



Uptake and Clinical Utility of prenatal cell-free DNA at Early Gestational Age in the REI Setting

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PURPOSE & OBJECTIVES

- In November of 2024, a whole genome sequencing based prenatal cell-free DNA (pcfDNA) aneuploidy screening assay utilizing fetal fraction (FF) amplification became clinically available at an early gestational age (EGA=8w0d – 9w6d gestation).
- Traditionally, Reproductive Endocrinology and Infertility (REI) clinicians do not order pcfDNA because they often transfer patient care at 8-9 weeks gestation.
- Prior to the launch of EGA, pcfDNA was not readily available before that transfer window.
- To better understand pcfDNA use at EGA in the REI setting, we characterized the clinicians ordering pcfDNA at EGA and identified those most likely to shift testing from >10 weeks gestation to EGA.

METHODS

- Retrospective analysis was performed on pcfDNA screening orders (Prequel, Myriad Genetics) between 12/2024-3/2026 in patients aged ≥18 years and compared to orders received in the prior 16 months.
- Ordering clinician types examined:
 - Obstetrics & Gynecology (Ob/Gyn)
 - Family Medicine
 - Primary Care
 - Maternal Fetal Medicine (MFM)
 - Genetics
 - REI
- Total ordering volume of pcfDNA at EGA and the ratio of pcfDNA screening ordered at 10+ weeks vs at EGA were compared across clinician types.
- Select REI clinicians were contacted for their viewpoint on pcfDNA screening in the REI population.

RESULTS

- Ob/Gyns had the highest volume of pcfDNA screens at EGA, making up ~90% of all EGA screens ordered (Figure 1).
- Although REI clinicians accounted for a very small proportion of pcfDNA orders both prior to and after EGA launch (0.19% and 0.88%), they demonstrated the largest increase in proportion of EGA orders (>4- fold) (Figure 1).
- REI clinicians also had the highest percentage of pcfDNA screens ordered at EGA at 65.1% (Figure 2).
- A subset of REI clinicians were interviewed about their utilization of pcfDNA screening at EGA (see grey boxes)

Figure 1: Percentage of total pcfDNA ordered at EGA by clinician type

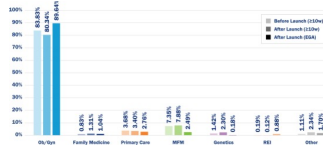
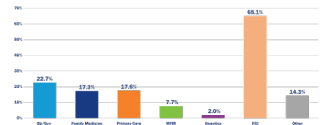


Figure 2: Percentage of pcfDNA screens ordered at EGA by clinician type.



“Offering pcfDNA at ≤10 weeks allows earlier follow-up, including timely referral to MFM for diagnostic testing or more detailed ultrasound surveillance.”

CONCLUSIONS

- This study provides a preliminary analysis of ordering patterns of pcfDNA at EGA, especially in the REI space.
- Surveyed clinicians conveyed the benefit of ordering pcfDNA screening in the REI space.
- The ordering patterns observed here, together with initial clinician feedback, highlight the need for further research to define the clinical value of EGA and pcfDNA in REI and their impact on clinical workflows, diagnostic decision-making, and patient experience.

“Working in an REI clinic puts us in a unique position—we often know the exact timing of conception and can identify pregnancies earlier than in spontaneous cases.... our patients typically transition out of care around 10 weeks’ gestation; we aim to order pcfDNA screening at the earliest appropriate time.”

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