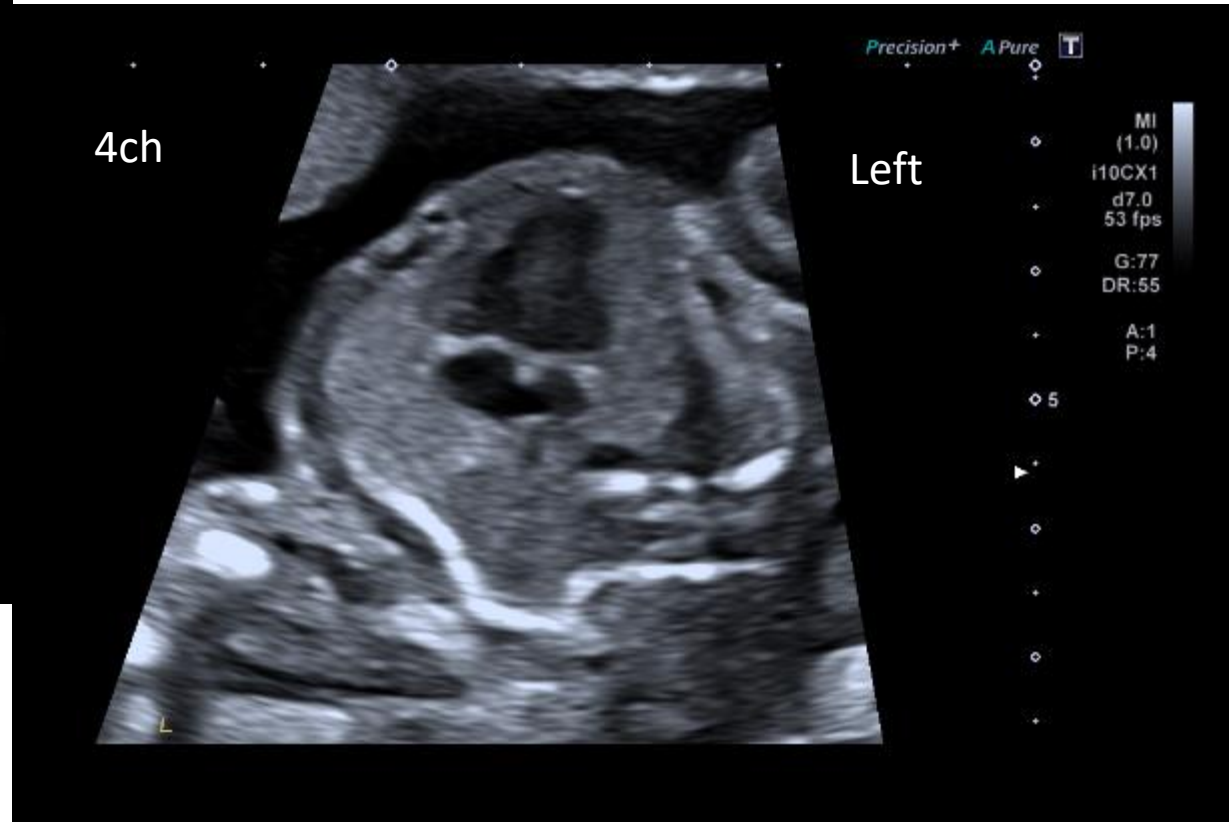
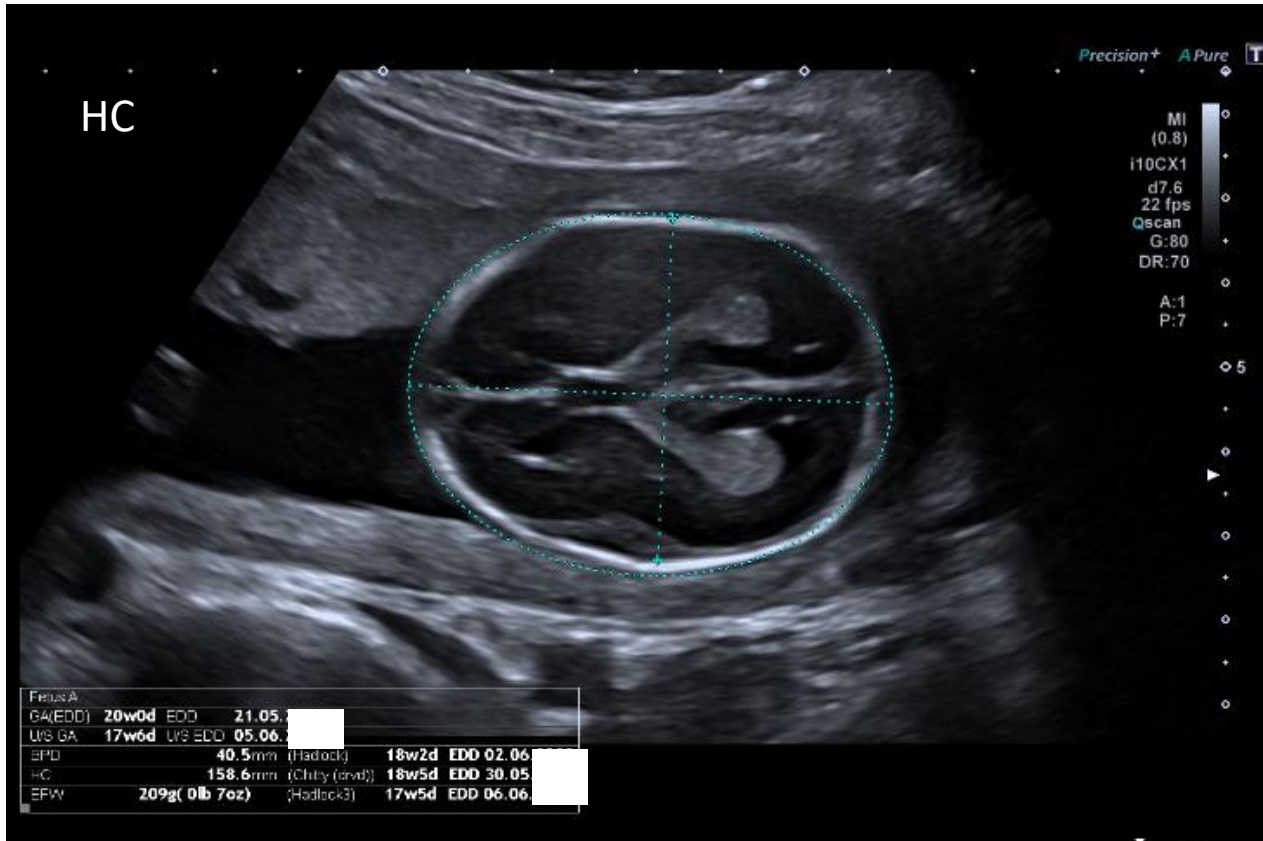
