4750: **Prenatal diagnosis, association and outcomes of micrognathia detected in the first trimester scan (FTS) and audit of the "missed cases" detected subsequently**

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Introduction : FTS provides a good opportunity to assess facial abnormalities. Facial profile can easily be studied on FTS as the view is the same for an optimal NT image. Careful examination of the facial profile and study of alignment of the bony landmarks while taking measurement for Nuchal Translucency (NT) can aid in suspecting micrognathia in the first trimester.

Objectives:

- Primary Objective: Association of micrognathia with structural defects and abnormal genotype
- Secondary Objective: Audit the "missed cases" in the FTS on subsequent scans or on postnatal evaluation

Methodology:

- Retrospective comparative study of prospectively collected data from a single tertiary fetal care referral centre during Jan 2005 to Dec 2022
- 14,439 singleton pregnancies who underwent first trimester (1T) screening between 11^{+0} – 13^{+6} weeks with known follow ups were included All scans were performed by FMF certified operators and were documented on Astraia fetal database software
- A re-audit was done to assess the impact of changes in the FTS protocol in detection of micrognathia
- All cases of micrognathia detected after FTS were considered as "missed cases"

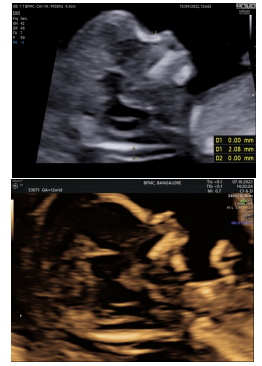

Fig 1 (a) : Normal profile, (b) Micrognathia

Results

Major defects : 1,125/14,439 (7.8%)
Incidence : 23/1,125 (2.0%)
1T Diagnosis : 14/23 (60.9%)

Phase wise DR in 1 T
P1 = 66.6%, P2 = 55.5%, P3 = 63.6%

Invasive testing : opted by 6/14 (42.8%)
Abnormal genotype - 3/6 (50% (T21- 2, Triploidy 1)
"Missed cases" of micrognathia in the FTS - 9/23 (39.1%) on MTAS

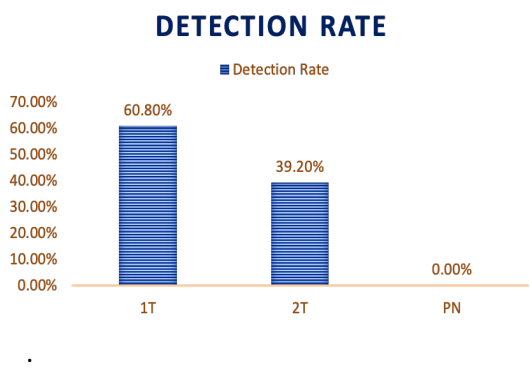

Fig 2 : Overall DR for micrognathia

Parameters (n = 14439)	Micrognathia (n = 14/23, 60.9%) (Phase 1 – 2, Phase 2 : 5 , Phase 3 :7)
1st T	Isolated – 8/14, **57.1%** Additional defects, 6/14, **42.9%**
Increased NT	4/14, **28.5%**
Outcomes	Terminations - 14/14, **100%**
Invasive	6/14, (**64.2%**) (3/6 – Normal, 3/6 – Ab ; 2 – T21, 1 Triploidy)

Table 1 : Characteristics of fetuses with FT diagnosis of micrognathia

2T	Micrognathia (9)
Phase 1 (1)	a/w Arnold Chiari malformation - 1
Phase 2 (4)	Isolated - 1 a/w – Corpus Callosal agenesis (3)
Phase 3 (4)	Isolated 2 a/w CLAP – 1, VSD - 1

Table 2: Characteristics of "missed cases" on FTS

Conclusions:

- Careful examination of NT plane aided in detection of 60% of cases in the FTS
- Diagnosis of "micrognathia" in 1T scan should mandate detailed examination for additional defects (>40% cases)
- Important to offer genetic workup as 50% had abnormal genotype
- Regular audit and modification of one's practice can improve DR in FTS for "micrognathia"

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