P. 637: Single center experience using single-gene-NIPT to evaluate fetal risk of autosomal recessive conditions

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Background

- Reflex single gene NIPT (sgNIPT) can determine fetal risk of cystic fibrosis (CF), spinal muscular atrophy (SMA), and hemoglobinopathies when maternal carrier status is detected.
- This novel method of fetal risk assessment for common autosomal recessive conditions performs sequencing of placental DNA in maternal blood to provide a low or high fetal risk assessment.
- However, sgNIPT is a screen, and all parents are offered follow up carrier assessment of the other parent and amniocentesis for diagnosis.

Objective

- The objective of this study is to evaluate uptake of partner screening and diagnostic testing with CVS or amniocentesis when reflex sgNIPT is positive for maternal carrier status and placental DNA suggests there is a high fetal risk of an autosomal recessive condition.

Methods

- Retrospective cohort study of patients undergoing carrier screening with sgNIPT in a single tertiary care center from March 2020 to July 2022.
- All patients received pre- and post-test counseling by a certified prenatal genetic counselor.
- All patients were offered diagnostic testing to follow up on screening results.
- Data were abstracted from an electronic medical record, including:
  - maternal demographic characteristics
  - results of sgNIPT screening
  - results of diagnostic testing (chorionic villus sampling or amniocentesis) if performed
  - results of partner carrier screening if performed
- Data were analyzed using descriptive statistics.

Results

- 229 patients had sgNIPT during the study period.
- Maternal demographic characteristics (Table 1).
- Median gestational age of test performance was 90 days or 12 weeks 6 days IQR [79,106 days].
- All pregnancies were singleton gestations.

Results

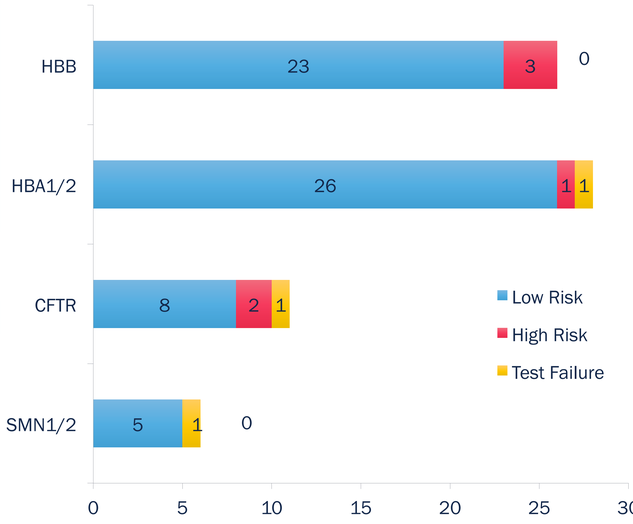
Table 1. Cohort Demographic Characteristics

Characteristic	n=229
Maternal Race/ethnicity	69 (36)
Caucasian	65 (34)
African or African American	35 (18)
Hispanic/Latine	7 (4)
Asian	14 (7)
Other	39 (17)
Not reported	
Gestational age (days) (median, IQR)	90 (79,106)
Singleton	229 (100)
Weight kg (mean, SD)	84.3 (24.6)

*Data shown as n(%) unless noted otherwise

- Sixty-two (27%) individuals were identified as carriers of at least one of the tested conditions.
- Nine were carriers of more than one condition.
- Reflex to sgNIPT was performed 71 times.
- Of those, 94% provided a fetal risk assessment.
- Low and high risk results shown in Figure 1.
- Of the cohort, five individuals (8.1%) elected to have partner screening.
- Fifty-six individuals had low risk fetal results; 3 (5%) had partner carrier screening and 0 had diagnostic testing.
- Six individuals had high-risk fetal results: 2 had partner carrier screening and 1 had diagnostic testing.

Figure 1. Fetal risk after reflex sgNIPT for AR conditions by gene of interest



Conclusions

- Few patients who utilized sgNIPT completed follow up paternal screening or fetal diagnostic testing despite pre- and postnatal genetic counseling and even in setting of a NIPT result that showed a high risk of an affected fetus.
- Additional work to assess patient understanding of screening test results and how parental knowledge of genetics and attitudes towards genetic testing is needed.