AVALIAÇÃO DA CONTRIBUIÇÃO DO CARIÓTIPO E DA ANÁLISE MOLECULAR PARA DETECÇÃO DE ANEUPLOIDIAS EM FETOS POLIMALFORMADOS DO HCPA

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Introduction: Chromosomal anomalies are reported as the most common genetic condition in humans, indicating that cytogenetic analysis is fundamental for the investigation of malformation syndromes. Prenatal diagnosis, for detecting fetus chromosomal aberration, has become routinely applied. A fetus with multiple malformations has a great probability of having abnormal chromosomes. Objectives: Although karyotyping has proved to be a highly reliable test, it has some limitations, mainly time consuming and culture failure. In order to overcome these problems, we propose to apply a molecular technique, such as QF-PCR, which does not depend on cell culture. Material and Methods: Cell culture, and routinely karyotype procedure. Also DNA extraction and PCR techniques to apply specific probes for detecting the most common aneuploidies. Results: DNA was extracted from 50 fetal samples for applying Q-F PCR, and from those we got 20 complete analysis (detection of 13, 18, 21, X or Y chromosomes) and 10 partial results (XX or XY). Also, when it was difficult to obtain amniotic fluid, because of multiple malformations, we collected fetal samples from different sources (urine, cystic hygroma, intraperitoneal, dysplastic kidney, lung or cerebrospinal fluids). From those fetal materials (13 samples) we obtained a 100% of successful karyotyping. Conclusion: The importance of this study remains in giving different alternatives for a final diagnosis to fetus with

multiple malformations. In this way, it is possible to obtain cytogenetic information, which is very important for genetic counseling and reproductive decisions in the family.