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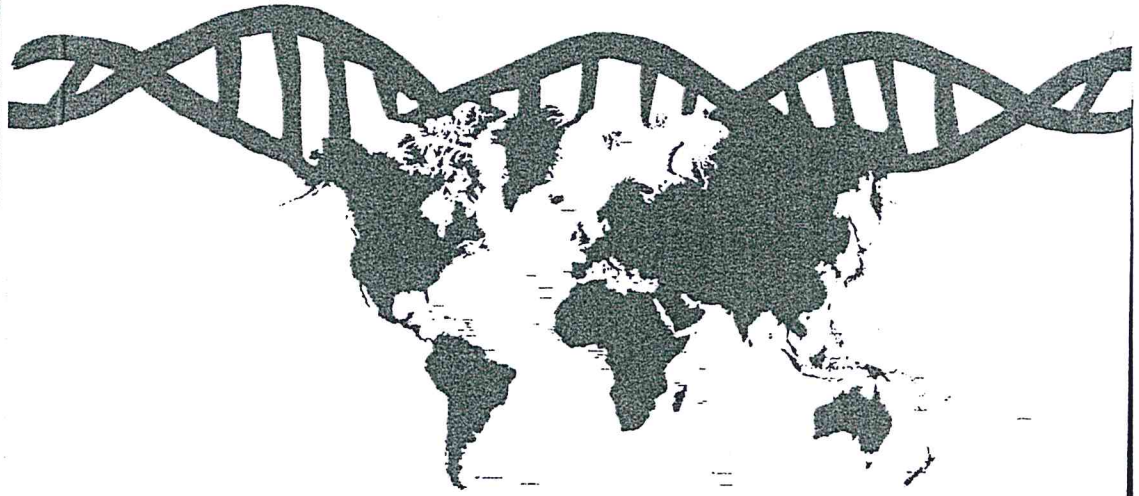
Abstract Book



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CYTOGENETIC ANALYSIS RELIABLE METHODS FOR DIAGNOSIS ANEUPLOIDIES IN PRENATAL DIAGNOSIS

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Introduction: Prenatal diagnosis of cytogenetic abnormalities is now widely recognized as a reliable method with an acceptable risk for couples at high risk of giving birth to a child with clinically significant chromosome abnormalities. Microscopic chromosome analysis of cultured cells has been regarded as the standard method for prenatal cytogenetic diagnosis.

Aim: Prenatal diagnosis employs a variety of techniques to determine the health and condition of an unborn fetus. Without knowledge gained by prenatal diagnosis, there could be an untoward outcome for the fetus or the mother or both.

Materials and methods: Prenatal cytogenetic analysis of chorionic villi obtained material or amniotic fluid was performed by culturing the cell culture from isolated cells, followed by the bendiranje metaphase chromosomes. However, this method is faced with a high degree of failure of cell culture, infection and contamination of the sample from the mother leading to inaccurate results or denial of a result. Additionally, conventional cytogenetic analysis (karyotyping, FISH) techniques are expensive and require a long time to get a result. Recently it was shown that fluorescence quantitative polymerase chain reaction (QF-PCR) is a useful and reliable method to diagnose aneuploidies in prenatal diagnosis. Also, QF-PCR analysis provides information of parent and origin of the aneuploidies, allowing adequate genetic counseling.

Results: Congenital anomalies account for 20 to 25% of perinatal deaths.

Conclusion: This report confirms the importance of screening and the cytogenetic diagnosis in the identification of the numerical chromosomal abnormalities.

Keywords: Prenatal diagnosis, chromosome analysis, abnormalities, aneuploidies

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