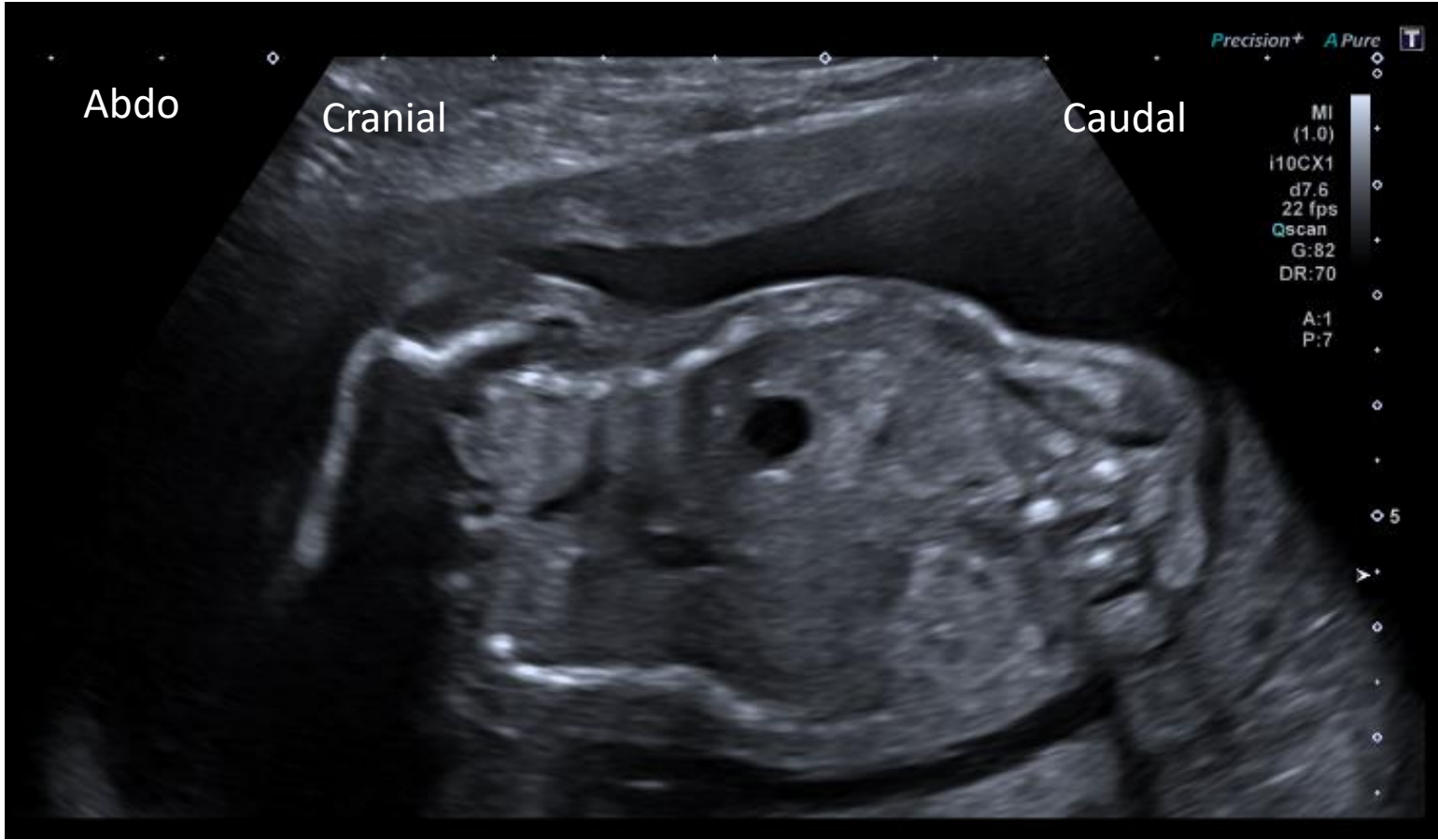


