



# Fetal Focus

A Newsletter from the Fetal & Women's Center of Arizona

## Managements of Myelomeningocele Study



The Fetal and Women's Center of Arizona (FWC) was recently featured on the Channel 3 news and in the East Valley Tribune for its participation in the Management

of Myelomeningocele Study (MOMS), which was a research study designed to compare the effectiveness of prenatal versus post-natal surgery in babies with spina bifida. With the help of FWC, Jody Simmer was able to be admitted into the randomized study and have prenatal surgery after the baby was diagnosed with spina bifida in 2004 by Dr. Nyberg. With clinical coordination the company was able to get the family connected with the closest study group in California. Today, Jody has a 6 year old daughter and is also expecting another in the upcoming months. FWC has had other patients that have participated in the study as well, one received surgery after the child's birth and the other patient had prenatal surgery in January, and is awaiting the birth of her child in the near future.

The trial was stopped for efficacy of prenatal surgery after the recruitment of 183 of a planned 200 patients. This report is based on results in 158 patients whose children were evaluated at 12 months. The first primary outcome occurred in 68% of the infants in the prenatal-surgery group and in 98% of those in the postnatal-surgery group. Actual rates of shunt placement were 40% in the prenatal-surgery group and 82% in the postnatal-surgery group. Prenatal surgery also resulted in improvement in the composite score for mental development and motor function at 30 months and in improvement

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in several secondary outcomes, including hindbrain herniation by 12 months and ambulation by 30 months. However, prenatal surgery was associated with an increased risk of preterm delivery and uterine dehiscence at delivery (New England Journal of Medicine).

About 80,000 babies are born in Arizona each year. Of these, we can expect at least 30 new cases of spina bifida. In experienced hands, an ultrasound should be able to detect 100% of significant open spina bifida cases, compared to 80% for AFP screening alone.

Please see our website [www.fetalcenter.com](http://www.fetalcenter.com) to learn how prenatal surgery now offers additional hope for families with a fetus with spina bifida, and to hear how FWC participated in the important MOMS trial which has helped pave the road for fetal surgery becoming a standard of care for spina bifida.

## Featured Patient



“At 19 weeks gestation, I learned from Dr. David Nyberg that my unborn daughter had a severe heart

defect, proving fatal without immediate intervention following birth. Dr. Nyberg spent time explaining his findings and educating my husband and I on what to expect in the weeks and months to come. After receiving this shocking news, The Fetal & Women’s Center coordinated our care with a wonderful local fetal cardiologist, a world-renown cardiac care team and continued to follow myself and my unborn daughter as patients, to term.

Today, after two open heart surgeries, my daughter, Zoe, is not only surviving, but thriving. Thank you, Dr. Nyberg and your entire staff for your expertise and taking my family to heart.

Follow Zoe’s journey and be inspired by our miraculous child at [www.thelihns.blogspot.com](http://www.thelihns.blogspot.com) and learn about other similar families through our non-profit organization, Sisters by Heart, [www.heartsisters.blogspot.com](http://www.heartsisters.blogspot.com) – an organization dedicated to providing support to newly diagnosed Hypoplastic Left Heart Syndrome (HLHS) families.” – Stacey Lihn, Mom to Zoe (Age 1)

*Would you like to share your story of how The Fetal and Women’s Center helped you? Call 1-877-FETAL-AZ and speak with our patient care coordinator*

## Did you know?

- FWC is the largest screening and diagnostic center of its kind in the southwest.
- Fetal abnormalities affect 2-3% of ALL pregnancies, regardless of maternal age, paternal age, family history, prior history or lifestyle.
- Please see the summary of birth defects (Insert) seen at FWC over the last 2 years. This includes over 150 fetuses with heart defects.
- Screening is not just about Down syndrome. Down syndrome is only one of many birth defects that affect about 1 in 500 pregnancies. Unlike most birth defects, the specific risk for Down syndrome varies with maternal age.
- Most patients mistakenly believe that birth defects are extremely rare, or because they are young and healthy, they are not at risk. The reality is we don’t know which patients are carrying a fetus with a serious birth defect until after a detailed ultrasound and genetic sonogram.
- Prenatal detection of birth defects can improve postnatal outcome and help optimize patient management for a variety of conditions, but especially for heart defects, diaphragmatic hernia, thoracic or neck masses, spinal defects, cleft lip/palate, urinary abnormalities, and club feet.
- We cannot predict what choices a couple will make until presented with specific information. For example, we see some patients who think they are not interested in prenatal screening, but change their minds when an ultrasound detects abnormalities suspicious for Down syndrome; they often choose genetic amniocentesis and may even choose to terminate the pregnancy. Other couples believe they would not consider pregnancy termination for any reason.
- In Down syndrome, a genetic sonogram at FWC can be used to detect one or more abnormalities in approximately 85% of fetuses. That is equivalent to first trimester screening. Please see abstract (Pg 3) “Why a Genetic Sonogram Is an Important Part of a Detailed 2nd Trimester Ultrasound”.

“Fetal abnormalities affect 2-3% of ALL pregnancies regardless of maternal age, paternal age, family history, prior history or lifestyle”

# 2nd Trimester Genetic Sonogram in the Era of First Trimester Screening



We reviewed our experience with screening and genetic sonography in 90 cases of fetal Down syndrome seen at a single community-based screening center over a 6 year period. Fifty eight patients underwent first trimester screening and of these 51 had a positive screen, including 38 patients with a risk of 1 in 100 or greater. 13 patients in the highest risk group had CVS or early amniocentesis and all

of those patients elected to terminate the pregnancy.

