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Fetal OEIS complex in second trimester- a Case Report

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Introduction : The normal anatomy of the bowels herniation most prominent at 9-10 weeks of gestations, and it should no longer be visible by 12 weeks.

Case Report: A 32-year-old female, G3P1A1 (blighted ovum), who had prenatal care in our department since pregnancy. Unfortunately, the ultrasound showed a cystic tumor protruding from the low abdomen of the fetus at her 14 weeks of gestation. After series of ultrasound follow-up, the cystic tumor became larger in size about 5cm in her 15 weeks. After discussion with the patient and family, they decided to have termination induction. Prostaglandin E2, cytotec, syntocin and extraamniotic foley traction were used during induction. The gross of the baby was a cystic bag protruding from the low abdomen with bowels protrusion, ambigues of sex and imperforate anus, and spinal deformities. According the gross of this baby, we prefer it is an OEIS complex syndrome.

Conclusion

Other anterior abdominal wall defect included the bladder exstrophy, cloacal exstrophy, limb-body wall complex (amniotic band syndrome), ectopia cordis, gastroschisis, omphalocele and urachal cyst.

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The Pitfall of Prenatal Screening of Thalassemia Disorder—Case report

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Introduction:

Thalassemia is a hereditary disorder with defects in the synthesis of hemoglobin. It is the most common genetic disease among the Chinese population in Southeast Asia. Currently, antenatal screening done for thalassemia disease in Taiwan is by simple blood examination of hemoglobin, mean corpuscular volume and mean cell hemoglobin level test. We report two individual cases which were not prenatally screened by traditional diagnostic test , including mean corpuscular volume, testing(MCV) and mean cell hemoglobin(MCH).

Case(s) report:

Two cases with, past obstetric history of G2P2, prenatal screening test of normal complete blood count (CBC) examination including >80fl delivered IIb Barts fetus and severe type B-thalassemia infant. Fetal diagnosis of thalassemia was retrospectively proved due to thalassemia trait of the both cases were not detected during antenatal care.

Discussion:

Thalassemia is the most common hereditary disease in Southeast Asia. Its severity in patients varies from being carrier, to early onset of severe anemia requiring regular blood transfusion or in the worst scenaria with intrauterine fetal death. An effective screening test of identifying couples at risk of producing a fetus with severe thalassemia is needed. Currently, in Taiwan, maternal blood sample is withdrawn during her first antenatal care visit. When MCV >80fl, the doubt of carring thalassemia disease is excluded. If initial serum testing reveals <80fl, paternal blood examination is performed. When both of the screening test reveal MCV <80fl, further evaluation is done . However, in our cases, initial maternal screening test did not show microcytic anemia of MCV <80fl. As a result, hydrops fetalis was diagnosed at gestational age 26wks in one case and another infant with severe anemia, later confirmed to be thalassemia disease, detected after delivery at term gestational age. Eventhough, screening test with routine blood count test has been used in decades, we must be careful in interpretation of the serum result. When a woman is detected with normal ranged "low" MCV, exclusion of thalassemia disease might be necessary for her spouse.

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Successful regression of antenatally diagnosed CCAM type III in contrast to formally reported regression rate

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Objective:

With the increasing usage of ultrasonography and higher resolution ultrasound imaging platforms at prenatal examination, the antenatal diagnosis of fetal lung tumors, such as congenital cystic adenomatoid malformation (CCAM) has become more frequent. The prognosis for fetuses with large congenital cystic adenomatoid malformations remains uncertain or unfavored, however, in the more recent published series, spontaneous in utero regression of CCAMs is now recognized as a more common event. Still, much less is known about the outcomes of fetuses who are initially diagnosed with large CCAMs, such lesions that are sizeable enough to cause significant mass effect on adjacent thoracic structure. Fetuses with large CCAMs are believed to be at considerable risk for hydrops or in utero demise. In this study, we reviewed our own experience with large fetal CCAMs in which we can better counsel the expectant parents in the future.

Case Reports:

In the second half of 2007, we studied eight cases of antenatally diagnosed type III CCAMs. Seven of the fetus had been diagnosed CCAMs with size average at 4.0 x 2.0 cm at gestational age around 18 weeks. All seven of the fetus had in utero regression, and had normal delivery. After the delivery, the fetus had been discharged without any surgery, and return home with regular follow-up. Unfortunately, one of the eight fetus had hydrop, which had been terminated at gestational age of 19 weeks.

Conclusions:

Besides of the better resolution rate of type III CCAM, we also raised a question that the diagnosis of CCAM. Since CCAM is mostly diagnosed by ultrasound and almost without any histology studies, the criteria might suggest differential diagnosis with simple lung obstructions. This might be another study we can look into in the future, which might help giving consult to the expectant parents

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Novel heterozygous missense mutation G404S of *ITGA9* is associated with poor prognosis in fetuses with chylothorax treated by antenatal OK-432 pleurodesis

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Abstract

Congenital chylothorax is a genetically heterogenous assembly of lethal pleural lymphatic disorders. Prenatal therapy of chylothorax by fetal OK-432 pleurodesis is feasible but is futile in certain group. To investigate whether mutations harbored in possible candidate genes are associated with treatment response of fetuses with chylothorax receiving OK-432 pleurodesis *in utero*, a total of 17 affected unrelated fetuses (including 12 received OK-432 pleurodesis) were genotyped for the reported susceptible loci *ITGA9*, *ITGB1*, *VEGFR3* and *FOXC2*. Two novel missense mutations in *ITGA9*, c.1210G>A (G404S) and c.1520G>A (G507E), were identified in a subset of fetuses. The G404S was found in heterozygous status in four of the five fetuses failed to respond to OK-432, and, notably, three of the four G404S carriers have family history of antenatal chylothorax. This variant is assumed to be pathogenic since it is not present in 100 controls, and the affected amino acid glycine is located within a conserved domain of integrin proteins and is highly conserved among human, chimp and dog. On the contrary, the G507E was found in homozygous status in two affected fetuses and their healthy parents, and seems to be neutral since the residue Gly507 has been recorded as a SNP in the HapMap dbSNP database. To the best of our knowledge, this is the first insight into the possible link between *ITGA9* and congenital chylothorax in human fetuses. The identification of pathogenetic mutations in this disorder should be useful for genetic counseling, especially for the clinical treatment of OK-432 in those mutation carriers.