4738 : Spectrum of cardiac abnormalities in fetuses with increased and normal NT at 11^{+0} to 13^{+6} weeks of gestation

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Introduction: Cardiac defects are the most common birth defects, with a prevalence of up to 8 - 12 per 1000 live births, half of them being major cardiac defects. A structured protocol including 4CV and OFT views for assessment of fetal cardia in the 1T can achieve high detection rates for major CHDs.

Objective: To study the spectrum of cardiac abnormalities in fetuses with increased and normal NT at 11^{+0} to 13^{+6} weeks of gestation in chromosomally normal and abnormal fetuses

Method

- Retrospective study of prospectively collected data from a tertiary fetal care centre from January 2005 to December 2022
- Singletons with increased and normal NT and completed outcomes were analyzed
- NT measurement was performed in 1T according to FMF protocol
- Detailed structural anatomical survey including assessment of fetal heart according to existing unit protocol was performed in all fetuses
- TR/reversed 'a' wave in DV/ARSA were considered as cardiac markers and were excluded in absence of cardiac anomalies along with those with extracardiac fetal abnormalities
- Fetal karyotyping was offered in all cases with increased first trimester risk

Fig 1. Fetal NT measurement

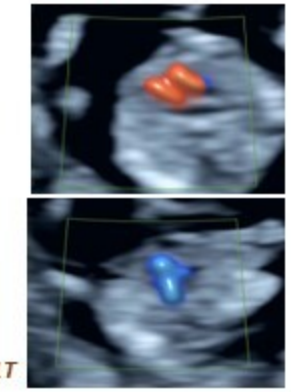
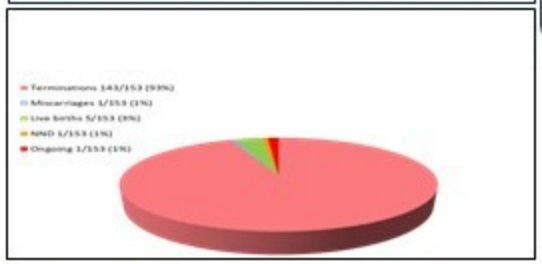


Fig 2. Cardiac exam in 1T

Results

Outcomes of isolated CHDs detected in 1T = 153



Chromosomal abnormalities	Most frequently seen major CHDs
Trisomy 21	AVSD - 8/12 (66.67%), VSD - 4/12 (33.33%)
Turners Syndrome	HLHS - 3/3 (100%)
Trisomy 18	HLHS - 5/7 (71.4%), AVSD - 1/7 (14.3%), VSD - 1/7 (14.3%)
Trisomy 13	HLHS – 2/3 (66.67%), AVSD – 1/3 (33.33%)
Wolf Hirschhorn Syndrome	VSD 1/1 (100%)

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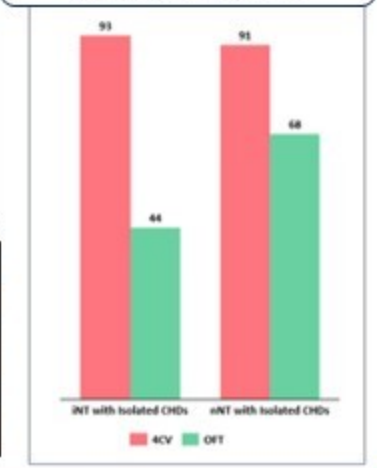
Detection rate of isolated CHD
iNT-93% (4CV), 44% (OFT)
nNT-91% (4CV), 68% (OFT)



Fetuses with isolated cardiac defects and available karyotype
65/ 153 (42.5%)

iNT 45/65 (69.2%)		nNT 20/65 (30.8%)	
Normal KT/CMA 19/45 (42.2%)	Abnormal KT/CMA **26/45 (57.8%)**	Normal KT/CMA 19 / 20 (95%)	Abnormal KT/CMA **1/20 (5%)**
HLHS - 7/19 (36.8%) VSD - 7/19 (36.8%) AVSD - 3/19 (15.8%)	HLHS - 10/26 (38.5%) AVSD - 10/26 (38.5%)	HLHS - 4/19 (21.1%) TGA - 3/19 (15.8%%) DORV - 3/19 (15.8%)	VSD - 1/1 (100%)

Conclusions

- More than half (69.2%) of fetuses with isolated cardiac defect had increased NT
- 42.2% of euploid fetuses with iNT had major CHDs
- 26/45 (57.8%) of fetuses with iNT and isolated cardiac defect had an abnormal karyotype, while 1/20 (5%) in the nNT group was affected with Trisomy 21
- The detection rate of isolated CHDs with 4 CV and OFT views in increased and normal NT groups was comparable
- Most common CHD in both groups, irrespective of euploid status, was HLHS and AVSD

References:

Galindo A, Comas C, Martínez JM, Gutiérrez-Larraya F, Carrera JM, Puerto B, Borrell A, Mortera C, de la Fuente P. Cardiac defects in chromosomally normal fetuses with increased nuchal translucency at 10-14 weeks of gestation. J Matern Fetal Neonatal Med. 2003 Mar;13(3):163-70.

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