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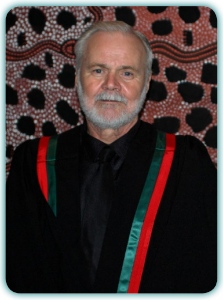

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Prenatal screening with first trimester biochemistry

Prenatal screening for assessment of fetal development has been established for almost half a century. Our first trimester screening program includes four (4) biochemical markers: PAPP-A, FβhCG, PlGF and AFP. Markers were quantified (DELFIA® DXpress; PerkinElmer) and marker MoM values calculated (LifeCycle v2.2; Perkin Elmer), using lot specific derived polynomial regression curves. Pregnancies with known outcomes included: (1) Aneuploid: Trisomy 21, Trisomy 18, Trisomy 13, Triploid, Sex chromosome aneuploidies, (2) Failed pregnancies, (3) Fetal structure: Anencephaly, Fetal growth restriction, and (4) Maternal Health: Gestational trophoblastic disease, Sub-chorionic bleeding. Reduced circulating PAPP-A level is cause for concern. Low PAPP-A level is associated with aneuploidy (Trisomy/13/18/21, Triploidy) and poor pregnancy prognosis. Similarly, a low FβhCG level is also associated with compromised pregnancy. However, an elevated FβhCG is indicative of Trisomy 21. Excessively elevated FβhCG, even in presence of normal PAPP-A levels, warrants further investigation of trophoblast cell physiology. Elevated levels of fetal derived marker (AFP) may indicate increased transplacental exchange or presence of fetal structural abnormality. Biochemical profile of viable aneuploidies such as, sex chromosome aneuploidies are not distinguishable from profiles of normal pregnancies. However, NT measurements in such pregnancies are markedly increased and may present as a cystic hygroma. PlGF, a marker of placental angiogenesis, levels are reduced in first trimester pregnancies which subsequently develop early onset pre-eclampsia and/or reduced fetal development. Whilst the combination of feto-placental derived biochemistry with ultrasonographic biometry, improves Trisomy 21 detection (to about 92-93%), the inclusion of PlGF has introduced another dimension to screening because now we can simultaneously screen for both fetal and maternal wellbeing. Over the past two decades, non-invasive prenatal screening has progressed from screening for targeted fetal abnormalities to combined prenatal assessment of feto-maternal wellbeing. Each screening modality (biochemistry, ultrasonography, cfDNA) offers benefits only attained in a holistic screening offering.

Biography

Michael J Sinosich established the Division of Prenatal Testing within Sonic Clinical Institute in 2002. Michael is an internationally respected investigator in early pregnancy well-being. He has published extensively in peer reviewed journals and presented numerous papers at local, national and international forums.

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