

Summary

Prenatal diagnosis and medical termination of pregnancy are activities in which quality assessment is crucial. Various methods of quality assessment have been defined, based mainly on assessing the quality of ultrasound. Another process to assess the quality of prenatal diagnosis is the comparison between prenatal diagnosis and post-mortem diagnosis.

In **Chapter 3** the **correlation between prenatal diagnosis and post-mortem diagnosis** was made in cases of elective termination of pregnancy due to fetal causes. The comparison between the pre-natal and post-mortem findings showed complete agreement in 61.1% of the cases, and there was no case of total disagreement. The autopsy resulted in the change of the risk of recurrence in 32% of cases.

In **Chapter 4** the **contribution of the autopsy in cases of termination of pregnancy due to chromosomal abnormalities** was evaluated. In these cases of termination of pregnancy there is already a definitive diagnosis and therefore autopsy does not alter the primary diagnosis or its implications. But the autopsy identified major anomalies not previously recognized in 12.2% of cases, and in 14% the anomaly previously detected by ultrasound was not confirmed. Cardiac anomalies was the group of anomalies in which there was a greater increase in information. Therefore, it is important to perform an echocardiogram before the couples decide to interrupt the pregnancy.

In **Chapter 5** the **phenotypic variability in Down syndrome** was evaluated. The phenotype of Down syndrome can be recognized in a fetus as early as 14 weeks' gestation but fetuses in the second trimester may have not yet developed the typical abnormalities; indeed, some fetuses appear normal even to the experienced observer. In this case-control study, the only biometric parameters significantly different were the occipito-frontal diameter and nasal bone length. Craniofacial anomalies were found in 96% of fetuses with Down syndrome and when evaluated separately, small ears were the only craniofacial feature not significantly different.

In **Chapter 6** the **contribution of perinatal autopsy in congenital heart anomalies** was evaluated. Abnormal cardiac findings were identified in 13.6% fetuses submitted to autopsy. Associated anomalies were found in 67.7% of fetuses with cardiac anomalies. In cardiac anomalies, the correlation between the pre-natal and post-mortem findings showed disagreement in 8% of cases. When the prenatal evaluation was performed by a pediatric cardiologist there was less disagreement.

In conclusion, the systematic autopsy in cases of medical termination of pregnancy allows, in many cases, the establishment of a definitive diagnosis, the redefinition of the prognosis, and the contribution to the appropriate counseling of couples (Chapter 3 and 6).

In cases of chromosomal abnormalities, autopsy provides additional information to the morphological characterization of chromosomal abnormalities. It may also improve the knowledge of the evolving phenotype and natural history, at early gestational ages (Chapter 4 and 5).

In the particular case of Down syndrome, a better knowledge of the fetal phenotype may help to define new ultrasound markers that could improve prenatal identification (Chapter 5).

An autopsy with full evaluation of the heart is important for the detection and better characterization of cardiac anomalies (Chapter 4 and 6).