Aneuploidy screening protocol for high risk women

Candidates
- Maternal age ≥ 35 at EDD (singleton) or ≥ 32 at EDD (twins)
- Family history Down syndrome
- First or second trimester screen positive for Down syndrome
- Ultrasound finding suggestive of aneuploidy

NIPT test performance in a high risk population

<table>
<thead>
<tr>
<th></th>
<th>Detection rate</th>
<th>False positive rate</th>
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</thead>
<tbody>
<tr>
<td>Trisomy 21</td>
<td>99%</td>
<td>0.2%</td>
</tr>
<tr>
<td>Trisomy 18</td>
<td>99%</td>
<td>0.2%</td>
</tr>
<tr>
<td>Trisomy 13</td>
<td>79-92% (limited data)</td>
<td>1.0%</td>
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</tbody>
</table>

“Review screening and testing options” visit with a genetic counselor at 11-12 weeks gestation

Diagnostic testing:
- CVS (10-13 6/7)
- Amniocentesis (>15 weeks)

If declines diagnostic testing

Non-invasive prenatal testing (NIPT) for Down syndrome, trisomy 18, and trisomy 13 (>10 weeks)
- Nuchal translucency>3mm
- Cystic Hygroma or other structural abnormalities
- Ultrasound for dating, nuchal translucency evaluate for multiple gestation and structural anomalies

Positive Screening results
- Positive (Increased risk of T 21 by 490x)
  - Confirm with amniocentesis or CVS
- Sample failure (estimated 0.8%)
  - Offer diagnostic testing or repeat sample
- Negative (Decreased risk of T 21 by 79x)
  - No further testing

Genetic consultation to follow-up regarding results

Traditional screening:
- First trimester combined screening (11-13 6/7)
- Quad screening (15-22 6/7)


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Notification to Users

These algorithms are designed to assist the primary care provider in the clinical management of a variety of problems that occur during pregnancy. They should not be interpreted as a standard of care, but instead represent guidelines for management. Variation in practices should take into account such factors as characteristics of the individual patient, health resources, and regional experience with diagnostic and therapeutic modalities. The algorithms remain the intellectual property of the University of North Carolina at Chapel Hill School of Medicine. They cannot be reproduced in whole or in part without the expressed written permission of the school.

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