4752: **Prenatal diagnosis, association and outcomes of Cleft Lip and Palate (CLAP) detected in the first trimester scan (FTS) and audit of the "missed cases" detected subsequently**

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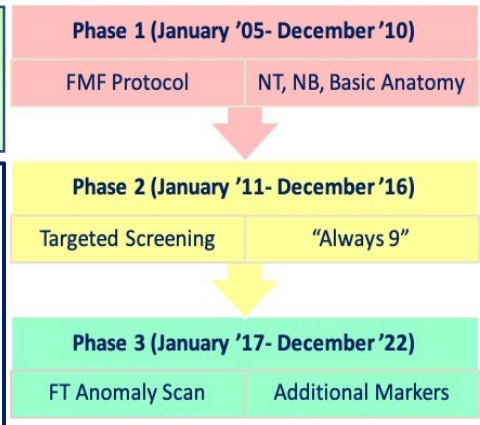
Introduction : First Trimester scan provides a good opportunity to assess facial abnormalities. Identification of CLAP should prompt detailed evaluation of other systems to rule multi-systemic involvement. Invasive testing for a thorough genetic workup should be offered to all to optimise pregnancy outcomes and assess the risk of recurrence.

Objectives:

- Primary Objective: Association of CLAP 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 CLAP
- All cases of CLAP detected after FTS were considered as "missed cases".



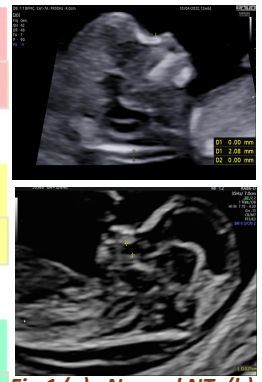


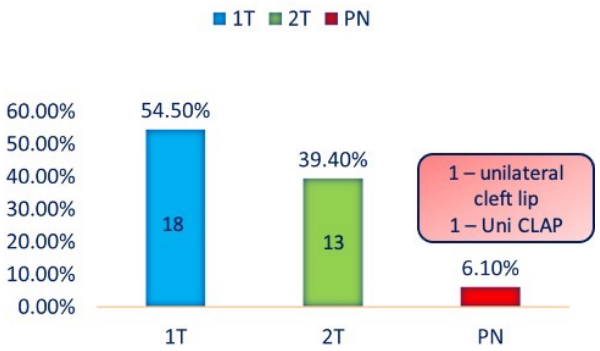
Fig 1 (a) : Normal NT, (b) "Maxillary Gap" - CLAP

Results

Major defects : 1,125/14,439 (7.8%)
Incidence of CLAP : 33/1,125 (2.9%)
1T Diagnosis of CLAP : 18/33 (5.4%)
Increased NT – 269/613 (43.8%)

Phase wise DR of CLA in 1 T
P1 = 0%, P2 = 60%, P3 = 60%

Invasive testing : opted by 5/18 (27.7%) - 3/5 (60%) had abnormal genotype (T21- 1, T13 – 1, Abnormal CMA -1)
"Missed cases" of CLAP in the FTS - 13/33 (39.4%) on MTAS & 2/33 (6.1%) postnatally



Overall DR of CLAP in 1T : 18/ 33 (54.5%)

Parameters (n = 14,439)	CLAP (n =18/ 33) (Phase 1 : 0, Phase 2 : 6, Phase 3 : 12)
1st T	Isolated – 8/18, **44.4%** Additional defects -10/18, **55.6%**
Increased NT	6/18, **33.3%**
Outcomes	Terminations - 18/18, **100%**
Genetic Workup	Available for - 5/18, **27.7%** **Abnormal 3/5, 60%** (T21 -1, T13 -1, gain of 9p on CMA – 1)

Table 1 : Characteristics of fetuses with FT diagnosis of CLAP

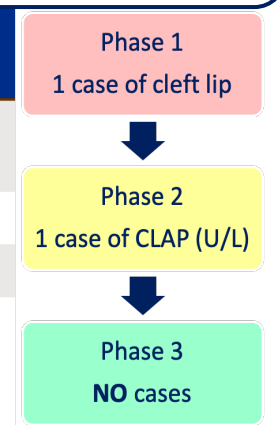


Fig 2 : Missed Cases (Postnatal)

Conclusions:

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

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