Correlation between fetal autopsy and prenatal diagnosis by ultrasound: A systematic review

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Objective: The objective of this study was to review literature about the correlation between fetal autopsy and ultrasound findings of fetal malformations.

Methods: Search in PubMed, Medline, EMBASE, Clinicl trials.org, reference list was performed. Inclusion criteria for studies selection were: fetal autopsy performed after termination of pregnancy (TOP) or stillbirth, TOP for fetal anomalies, prenatal diagnosis of malformations, data reported as proportional rates, case reports, non-English language and data reported in graphs or percentage. From each article: sample size, type of malformation, indication for TOP, autopsy findings was collected. Fetal anomalies were grouped in central nervous system (CNS), genitourinary (GU), congenital heart defects (CHD), gastrointestinal (GI), thorax, limbs, skeleton, genetics (TOP for abnormal karyotype) and multiples (TOP for multiple severe malformations for which a single indication for TOP/stillbirth could not be identified). Correspondence between autopsy and ultrasound was defined as agreement (same diagnosis), additional (additional findings undetected by ultrasound) and unconfirmed (false positive and false negative ultrasound). PRISMA guidelines were followed.

Results: From 19 articles, 3534 fetuses underwent autopsy, which confirmed prenatal ultrasound in 2401 (68.0%) fetuses, provided additional information in 794 (22.5%) fetuses, and unconfirmed prenatal ultrasound in 329 (9.2%) fetuses. The latter group consisted of 3.2% false positive and 2.8% false negative cases. The additional findings changed the final diagnosis in 3.8% of cases. The most frequent indication for TOP/stillbirth was CNS anomalies (36.3%), whereas thorax anomalies represented the less frequent indication (1.7%). The highest agreement between autopsy and prenatal ultrasound 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: In spite of the high agreement between prenatal ultrasound and autopsy, fetal examination is mandatory because in a minority of cases it discloses additional findings or changes the final diagnosis and genetic counseling.

Biography
Cristina Rossi was born in 1974 and got her medical degree in 1998 and Residency in Obs/Gyn in 2003. She is both author and reviewer of peer-reviewed journals. She is mainly interested in prenatal diagnosis and twin pregnancy. She is actually consultant in Obs/Gyn at Ospedale della Murgia (Bari, Italy).
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