



# Perinatal Gazette

Newsletter of the Regional Perinatal Center  
Westchester Medical Center



Volume 2, Issue 5

September 2002

## The Regional Perinatal Center at Westchester Medical Center

Department of Health Regional Health Forum

**SAVE THE DATE!**

**Thursday, October 24, 2002**

**8am – 5pm**

At the Medical Educational Center Auditorium at

New York Medical College

Some of the topics that will be presented:

**Neonatal Epidemiology**

**Environmental Exposures in Breast Milk**

**Innovation in Fetal Diagnosis and Intervention**

**Prenatal Detection & Management of Congenital Heart Disease**

**Regional Database Management and Benchmarking**

For more information call (914) 493-8590

## Newborn Screening for Cystic Fibrosis Comes to New York State

After years of discussion, New York State is adding Cystic Fibrosis (CF) to the growing list of genetic and metabolic disorders tested for in all newborns. In just a few months, pediatricians and family physicians will start receiving phone calls from the New York State Laboratory in Albany that one of their patients has screened positive for CF. Here is a very brief summary of vital information about CF and newborn screening:

### Basic Facts about CF:

- About 1 in 31 people carry one mutation of the CF gene, and are totally asymptomatic (about 1 in 28 Caucasians).
- About 30,000 people in the United States have CF.
- Approximately 23,000 of these patients are being cared for at one of the 110 CF Care Centers accredited by the Cystic Fibrosis Foundation. These centers go through a rigorous inspection and review process and are required to be centers of excellence not only in the care of patients with CF, but also in CF research and education. The data is quite clear that patients at accredited CF Centers do better.
- Although a cure remains elusive, improved therapy has resulted in dramatic improvements in life expectancy, with median survival now approximately 31 years of age. This means that 38% of our patients are now over 18!
- Westchester Medical Center has the only accredited CF Care Center in the Hudson Valley. Named for our founder, The Armond V. Mascia, MD Cystic Fibrosis Center has been caring for patients of all ages with CF for over 35 years.

### Why do newborn screening for CF?

There is increasing evidence that early diagnosis is important in long-term outcome. While most patients with CF are diagnosed within the first year of life, there are many milder patients who go undiagnosed for many years or even decades. Data from other states and countries with established newborn screening programs have demonstrated that children diagnosed soon after birth gain weight and grow much better, and that children diagnosed late do not "catch-up."

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**If newborn screening is so great, what took so long?**

Unfortunately, newborn screening for CF is not straightforward. Although all patients with CF have two mutations of the CF gene (CFTR), there have been over 1,000 mutations described worldwide, and there is still no completely accurate blood test for CF. Newborn Screening will certainly identify many patients earlier and that is great. However, at least two very important problems are going to arise:

1. There will be many false positives. *A majority of infants that screen positive will turn out not to have CF.* This may create significant emotional hardship and confusion for a few weeks until the diagnosis of CF is definitively excluded and families understand this fully. I suspect even after that, many parents will carry scars from that frightening experience.
2. Many children that screened positive but don't actually have CF will turn out to be "merely" carriers. This may have important significance to these families in the future, and misperceptions may be common unless parents receive accurate and effective genetic counseling.

**What should primary care physicians do if their patient screens positive?**

- Some of the time, the diagnosis of Cystic Fibrosis will be made by the state lab before they call you. In that case I would urge you to contact an *accredited* Cystic Fibrosis Center, hopefully ours. That diagnosis must be confirmed.
- If the diagnosis is not made, you should emphasize to parents that most babies that screen positive do not actually have Cystic Fibrosis.
- Next the baby requires further evaluation, which may include either a sweat test or a blood test or both; and parents will need counseling regarding the meaning of a positive screen and interpretation of any subsequent tests.
- Simply call one of the physicians in our Division of Medical Genetics, and they will immediately work your patients and have their families in for testing and counseling.
- Alternatively, feel free to call me or any other physicians in our CF Center staff. We will also be pleased to help arrange for quick and appropriate testing and evaluation.
- **Key Point:** Don't trust a sweat test unless it is done at a center accredited by the Cystic Fibrosis Foundation (CFF). Fortunately in our region, most community hospitals understand how difficult it is to perform consistently accurate sweat tests, particularly in newborns, and therefore no longer perform them. I would urge all parents of babies screened positive to seek genetic counseling, again hopefully here at the Children's Hospital at Westchester Medical Center.

