IN THIS ISSUE:
PALLIATIVE CARE

The Perinatal Times

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THE PERINATAL OUTREACH PROGRAM

The Perinatal Outreach Program is a collaborative effort between SSM Maternity Care at St. Mary’s Health Center, SSM Cardinal Glennon Children’s Medical Center and Saint Louis University School of Medicine.

It is designed to improve outcomes for mothers and babies through educational programs and quality improvement activities for regional perinatal care providers in eastern Missouri and southern Illinois.

The SSM Cardinal Glennon Children’s Medical Center and SSM St. Mary’s Health Center are the designated Administrative Perinatal Center for Southern Illinois.

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The Perinatal Times welcomes comments on any of its articles and will consider such letters for publication. Suggestions for future topics of interest or announcements are encouraged.

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NRP IN THE KNOW

OXYGEN: HOW MUCH IS INDICATED? BY GLENN BARBER, RNC-NIC, BSN

A 39 5/7 week gestation infant is delivered by emergent cesarean section due to a Category III fetal heart rate pattern. The mother had an uncomplicated pregnancy and had clear amniotic fluid at delivery. Immediately after birth, the infant was non-vigorous. The baby was placed under a radiant warmer, dried, stimulated and positioned with the head slightly extended. The infant was cyanotic, flaccid, had gasping respirations and a heart rate of 90. Which of the following interventions would have been appropriate for this infant?

1. Give 100% supplemental oxygen (free flow) by holding the tubing close to the infant’s mouth and nose.
2. Provide positive pressure ventilations via bag/mask or T-piece resuscitator using room air.
3. Provide positive pressure ventilations via bag/mask or T-piece resuscitator using 60% oxygen.
4. Provide positive pressure ventilations via bag/mask or T-piece resuscitator using 100% oxygen.

CORRECT RESPONSE

Provide positive pressure ventilations via bag/mask or T-piece resuscitator using room air.

DISCUSSION

After the completion of the initial steps (warm, clear the airway if necessary, dry and stimulate), evaluation of the infant’s heart rate and respiratory effort are needed to determine if resuscitation is indicated. If the heart rate is less than 100 or the infant is gasping or apneic, positive pressure ventilation via bag/mask or T-piece resuscitator is indicated. The 2010 International Consensus on Neonatal Resuscitation and the Neonatal Resuscitation Program, 6th edition, both recommend starting with room air during resuscitation of term infants and then using an FiO₂ (blended oxygen & air) guided by oximetry. 1, 2 This recommendation is supported by two systematic reviews and meta-analyses which suggested that starting with room air is not only superior to 100% oxygen, but may result in a lower mortality rate. 3, 4 The goal is to achieve oxygen saturations that are similar to healthy infants during normal transition. These are reflected in the NRP targeted pre-ductal SpO₂ during the first 10 minutes after delivery – see Table 1.

In the preterm infant at less than 32 weeks gestation, limited evidence is available regarding optimum oxygen saturations; therefore, the recommendation is to keep these infants in the same saturation range as the term infant. Resuscitation of this group with room air or 100% oxygen is more likely to result in saturations outside the targeted range. The recommendation in infants less than 32 weeks, although broad, is to initiate resuscitation using 30% to 90% oxygen and titrate to the targeted oxygen saturation. Most practitioners feel that starting at 30% to 40% oxygen and titrating up as necessary is a reasonable approach until more evidence is available.

Free flow or supplemental oxygen is rarely needed after a routine delivery of a normal term infant. However, it may be indicated in some infants that remain cyanotic and have a low targeted pre-ductal SpO₂. It has been shown that visual assessment of cyanosis is not very reliable in determining the need for supplemental oxygen; therefore, pre-ductal oximetry should be used to confirm SpO₂ and help guide the practitioner while delivering free flow oxygen.

Since the release of the Neonatal Resuscitation Program, 6th edition, oxygen management during neonatal resuscitation has changed dramatically. Instead of 100% oxygen, the recommendation is to start resuscitation with 21% in term infants and use an oxygen/air blender to deliver the therapeutic dose. Many consider oxygen to be a medication and giving the correct dose is important to improve neonatal outcomes. Just as with other medications, giving too little may not be beneficial and too much may be toxic. The goal is to give the recommended therapeutic dose.

