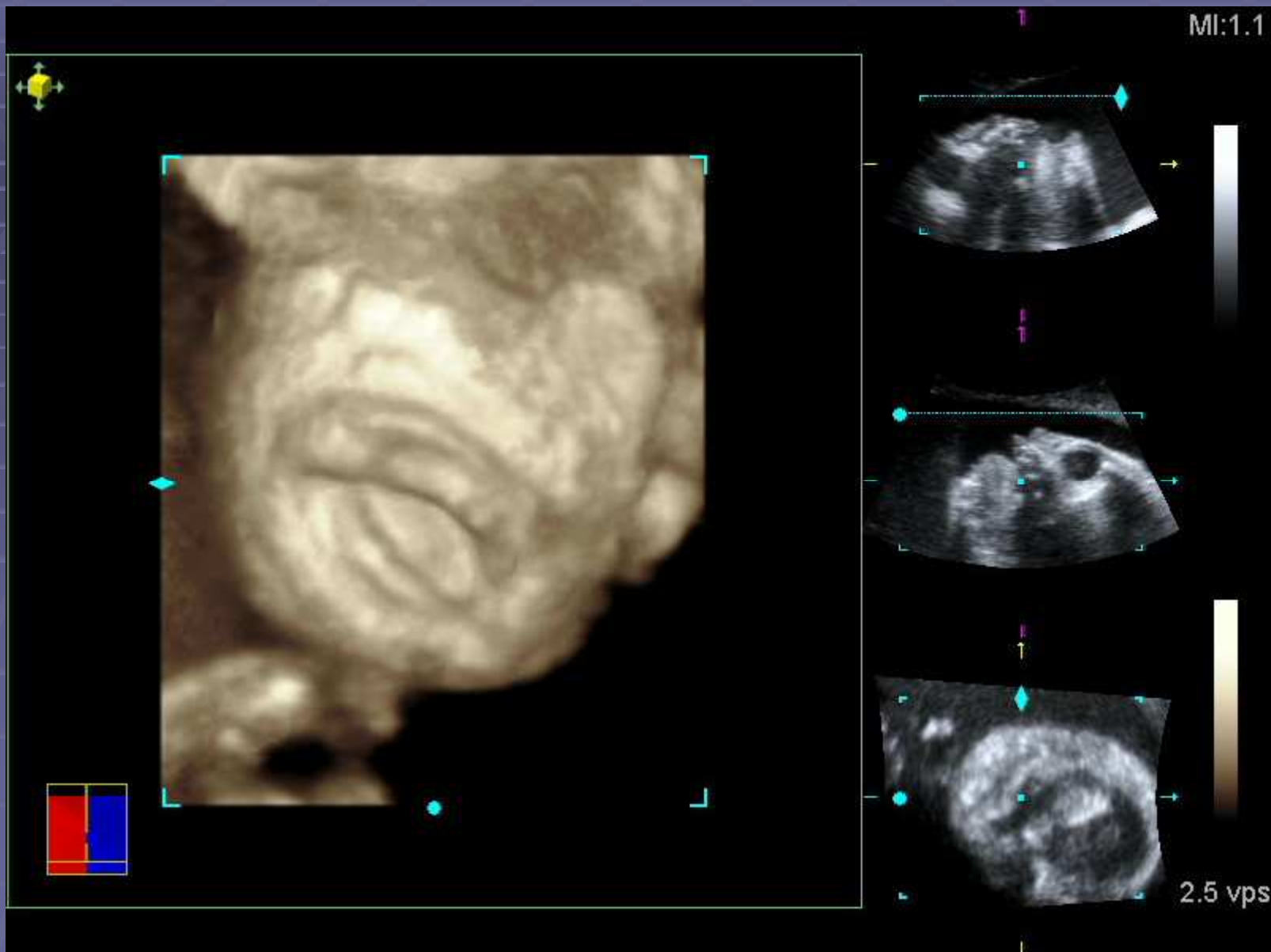


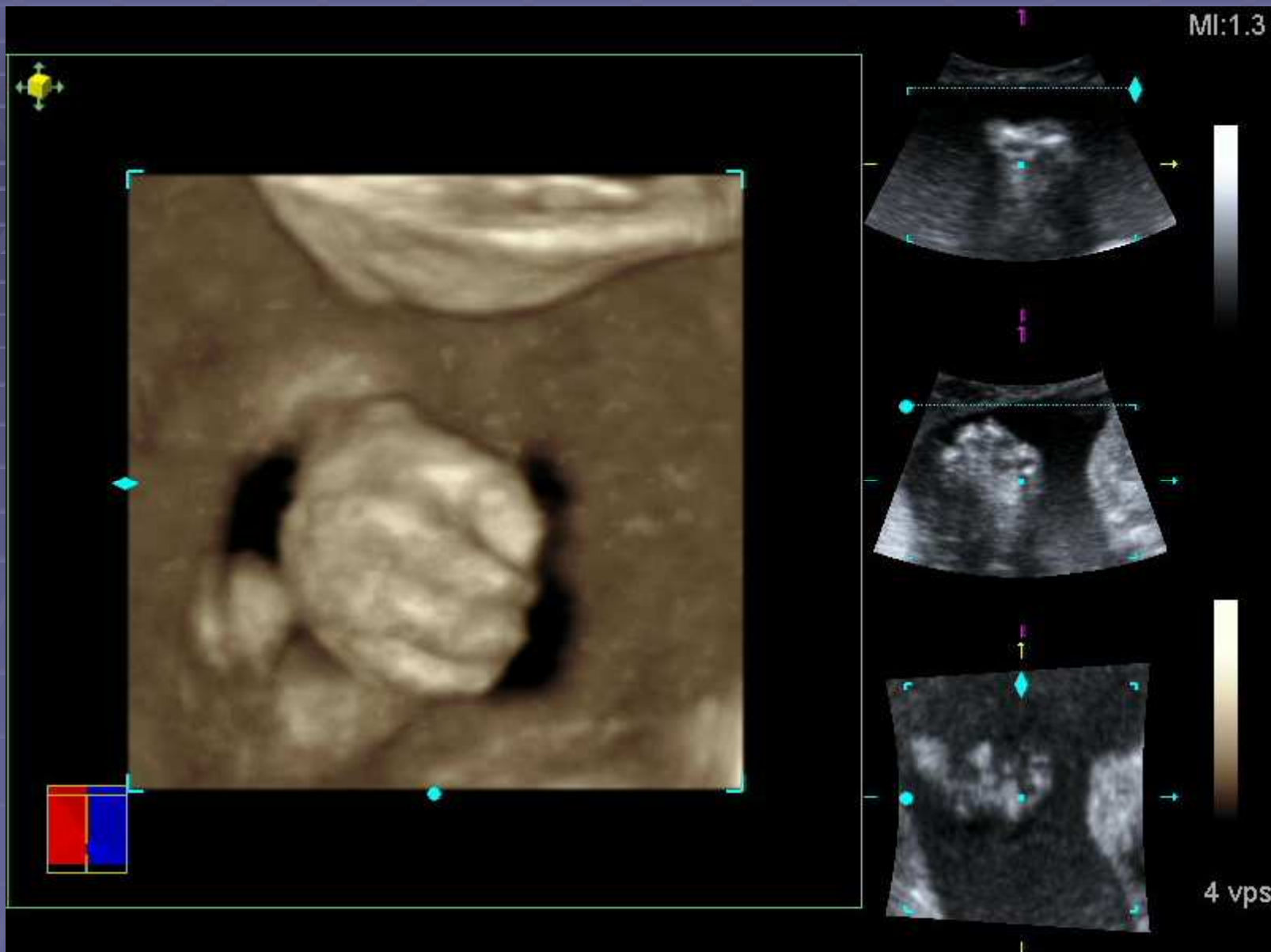






2.5 vps





DELIVERY

- Vaginal delivery at 37 weeks without fetal monitoring
- APGARS normal

NEONATAL FINDINGS

- Typical features of Harlequin Ichthyosis
 - Hyperkeratosis : triangular, diamond-shaped scaling of the skin
 - Large fissures oozing of bloody secretions
 - Typical clown-like open mouth with circumferential erythematous discoloration
 - Hands and feet swollen by circumferential constriction
 - Small ischaemic toes and fingers
 - Only ear canals visible
 - 2 masses above eyes : ? auricles



MANAGEMENT and OUTCOME

- Supportive management
 - Higher than normal IV fluids
 - No antibiotics given
- Neonate died day 2 after birth

DISCUSSION

- HI is the most severe form of congenital ichthyosis, which belongs to the group of genodermatosis
- Characterized by hyperkeratosis and desquamation of the epidermis caused by defective lipid transport that dramatically affects the function of the skin barrier
- Inherited as an autosomal recessive condition
- Several causative mutations identified (ABCA12)

DISCUSSION

- Prognosis poor
- Active management (antibiotics, intensive skin care and active fluid management) improves prognosis for survival
- Poor quality of life

CONCLUSION

- See one.....??
- ? Justifies late TOP?

■ THANK YOU