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Obstetrics Case 2: September 2021

26 y/o female

Combined dating scan and nuchal assessment

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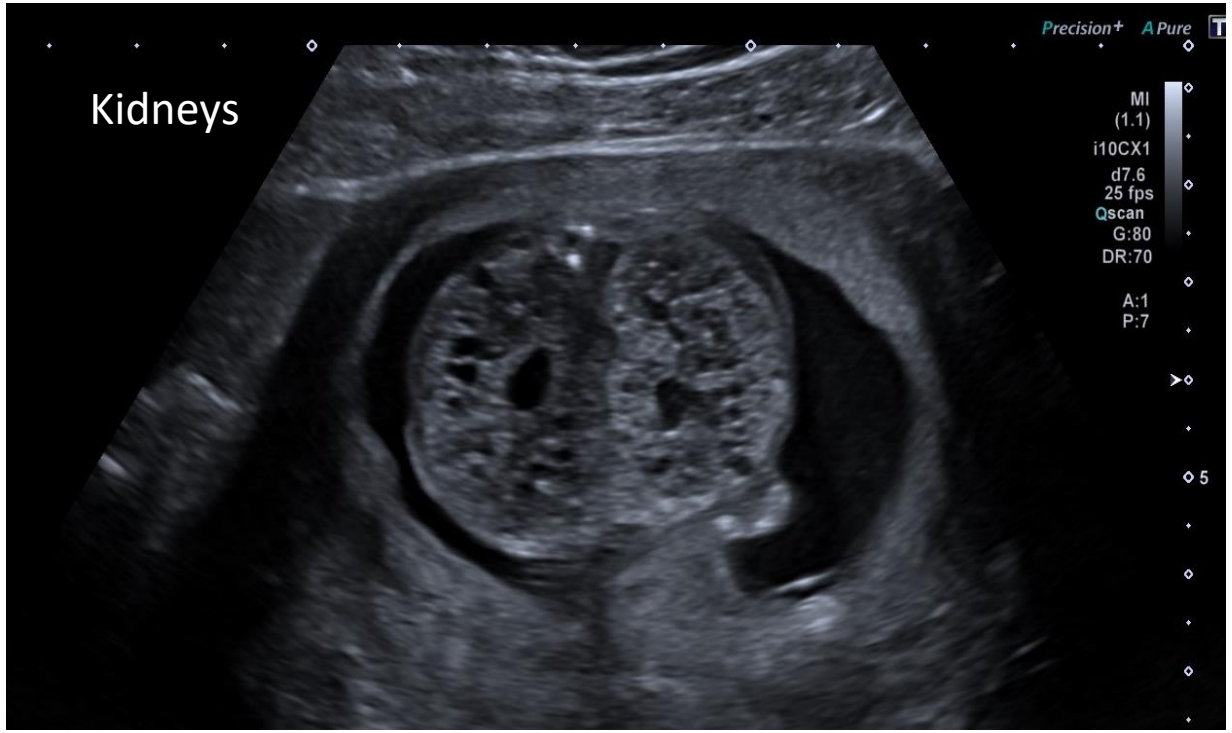
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MI