<table>
<thead>
<tr>
<th>Time (min)</th>
<th>Targeted Pre-ductal SpO₂ After Birth</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>60% - 65%</td>
</tr>
<tr>
<td>2</td>
<td>65% - 70%</td>
</tr>
<tr>
<td>3</td>
<td>70% - 75%</td>
</tr>
<tr>
<td>4</td>
<td>75% - 80%</td>
</tr>
<tr>
<td>5</td>
<td>80% - 85%</td>
</tr>
<tr>
<td>10</td>
<td>85% - 95%</td>
</tr>
</tbody>
</table>

REFERENCES


ABOUT THE AUTHOR

Glenn Barber is a Neonatal Outreach Educator in the Perinatal Outreach Department at SSM Cardinal Glennon Children’s Medical Center. Glenn is a Regional NRP Instructor, a Lead STABLE Instructor and a Master Trainer for the Helping Babies Breathe Program with almost 25 years of clinical experience. Contact Glenn at glenn_barber@ssmhc.com

TABLE 1
Despite advances in neonatal medicine and intensive care, there are some instances where extraordinary care to maintain life is not appropriate. Infants who are born too early, who are too acutely ill and not responding to intensive care therapies, those who are suffering to the point of therapies being a burden, or those with a diagnosis that is incompatible with life, may be offered the option of palliative care. It is an approach to care that encompasses the entire family beginning with the diagnosis and continuing throughout the infant’s life, death and beyond. It embraces physical, emotional, social and spiritual elements and focuses on enhancement of the quality of life for the infant and family. If the condition is diagnosed in the antepartum period, resuscitation plans should be discussed with the parents in advance, including the level of intervention and comfort measures they desire. This is regardless of the time of anticipated death, which could be in utero, in the delivery suite, neonatal unit or at home.

Palliative care includes, but is not limited to, the following:

1. **Physical comfort care to minimize distress:** positioning, skin care, mouth care, human contact including breast feeding and skin to skin contact.

2. **Review of medications:**
   - a. Discontinue medications that do not add to the comfort of the baby.
   - b. Some infants may require anticonvulsants to control seizures.
   - c. Consider pre-drawn medications of fentanyl or morphine for nasal or buccal administration for pain, restlessness, or respiratory distress. Doses may be increased by 30% over normal recommendations as needed to ensure the baby is comfortable. Versed may also be used buccally or subcutaneously.
   - d. If the infant is intubated, paralytics should be weaned off.

3. **Nutrition, feeding, and GI Symptoms:** the goal is to provide comfort and reduce distress from hunger. Breast feeding may be comforting for the baby and the mother. Parenteral fluids and NG feeding are rarely indicated.

4. **Vomiting:** reduce feedings to the level the baby will tolerate; anti-reflux medications may be considered if reflux is present.

5. **Constipation:** opiates may constipate so consider glycerin suppositories.

6. **Respiratory secretions:** treat secretions only if distressing the baby.
**MEDICATION DOSING:**

**Seizure Control:** Midazolam 0.1 mg/kg IV every 15 minutes as needed or Phenobarbital 20 mg IV Loading dose then 5 mg/kg IV or PO maintenance

**Reduction of secretions:** Glycopyrrolate 0.02 -0.04 mg/kg PO Q6-8 hours

**Dry mouth or lips:** petroleum jelly /moist swabs

**Pain/sedation:** Some studies have looked at utilizing intranasal fentanyl for neonatal palliative care. It is a highly potent opioid so only small volumes are needed. It is highly lipophilic so it is absorbed readily through the mucosal membranes and blood-brain barrier. Therapeutic levels of intranasal fentanyl can be seen as quickly as 2 minutes and it is not irritating to the nasal mucosa. For specific medication dosing, see Tables 1 & 2 on page 5.

