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pcfDNA Screening with Fetal Fraction Amplification Between 8-10 Weeks: Laboratory Experience with >28k Samples

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Disclosures

All authors were employees of Myriad Genetics, Inc. at the time of this study and received salary and stock as compensation

Objective

- Explain benefits of fetal fraction amplification (FFA) in prenatal cell-free DNA (pcfDNA) screening
- Review early laboratory experience with pcfDNA screening with FFA between 8w0d and 9w6d gestation

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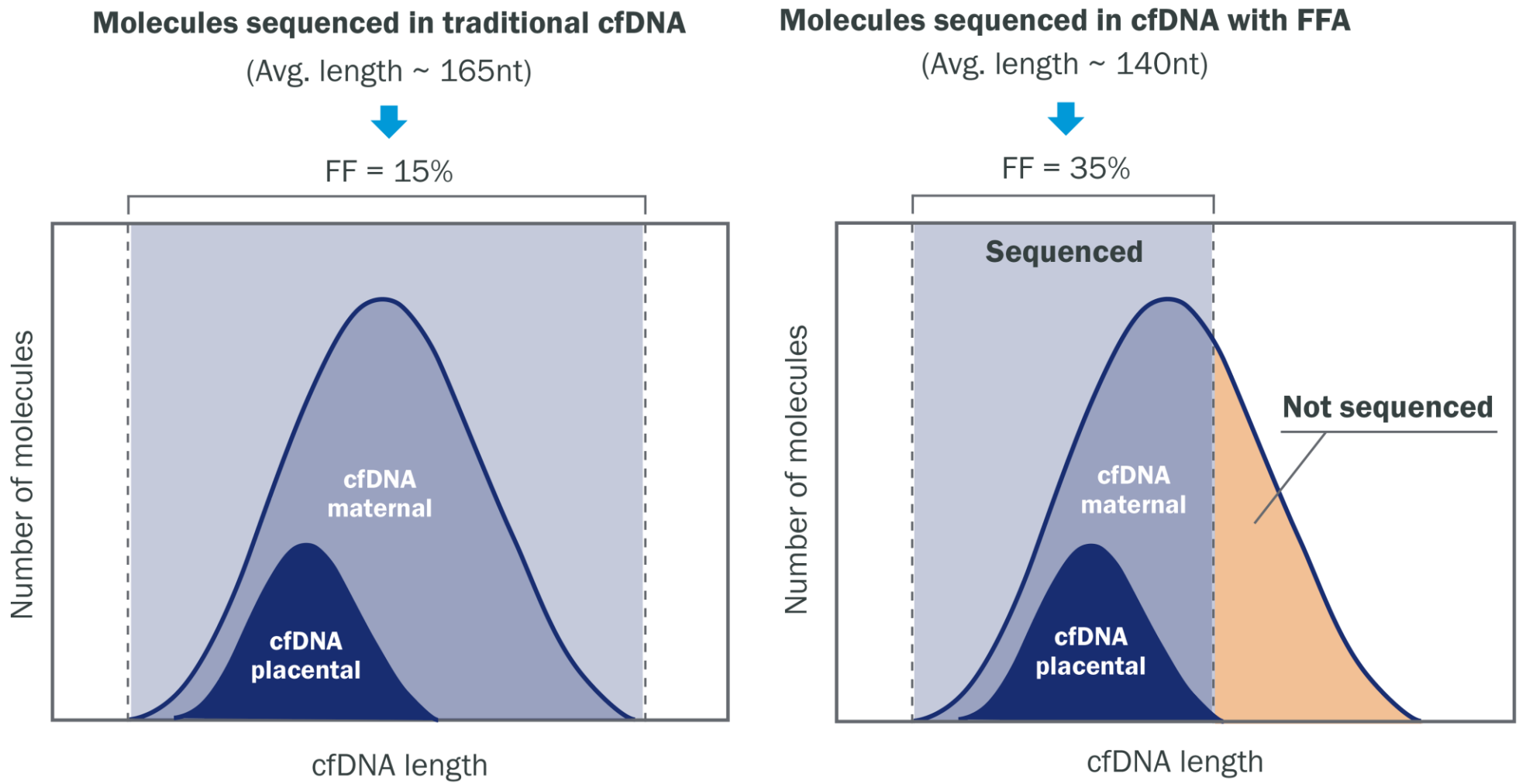
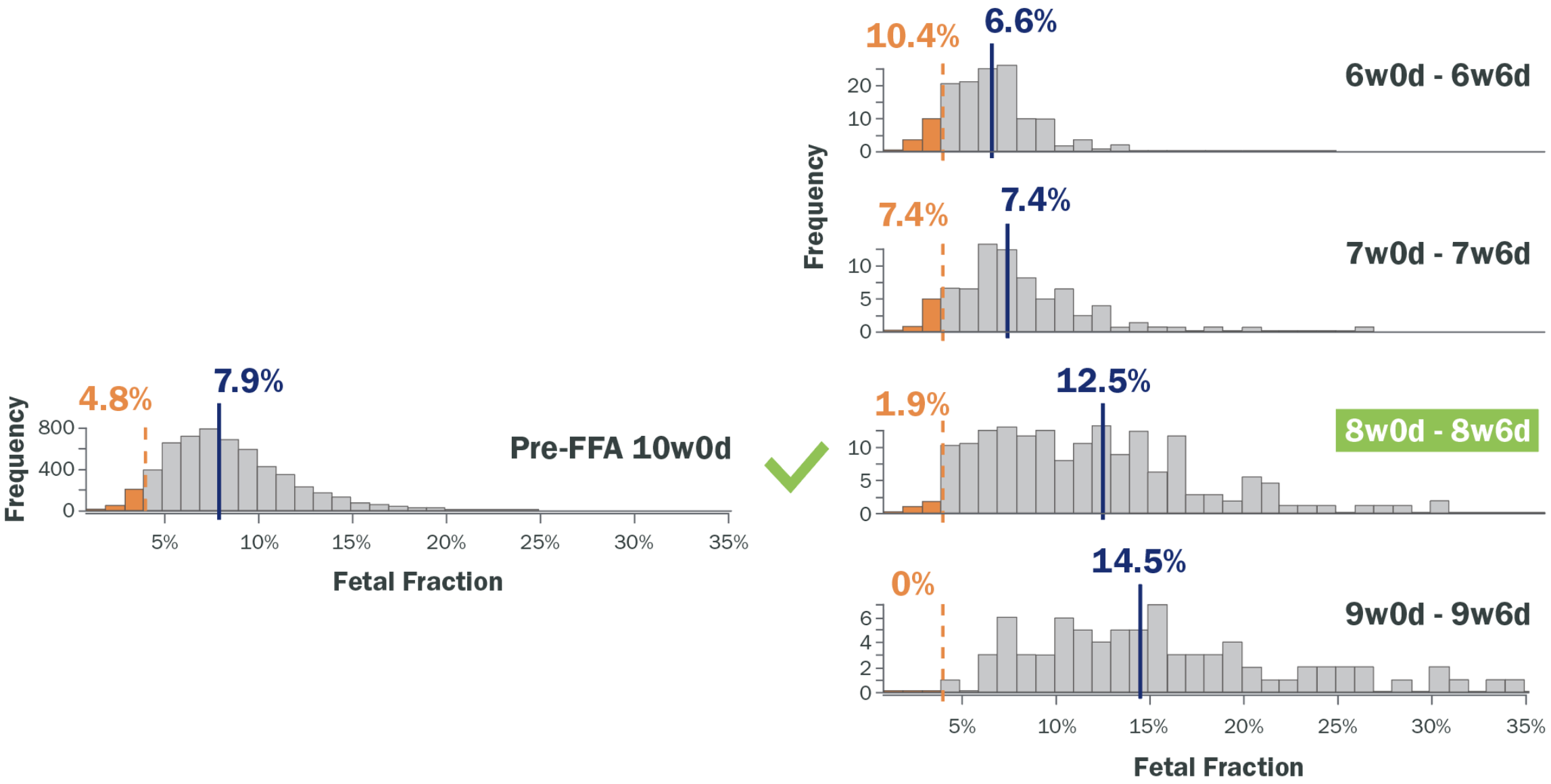
Dugoff L.D., Mabey B., Bordwell J. et al. Evaluation of prenatal cell-free DNA screening with fetal fraction amplification at 6-9 weeks gestation (in prep)

