5598: Chromosomal microarray (CMA) or conventional karyotype (KT) in fetuses with increased Nuchal translucency – making the right choice to optimize the pregnancy outcome

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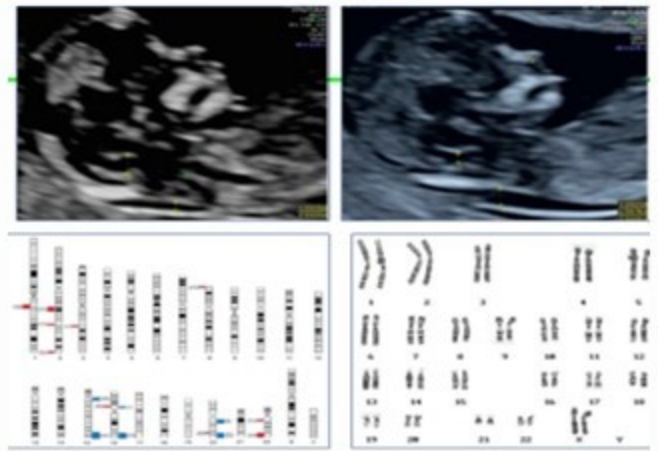
Introduction: Increased NT can be associated with sub microscopic chromosomal abnormalities that are typically missed on conventional KT and are picked by chromosomal microarray.

Objectives: To compare the diagnostic efficacy of chromosomal microarray (CMA) with conventional karyotype (KT) in fetuses with increased Nuchal translucency (iNT)



Methodology:

- Retrospective comparative study of prospectively collected data from a single tertiary fetal care referral centre during Jan 2002 to Dec 2022.
- NT was measured as per FMF guidelines & iNT was defined as NT > 95th centile for the GA.
- Risk calculation was done on the FMF software and all invasive procedures were performed by trained and FMF certified operators.
- Records were maintained on the Astraia software.
- Past natal outcomes were obtained by telephonic interview with the parents

Results

Table 1

Distribution of cases

Total cohort	14562
Increased NT (iNT)	1391(9.5%)
Normal NT	13171(90.4%)

Table 2 Karyotypic abnormalities in increased NT

Karyotype Results	Karyotype Normal	KT-Aneuploidies	KT-Translocations/ mosaicism
Isolated increased NT -791	722/791 (91.3%)	57/791 (7.2%)	12/791 (1.5%)
iNT with defects-96	58/96 (60.4%)	35/96 (36.4%)	3/96 (3.1%)
Normal NT with defects - 26	15/26 (57.6%)	11/26 (42.3%)	0

Table 3 CNVs in CMA of increased NT

CMA Results	Normal	Numerical aberrations	Pathogenic CNVs	VOUS
Isolated iNT-45	36/45 (80%)	6/45 (13.3%)	2/45 (4.4%)	1/45 (2.2%)
iNT with defects -11	5/11 (45.4%)	2/11 (18.2%)	1/11 (9.1%)	3/11 (27.2%)
Normal NT with defects - 4	1/4 (25%)		3/4 (75%)	

Table 4 Individual cases with pathogenic CNVs in iNT

Maternal age (years)	GA	NT	Defects	Defects-1st T/2nd/3rd/ PN	CMA- Results	Visible by KT (Y/N)	Outcome	Outcome comments
38	13	2.9mm	-	-	Gain on 15q11.2	N	Live birth	1.8years, doing well
22	12	3.1mm	Short long bones, Cardiomegaly	2nd T	22q11.2 microdeletion	N	Termination	
29	13	4mm	-	PN	(13.92 Mb deletion) on 10q26.11	N	Live birth	Hypertelorism, syndactyly, PDA VSD ,developmental delay-Genetic syndrome

Conclusion-

- **The diagnostic yield of CMA in isolated iNT was 4.4% and yield was increased to 9.1% in the presence of defects.**
- **The diagnostic yield of KT in isolated iNT was 1.5% to identify translocation, and mosaicism that cannot be detected by CMA and yield increased to 3.1% in the presence of defects.**
- **Our study highlights the importance of carefully selecting the indications when CMA will give additional information as opposed to KT and vice versa.**

Egloff et al. Diagnostic yield of chromosomal microarray analysis in fetuses with isolated increased nuchal translucency: a French multicenter study. doi.org/10.1002/uog. 18928.Ultrasound in Obstetrics and Gynecology.2017

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