The main goal of palliative care is to keep the infant comfortable, whether it be for minutes, hours or days after delivery. Medications are given to relieve distressing symptoms such as seizures, respiratory distress and to provide analgesia. Medication management needs to address the symptoms with choices that are noninvasive and minimally burdensome so the family has optimal bonding time. Intravenous line placement may take time the infant could be snuggling with the parents. Some parents may worry that drugs may hasten death, but literature supports that opioids administered in doses proportional to the degree of distress do not quicken death but may in fact delay death. Parents should be supported and reassured they are providing the best level of care to enhance the precious time with their infant.

**REFERENCES**


Palliative Care for Newborns and Infants. National Association of Neonatal Nurses


**ABOUT THE AUTHOR**

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PALLIATIVE CARE
FOR THE INFANT AND FAMILY
REBECCA L. HUNT, RN, MSN, NNP-BC, CCNS

Advances in technology have changed modern day medicine. We, as a healthcare team, are better able to prolong life; however, we are also more often in a position to make deliberate decisions about when and how death will occur. Despite the technological advances in neonatal care made in recent decades, two thirds of infant deaths occur in the first month after birth due to preterm delivery or birth defect. Palliative care can, and should, occur from the earliest recognition of a life threatening condition. The focus is to provide all resources available to ensure the patient’s comfort while offering support to the family.

The process of transitioning to palliative care requires a careful assessment and mutual decision making. This process should include communication about, documentation of, and respect for family goals concerning palliative care. Special attention should also be given to the family’s psychosocial, cultural, and spiritual needs.

WHO SHOULD RECEIVE PALLIATIVE CARE

While many aspects of palliative care should be integrated into the care of all newborns, there are infants born who the parents and health care team believe that palliative care is the most appropriate form of care. This group of infants is very diverse in pathology and includes:

• Genetic problems: Trisomy 13, 15, or 18, Triploidy, Thanatophoric dysplasia, lethal forms of osteogenesis imperfecta, and errors of metabolism expected to be lethal even with available therapy
• Renal problems: Potter’s syndrome, renal agenesis with severe lung hypoplasia, and some cases of polycystic kidney disease
• Central Nervous System abnormalities: Anencephaly, acrania, congenital severe hydrocephalus with absent or minimal brain tissue, and neurodegenerative diseases requiring mechanical ventilation
• Cardiac problems: Inoperable heart anomalies and ectopia cordis
• Structural anomalies: Giant omphaloceles; severe cranio-facial anomalies, and severe congenital diaphragmatic hernia
• Extreme Prematurity: If at birth, an infant is considered non-viable or extremely immature, it may be decided to provide palliative care.
• Overwhelming illness: Includes infants, regardless of gestational age, who may have exceeded the benefits of neonatal intensive care (including no response to aggressive resuscitation, severe cases of perinatal brain injury, severe asphyxia, hypoxic-ischemic encephalopathy, and overwhelming sepsis).

PALLIATIVE CARE DISCUSSIONS

Unfortunately, neonatal palliative care has rarely been a topic of public discussion. Therefore, as soon as a potentially lethal problem is identified, options should be discussed with the family. All major problems should be reviewed with the family clearly, accurately, and with empathy (family includes parents, grandparents, and other relatives and support people that the family indicates). All high-risk families should be honestly counseled on their options and allowed to define their goals and hopes for treatment.

When discussing palliative care, the family should know that they will not be abandoned. Terms such as “withdrawal of treatment” or “withdrawal of care” should be avoided; instead language such as, “We will continue to provide the most appropriate medical care for your infant” should be used. If indicated, words such as “death” and “dying” should be used and euphemisms avoided. The family should also be reminded that even though the healthcare team can not change the situation, their infant will be treated with compassion and dignity. The healthcare team should acknowledge that not beginning interventions and/or discontinuing interventions that cause suffering is a brave and loving action for parents to take for their child.

INFANT CARE

A consensus has not been achieved about how best to monitor infants who are receiving palliative care. Frequent assessments by physicians and the nursing staff will continue in order to ensure the infant’s pain control. However, many clinicians feel that monitoring (vital signs, pulse oximetry, etc.) is not necessary but may be used if it is helpful for the parents to better understand their infant’s condition.