AMPLIFY™ technology increases fetal fraction by leveraging differences in cfDNA length



Welker N.C., Lee A.K., Kiolby R.A.S. et al. High-throughput fetal fraction amplification increases analytical performance of noninvasive prenatal screening. Genet Med (2020). https://doi.org/10.1038/s41436-020-01009-5

Background: Early Gestational Age (EGA) Validation



Dugoff L.D., et al. Manuscript under review.

Methods

- pcfDNA screens ordered between 11/19/2024 and 07/21/2025 considered for analysis
- Number of orders received, screen-positive results, and screen failures calculated
- Fetal fraction (FF) medians and first and third quartiles (Q1, Q3) for the 8- and 9-week gestational age period calculated
- BMI information analyzed when provided

Dugoff L.D., et al. Manuscript under review.

Results of weeks 8 and 9

- 28,446 orders were placed for patients between 8w0d and 9w6d

Result type	702 screen positive reports (2.47% screen positive rate)
Aneuploidies	
Trisomy 21	**140** (19.94%)
Trisomy 13	**48** (6.84%)
Trisomy 18	**39** (5.55%)
Sex chromosome aneuploidy (SCA)	**234** (33.33%)
Rare autosomal aneuploidy (RAA)	**292** (41.60%)
Microdeletions	
1p36	**8** (1.14%)
22q11.2	**4** (0.57%)
15q11.2	**3** (0.43%)
5p	**4** (0.57%)
4p	**2** (0.29%)

*25 were screen positive for two or more abnormalities

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Screen failures

Reason for failure	**61 total no call reports** (0.2% screen failure rate)
Insufficient FF	**19 Samples** (0.06%)
Other QC metrics	**42 Samples** (0.14%)

Week	Rate
Week 8	**37 Samples** (0.13%)
Week 9	**24 Samples** (0.08%)

