OBJECTIVE. To review literature about the correlation between fetal autopsy and prenatal diagnosis (PD) of fetal malformations.

MATERIALS. Search in PubMed, Medline, EMBASE, reference list. Inclusion criteria: fetal autopsy performed after TOP/stillbirth, TOP for fetal anomalies, PD of malformations by ultrasound. Exclusion criteria: case reports, data in graphs/percentage. From each article, sample size, type of malformation, indication for TOP, diagnosis by autopsy. Correspondence between autopsy and PD was defined as agreement (same diagnosis), additional (additional findings undetected by PD), unconfirmed (false positive and false negative US). PRISMA guidelines were followed.

RESULTS. From 20 articles, 3625 fetuses underwent autopsy. Autopsy findings confirmed PD in 2467 (68%), provided additional information in 817 (22%), unconfirmed PD in 331 (9%) fetuses. The latter group consisted of 3.2% false positive and 2.8% false negative cases. Additional findings changed the final diagnosis in 4.5% of cases. The highest agreement between autopsy and PD was observed in CNS (79.4%) and genetics (79.2%), followed by GU anomalies (76.6%), skeleton (76.6%), CHD (75.5%), thorax (69.7%); GI (62.6%), multiple (37.0%), limbs (23.3%).

CONCLUSION. Fetal autopsy provides additional information in 22% of cases. In 3% of cases, PD leads to unjustified TOP.