The literature is also inconclusive regarding obtaining laboratory specimens. Some clinicians in the health care field believe laboratory specimens should only be obtained if the results may lead to treatment that will improve the infant’s quality of life. However, there are also clinicians who feel that laboratory collections should be obtained, especially if confirmation of the diagnosis is needed postnatally or that further information could help the family when considering future pregnancies. It’s important to remember that some treatments that may be viewed as invasive, such as antibiotics, oxygen, anticonvulsants, and insertion of VP shunts can have a role in symptom control.

FAMILY CARE

The most important element in communicating with families is listening to them. The more complex the medical situation, the more crucial it is that families receive consistent information, perhaps from a single designated person on the
health care team. Usually, prenatal diagnosis of anomalies gives parents time to adjust to their child’s condition before birth or before decisions have to be made. Parents should be allowed to spend as much time with the infant as possible. The parents may need to be given choices on how they would like to spend their time with their baby as many will rarely have any idea of what they can or want to do. Many parents will be grateful if the staff suggests they start to create and collect mementos as soon as possible. These mementos will be the only tangible evidence of their infant’s existence and their time together and may be very important in the future. Siblings should also be allowed to visit and bond with the infant (in accordance with the parents wishes).

Parents should be offered a choice of whether they would like to have a religious or spiritual ceremony. All of the families wishes should be documented and clearly communicated between shift changes, as this will not only help care givers provide exceptional care but also allow the family to avoid being asked the same question(s) multiple times.

PAIN CONTROL
Once a decision is made to either limit or withdraw life-sustaining medical treatment, relieving an infant’s pain and agitation should become a primary goal regardless of the risks of undesired consequences, such as hypotension and respiratory depression. Intravenous (IV) access may need to be established or remain in place to give medication for symptom relief. Infants may need pain relief, relief for labored respirations, or treatment for seizure activity. Medication doses should be sufficient to relieve pain (Table 1) and provide comfort and sedation (Table 2).

SUMMARY
Palliative care should be offered as supportive, compassionate care to all high-risk newborns. The palliative plan of care should cover all foreseeable outcomes but should also be flexible enough to allow for changes in the condition of the infant or in the parents’ views and wishes. Palliative care should emphasize that each day is precious and honors the life of an infant, even if for a short time, and can be a meaningful experience for a family and the healthcare team.

### Table 1: Medications for Pain

<table>
<thead>
<tr>
<th>Medication</th>
<th>Starting Dose</th>
<th>Frequency</th>
<th>Infusion</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Morphine</td>
<td>0.02 - 0.1 mg/kg IV/IM</td>
<td>Q 2 - 4 hours</td>
<td>0.02 - 0.1 mg/kg/hour</td>
<td>Opioid</td>
</tr>
<tr>
<td>Fentanyl</td>
<td>1 - 3 mcg/kg/dose IV/Intranasal</td>
<td>Q 1 - 2 hours</td>
<td>1 - 3 mcg/kg/hour</td>
<td>Short acting opioid</td>
</tr>
<tr>
<td>Methadone</td>
<td>0.05 - 0.2 mg/kg IV/PO</td>
<td>Q 4 - 24 hours</td>
<td>Long acting opioid</td>
<td></td>
</tr>
<tr>
<td>Acetaminophen</td>
<td>10-15 mcg/kg PO 20 mg/kg PR</td>
<td>Q 6 hours</td>
<td>Analgesic</td>
<td></td>
</tr>
<tr>
<td>Oral Sucrose (24%)</td>
<td>&lt;1 kg: 0.1 mL 1 - 2 kg: 0.5 mL &gt;2 kg: 1-2 mL</td>
<td>PRN</td>
<td>Analgesic</td>
<td></td>
</tr>
</tbody>
</table>