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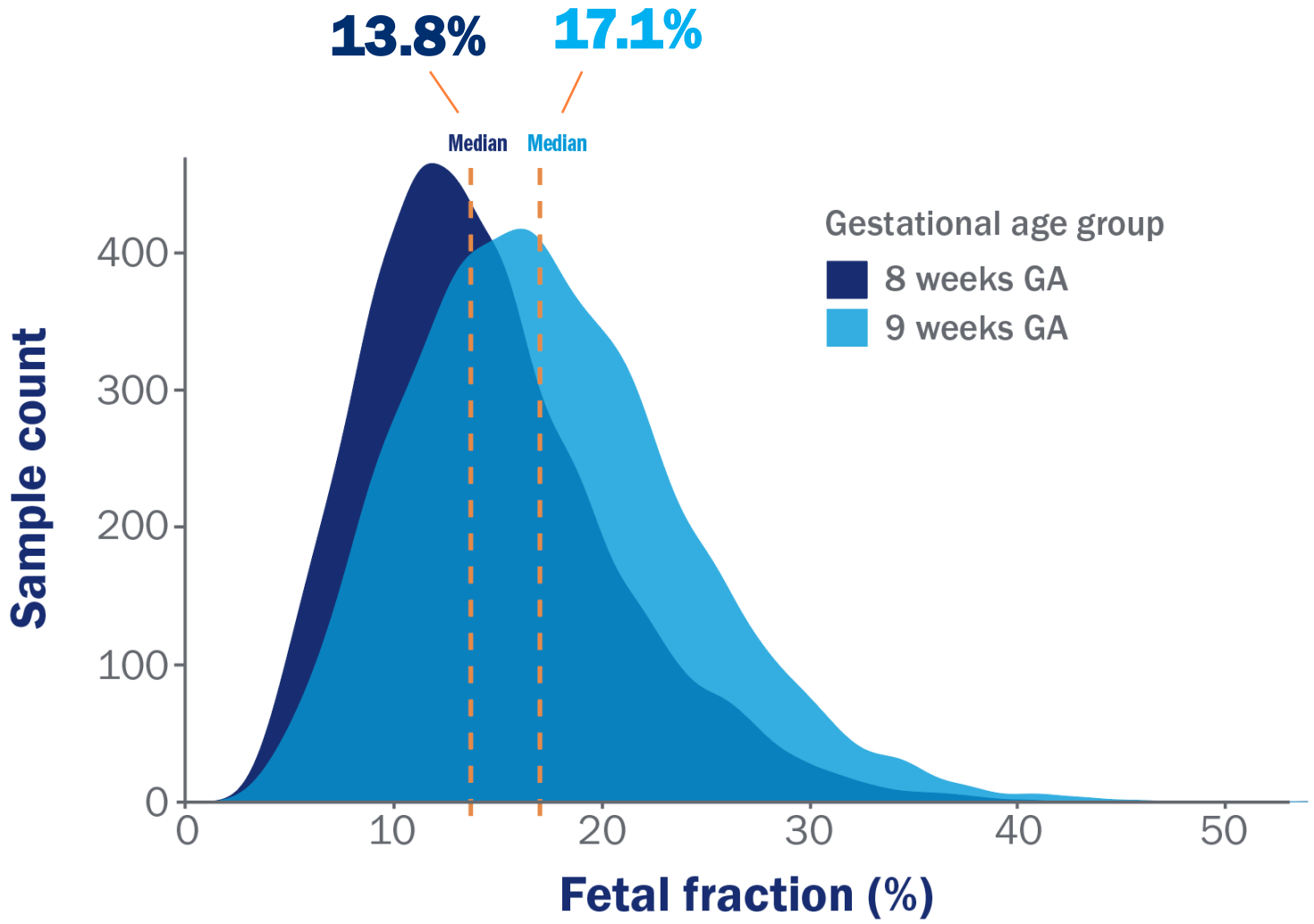
Median FFs

- **Across the cohort:**
  15.4% (Q1=11.4%; Q3=20.2%)
- **8th week:**
  13.8% (Q1=10.3%; Q3=18.2%)
- **9th week:**
  17.1% (Q1=12.9%; Q3=22.0%)



