

ULTRASOUND

in Obstetrics & Gynecology

Editor in Chief: Professor Basky Thilaganathan

The Official Journal of
the International Society of Ultrasound
in Obstetrics and Gynecology



CONTENTS

EDITOR'S NOTE

- 1 Ushering in a new dawn in obstetrics and gynecology: the industry of cell-free DNA testing
B. Thilaganathan

EDITORIAL

- 4 Prenatal exome sequencing for fetuses with structural abnormalities: the next step
S. C. Hillman, D. Willams, K. J. Carss, D. J. McMullan, M. E. Hurles and M. D. Kilby

OPINION

- 10 Clinical implementation of cell-free DNA-based aneuploidy screening: perspectives from a national audit
L. Hui, M. Teoh, F. da Silva Costa, P. Ramsay, R. Palma-Dias, Z. Richmond, S. Piessens and S. Walker, on behalf of the Australian NIPT collaboration

SYSTEMATIC REVIEWS

- 16 Procedure-related risk of miscarriage following amniocentesis and chorionic villus sampling: a systematic review and meta-analysis
R. Akolekar, J. Beta, G. Picciarelli, C. Ogilvie and F. D'Antonio
- 27 Array comparative genomic hybridization and fetal congenital heart defects: a systematic review and meta-analysis
F. A. R. Jansen, Y. J. Blumenfeld, A. Fisher, J. M. Cobben, A. O. Odibo, A. Borrell and M. C. Haak

ORIGINAL PAPERS

- 36 Screening for trisomies 21, 18 and 13 by cell-free DNA analysis of maternal blood at 10–11 weeks' gestation and the combined test at 11–13 weeks
M. S. Quezada, M. M. Gil, C. Francisco, G. Or6sz and K. H. Nicolaides
- 42 First-trimester contingent screening for trisomies 21, 18 and 13 by fetal nuchal translucency and ductus venosus flow and maternal blood cell-free DNA testing
K. O. Kagan, D. Wright and K. H. Nicolaides
- 48 A unified approach to risk assessment for fetal aneuploidies
D. Wright, A. Wright and K. H. Nicolaides
- 55 Non-invasive prenatal testing for fetal aneuploidies in the first trimester of pregnancy
Y. Song, S. Huang, X. Zhou, Y. Jiang, Q. Qi, X. Bian, J. Zhang, Y. Yan, D. S. Cram and J. Liu
- 61 Performance of screening for aneuploidies by cell-free DNA analysis of maternal blood in twin pregnancies
E. Bevilacqua, M. M. Gil, K. H. Nicolaides, E. Ordo6nez, V. Cirigliano, H. Dierickx, P. J. Willems and J. C. Jani

- 67 UK NHS pilot study on cell-free DNA testing in screening for fetal trisomies: factors affecting uptake
M. M. Gil, G. Giunta, E. A. Macalli, L. C. Poon and K. H. Nicolaides
- 74 Cell-free fetal DNA screening in the USA: a cost analysis of screening strategies
M. I. Evans, J. D. Sonek, T. W. Hallahan and D. A. Krantz
- 84 Costs and benefits of non-invasive fetal RhD determination
L. Teitelbaum, A. Metcalfe, G. Clarke, J. S. Parboosingh, R. D. Wilson and J. M. Johnson
- 89 Differentiation of genetic abnormalities in early pregnancy loss
S. T. Romero, K. B. Geiersbach, C. N. Paxton, N. C. Rose, E. F. Schisterman, D. W. Branch and R. M. Silver
- 95 Chromosomal microarray in fetuses with increased nuchal translucency
I. C. B. Lund, R. Christensen, O. B. Petersen, I. Vogel and E. M. Vestergaard
- 101 Fetal fraction of cell-free DNA in maternal plasma in the prediction of spontaneous preterm delivery
M. S. Quezada, C. Francisco, D. Dumitrascu-Biris, K. H. Nicolaides and L. C. Poon
- 106 Maternal plasma cell-free DNA in the prediction of pre-eclampsia
D. L. Rolnik, N. O'Gorman, M. Fiolna, D. van den Boom, K. H. Nicolaides and L. C. Poon

LETTERS TO THE EDITOR

- 112 Performance of non-invasive prenatal testing when fetal cell-free DNA is absent
T. Takoudes and B. Hamar
- 113 Uptake of non-invasive prenatal testing by Japanese women
R. Akaishi, T. Yamada, S. Kawaguchi, T. Kojima, T. Koyama, T. Umazume, M. Morikawa, K. Cho and H. Minakami
- 114 Prenatal diagnosis of androgen insensitivity syndrome using cell-free fetal DNA testing
D. Zilberman, L. I. Parikh, M. Skinner and H. J. Landy
- 115 Non-invasive prenatal testing – it's all a matter of timing
Y. Zalel

CORRESPONDENCE

- 117 A flawed challenge but valid recommendation: a response to Takoudes and Hamar
G. E. Palomaki, E. R. Ashwood and K. E. Weck