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Topic of the presentation: cfDNA screening in clinical practice and results of an updated metaanalysis

Biography: Mar qualified in Medicine and specialized in Obstetrics and Gynecology in Madrid, Spain. From 2012 to 2015 she was a Research Fellow at Harris Birthright Research Centre for Fetal Medicine, King's College Hospital, London. Currently, she is a Consultant in Fetal Medicine and Obstetrics in Torrejon University Hospital, Madrid. From 2012 to date she has published, under the supervision of Professor Kypros Nicolaides, a series of clinical validation and implementation studies as well as meta-analysis on cell free DNA analysis of maternal blood for early screening of aneuploidies. She has participated in several specialist meetings on this field, which also constituted her PhD.

ABSTRACT: Prenatal diagnosis of fetal aneuploidies like Down syndrome requires chorionic villous sampling or amniocentesis, but these are associated with risk of miscarriage.

In the last 45 years screening for aneuploidies has evolved from maternal age alone to second trimester serum biochemical testing and finally, first-trimester combined test which detects about 90% of cases of Down syndrome at a false positive rate of 5%.

Recently, analysis of cell-free DNA (cfDNA) in maternal plasma has become possible and this has led to a major increase in detection rate and decrease in false positive rate of fetal trisomies.

In this lecture we will discuss the main concerns for the introduction of this method in fetal medicine practice and the results of an updated meta-analysis.