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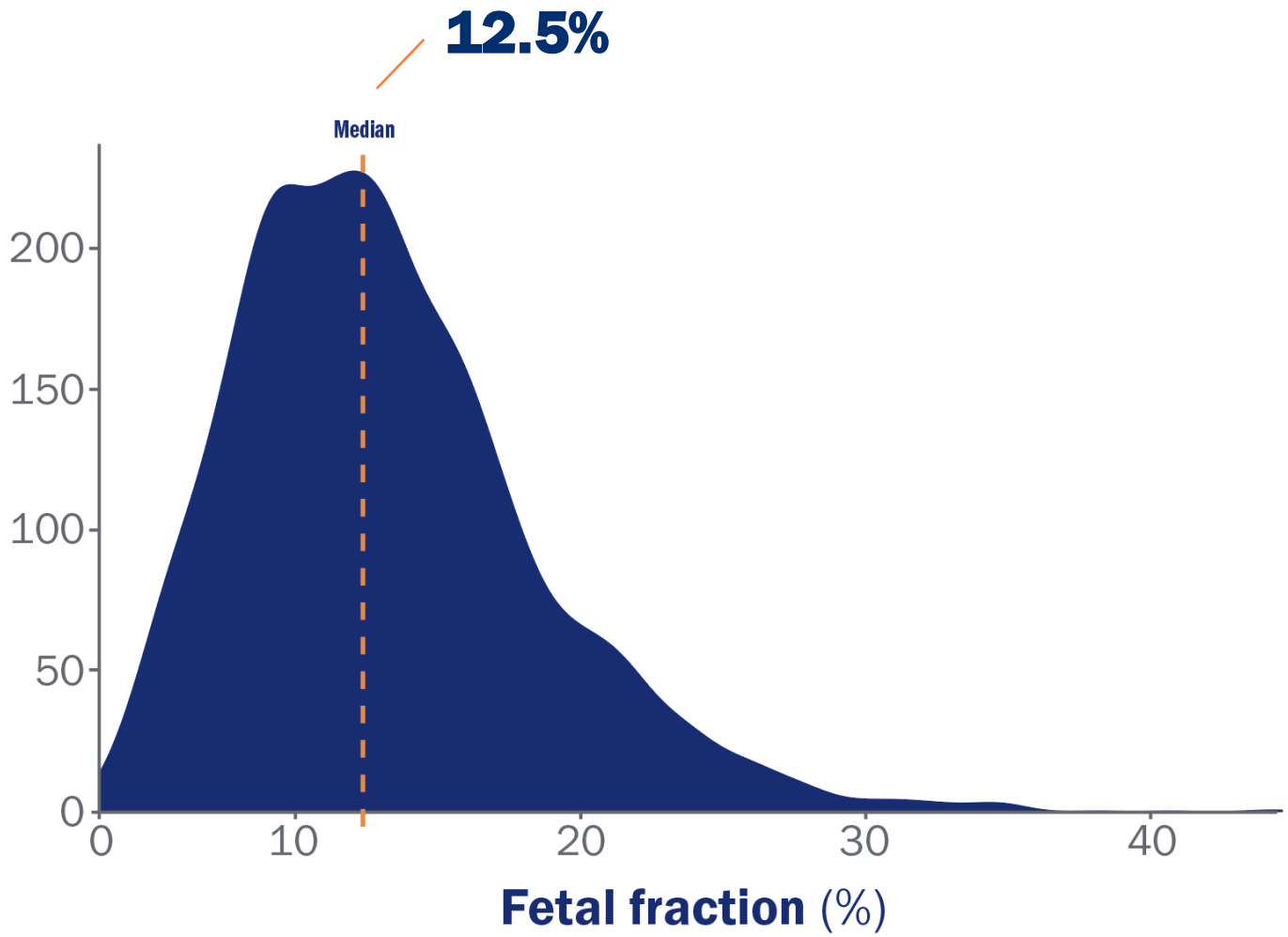
Fetal Fraction distribution at high BMI

- BMI ≥30: 35.5% (n = 6,246)
- Median FF 12.5%

  (Q1=9.3%; Q3=16.1%)



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Clinical Utility

- Screening at earlier gestational ages facilitates genetic counseling and pregnancy management, such as earlier diagnostic testing

- pcfDNA screening at 8 weeks aligns with initial prenatal visits that commonly occur at approximately 8 weeks gestation, avoiding the need for a later blood draw

- Earlier screening has the potential to reduce anxiety and uncertainly earlier in pregnancy

Conclusions

- Laboratory experience with pcfDNA screening for pregnancies between 8w0d-9w6d weeks gestation:
  - Demonstrated sufficient FF for analysis
  - Maintained a low screen-failure rate across a broad population, including patients with high BMI
- Screening for fetal aneuploidy at an earlier gestational age allows patients and providers to gain clinical insights earlier, leading to earlier decision making

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Thank you

Questions?

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