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Is abdominal circumference accurate for diagnosing fetal growth restriction and its neonatal complications?

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Fetal Growth Restriction (FGR) is associated with adverse neonatal, post-natal and paediatric outcomes. Early detection via FGR screening allows adequate risk management with emphasis placed on early prenatal care. This review aims to evaluate the usefulness of Abdominal Circumference (AC) in FGR diagnosis and in prediction of associated adverse outcomes. Pubmed, Cochrane and Scopus were systematically searched to find 8 appropriate articles for review. Evidence determined AC to be a useful technique to elucidate fetal growth independently but to be even more accurate in combination with other parameters. This data will inform the practice of obstetricians; selecting those fetuses at risk is necessary for allocation of antenatal testing, appropriate treatment and timely delivery. Limitations of this review included varying population selection, use of retrospective data and the inclusion of both twin and singleton studies. A combined approach of AC and estimated fetal weight (EFW) in FGR diagnosis appears to be the most sensitive and specific criteria. Future research into individualisation of fetal growth study and inclusion of biochemical markers appears promising.

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Mutations in SCN3A gene cause early infantile Epilepsy

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Neurons allow the brain and body to communicate through electrical signals. Voltage-gated ion channels like sodium, calcium, potassium, and chloride channels are critical in electrical signaling. Sodium channels play an important role in the generation and propagation of the signals. Under normal conditions, these signals allow neurons to communicate, however, abnormal, and excessive excitation of neurons may lead to epileptic encephalopathies. The SCN3A gene, encoding the type 3 sodium channel, Nav1.3, is highly expressed in the brain starting from 16 weeks of fetal life. We have discovered that the mutations in SCN3A result in the gain of function by altering gating properties of the channel, leaving the ion channel stuck open that in turn causes current flood leading to electric sparking, a signature of epilepsy. Magnetic resonance imaging (MRIs) and neurological evaluations further revealed that the epileptic patients carrying mutations in SCN3A also exhibit malformation of cortical folding indicating its possible role in brain development. This study reinforces the role of variants in SCN3A as a cause of neurodevelopmental disorders along a spectrum of severity that includes epilepsy and polymicrogyria and suggests that gain of channel function is an important mechanism of disease pathogenesis. Manipulating epileptic genes in utero could be used to analyze roles of genes in embryonic development and intellectual disability. These are still early days, but with precision medicine, early prenatal diagnosis, SCN3A gene manipulation during the critical window, may help prevent brain malformations in babies.

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Obstetric outcomes of assisted reproduction in advanced maternal age

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Statement of the Problem: In recent years, the number of women who give birth for the first time in advanced maternal age (AMA) after assisted reproduction (ART) has increased significantly. Pregnancy and childbirth in this population usually proceed with a large number of complications and require special tactics of follow-up and delivery. The purpose of this study is to investigate the impact of AMA and ART on obstetric outcomes of nulliparous singleton cephalic pregnancies and determine possible measures for the prevention of obstetric complications in AMA women after ART

Methodology & Theoretical Orientation: A retrospective statistical analysis of medical recorders of live-born singletons, cephalic, non-anomalous nulliparous pregnancies was done. The ART conception and spontaneous conception groups were compared and contrasted.

Findings: Intrauterine growth restriction, nonreassuring fetal state, induction of labor, operative vaginal delivery, C-section and massive obstetric hemorrhage (MOH) are authentically more common for the nulliparous ART-conception AMA patients, especially in the subgroup aged of 40-45 years.

Conclusion & Significance: Nulliparous AMA women after ART have significantly higher risks of the adverse obstetric complications, and therefore require dynamic monitoring of the mother`s and fetal conditions, timely prevention and correction of disorders during pregnancy and delivery.

Recommendation: These made to the maternity hospitals to consider this population as highly threatening for C-section and MOH and use uterotonics and hemostatics for prevention of the complications. Taking into account that the risk of complications significantly increases in patients aged 40 years and older, the option of the elective term C-section also should be considered for this subgroup of patients.

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Utility of circulating cell-free DNA in assessing microsatellite instability and loss of heterozygosity in breast cancer using human identification approach

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The diagnostic and prognostic utility of circulating cell-free DNA (cfDNA) in breast cancer (BC) patients was recently reported. Here, we investigated the use of cfDNA to examine microsatellite instability (MSI) and loss of heterozygosity (LOH) for early BC diagnosis. cfDNA and genomic DNA from 41 female BC patients and 40 healthy controls were quantified using NanoDrop spectrophotometry and real-time PCR. The stability of genomic and cfDNA was assessed using a high-resolution AmpFlSTR MiniFiler human identification kit. Significant increases in cfDNA plasma concentrations were observed in BC patients compared to controls. The genotype distribution of the eight autosomal short tandem repeat (STR) loci D7S820, D13S317, D21S11, D2S1338, D18S51, D16S539, FGA, and CSF1PO were in Hardy–Weinberg equilibrium. Significant differences in the allele frequencies of D7S820 allele-8, D21S11 allele-29, allele-30.2, allele-32.2, and CSF1PO allele-11 were seen between BC patients and controls. LOH and MSI were detected in 36.6% of the cfDNA of patients compared to genomic DNA. This study highlights the utility of plasma-derived cfDNA for earlier, less invasive, and cost-effective cancer diagnosis and molecular stratification. It also highlights the potential value of cfDNA in molecular profiling and biomarkers discovery in precision and forensic medicine.

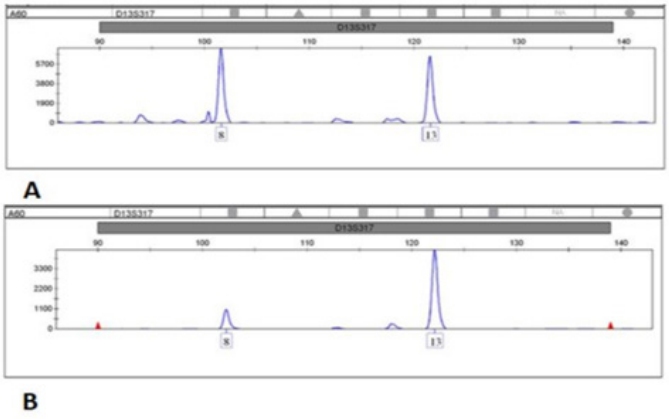

Figure 1. Loss of Heterozygosity observed at locus D13S317. Panels (**A**) represents normal genotypes in genomic DNA, while Panel (**B**) is a representative of MSI in cfDNA.

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