### Table 2: Medications for Sedation

<table>
<thead>
<tr>
<th>Medication</th>
<th>Starting Dose</th>
<th>Frequency</th>
<th>Infusion</th>
<th>Comments</th>
</tr>
</thead>
<tbody>
<tr>
<td>Clonidine</td>
<td>3 - 10 mcg/kg/day PO</td>
<td>Q 6 – 12 hours</td>
<td>Alpha agonist</td>
<td></td>
</tr>
<tr>
<td>Midazolam</td>
<td>0.05 - 0.1 mg/kg IV/IM 0.2 mg/kg SL/Intranasal</td>
<td>Q 1 – 4 hours</td>
<td>1 – 2 mcg/kg/min</td>
<td>Very short acting Benzodiazepine</td>
</tr>
<tr>
<td>Lorazepam</td>
<td>0.05 - 0.1 mg/kg IV/IM/PO/PR</td>
<td>Q 2 – 4 hours</td>
<td>Benzodiazepine</td>
<td></td>
</tr>
<tr>
<td>Chloral Hydrate</td>
<td>25 – 75 mg/kg PO/PR</td>
<td>Q 4 – 12 hours</td>
<td>Use for sedation</td>
<td></td>
</tr>
</tbody>
</table>

REFERENCES

ABOUT THE AUTHOR
Rebecca L. Hunt received her master’s degree in nursing from Duke University, specializing as a neonatal nurse practitioner and a neonatal clinical nurse specialist. She has 11 years clinical experience and is currently working as an NNP in the NICU at SSM Cardinal Glennon Children’s Medical Center in St. Louis, Missouri.
Fetal tachycardia is defined as a sustained fetal heart rate (FHR) greater than 160 bpm, but in most cases it will be greater than 180 bpm. The causes of fetal tachycardia include sinus tachycardia, supraventricular tachycardia (SVT), atrial flutter, atrial fibrillation and ventricular tachycardia. While the most common type of fetal arrhythmia is an irregularity of rhythm such as premature atrial contractions or premature ventricular contractions, tachyarrhythmias are the next most common and represent approximately 10% of fetal arrhythmias with an incidence of 0.4-0.6% of all pregnancies (Murray, 2007). Sustained tachyarrhythmias can be life-threatening and are accompanied by anatomic abnormalities in 6-7% of cases.

**SINUS TACHYCARDIA**

In sinus tachycardia, the sinus node is firing faster than its inherent rate with a normal conduction of the impulse. The onset and offset is usually gradual with the FHR, ranging from 160-200 bpm. The increase in the rate is usually a response to a demand by the body for an increase in cardiac output (Murray, 2007). Causes may include hypoxia; maternal pain and/or anxiety; maternal infection and/or fever; amnionitis; hyperthyroidism; and drugs such as cocaine, amphetamines or beta-adrenergic drugs such as terbutaline. Treatment of sinus tachycardia includes identifying and treating the cause. If the cause is hypoxia, intrauterine resuscitation is required.

**SUPRAVENTRICULAR TACHYCARDIA**

Supraventricular tachycardia (SVT) is a global term for a tachyarrhythmia in which extra cardiac impulses are conducted to or from the atria to the atrioventricular (AV) node. Fetal SVT includes atroventricular reentry tachycardia, atrial ectopic tachycardia, junctional ectopic tachycardia, atrial flutter and atrial fibrillation (Murray, 2007). Fetal SVT usually includes a FHR in the range of 240-280 bpm with a 1:1 AV conduction (Murray, 2007). Atrial flutter is rare with an atrial rate of 300-500 bpm and is commonly associated with a 2:1 AV block with only half of the impulses being conducted resulting in a ventricular rate of 150-250 bpm (Murray, 2007). Atrial fibrillation is rarely identified in the fetus and would include an atrial rate of 400-500 bpm with a ventricular rate of 200-250 bpm (Murray, 2007). SVT starts abruptly, i.e., with one beat of the heart. A sustained fetal SVT is associated with compromised cardiac output, congestive heart failure, hydrops, and intrauterine growth restriction. Distinguishing the different tachyarrhythmias by ultrasound can be challenging, thus the patient should be referred for a detailed fetal echocardiogram. When a sustained SVT has been
diagnosed and the fetus has been evaluated, a treatment plan will be developed. Management varies depending on gestational age and fetal status. If the fetus is preterm and demonstrates evidence of compromise, treatment will likely include an antiarrhythmic drug that will be given to the mother or directly to the fetus.

