What is your diagnosis?

A patient who was 30 years old was referred to our clinic at the 16th week of gestation. It was her first IVF pregnancy. Ultrasonographic examination showed an anechoic image that measured 80x59 mm (Figure 1). What is your diagnosis?

Figure 1. Cystic mass in the uterine cavity
Ultrasound examination revealed a fetal part near the anechoic cyst. We realized a calvarium and that the fetal heart was not beating (Figure 2). A previous ultrasonographic image at 12 weeks of pregnancy showed that the fetal bladder was 31x18 mm length and showed hyperechogenic kidneys (Figure 3). This case was related with fetal megacystis. Megacystis is an anomaly that is visible at any week of gestation. The incidence of this anomaly is not known. Megacystis is the longitudinal size of a fetal bladder greater than 7 mm in the first trimester (1). The most common cause of megacystis is lower urinary tract obstruction (2). Megacystis can also be associated with chromosomal anomalies (mostly with trisomy 13 and 18) (3). Genetic examination should be done in the cases of megacystis. Urethral atresia, posterior urethral valve, megalourethra, prune belly syndrome, and megacystis-microcolon-intestinal hypoperistalsis syndrome can be other reasons of megacystis. The definition of megacystis in the second and third trimester is more subjective than the first trimester. Enlarged bladder in 45 minutes during an examination can be used to diagnose for megacystis in the second and third trimester (1). In the second and third trimester, lower urinary tract obstruction (LUTO) frequently can be caused by posterior urethral valve (PUV) in male fetuses with megacystis. Keyhole sign in sonographic examination is important for the diagnosis of PUV (4). Survival of fetuses with megacystis in the second and third trimester is better than in the first trimester (1). Bladder drainage not only reduces the risk of pulmonary hypoplasia but also increases survival. Vesicocentesis and vesico-amniotic shunting may be used for the drainage of bladder (1).

In our case, the patient was offered chorionic villus sampling and termination of the pregnancy when we diagnosed megacystis, oligohydroamnios, and bilateral renal dysplasia in the first trimester. But, the family did not accept. We observed the fetus with a megacystis 80 mm wide and no heart beating 4 weeks later. After informed consent, we terminated the pregnancy (Figure 4).