The Effect of Commercial Tests for Aneuploidy Screening Using Cell-free Fetal DNA on Rates of Invasive Testing in Clinical Practice

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ABSTRACT:

Objective: TTo determine the effect of new commercial tests for aneuploidy screening using cell free fetal DNA (cffDNA) on rates of invasive testing.

Study Design: Commercial aneuploidy screening tests using cffDNA were first offered at Lehigh Valley Health Network Maternal Fetal Medicine in late December 2011. All patients offered cffDNA testing were at high risk for fetal aneuploidy defined as advanced maternal age, abnormal aneuploidy screening, abnormal ultrasound findings and/or personal/family history. Data regarding cffDNA test rates, invasive testing rates (amniocentesis and CVS) and indication for testing were collected. Invasive testing indications were further categorized into testing for aneuploidy or for other indications. Chi square was used to compare rates of invasive testing before and after introduction of cffDNA testing.

Results: 365 patients underwent cffDNA testing from 12/1/2011 to 12/30/2012. The rates of invasive testing overall and invasive testing for aneuploidy both decreased significantly after introduction of cffDNA testing as compared to before (P<0.001). The difference in the number of invasive tests over time was due to a decrease in procedures for aneuploidy testing (Figure 1). The number of cffDNA tests increased significantly from the first half of 2012 to the second half of 2012 (P<0.001).

Conclusion: The new commercial tests available for aneuploidy screening using cffDNA were rapidly incorporated into clinical practice and are associated with decreased use of invasive tests.

INTRODUCTION:

In the past two decades, numerous strategies for the detection of fetal aneuploidy have been developed, although definitifve diagnosis is possible only through the use of invasive procedures. In the fall of 2011, non-invasive tests for the diagnosis of common aneuploidies first became commercially available for the diagnosis of common trisomies and sex chromosome abnormalities. These tests work by identifying cffDNA in maternal blood with a high degrees of sensitivity and specificity, and have the advantage of providing the patient with highly accurate information without the discomfort or risk of an invasive procedure. The purpose of this study is to characterize the use of cffDNA testing at our institution and to see how the introduction of these tests has affected the rate of invasive procedures performed over time.

METHODS:

- Retrospective cohort study of all women undergoing cffDNA testing and/or invasive testing for aneuploidy through Lehigh Valley Health Network Maternal Fetal Medicine practice from 12/1/2011 – 12/30/2012.
  - Consistent with ACOG guidelines, invasive testing for aneuploidy (amniocentesis and CVS) is available to all women.
  - CffDNA testing became available in late December 2011 in our practice and was offered only to women at high risk for aneuploidy based on maternal age, abnormal screening results, abnormal ultrasound findings, and/or family/personal history of aneuploidy.
  - All women underwent genetic counseling prior to invasive or cffDNA testing.
  - Women with positive cffDNA results were strongly encouraged to undergo invasive testing to confirm results.
- Women undergoing invasive procedures and cffDNA testing during the study period were identified by review of a MFM clinical and ultrasound databases. Only women undergoing invasive testing for aneuploidy detection were included in the analysis.
- Medical records were reviewed for baseline characteristics, indications for testing, test results and pregnancy outcomes. Neonatal records were reviewed when available.
- Descriptive statistics were generated for baseline characteristics and test indications. Chi square was used to compare the rate of change in invasive procedures and utilization of cffDNA over time.

RESULTS:

- 365 patients underwent cffDNA testing from 12/1/2011 to 12/30/2012.
- The number of cffDNA tests increased significantly from the first half of 2012 to the second half of 2012 (143 vs. 222, P<0.001).
- Rates of invasive testing for aneuploidy decreased significantly from 2011 (before cffDNA) to 2012 (after cffDNA), p<0.0001.

Table 1. Baseline Characteristics of Women Undergoing Cell-free Fetal DNA Testing*

Mean maternal age (years)	32.3±6.5
Advanced maternal age	52%
Ethnicity	
Caucasian	72%
Black	7%
Asian/Indian	6%
American Indian	2%
Arabic	1%
Mixed	2%
Unknown	11%
Non-hispanic	83%
Multiparous	64%
Married	58%
Private obstetrician	77%
Private insurance	51%
Singleton gestation	95%
High school graduate	89%
College graduate	42%
Median gestational age (weeks)	18.64 (10.29 – 28.43)
Trimester	
First	25%
Second	71%
Third	4%

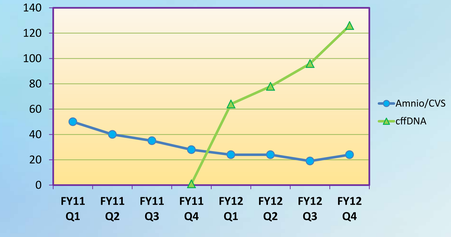
* Not all characteristics available for all subjects
Data in n (%), mean +SD or median (range). GA, gestational age.

Table 2. Primary Indications for Cell-free Fetal DNA Testing

AMA only	18%
Abnormal screen only	29%
Abnormal ultrasound only	37%
Abnormal screen and abnormal ultrasound	13%
History indicated	2%
Unspecified	1%

Data are in n (%).

Figure 1. Volume of invasive testing for aneuploidy and cell-free fetal DNA diagnostic tests for first year cell-free fetal DNA testing was offered



CONCLUSIONS:

Our study provides some information on the use of new commercial tests for aneuploidy screening using cffDNA in clinical practice in a non-research setting. The new commercial tests available for aneuploidy screening using cffDNA were rapidly incorporated into clinical practice and are associated with decreased use of invasive testing for aneuploidy.