Obstetric Case 9
August 2023

37 y/o female
Anomaly scan. 20 weeks 0 days.





Upper limbs

Precision+ A Pure

MI (1.1)
i10CX1
d7.6
22 fps
Qscan
G:80
DR:70

A:1
P:7

5

Fetus A
GA(EDD) 20w0d EDD 21.05.20
WS GA 17w6d WS EDD 05.06.20
RAD 21.6mm (Chity) 17w6d EDD 05.06.20

Upper limbs

Precision+ A Pure

MI (1.1)
i10CX1
d7.6
22 fps
Qscan
G:80
DR:70

A:1
P:7

5

Fetus A
GA(EDD) 20w0d EDD 21.05.20
WS GA 17w6d WS EDD 05.06.20
PL 23.8mm (Chity) 17w1d EDD 10.06.20

FL



Fetus A
GA(EDD) 20w0d EDD 21.05.20
US GA 16w0d US EDD 18.06.20
FL 20.1mm (Chity) 16w0d EDD 18.06.20



Fetus A
GA(EDD) 20w0d EDD 21.05.20
US GA 16w0d US EDD 18.06.20
Foot 28.1mm (Chity) 18w4d EDD 31.05.20

Try to answer the following questions:

1. What abnormality is visible?
2. What ultrasound features led to this conclusion?
3. Why is there such a poor prognosis associated with this condition?

Do not progress to next slide until you have attempted to answer the previous questions.

Question 1

- Lethal skeletal dysplasia -
Thanatophoric dysplasia type 1

Question 3

- Pulmonary hypoplasia due to
underdevelopment of the thorax

Question 2

- Small head
- Small long bones
(femur, humerus, radius, ulna)
- “Telephone receiver” femur
- “Champagne cork” appearance of
the abdomen
- Small thorax – normal sized heart
looks enlarged due to thorax size

Thanatophoric dysplasia type 1 Fact File

- Thanatophoric dysplasia is autosomal dominant and caused by mutations in the *FGFR3* gene
- Rare – 1 in 20,000 to 50,000 births
- Two subgroups:
 - Type 1 - the bones of the skeleton are poorly developed and there is bowing of the limbs. In this subgroup, the shortness of the limbs is more marked
 - Type 2 - the limbs may not be as short but often there is a 'cloverleaf' formation of the skull, which may result in brainstem compression
- Very poor prognosis due to pulmonary hypoplasia