VENTRICULAR TACHYCARDIA

Fetal ventricular tachycardia is far less common than SVT, and it is commonly fatal when sustained. It usually manifests with fetal rates between 180 and 300 bpm and is usually accompanied by poor ventricular function. Fetal ventricular tachycardia has been associated with tumors, structural heart disease, prolonged QT interval, and fetal distress or acidosis (Sklansky, 2009). In some cases, pharmacologic treatment may be attempted, but the prognosis is guarded.

The following fetal monitor tracings are from a 30 y/o, G3P1011 admitted at 32 weeks gestation for evaluation of a fetal tachycardia that was identified in the physician’s office during a routine prenatal visit. Fetal evaluation at the Perinatal Center confirmed the diagnosis of fetal SVT along with the presence of fetal ascites, pericardial effusion and hydrocele. No structural anomalies were identified.

The umbilical artery Doppler flow studies were within normal limits. The biophysical profile score was 8/10 with a normal amniotic fluid volume. The patient was treated with antiarrhythmic drugs including digoxin and flecainide. The second tracing demonstrates the conversion from SVT to normal sinus rhythm. The fetus continued to have nonsustained episodes of SVT, but over the course of about a week the fetal hydrops resolved. Labor was induced at 37 weeks gestation resulting in a spontaneous vaginal delivery. Mom and baby did well. The infant continued to receive an antiarrhythmic medication and was followed closely by pediatric cardiology.

**REFERENCES**


**ABOUT THE AUTHOR**

Sharon Rector is an Education Consultant specializing in perinatal nursing at SSM Health Care in St. Louis.
Since 2011, a new technology that uses the presence of DNA fragments in a pregnant woman’s blood stream to screen for fetal chromosome abnormalities has become available. The cell-free portion of human blood (serum) is known to contain relatively large amounts of genetic information or DNA which can be isolated and tested. During pregnancy, this cell-free DNA comes from both the mother and the fetus. This article will discuss the basic technology used for this testing, the benefits and limitations of this testing, and some specific information about how this test can be used clinically.

Individuals who provide obstetrical care are familiar with the fact that the chance of giving birth to a baby with a chromosome abnormality such as Down syndrome (trisomy 21), increases with the advancing age of the mother. In 2007 the American College of Obstetrics and Gynecology issued a revised practice bulletin that recommended all pregnant women, regardless of their age, be informed of the chance they could give birth to a baby with Down syndrome, and that they be provided with information regarding the availability of diagnostic testing, including chorionic villus sampling in the first trimester and amniocentesis in the second trimester. In addition, all women should be provided with information regarding the first and second trimester tests that screen for the other common chromosome abnormalities, trisomies 13 and 18. These screening options include first trimester ultrasound assessment of nuchal translucency, and first and second trimester maternal serum screening. The differences between screening and diagnostic tests, their risks, limitations and benefits were to be reviewed, allowing women to make an informed choice regarding testing.

The introduction of prenatal screening using the DNA found in the cell-free portion of human blood presents care providers with yet another option to discuss with certain patients (see below). This DNA is not found as complete chromosomes, but fragments of chromosomes. The number of fragments from each chromosome usually corresponds to the size of that chromosome, relative to the others. For example, chromosome #3 makes up about 6% of the total genomic DNA while chromosome #21 makes up about 1.7% of the total. In the usual situation, fragments from chromosomes 3 and 21 are isolated in a ratio of about 5 to 1. Advances in technology have made it possible to isolate these cell-free DNA fragments, determine which chromosomes they come from, and compare the relative amount from any two chromosomes of interest. Additional developments allow laboratories to compare DNA samples from different individuals and distinguish one from the other using natural variations in the genetic code. These natural variations in the genetic code are called “polymorphisms”. Testing for chromosome abnormalities using this cell-free fetal DNA (cff DNA) are often referred to as non-invasive prenatal tests or NIPT.

