Effectiveness of Prenatal Screening Tests on Predicting Cardiac Abnormalities

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Introduction

Congenital heart disease (CHD):
- Most common birth anomaly - 28% of major congenital anomalies[1]
- Infant mortality rate of 30-50%[2]
- Current prenatal screening tests detect chromosomal anomalies - trisomy 13, 18, and 21
- Minimal knowledge on screening's detection for CHD

Screenings:

Anatomy Ultrasound (US)
- 18-20th week
- Sensitivity (44.0%) vs. Specificity (99.9%)[3]

Nuchal Translucency (NT) Screening
- 11-14th week
- Measure thickness of fluid collection behind fetus' neck
- Sensitivity (44.4%) vs. Specificity (94.5%)[4]

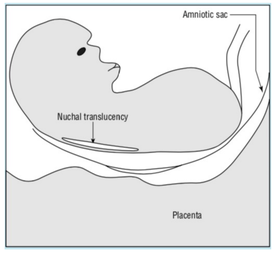

Figure 1: Depiction of fetus showing measurement of fetal nuchal translucency thickness[5]

Cell-free DNA (cfDNA) Test
- Optimal detection at 10th week
- Maternal and placental DNA and miRNA biomarkers in maternal blood[6]
- Not previously studied for CHD detection, thus lacks data on its accuracy for detecting cardiac defects

Goals of this research:
- Compare accuracy of three prenatal screening tests in detecting a cardiac defect
- Provide obstetricians with better knowledge of whether CHD can be detected earlier than the standard 18-20th week anomaly ultrasound
- Earlier prenatal diagnosis can decrease postnatal morbidity and mortality

Hypothesis: cfDNA has an increased accuracy in detecting CHD in earlier gestation, followed by NT screening then the anomaly ultrasound.

Aims and Objectives

- Identify and compare the specificity and sensitivity of the three prenatal screening tests - anatomy ultrasound, NT screening, and cfDNA test - in identifying an increased risk of fetal cardiac anomalies

Methods

- Retrospective chart review
- Convenience sample
- Infants with chromosomal abnormalities were excluded because CHD are usually associated with chromosomal abnormalities

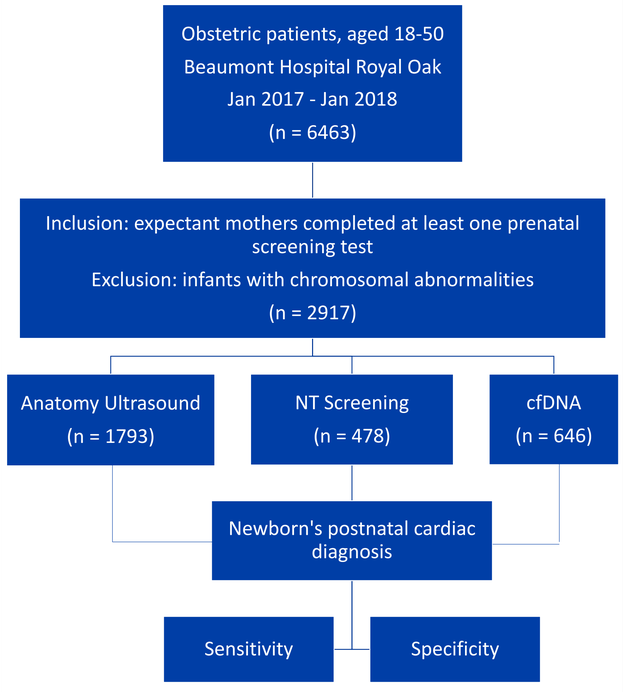

Figure 2: Flow diagram of included participants and research methodology

Results

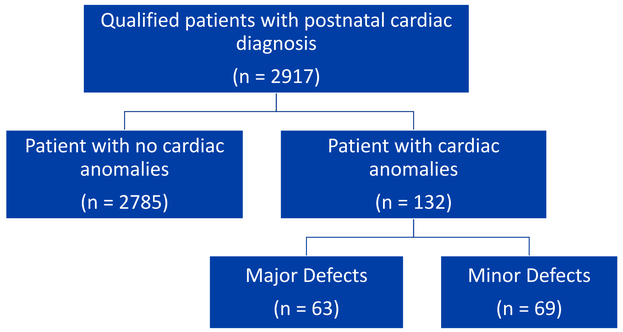

Figure 3: Participants screened with major and minor cardiac defects

Table 1: Major and Minor Cardiac Defects

Major Cardiac Defects	Minor Cardiac Defect
• Continuous follow-up or surgery • Atrial septal defect • Ventricular septal defect • Vascular ring	• Do not affect quality of life • Resolves within a year without intervention • Functional murmur • Small patent foramen ovale • Patent ductus arteriosus

- Sensitivity and specificity are calculated with a 95% confidence interval and a p-value < 0.05
- Accuracy of all three screenings were statistically significant
- Anatomy ultrasound has the highest sensitivity at 20.0% and highest specificity at 99.7%, followed by NT then cfDNA (Table 2 and 3)

Table 2: Sensitivity of Prenatal Screening Tests

Prenatal Screening Tests	Sensitivity	95% Confidence Interval
Anatomy Ultrasound	20.0%	7.7% - 38.6%
NT Screening	18.8%	4.0% - 45.7%
cfDNA Test	5.6%	0.1% - 27.3%

Table 3: Specificity of Prenatal Screening Tests

Prenatal Screening Tests	Specificity	95% Confidence Interval
Anatomy Ultrasound	99.7%	99.2% - 99.9%
NT Screening	98.0%	96.2% - 99.1%
cfDNA Test	97.4%	95.8% - 98.5%

Conclusions

- Anatomy US - most accurate at detecting CHD
- NT screening is comparable to anatomy ultrasound
- Small sample population - low sensitivity of ultrasound and NT screening
- Limitation: studies performed outside of Beaumont Hospitals

Research Impact:
- Congenital heart disease vs chromosomal anomalies
- Utilize earlier prenatal screening tests before standard 18-20th week ultrasound

Future Research:
- Multicenter data collection
- Combination of screening tests
  - Additional 20.2% of structural abnormalities detected with NT and cfDNA tests combined[7]

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