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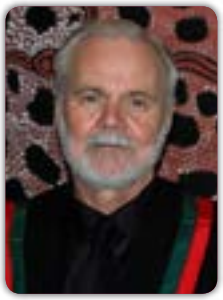

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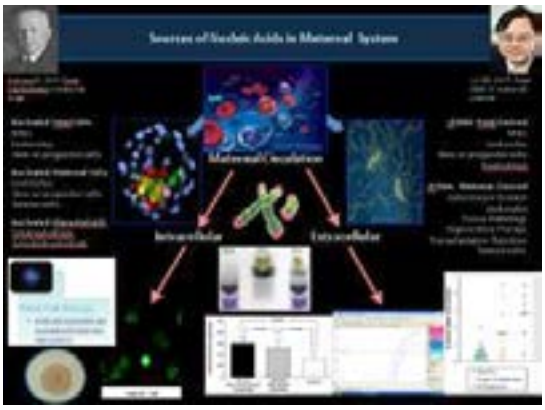
Non-invasive prenatal screening and then some?

By definition, non-invasive prenatal screening (NIPS) encompasses all testing modalities applied prenatally in order to identify a subset of obstetric population deemed to be at increased risk for disorder being screened. So, if we focus on screening for fetoplacental wellbeing, technically, this includes ALL current screening modalities: proteomic (PAPP-A, FβhCG, AFP), ultrasonographic (NT), extracellular (cf) DNA and fetal cells. Patients with positive screening results should be counseled and advised to seek confirmatory testing, prior to any definitive management action. In figure are shown the various sources of extracellular (cf) and intracellular (g) DNA in the mother. Whilst efficacy of cfDNA screening for trisomy 21 is well established, the physiology and trophoblastic source of cfDNA limits its screening application. More recently, NIPS has been applied to fetoplacental cells as a source of fetal genomic (g) DNA. Cellular targets include nucleated fetal red blood cells (NRBS), mononucleated cytotrophoblast (CT) and polynucleated syncytiotrophoblast (SCT) cells. Cell enrichment may be mediated immunologically or physically, at present fetal cell isolation is achieved by laser dissection. Whilst providing a pure fetal cell, this technology is labor intensive and limited in application to high risk populations. This technology is not applicable to population based screening. Non-invasive prenatal screening has progressed from proteomic, as current first trimester programmes, to ultrasonographic and, more recently, to cfDNA. Whilst limitations of cfDNA are understood and accepted, this has not hampered to uptake of cfDNA based screening. With improvements in molecular technology and application to single cells, we are on the verge of entering into the era of comprehensive prenatal fetal genotyping, including exosome sequencing, based on analysis of gDNA.



Biography

Michael J Sinosich has completed his PhD on Trophoblast Physiology and PAPP-A. His research interests include non-invasive assessment of fetomaternal wellbeing. He is the Director of Prenatal Testing (DHM Pathology) and serves as Consultant at Pictor Ltd, a developer and manufacturer of multiplexed microELISA assay platform. He has published/presented numerous papers in reputed journals and holds several patents.

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