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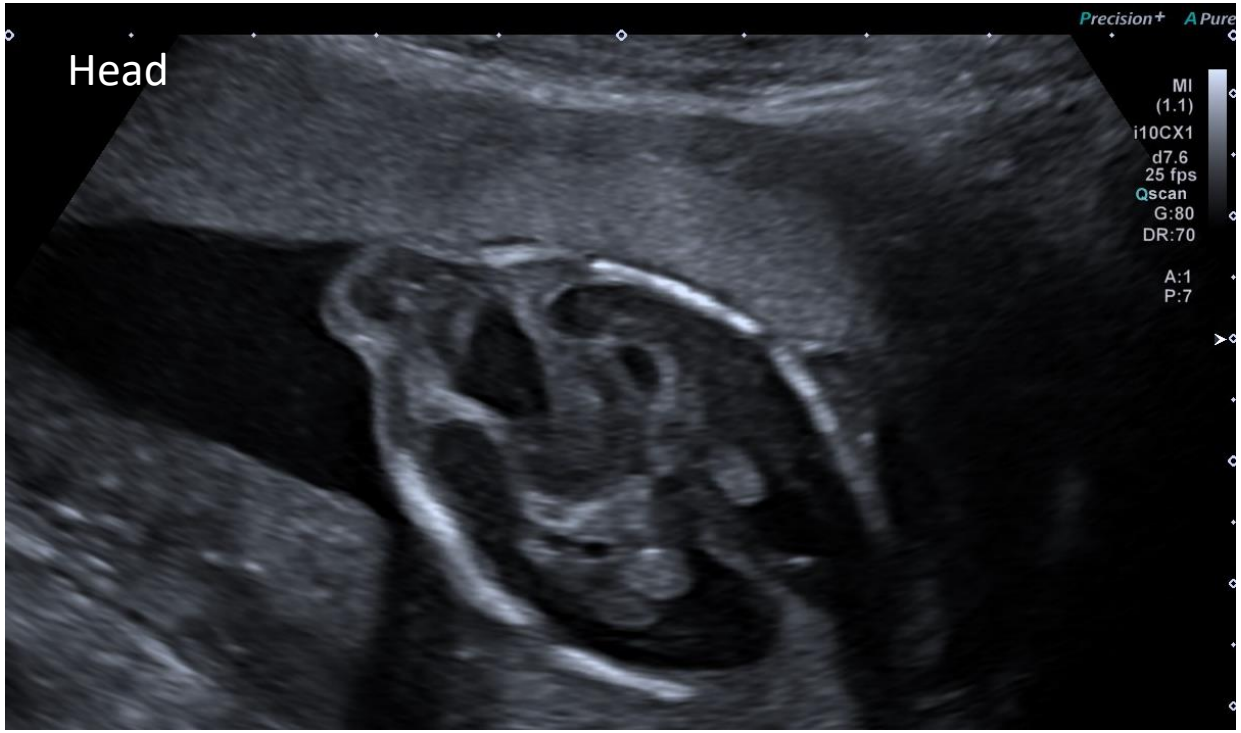
d7.6
25 fps
Qscan
G:80
DR:70
A:1
P:7
5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Precision+
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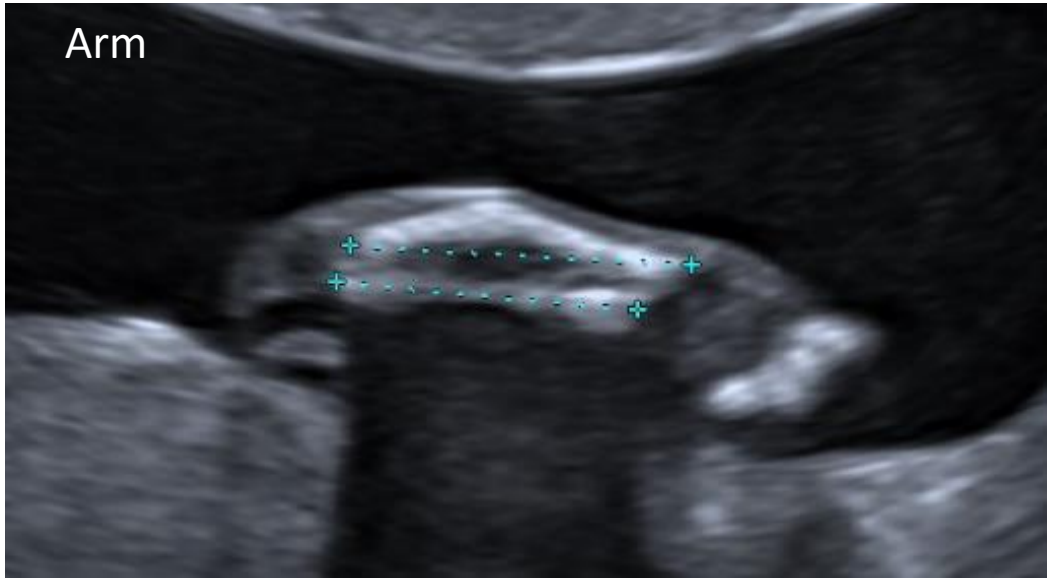