**How the staff of the Armond V. Mascia, MD Cystic Fibrosis Center can help.**

Our goal is to help you in anyway we can. Newborn screening for CF will no doubt create new challenges for all of us, and we will have to find our way together. Here are two very important phone numbers that give you direct access to a physician. In this way we can quickly facilitate the process for you:

**Medical Genetics:        914 – 347 – 3010 touch 3.**

Our team has tremendous experience and knowledge regarding screening for all newborn diseases, as well as interpreting Cystic Fibrosis tests for families and physicians.

**Cystic Fibrosis Center: 914 – 493 – 7585 touch 1.**

We have a large staff, including 7 full-time physicians, and are available 7 days a week, 24 hours day to assist you. **If there are any delays or difficulties arranging for timely sweat tests or patient evaluations, or any questions at all, please feel free to call us at any time.**

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## Perinatal Craniofacial Care

It is estimated that parents-to-be are faced with a 3 to 4% incidence of a major birth defect or genetic problem. Many anomalies of the craniofacial structures can be identified prenatally. These include clefts of the primary (lip to incisive foramen) and secondary (incisive foramen to uvula) palates and the craniosynostoses. Today, several diagnostic modalities exist to visualize and evaluate the fetus. Ultrasonography, amniocentesis, and chorionic villous sampling have been available for more than three decades. Today, it is possible to visualize the landmarks of the face as early as 12 weeks gestation with high-resolution transvaginal ultrasonography. And earlier visualization has allowed patients to bond with their children at an earlier age.

The efficacy of ultrasonography in identifying cleft lip and palate at one time approximated 20 to 30%. Today, 50% of patients with cleft lip/palate are diagnosed prenatally with routine ultrasound (Boyd, 1998) and over 70% when the fetal face is carefully inspected (Robinson, 2001). Few false positive results have been reported and the sensitivity will assuredly increase with the improvement in technology. In addition to standard ultrasonography, 3-D dimensional ultrasonography, alpha-fetoprotein levels, and ultra-fast MRI have become more popular owing to their attractiveness and the increase in availability.

The decision to aggressively pursue the antenatal diagnosis of craniofacial anomalies raises important concerns. While some diagnoses portend a poorer outcome, the majority do not. Several studies have shown that antenatal diagnosis has value in preparing the parents mentally for the coming birth (Davalbhaka, 2000). The anxiety of some families is apparent by the amount of information taken from the internet and other sources and brought to the child's first consultation. The most frequently encountered anomaly – cleft lip/palate – is certainly compatible with life and may afford the patient a relatively normal childhood. However, when faced with the decision to bring a child with cleft lip/palate into the world, some parents choose otherwise. Their decisions depend on the religious and moral backgrounds of those involved. In one study, 23 of 24 parents opted for termination of pregnancy at 13 to 16 weeks when informed that the fetus was diagnosed with a cleft lip/palate (Eisenman and Strauss, 1999).

Evaluation of the neonate with a craniofacial anomaly begins with a complete history and thorough search for associated problems. A review of the family history and prenatal history must be obtained. Key points include relatives with facial clefts, congenital anomalies, or parental consanguinity, and possible use of teratogens during the pregnancy.

For cleft patients, the physical examination should assess the nature of the cleft and the involved structures. Some clefts are incomplete and do not extend across the entire height of the lip and thus spare the floor of the nose. Inspection of the future tooth-bearing alveolar ridge should identify simple notching versus a complete cleft. Finally, examination of the palate should determine whether the hard palate, the soft palate, or both are involved.

Some patients have submucous clefts of the soft palate that are not readily identified on cursory inspection. These are identified by a trio of findings: 1) a bifid uvula, 2) a translucent line running down the middle of the soft palate, and 3) a splayed posterior nasal spine at the back of the hard palate (palpable with a gloved finger). In these patients, the mucosa is intact but the underlying muscles are malpositioned. The exact incidence of submucous cleft palate is unknown since many cases go undiscovered if speech develops normally.

Cranial vault deformities may be present at birth or develop afterwards. Congenital deformities result from premature closure of one or more of the cranial sutures. The most commonly affected suture is the midline sagittal suture producing a long and narrow skull (scaphocephaly). This is followed by the unilateral coronal suture, which produces an asymmetric appearance to the orbits. Remodeling of the vault is performed sometime after three months of age.

Secondary deformities result from either supine positioning or insertion of a ventriculoperitoneal shunt for hydrocephalus. The former has increased in incidence as a result of the “back to sleep” campaign and is managed nonsurgically with changing the position of the infant and/or helmet therapy. Rapid decompression of the ventricles often produces the most dramatic deformities but may be successfully treated with remodeling later in infancy.

