Comparison of prenatal ultrasound diagnosis to fetal autopsy in medical interruptions of pregnancies

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Objective
Congenital malformations are a major cause of perinatal mortality. The development of fetal ultrasound imaging has made available the majority of fetal malformations in prenatal diagnosis. However, in some cases, it may not be an accurate prenatal diagnosis, and the consequences are not exactly known. Therefore, autopsy and histological examination after abortion can provide essential information including a definitive diagnosis and management of subsequent pregnancies. The purpose of our work is firstly to compare sonographic and fetal autopsy findings, secondly to assess the sensitivity and specificity of ultrasound and its value for prenatal diagnosis and thirdly to assess the contribution of fetal autopsy in establishing a definitive diagnosis and management of a subsequent pregnancy.

Methods
This is a retrospective study of 193 cases of medical termination of pregnancy carried out in 2013 in the Center of Maternity and Neonatology of Tunis for congenital malformations who had a fetal autopsy after abortion and whose diagnosis was suspected prenatally by ultrasound.

Results
The average age of patients was 31.4 years [range 20-48]. Screening for malformations that have indicated the interruption of pregnancy was made by ultrasound. The mean gestational age at diagnosis was 21.6 weeks [range 12-34]. 21.8% of cases (n = 42) were diagnosed in the first trimester, 70.9% (n = 137) in the second trimester and 7.2% (n = 14) in the third trimester. The most common malformations were neurological malformations 49.7% (n = 96), second place come the amniotic fluid abnormalities 13.9% (n = 27) then renal and urinary tract malformations 8.8% (n = 17), the bone defects of the ends 7.2% (n = 14), gastrointestinal malformations 6.2% (n = 12), cardiac malformations 5.1% (n = 10), the cystic hygroma of the neck 4.66% (n = 9), increase nuchal translucency 3.1% (n = 6) and malformations of the face 2.5% (n = 5). There is a good correlation between fetal autopsy and ultrasound. In 76% of the cases, fetal autopsy confirmed the ultrasound findings. It concludes exactly the same sonographic findings in 16.6% of the cases (n = 32). In 69.4% of the cases (n = 134) there was not additional malformations suspected on the ultrasound. In 13.9% of cases (n = 27) there was a total mismatch between the data and ultrasound findings after final fetal autopsy. Ultrasound had a better sensitivity for the detection of neurological abnormalities (88.9%) with a specificity of about 97.8%, whereas it had a low sensitivity for the detection of malformations of the face (13.6%) and gastrointestinal malformations (24.4%).

Conclusion
The contribution of fetal autopsy and histological exam is very important in the multidisciplinary management of prenatal diagnosis. Prenatal ultrasound examination and fetal autopsy should be seen as complementary to increase our knowledge of the suggestive signs of fetal malformations. Indeed following the suspicion of congenital malformation should be multidisciplinary involving gynecologists, radiologists, neonatologists, geneticists, pathologists and histologists, and to allow a better approach to final diagnosis and better management of newborns and couples for decision to any subsequent pregnancy.