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Controversies and pitfalls in first trimester NIPT screening for sex chromosome aneuploidies (XO, XXX, XXY)

The role of NIPT in screening for chromosome aneuploidies Trisomy 21, 18, and 13 has revolutionized the assessment of the fetus in the 1st trimester of pregnancy. Detection rates of up to 99%, 99%, and 97% respectively have been reported in the screening of these chromosome aneuploidies. What is less clear are the detection rates and positive predictive values in the screening of sex chromosome aneuploidies XO, XXX, XXY, and XYY. There is also a wide variation in the efficacies of different NIPT platforms in screening for sex chromosome aneuploidies.

The lecture seeks to address important screening objectives, such that clinicians are equipped to apply the clinically relevant genetic and genomic knowledge, understand the indications, benefits and limitations of genetic testing, recommend the appropriate genetic test and demonstrate appropriate pre-test and post-test genetic counseling. Controversial issues such as appropriate interpretation of a NIPT no-call result for Monosomy X will be addressed. Other confounding factors in no-call results for sex chromosome aneuploidies such as maternal malignancy and maternal sex chromosome abnormalities will also be discussed.

Join our webinar!

Date: Thursday, July 29

Time: 6:00pm SGT

Register Here

This is a CME accredited event.

FEATURED SPEAKERS:

Dr Chang Tou Choong MBBS FRCS FRCOG MD FAMS

Dr. T.C. Chang is a Consultant Obstetrician and Gynaecologist based at Thomson Medical Centre. From 1998 to 2020, he was the Head of the Fetal Assessment Unit, Centre for Prenatal Diagnosis and Obstetric Ultrasound, at Thomson Medical Centre. He is presently the Head and Director, Fetal Assessment Centre, Singapore Women's Medical Group. His interests are in first trimester anatomy scanning, NIPT in challenging clinical scenarios, first trimester screening for pre-eclampsia and fetal growth restriction, ultrasound assessment of the at-risk fetus and management of high-risk pregnancies.

Dr Samantha Leonard MBBS BSc (Hons) MA PhD MRCPCH

Dr Samantha Leonard is the International Medical Director of Natera Inc. She has a unique background in clinical genetics and medical ethics and has practiced in both the UK and France. Her work on pan-European research projects in the area of translational genetic medicine made her well-positioned to meet the challenges of working at the forefront of clinical genetic testing technologies. Dr Leonard has a wide experience of teaching, presenting, and publishing in the field of clinical genetics and next-generation sequencing.