In the nursery or the neonatal intensive care unit. The most pressing issue for infants with cleft lip/palate is feeding. The ability to form an oronasal seal is diminished, yet virtually all infants are able to feed by mouth and many are able to breastfeed. If feeding difficulties are encountered, a nipple with a large bore opening should be recommended. Almost no infant should require placement of a gastrostomy tube for feeding.

It is important that patients born with a craniofacial anomaly receive treatment by a team of clinicians from various specialties with experience in treating such patients. Commonly, craniofacial teams are administered by plastic surgeons and/or pediatricians and may include specialists from audiology, ophthalmology, oral surgery, orthodontics, otolaryngology, pediatric dentistry, psychiatry, social work, and speech pathology. The team approach allows many different clinicians to see the children at each visit and creates a coordinated treatment plan. Team care should begin within a few days of life and continue until the physical growth of the patient has been completed – generally around 21 years of age.

Beyond the first year of life, other issues become important. Surgery to close the lip is performed around 3 months of age, while the palate is closed around one year of age. For infants with premature closure of one or more cranial sutures, release is performed between 6 to 12 months of age. Patients with cleft lip/palate are prone to ear infections, hypernasal speech, orthodontic problems and malocclusion. Each child should have an age-appropriate audiologic assessment for each ear within the first three months of age and be followed as indicated.

*Continues on page 4*

Continued from Craniofacial

The **Westchester Craniofacial Center** has an experienced team of clinicians dedicated to treating patients with a wide variety of craniofacial anomalies. The team meets Wednesdays to evaluate new patients and follow-up established patients.

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References:

- Boyd PA, Chamberlain P, Hicks NR. Six-year experience of prenatal diagnosis in an unselected population in Oxford, UK. *Lancet* 352: 1577, 1998.
- Davalbhakta A and Hall PN. The impact of antenatal diagnosis on the effectiveness and timing of counseling for cleft lip and palate. *Br. J. Plast. Surg.* 53:298, 2000.
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## Welcome to our new Neonatologist

Dr. Heather Brumberg will be joining our faculty as of September 1, 2002. She graduated from the Yale New Haven Medical Center's Neonatal Fellowship Program this past June. She has an M.P.H. from the Yale School of Public Health with honors.

Heather is excited about the possibilities of assisting us to coordinate several follow-up clinics and helping to establish databases for research. We await her arrival with anticipation and trust that her joining our faculty will be productive for all.

## Under Construction 8/02 Maria Fareri Children's Hospital/Trauma Center Project – Steel 75% complete



## 20<sup>th</sup> Year Celebration & Graduate Reunion Party



*Calling all staff that trained and/or worked here. Save the date: We've come along way!*

On Thursday, **September 19, 2002 from 2-5 PM** The Regional Neonatal Center will celebrate 20 years of providing care to families in the Hudson Valley Region. This will be a "celebration of life" for the babies and their families who required intensive care at birth or soon after birth. More than 13, 00 babies have graduated from Westchester Medical Center Neonatal Intensive Care Unit, which treats many of the sickest newborns in the New York State with cutting edge new technologies.

Our first baby was admitted as a transport in May 1982, we now have 200 transports each year. In August 1982 our first baby was admitted who was delivered at Westchester. There is great excitement in scheduling this event amongst the staff. Many families whose babies were born in 1982-1984 have been contacted and they are planning to attend some even coming from college for the celebration. We hope that all staff and families will join us to mark this milestone for The Regional Neonatal Center.

For details:

**Please contact Natalie Dweck, RN (914) 493-8998**

## Pediatric Grand Rounds Schedule for September 2002

Location: Westchester Institute for Human Development  
Baird Auditorium  
Wednesday 8:00 AM

- 9/4 **NO GRAND ROUNDS**  
9/11 **"BIO-TERRORISM – AN EMERGING PUBLIC HEALTH THREAT"**  
9/18 **"IMPACT OF RSV INFECTION, AND PROPHYLAXIS"**  
9/25 **MORBIDITY AND MORTALITY CONFERENCE**

## Department of Obstetrics/Gynecology Schedule for September 2002 Grand Rounds

Location: Learning Center  
Thursday at 8:00

- 9/12 **CHROMOSOMAL ABNORMALITIES IN PRENATAL DIAGNOSIS**  
9/19 **SALINE INFUSION SONOHYSTEROGRAPHY**  
9/26 **LIVER DISEASE IN THE FEMALE PATIENT**

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*We are interested in providing you with a newsletter that is relevant and of interest to you. Please contact us with perinatal topics you would like to see addressed.*

*For a copy of our newsletter or to be placed on our mailing list contact us by phone or e-mail.*

Please see below the NYMC neonatal web site address to locate other issues of The Gazette:

<http://www.nymc.edu/depthome/peds/neonatology/index.htm>

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