REVIEW OF PRENATAL KARYOTYPES AND ARRAYS IN THE REFERENCE HOSPITAL OF GIRONA AREA

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Abstract Body

INTRODUCTION: Chromosomal microarray (CMA) in prenatal diagnosis allows a significant increase in the number of diagnoses in pregnancies with ultrasound abnormalities, nuchal translucency >P99 or fetal loss, with normal QF-PCR/karyotype results.
METHODS: We present a review of the results of conventional karyotyping and CMA performed in prenatal diagnosis in the reference hospital of Girona Area (January 2015 - July 2017). A total of 395 prenatal samples were received. CMA was performed in 196 cases with normal QF-PCR/karyotype. The distribution of indications for CMA was: 34.2% ultrasound abnormalities, 17.3% positive first trimester screening, 19.4% nuchal translucency >P99, 10.7% fetal loss and 18.4% others.
RESULTS: Abnormal karyotypes were observed in 42 cases (10.6%). Four of them were balanced rearrangements, referred for familial chromosome rearrangements. Among the 196 cases that underwent CMA, 12 presented abnormal results (6.1%). Eleven were considered pathogenic and 1 variant of unknown significance. Nine of the imbalances were detected in fetuses with ultrasound abnormalities (diagnosed in the late second trimester). All abnormalities detected by CMA were cryptic, with the exception of a mosaic trisomy 22 diagnosed in a fetal demise. Eight of the abnormalities corresponded to well-described microduplication or microdeletion syndromes. All but 3 of the fetus with abnormal CMA results had normal first trimester screening.
CONCLUSIONS: Obtained detection rates for chromosome abnormalities are in agreement with published literature. Ultrasound abnormalities are the indication with a higher positive predictive value (13.4%). Parameters evaluated in the first trimester screening are usually within the normal range in fetuses with cryptic imbalances. Thus, suspicion of cryptic imbalances in pregnancies without ultrasound abnormalities or known family rearrangements remains a challenge. Another challenge is to define the etiology of ultrasound findings in the scenario of normal CMA results. Whole-exome sequencing will probably shed some light in this field in the near future.