In most women, after the first trimester, over 5% of the cell-free DNA is of fetal origin. The percentage of DNA that is from the fetus is referred to as the “fetal fraction”. This DNA is believed to come from cells within the placenta. Using chromosomes 3 and 21 as our example, if a woman is carrying a baby who has two copies of chromosomes 3 and 21, the ratio of fragments in her blood stream from these two chromosomes will be about 5 to 1, as expected. However, if a woman is pregnant with a baby who has Trisomy 21, there will be relatively more fragments from chromosome 21 than expected relative to chromosome 3, usually reflecting a ratio of closer to 4 to 1. This change in ratio can be detected, allowing the laboratory to report that this pregnancy is at higher risk to have Down syndrome. Similarly, the relative number of DNA fragments...
can be determined for chromosomes 13, 18, X and Y. Tests that rely on differences in the relative amounts of different chromosomes use what is called the “counting method”. The ability to detect chromosome abnormalities, or the sensitivity of the test, is dependent upon the fetal fraction. Fetal fraction tends to increase as pregnancy advances. Fetal fraction tends to be lower in women with higher body weights. Chromosome fragments from certain chromosomes, such as chromosome 13, may not be as reliably measured using the counting method, so the detection rate may not be as high.

An alternative method of NIPT uses the presence of changes in DNA sequences, or polymorphisms, to read the genetic code of the DNA fragments in the mother’s blood stream. This method reads differences in the codes between the mother and the baby. A simple cheek swab from the baby’s father can also be sent with the sample, and may help with interpretation of results. The ability to detect different chromosome abnormalities using this method is the same for all chromosomes.

There are a few things that are important to remember about these new blood tests. First of all, the research that has been performed has only tested women who are at high risk to be carrying a baby with a chromosome abnormality. Current ACOG guidelines indicate that this testing is not recommended for women at “routine risk” usually defined as a risk of less than that of a 35 year old which is about 1/270.

NIPT is not considered a replacement for chorionic villus sampling and amniocentesis. Both false positive and false negative tests have been reported. No irreversible action should ever be taken based upon an NIPT result without CVS or amniocentesis for confirmation. A family should be informed, however, that if an NIPT indicates a high risk, the likelihood that the baby is affected is generally greater than 90%. If an NIPT indicates a low risk, the likelihood that the baby is affected is generally much less than 1 in 1000.

These tests only provide information for very specific abnormalities. There are many situations in which a baby may have genetic abnormalities that are not detected by these tests and NIPT may not be the best option. For example, if a baby has a major structural birth defect, such as a heart defect, there is an increased risk for chromosome abnormalities involving chromosomes other than chromosomes 13, 18 and 21 which would not be detected by NIPT. If a family is at risk to have a baby with a genetic disorder that involves a gene change, such as cystic fibrosis, it will not be detected by NIPT. The number of conditions that are detectable using NIPT is changing rapidly, and consultation with a genetic counselor may be helpful in deciding which tests to offer.

Up to 5% of the time, a woman will not receive a result, or will receive only a partial result, from the NIPT test. This is often due to a low fetal fraction. We recommend waiting at least a month before sending a second sample to increase the likelihood that it will return a result. If a woman wants test results sooner, then either traditional screening with maternal hormones and/or ultrasound or a diagnostic test should be considered. If a woman weighs over 200 pounds, delaying the test until after 14 weeks should also be considered. In general, NIPT returns a result within 1-2 weeks, which is longer than either CVS or amniocentesis.

Within SSM Health Care - St. Louis, a genetic counselor is available 5 days a week to meet with families and discuss options for prenatal testing. For questions about prenatal testing, or to speak to a genetic counselor, call 314-768-8673. The scope of prenatal testing that will become available using cell-free fetal DNA in the maternal circulation will expand exponentially over the next few years. Geneticists can provide the most up to date information regarding test options and availability.

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CONTINUING EDUCATION OPPORTUNITIES

Many continuing education opportunities, including traditional lectures, hands-on practice as well as online presentations, are available for perinatal professionals in eastern Missouri and southern Illinois. These are offered through SSM St. Mary’s Health Center, SSM Cardinal Glennon Children’s Medical Center, Saint Louis University School of Medicine and the Perinatal Outreach Program. Most programs offer nursing contact hours and/or CMEs.

For course calendars or more specific information on programs, please go to www.cardinalglennon.com and click on the “For Professionals” tab or call the Perinatal Outreach Program at 314-577-5317.