Objective  To study the imaging features of fetuses diagnosed as abdominal wall defect and the proportion of the chromosomal abnormalities and pathogenic CNVs among these fetuses.

Methods  A cohort of 45 fetuses with abdominal wall defects, which were detected by prenatal ultrasound and confirmed by specialist consultants, were collected. MRI scanning were performed in a proportion of these fetuses. The available tissues, including fetal umbilical cord from live-born fetuses or thigh muscle from aborted fetuses, were subject to chromosomal microarray analysis (CMA) and low whole-genome sequencing (WGS), to identify chromosomal anomalies and microduplication/deletions (CNV).

Results  There are 23 male and 22 female fetuses. 5 fetuses were diagnosed as abdominal wall defects below 14 gestational weeks; 16 were between 14 and 17 Weeks; 13 were between 18 and 23 weeks; 7 between 24 and 28 weeks; 4 were after 28 weeks. In these 45 fetuses with abdominal wall defects, 24.4% (11/45) of fetuses had chromosome anomalies, and 2.2% (1/45) of fetuses had a pathogenic CNV, with an overall diagnosis rate of 26.7% (12/45). The chromosome anomalies included 13-trisomy (3 cases), 18-trisomy (5 cases), and 45XO (3 cases); the pathogenic CNV were 1.9M microdeletion on 3q26.3. In this study, 25 fetuses were diagnosed as omphalocele, with 10 cases having chromosome anomalies (40%, 10/25) and 1 having pathogenic CNV (4%, 1/25); 11 fetuses were found to have body stalk anomaly, with 1 case having chromosome anomalies (9.1%, 1/11) and no fetuses having pathogenic CNV (0%, 0/11); no chromosome anomalies or pathogenic CNV were detected in the 5 fetuses with Amniotic band syndrome, 3 fetuses with Gastrochisis, and 5 fetuses with Cantrell.

Conclusions  In fetuses with abdominal wall defects, the detection rate of chromosome anomalies is much higher than the rate of pathogenic CNVs.