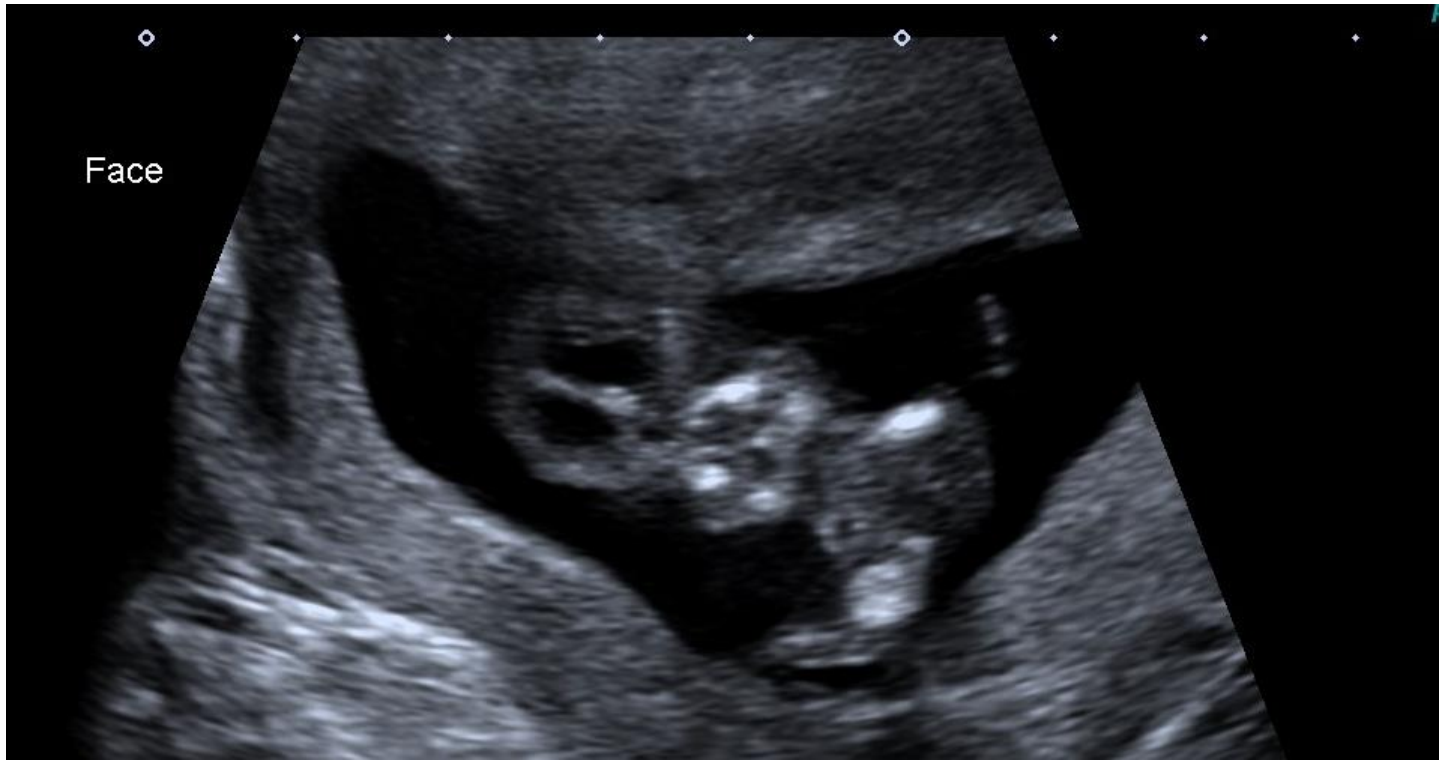

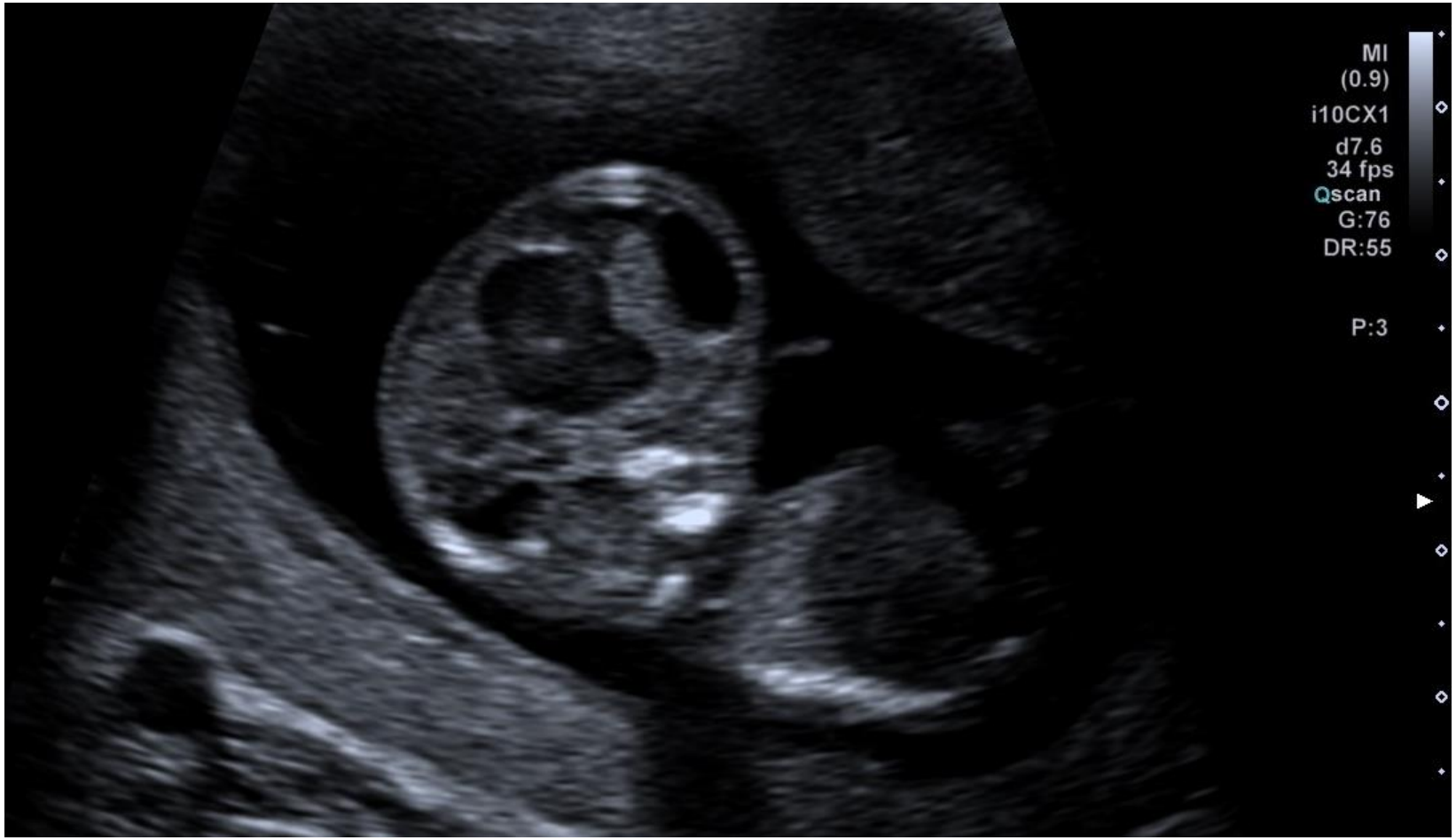
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Please consider the following questions:

1. What fetal abnormalities are present?

2. What is the diagnosis and is there any association with chromosomal conditions?

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

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Question 1

- Bilateral multicystic dysplastic kidneys
- Occipital encephalocele
- Campomelic dysplasia of the ulna
- Facial cleft

Question 2

- Mekel-Gruber Syndrome
- Yes chromosomal linked

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Mekel-Gruber Syndrome Fact File

- Autosomal recessive disorder, therefore a gene has to be inherited from each parent for the child to be affected. A child with one normal and one altered gene will be a carrier. Risk of phenotypical display is 25%, risk of becoming a carrier is 50% and the chance of receiving normal genes from both parents is 25%.
- Mekel-Gruber can affect the central nervous system, pulmonary system, and renal system as well as affecting the limbs and heart. Multiple abnormalities are often present however not **all** abnormalities will be present.
- The most common anomalies seen on ultrasound are multicystic dysplastic kidneys, occipital encephalocele and facial abnormalities.
- The condition is not compatible with life.