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First trimester screening based on ultrasound and cfDNA vs. first-trimester combined screening - a randomized controlled study

Kagan KO, Sroka F, Sonek J, Abele H, Lüthgens K, Schmid M, Wagner P, Brucker S, Wallwiener D, Hoopmann M. Ultrasound Obstet Gynecol. 2017 Sep 19. doi: 10.1002/uog.18905.

Summary and Key Points

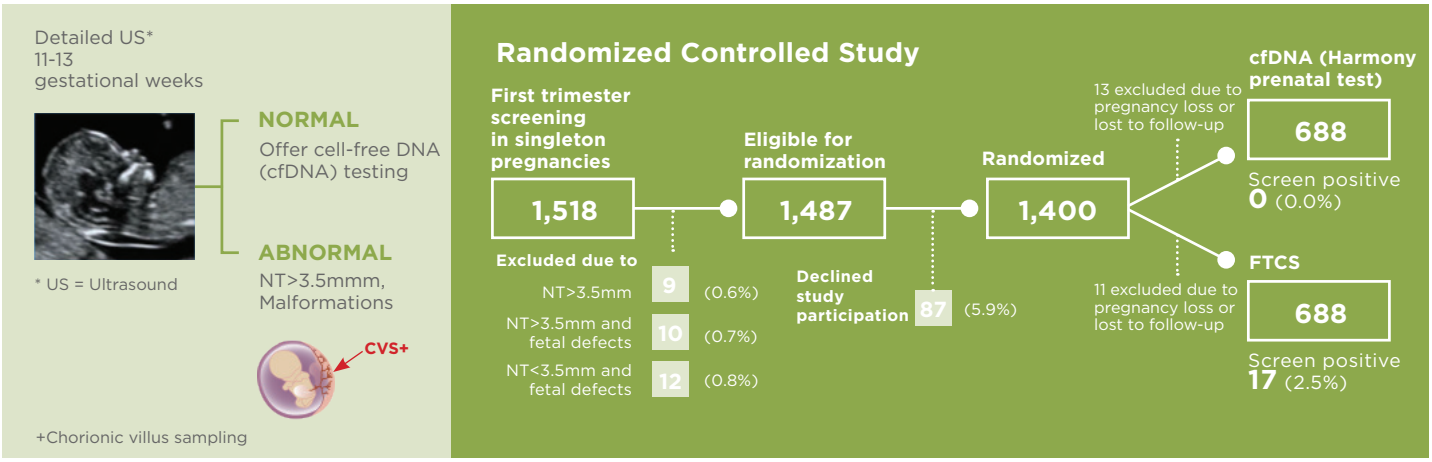
Purpose: To compare the false positive rates of First Trimester Combined Screening (FTCS) against a combination of ultrasound examination with cfDNA (Harmony prenatal test) analysis

Results: cfDNA analysis using the Harmony prenatal test in combination with first trimester ultrasound examination led to significantly lower false positive rates for trisomy 21 as compared to FTCS.

Study Population

1,400 singleton pregnancies with normal first trimester ultrasound were randomized into two groups: FTCS or cfDNA screening.

FTCS includes: maternal and gestational age, fetal nuchal transluscency (NT), maternal serum pregnancy-associated plasma protein A (PAPP-A) and free beta human chorionic gonadotropin (hCG). First trimester ultrasound protocol followed ISUOG recommendations. Pregnancies with fetal defects and/or increased nuchal translucency noted during ultrasound were excluded from randomization and counseled regarding follow-up testing options. Only pregnancies with complete outcome information were included in the study results.Median maternal age: 33.9 years; Median gestational age: 12.7 weeks.



Conclusion/Discussion:

No false positives were seen in the group receiving cfDNA screening; 2.5% of cases in the FTCS group were false positives.

Authors discuss the superior detetection of cfDNA screening for Down syndrome as compared to FTCS and contingent screening models.

Authors suggest implementation of a primary screening approach using cfDNA analysis and first trimester ultrasound. Benefits of this approach:

- Excellent detection rate of rare and common trisomies as well as fetal structural abnormalities
- Low false positives leading to reduction in unnecessary anxiety and follow-up testing
- Less complicated protocol than the contingent screening model, with almost all patients getting clear results from the first blood draw

Full article: **http://onlinelibrary.wiley.com/doi/10.1002/uog.18905/pdf**