

PRENATAL DIAGNOSIS FOR FETAL CHROMOSOMAL ABNORMALITIES: REPORT OF 18-YEAR EXPERIENCE IN A BRAZILIAN PUBLIC HOSPITAL

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**Introduction:** The study of fetal karyotype became a very important tool for fetal diagnosis of genetic diseases. Although this test is very restricted in Brazil, we analyzed 905 fetal karyotypes since 1989. Objectives: To describe the most frequent indications for karyotyping the fetus in our social-economic conditions; to estimate the frequency of most common aneuploidies in the population of Hospital de Clínicas de Porto Alegre; to report the results obtained with alternative samples. Method: Cytogenetic findings were reviewed from 1989 to 2007 in 905 women. Fetal material was cultivated in long term cell cultures. Standard cytogenetic techniques were used. Results: Although advanced maternal age was the most frequent indication, the majority of aberrant karyotypes was found when the indication was fetal malformation. 805 (91.5%) samples showed normal karyotypes, and 74 (8.5%) had abnormal karyotypes. When obtaining amniotic fluid was difficult, alternative fetal materials (urine, hygroma cystic fluid, lung fluid, or cerebro-spinal fluid) were collected and 100% of success was achieved. Conclusion: Prenatal cytogenetic analysis is recommended in high risk pregnancies to improve genetic counseling. The use of "alternative" fetal samples for karyotyping may be considered when amniotic fluid or fetal blood is difficult to obtain. In selected cases, this approach avoids unnecessary risk of additional invasive procedures, and provides a karyotype result to high-risk families. This report suggests that prenatal cytogenetic analysis should be performed specially in high-risk pregnancies, but should be established through a public health policy for prenatal diagnosis.