4762: Increased Nuchal Translucency (iNT) and aneuploidies – revisiting the traversed path.

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Introduction: Increased fetal nuchal translucency thickness at 10–14 weeks of gestation is a common phenotypic expression of fetal chromosomal defects, structural abnormalities and genetic syndromes. There is also increased incidence of adverse pregnancy outcome. However, once chromosomal abnormalities have been excluded, majority of cases are known to have a good outcome

Objective

To assess the prevalence of aneuploidies in fetuses with isolated iNT and compare with those with additional single and additional multiple marker(s) in structurally normal fetuses

Methodology

- Retrospective comparative study of prospectively collected data from a referral centre during Jan 2005 to Dec 2022)
- 13,992 singleton pregnancies who underwent first trimester screening between 11^{+0} – 13^{+6} weeks with known follow ups were included. Multi fetal pregnancies and fetuses with major structural anomalies were excluded from our study.
- All scans were performed by FMF certified operators, as per FMF criteria and were documented on Astraia fetal database software
- Outcomes were obtained by telephonic interview and examination of delivery details in hospital records.

Results

	Number of fetuses	Abnormal karyotype 107/13992 (0.76%)
No markers	12515	37 (0.29%)
Isolated iNT	884 (84.1%)	38 (4.3%)
iNT + TR	72 (6.85%)	3 (11.3%)
iNT + aDV	38 (3.6%)	6 (15.8%)
iNT + ANB	26 (2.47%)	13 (18.05%)
iNT + 2 markers	30 (2.85%)	10 (33.33%)

Prevalence of aneuploidy increases as the number of markers seen increases

	T21	T18	Turner's	Others
No markers	13	1	0	23
iNT + ANB	10	2	1	0
iNT + aDV	2	1	2	1
iNT + TR	3	0	0	0
iNT + 2 markers	8	1	0	1

Most common aneuploidy seen is Trisomy 21

	N=14650	Without anomaly	Major anomaly
Increased NT	1359 (9.3%)	1050	309 (22.7%)
Normal NT	13291 (90.7%)	12942	349 (2.6%)

Fetuses with iNT had 9 times higher chance of having a major structural anomaly

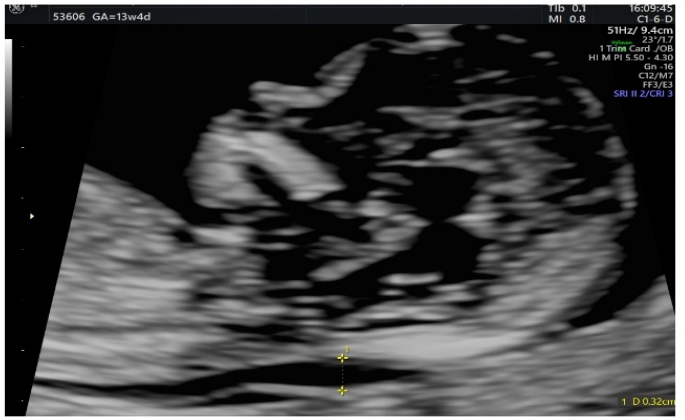

Fig 1: Measurement of fetal NT as per FMF criteria

Conclusions:

- Prevalence of aneuploidy is higher in iNT group in comparison to no markers' group and this increases further when increased NT is associated with other markers
- Prevalence of structural anomalies is increased in iNT group in comparison to normal fetuses & warrants for a detailed first trimester anatomy check

- Increased NT should provoke search for possibility of associated markers
- Invasive testing should be offered to all
- A detailed first trimester anomaly scan in all fetuses with increased NT is mandatory

Reference: Souka, A.P et al Defects and syndromes in chromosomally normal fetuses with increased nuchal translucency thickness at 10–14 weeks of gestation. Ultrasound Obstet Gynecol, 11: 391-400

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