Prenatal Screening for Aneuploidy: Should cfDNA Replace Traditional Methods?

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Prenatal Screening for Aneuploidy: Should cfDNA replace traditional methods?

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Abstract
In 2011, advances in research in medical genetics led to the advent of prenatal cell-free DNA (cfDNA). This screen consists of analysis of placental DNA circulating in maternal blood. cfDNA has had a major impact on prenatal screening for aneuploidy. cfDNA allows for analysis of higher risk pregnancies and is less invasive than amniocentesis. This screen is used for high risk populations and in low-risk populations it is not as reliable. The purpose of this paper is to compare and contrast cfDNA to other screening methods for fetal aneuploidy.

Research Question
Why should prenatal screening be offered? What are the current options for screening for fetal aneuploidy? Should NIPS replace standard screening for aneuploidy?

Literature Review
Why should prenatal screening be offered?
It is well documented that the risk of carrying a fetus with a chromosome alteration increases with maternal age. 

Introduction
Screening for chromosome abnormalities prenatally is a complex topic and should be employed as an essential component of comprehensive obstetrical care. Various screening options are available to screen for the most common fetal aneuploidies, namely Trisomy 21 (Down syndrome), Trisomy 18, and Trisomy 13. Each screening tool has advantages and disadvantages, and informed decision making between patient and provider is essential for selection of which screen suits each patient.

Table 1: Risk of Chromosomal Abnormalities Based on Maternal Age at Test
<table>
<thead>
<tr>
<th>Age at Test</th>
<th>Risk of Trisomy 21 (%)</th>
<th>Risk of Any Chromosomal Abnormality (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>22-23</td>
<td>1.16-1.64</td>
<td>1.08-1.43</td>
</tr>
<tr>
<td>24-25</td>
<td>1.17-1.63</td>
<td>1.08-1.44</td>
</tr>
<tr>
<td>26-27</td>
<td>1.17-1.65</td>
<td>1.08-1.44</td>
</tr>
<tr>
<td>28-29</td>
<td>1.17-1.69</td>
<td>1.08-1.43</td>
</tr>
<tr>
<td>30-31</td>
<td>1.17-1.68</td>
<td>1.08-1.44</td>
</tr>
<tr>
<td>32-33</td>
<td>1.17-1.69</td>
<td>1.08-1.44</td>
</tr>
<tr>
<td>34-35</td>
<td>1.17-1.69</td>
<td>1.08-1.45</td>
</tr>
<tr>
<td>36-37</td>
<td>1.17-1.71</td>
<td>1.08-1.45</td>
</tr>
<tr>
<td>38-39</td>
<td>1.17-1.73</td>
<td>1.08-1.45</td>
</tr>
</tbody>
</table>

The traditional methods of screening will be compared to NIPS including gestational age at which these screens may be performed, sensitivity and specificity for Trisomy 21, 18, and 13, and for which populations these screens are most appropriate.

Statement of the Problem
With an increase in the amount of options for screening for fetal aneuploidy, awareness and understanding of the disadvantages and advantages of each screening tool is necessary in providing comprehensive obstetrical care. Patients must first be counseled on the existence and possibility of fetal chromosome abnormalities; and informed decision making between patient and provider should ensue regarding which, if any, screening tool best suits the patient.

Table 2: Characteristics, Advantages, and Disadvantages of DifferentScreening Tests for Aneuploidy

<table>
<thead>
<tr>
<th>Screening Test</th>
<th>Characteristics</th>
<th>Advantages</th>
<th>Disadvantages</th>
</tr>
</thead>
<tbody>
<tr>
<td>First trimester screening</td>
<td>Maternal serum hCG and PAPP-A + nuchal translucency ultrasound at 11 to 14 weeks</td>
<td>High detection rates for the most common aneuploidies, Can be performed any time after 10 weeks of gestation</td>
<td>Only one blood draw needed, Mixed data on PPV in non high-risk populations, Unreliable with multiples</td>
</tr>
<tr>
<td>Second trimester screening</td>
<td>Quad screen analyzes four biochemical markers which include hCG levels, alpha-fetoprotein, unconjugated estradiol, and dimeric inhibin A.</td>
<td>High detection rates for the most common aneuploidies, Can be performed any time after 10 weeks of gestation, Only one blood draw needed</td>
<td>Mixed data on PPV in non high-risk populations, Unreliable with multiples</td>
</tr>
<tr>
<td>First trimester integrated/sequential screening</td>
<td>First trimester nuchal translucency study with ultrasound, hCG and PAPP-A levels, as well as a second trimester quad screen</td>
<td>High detection rates for the most common aneuploidies, Can be performed any time after 10 weeks of gestation</td>
<td>Only one blood draw needed, Mixed data on PPV in non high-risk populations, Unreliable with multiples</td>
</tr>
</tbody>
</table>

Advantages of NIPS:
• High detection rates for the most common aneuploidies
• Can be performed any time after 10 weeks of gestation
• Only one blood draw needed

Disadvantages of NIPS:
• Only detects Trisomy 21, 18, and 13
• Mixed data on PPV in non high-risk populations
• Unreliable with multiples

Discussion: Should cfDNA replace traditional methods?
According to a survey published by the Journal of Maternal-Fetal and Neonatal Medicine in September of 2016, obstetric care providers identified NIPS as clinically superior to other screening tools (Brewer, Demers, & Musci, 2016). The survey stated that 81.5% of respondents believed that NIPS is a superior test in screening for aneuploidy regardless of maternal age. The survey also stated that most respondents would like ACOG to formally recommend this screening tool to all pregnant women, regardless of age.

Applicability to Clinical Practice
The research as stated in this review is highly applicable to clinical practice due to the fact that offering prenatal screening for aneuploidy to all women is an important part of providing comprehensive obstetric care. Physicians Assistant in both the primary care setting and women’s health setting may be providing obstetric care to clients. Knowledge of not only the existence of chromosome alterations but how to screen for them is of utmost importance in providing quality care. ACOG recommends offering screening to every pregnant patient.

References

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