EP11.10
First trimester diagnosis of Meckel–Gruber syndrome
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A 28-year-old G5 P1031, in a consanguineous marriage, referred for sonography at 14w1d (by dates) due to a suspected head abnormality. Composite fetal biometry was consistent with 12w6d. Past history was notable for early second trimester losses of anomalous fetuses.

On ultrasound, the fetus measured 67.9 mm and had an NT of 1.1 mm. A clear butterfly was noted and the orbits, nasal bone, maxilla and mandible were seen. The fetal stomach, bladder, cord insertion and spine were seen as well. However, there was an occipital encephalocele, the kidneys were enlarged and polycystic, and the digits demonstrated polydactyly. On assessing the fetal heart, an atrioventricular septal defect (AVSD) was noted. Differential diagnosis included Trisomy 13 and 18, autosomal dominant polycystic kidney disease and Meckel–Gruber syndrome (MGS).

In our case, the fetus manifested the classic triad of MGS: an occipital encephalocele, polycystic kidneys and polydactyly. In addition there was an AVSD. Though Trisomy 13 and 18 and autosomal dominant polycystic kidney disease were in the differential, the fact that this was a consanguineous couple, with a prior history of anomalous fetuses, made MGS the most likely diagnosis. The family elected termination of pregnancy. They declined a postmortem examination or a genetic workup.

Given its lethality, early diagnosis of MGS, as well as other such lethal anomalies, has a tremendous impact on counselling the family particularly when termination of pregnancy may be a consideration.

Supporting information can be found in the online version of this abstract

EP11.11
Key points of fetal malformation in level I ultrasound examination during late pregnancy
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Objectives: To investigate the key points and skills of level I ultrasonography during late pregnancy.

Methods: From Jan 2013 to Dec 2017, 17170 cases received level I ultrasonography during late pregnancy (28-42 weeks). All physicians were operated according to The Guidelines for Prenatal Ultrasound (2012). The results of ultrasound were compared with the results after delivery.

Results: 35 cases were detected by ultrasound (detection rate 0.2%). The positive results were as follows:

- Abnormal urinary system: 7 cases of renal pelvis separation (>10 mm)—all were normal after birth; 4 cases of renal cystic dysplasia, 3 cases of unilateral renal did not show and 1 case of giant bladder. All were consistent with the results of postpartum re-examination.
- Cardiac malformation: 1 case of complete transposition of great arteries,ToF, ASD and aortic root thin with VSD. All were consistent with the results after birth.
- Central nervous system malformation: 1 case of cyst in the posterior part of the 3rd ventricle. 3 cases of lateral ventricle widening (>10 mm). Postnatal MRI revealed: 1 case of simple cyst in the 3rd ventricle, lateral ventricle widening and abnormal brain development, and 2 cases of ventricular widening.
- Digestive tract malformation: duodenal atresia in 1 case, consistent with the results after birth. 3 cases of dilatation of intestine (>18 mm). No abnormalities were found after delivery.

There were 2 cases of cleft lip and limb short, 1 case of diaphragmatic hernia, chest wall mass and right pleural effusion. All were consistent with the results after birth.

1 case was misdiagnosed. Ultrasound found NF was thickened. Newborn was macrosomia (weight 5300 g), and the structure was not abnormal.

Conclusions: Our study confirmed that abnormal urinary system is highly sensitive to level I ultrasonography in late pregnancy. Heart and digestive system abnormalities are high detection rate and severe postpartum consequences. Through observing the emphasis, it can help the discovery deformity, help the clinician evaluate the fetus, make intervention plan, and improve prognosis.

EP11.12
Antenatal diagnosis of Miller-McKusick-Malvaux (3-M) syndrome: a case report
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The Miller-McKusick-Malvaux (3-M) syndrome is a rare primordial growth disorder characterised by low birth weight, reduced birth length, severe postnatal growth restriction, and a spectrum of minor anomalies, including facial dysmorphism. Although individuals with 3-M syndrome have short stature, and skeletal abnormalities, their intelligence is not affected. 3-M syndrome is inherited in autosomal recessive manner, and to date, there are only about 100 cases of affected individuals that have been identified worldwide. In the antenatal period, individuals with 3-M syndrome typically have long bones which are below the 3rd centile, and they are generally small for gestational age. This slow growth continues throughout childhood and adolescence. They have low birth weight and length, and remain much smaller than others in their family, growing to an adult height of approximately 120-130 cm. The majority of 3-M cases are diagnosed postnatally.

We present a case of 3-M syndrome which was diagnosed during the antenatal period. This is the first 3-M case diagnosed prenatally which led to a live birth. Due to the fact that 3-M syndrome is inherited via an autosomal recessive pattern, early diagnosis in a fetus can aid in the genetic counselling for the current and future pregnancies. Antenatal detection is possible by analysis of DNA extracted from fetal cells by chorionic villus sampling or amniocentesis but in the absence of a family history, prenatal diagnosis is often difficult, and 3-M joins a long list of possible causes of shortened long bones, such as include Silver–Russell syndrome, Bloom syndrome, Dubowitz syndrome, Rubinstein-Taybi syndrome, Floating-Harbor syndrome, Mulibrey nanism and fetal alcohol syndrome.

3-M syndrome is a rare genetic disorder that should be considered in the differential diagnoses when assessing fetuses which present with intrauterine growth restriction and short long bones.