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# ROLE OF SECOND TRIMESTER ULTRASOUND ISOLATED SOFT MARKERS IN THE ERA OF FETAL CELL-FREE DNA

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**Objectives:** This study had two aims. First, to evaluate the prevalence of each second trimester soft marker after a first trimester screening based on combined screening and fetal cell-free DNA (cfDNA). Second, to assess the predictive value of second trimester soft markers for the detection of fetal aneuploidy after first-trimester screening with contingent use of cfDNA.

## Methods:

- This was a retrospective descriptive study including 4632 women with a singleton pregnancy who underwent first-trimester screening at our Prenatal Diagnostic Unit between January 2019 and December 2021.
- First-trimester screening was carried out according to the Pregnancy Follow-up Guideline in Catalunya, and results were classified as: low-risk (no other testing is offered), intermediate-risk (cfDNA is offered), high-risk (invasive technique or cfDNA is offered).
- The results of first- trimester prenatal screening and result of the second trimester ultrasound were reviewed.
- All second-trimester soft markers are routinely examined at the time of the anatomy scan, and first-trimester risk is reassessed if those markers are detected.
- In this study, results of the first-trimester screening were compared with the second-trimester risk-reassessments based on the finding of isolated Soft Markers.
- Variations in the risk level were evaluated, as well as the indication or not to perform additional fetal studies (invasive techniques), and also the genetic results and perinatal results of all pregnant women with isolated "Soft Markers".

## Results:

- At the time of first-trimester combined screening, a total of 84.2% women (n=3899) were classified as low risk, 11% (n=508) as intermediate risk and 4.9% (n=225) as high risk.
- A total of 4335 women (94%) underwent second trimester ultrasound in our center and, of these, 232 (5,3%) had an isolated soft marker.

| Recalculation of risk with 2nT sort markers n=232 | | First trimester screening | | |
|---|---|---|---|---|
| | | Low risk N=213 | Intermediate risk N=11 | High risk N=8 |
| Low risk | 201 (86.6) | 193 (90.6) | 5 (45.4) | 3 (37.5) |
| Intermediate risk | 27 (11.6) | 15 (7) | 2 (18.2) | 0 (0) |
| High risk | 4 (1.7) | 5 (2.3) | 4 (36.4) | 5 (62.5) |

*Figure. Recalculation of risk when taking into account the second trimester soft markers*

- Among the patients who had a soft marker detected in the second trimester ultrasound no aneuploidies were detected after invasive techniques or cfDNA testing on patients who were at increased risk because of soft marker finding.

**Conclusions:** In this study, the isolated finding of a second-trimester soft marker with aneuploidy risk-reassessment and subsequent prenatal testing did not detect any additional anomalies, after a first-trimester protocol including both invasive and non-invasive testing. Our results suggest that, in that context, the finding of isolated second-trimester soft markers is not likely to improve aneuploidy detection.