- *Of the 90 cases of fetal Down syndrome, 67 patients had a 2nd trimester ultrasound and genetic sonogram, including 43 patients who had a prior first trimester screen.*
- *Of these, 9 had a normal ultrasound and 58 (86.6%) showed one or more abnormalities or ultrasound markers. The single most common ultrasound marker was an absent or hypoplastic nasal bone, seen in 33 (49%). An echogenic intracardiac focus (EIF) was seen in 16 patients (24%), and structural abnormalities were seen in 11 cases (16%).*

- *Of the 9 cases with a normal ultrasound, 5 had an abnormal first or 2nd trimester screen but due to the degree of risk, all patients remained high risk following the ultrasound, even after reducing the risk by a factor of 3x fold for a normal ultrasound. Among 10 patients with normal first trimester (7) or 2nd trimester biochemical screening (3), 7 had one or more ultrasound findings and 3 patients had a normal ultrasound.*

We conclude that in our community, the majority of patients with fetal Down syndrome undergo detailed 2nd trimester ultrasound, even when they have abnormal first trimester screening results. Also, although most patients who prove to have fetal Down syndrome underwent first trimester screening, 47% did not. For those patients who underwent first trimester screening, 2nd trimester ultrasound detected abnormalities or markers in 86.5%, and 70% of those cases missed by other screening including first trimester screening. We conclude that a genetic sonogram should be performed at the same time as a detailed second trimester ultrasound, and this will improve the detection of fetal Down syndrome above that offered by first trimester screening alone.

## An Introduction to Genetic Counseling in the Prenatal Setting



Genetic counseling is a service to help individuals and families translate scientific knowledge into practical information. A genetic counselor works with a person or family that may be at risk for an inherited disease or abnormal pregnancy outcome, discussing their chances of having children who are affected. Genetic counselors often

work as part of a health care team in conjunction with specially trained physicians, social workers, nurses, medical geneticists, or other specialists to help families make informed decisions about their health. They also work as patient advocates, helping individuals receive additional support and services for their health care needs.

One third (33%) of patients referred for genetic counseling have other genetic issues in their families that need to be addressed. These issues are identified by taking a detailed family health history. The genetic counselor will talk to patients and their families about any inherited disorders that arise in the process of diagramming the family history and their chance of passing these disorders on to their children. The counselor will also ask about any toxic substances the patient may have been exposed to or medications she may be taking or have taken during her pregnancy. Genetic counselors can help patients understand the risks, benefits, and limitations of any prenatal tests they may be considering and the results of tests patients have already taken. Consequently, genetic counselors play an integral role in healthcare today. The genetic counselors at The Fetal and Women's Center of Arizona are dedicated to providing excellent patient care and acting as a valuable resource for referring physicians and the public.



## Eight Convenient Valley Locations

### **Anthem**

4810 North Venture Drive  
Building EE , Suite 152  
Anthem, AZ 85086  
Phone: 480.467.2155

### **Chandler**

1910 West Frye Road  
Chandler, AZ, 85224  
Phone: 480.467.2145

### **Estrella**

10240 West Indian School Road  
Suite 100  
Phoenix, AZ 85037  
Phone: 623.243.5914

### **Gateway**

4864 East Baseline Road  
Suite 101  
Mesa, AZ 85206  
Phone: 480.467.2150

### **Glendale**

5605 West Eugie Avenue  
Suite 212  
Glendale, AZ 85304  
Phone: 602.942.6800

### **Mesa**

2242 West Southern Avenue  
Mesa, AZ 85202  
Phone: 480.491.1600

### **Phoenix** – Coming spring 2011

2942 North 7th Avenue  
Suite 102  
Phoenix, AZ 85013  
Phone: 602.942.0600

### **Scottsdale**

9440 East Ironwood Square Drive  
Scottsdale, AZ 85258  
Phone: 480.551.9700

[www.fetalcenter.com](http://www.fetalcenter.com)

[www.fetalscreening.com](http://www.fetalscreening.com)

**480-756-6000**



# Featured Employee



Melanie Kunz, director, clinical services for FWC, joined the practice in 2009 with a focus on developing and overseeing the

diabetes program that provides care and management of patients through education.

Melanie completed her bachelor's degree in Health Administration at the University of St. Francis in Joliet, Ill. She later attended the Women's Healthcare Nurse Practitioner program affiliated with San Jose State University in California. Melanie continues her education and is now enrolled at the University of Phoenix, where she is working on her masters in nursing in the Family Nurse Practitioner Program.

While managing The Perinatal Services for The Medical Group of Northern Nevada in Reno, she launched her own medical triaging company; working with physicians in both California and Nevada. In 2007, she launched MedEdCo, which is an education company for diabetes therapy that is utilized in the U.S., Europe and Thailand.

Her educational expertise in diabetes therapy was a perfect fit for the development and launch of the diabetes program at FWC.

The program assists the primary obstetrician in co-management of the diabetic patient. Ms. Kunz provides education, weekly reviews of the patient's diet, monitoring of blood glucose values, and recommendations to the primary obstetrician on times for specific labs, tests, and ultrasounds, to help facilitate a safe pregnancy with an optimal outcome. The program is currently available throughout metro-Phoenix.

Melanie is an active member of the Association of Women's Health, Obstetric and Neonatal Nurses (AWHONN), a nonprofit membership organization that promotes the health of women and newborns.

## March of Dimes

The Fetal and Women's Center of Arizona pledged its support to the March for Babies and helped participate in fundraising and walking for the event April 9th, 2011. While we strive to provide the best possible care and service to women and expecting parents throughout Arizona, that is not enough. Our team participated in the March for Babies because we wanted to do something more; not only to help raise awareness, but to raise funds in the fight against fetal abnormalities, premature birth and infant mortality.

FWC was able to raise a total of \$5,510 for this year's event and will continue to participate in the March for Babies in the future years as well.