False positive NIPS results: Do fibroids play a role?

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INTRODUCTION

- Noninvasive prenatal screening (NIPS) evaluates cell-free fetal DNA (cfDNA) from the maternal circulation[1]
- NIPS has a high sensitivity and specificity for trisomy 21, trisomy 18, trisomy 13, and sex chromosome aneuploidies[1]
- Discrepancies between fetal karyotype and NIPS results can be caused by a vanishing twin, demise of a twin, low fetal fraction, maternal chromosome abnormality and maternal disease[2,3,]
- It is estimated that the prevalence of fibroids, benign uterine tumors, varies by ethnicity and affects 1:10.4 people[4]
- Previous studies have attributed high risk NIPS results to maternal fibroids[5,6]

AIM

- Assess whether the presence of maternal fibroids has an influence on high-risk noninvasive prenatal screening (NIPS) results

METHODS

- A retrospective cohort study was conducted to compare the accuracy of the NIPS results to the presence of maternal fibroids
- Inclusion criteria were singleton pregnancies with high-risk NIPS results, and accessible pregnancy outcome information
- 11,880 prenatal patients received NIPS during the study period. 171 were identified as high risk via NIPS
- Total fibroid volume was calculated based on early gestational age sonograms
- NIPS results were then compared to diagnostic testing results, ultrasound findings, or postnatal diagnostic testing and physical exam

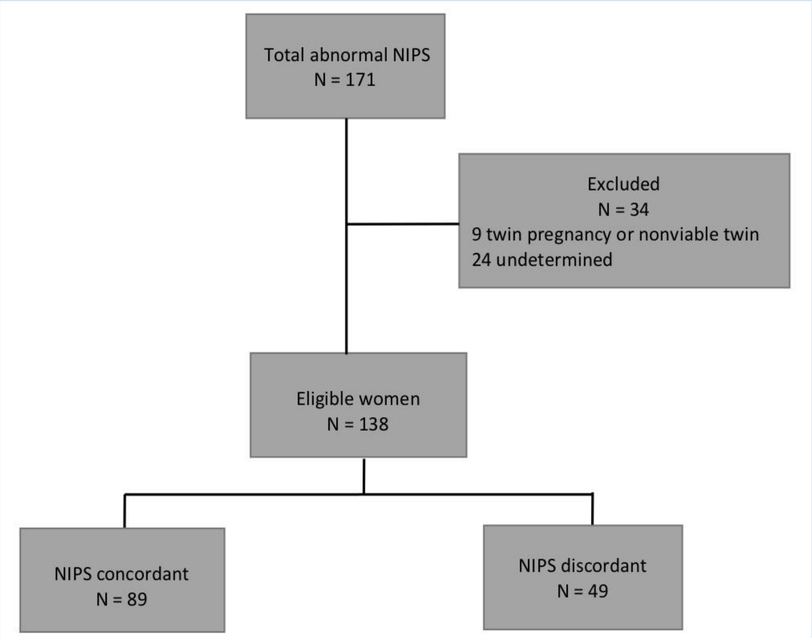

Figure 1: NIPS (noninvasive prenatal screening) study design

Characteristics	True positive NIPS N = 89	False positive NIPS N = 49	Total	p-value
Presence of fibroid n, (%)	8 (9.0%)	7 (14.3%)	15 (10.9%)	0.339
Total fibroid volume mean (SD)	8.3 (46.5)	25.7 (116.6)	14.4 (78.9)	0.216
GD/Diabetes	9 (10.1%)	8 (16.3%)	17 (12.3%)	0.288
HTN/pre-eclampsia	13 (14.6%)	8 (16.3%)	21 (15.2%)	0.788
Consaguinity	2 (2.2%)	1 (2.0%)	3 (2.2%)	1

Table 1: Medical history of women with true positive NIPS compared to women with false positive NIPS

RESULTS

- Between March 2014 and August 2019, 11,880 prenatal patients had NIPS during prenatal care at Montefiore Medical Center. Of those, 171 patients had high risk results and 138 were eligible for the study
- A total of 35.5% (49/138) of all high-risk NIPS results were false positive and those patients ultimately had chromosomally normal babies.
- The mean total fibroid volume in the concordant NIPS group was 8.3 cm^3 versus 25.7cm^3 total fibroid volume in the discordant NIPS group ($p = 0.216$)
- The odds of a false positive NIPS result among patients with fibroids was 3.486 (95% CI: 0.846, 14.354) times those without fibroids, adjusting for fetal fraction, maternal age, gestational age, BMI and race

CONCLUSIONS

- The presence of fibroids was associated with a higher rate of false positive NIPS results
- Additional studies are necessary to determine the relationship of presence and size of fibroids on the accuracy of NIPS results

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