A Noninvasive Prenatal Screen with >4% Fetal Fraction in All Samples: Clinical Laboratory Experience

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Objectives:

1. Describe fetal fraction amplification
2. Discuss the impact of fetal fraction amplification on fetal fraction levels and how this can impact test failure rates in noninvasive prenatal screening
3. Describe case examples in which fetal fraction amplification allowed for confident noninvasive prenatal screen results delivery rather than test failure

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Purpose

- To assess a whole-genome sequencing (WGS)-based Noninvasive Prenatal Screening (NIPS) that employs FF amplification (FFA) technology for all samples

Methodology

- We retrospectively analyzed results from 19,464 patients who underwent NIPS with FFA during a two-month period
- The FFA technology increases fetal fraction (FF) by preferentially sequencing short cell-free DNA (cfDNA) fragments, known to be enriched for fetal-derived cfDNA
- BMI data were available for 12,687 patients
- Several samples in the dataset included redraws from patients that had received a test failure from another laboratory

Results

- Median maternal age was 31 years and median gestational age was 12 wks
- No patients had FF results <4%, regardless of high BMI or early gestational age
- Ninety-nine percent of patients had FF >8.1%
- Patients who did not receive results from other labs due to low FF received confident results with FFA technology (Table)

	Gestational age	BMI	Other Lab result (FF)	FF with FFA	NIPS with FFA result
Patient A	10 wks	25	Failed (2%)	20%	Negative
Patient B	10 wks	39	Failed (3%)	9%	Negative
Patient C	11 wks	>40	Failed (3%)	12%	Negative
Patient D	12 wks	45	Failed (2%)	9%	Negative
Patient E	13 wks	25	Failed (3%)	6%	Positive (confirmed via amnio)

Conclusion

- FFA prevents unnecessary test failures, providing confident results to all patients regardless of high BMI or early gestational age