

# CONSULTS

NEWS FOR PHYSICIANS FROM THE UCONN HEALTH CENTER

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## NEW SCREENING TECHNIQUES INCREASE EARLY DETECTION OF BIRTH DEFECTS

Nearly 90 percent of Down's syndrome cases can be identified before birth, thanks to major advances in screening methods. The combined use of blood tests and ultrasound during the first and second trimesters can detect approximately 90 percent of Down's syndrome cases with a very low (3 percent) false positive rate, according to Peter Benn, Ph.D., who has done extensive research on the screening tests and is a professor of genetics and developmental biology at the UConn Health Center. "We have already demonstrated our ability to halve the number of invasive procedures while increasing the number of Down's syndrome fetuses detected antenatally," says Dr. Benn. The new screenings include:

- two biochemical tests (PAPP-A and hCG) and an ultrasound examination of the fluid accumulation behind the neck of the developing baby, called nuchal translucency, in the first trimester.
- AFP, hCG, uE3 and INHA blood tests in the second trimester.



Winston Campbell, M.D.

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## SYMPTOMS OF PULMONARY ARTERIAL HYPERTENSION MAKE DIAGNOSIS DIFFICULT

Symptoms of pulmonary arterial hypertension (PAH)—breathlessness, dizziness, fainting, chest pain, palpitations and fatigue—are similar to those of many other disorders. "That's why PAH can be a difficult disease to diagnose," says Raymond Foley, D.O., F.C.C.P., medical director of the Pulmonary Hypertension Program at the UConn Health Center and an assistant professor in the center's Division of Pulmonary and Critical Care Medicine. "The difficulty diagnosing the condition can delay treatment. One of our goals is to increase awareness of this disease in the medical community."



Raymond Foley,  
D.O., F.C.C.P.

The diagnosis of PAH relies on a variety of tests, including chest radiographs, pulmonary function tests, EKGs, echocardiograms, lung scans and ultimately cardiac catheterization. "The goal is to determine the precise pressure in the pulmonary arterial circulation and how well the heart is accommodating the increased resistance to blood flow through the lungs," says Dr. Foley, whose clinic is the only one in the state to offer comprehensive testing and care for PAH.

In patients with PAH, blood vessels have stiffened and are less flexible, and the right

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## CONTACT US

Want to learn more about the UConn Health Center?

**Please contact:**  
**Rick Daddario**  
**Physician Outreach**  
**Manager**  
**UConn Health Center**  
**860-679-1695 or**  
**1-877-676-1733**



Rick Daddario

# RECONSTRUCTION IS PART OF VICTORY FOR CANCER PATIENTS

After the shock of being told they have cancer, most patients have two questions: First, “Am I going to survive?” and second, “How am I going to get on with my life?”

Rajiv Chandawarkar, M.D., a plastic surgeon with the Carole and Ray Neag Comprehensive Cancer Center, uses powerful optics and other microsurgery tools to help answer the second question. “In many instances, we can restore form and function to a patient who has had significant tissue removed during cancer surgery,” says Dr. Chandawarkar. Using microsurgery, sutures half the diameter of a strand of human hair are used to stitch together thread-thin vessels that maintain the blood supply to transplanted tissue.

“We can rebuild breasts disfigured by mastectomy by using implants or by transplanting skin and fat from the lower abdomen to the chest. Whenever possible, using one’s own tissue is a great improvement over the use of implants to reconstruct breasts,” says Dr. Chandawarkar. “When we use a person’s own tissue for reconstruction, it ages as she ages, and it looks and feels more natural.”

Microsurgery is used to reconstruct other areas of the body as well, for example, in patients with head and neck cancer where loss of the jaw, tongue or the

throat can be functionally crippling as well as cosmetically destructive.

Dr. Chandawarkar recently helped reconstruct a man’s pharynx with an elliptical piece of skin removed from his thigh. Surgery to remove cancer from the pharynx left a gaping hole (12 cm. long and 8 cm. wide) in the patient’s gullet. “I was able to repair the gullet so he can eat and drink and even feel comfortable going out to a restaurant,” says Dr. Chandawarkar. “The reconstructive surgery left him with a thin, straight scar on his thigh which is not a bad thing for a 55-year-old man.

“When we treat someone who has cancer, we have to be concerned with more than basic survival. To really achieve victory for patients, we have to help them get their lives back as fully as possible. That, for me, is where the real satisfaction lies.”

**For more information about Dr. Chandawarkar’s services, call our Physician’s Referral Line at 1-877-676-1733. To speak with Dr. Chandawarkar, call 860-679-4444.**



Rajiv Chandawarkar, M.D.

## MOHS SURGERY FOR SKIN CANCER



James Whalen, M.D.

For the 100,000 Americans affected every year by basal and squamous cell carcinomas, Mohs surgery offers the most precise treatment, the highest cure rate and the lowest chance of relapse. “Mohs surgery has been shown to be a highly effective treatment for these types of skin cancers, with a cure rate of up to 99 percent,” says James Whalen, M.D., a board-certified Mohs surgeon and associate professor of dermatology at the UConn Health Center.

Mohs surgery is usually indicated for skin cancers that have recurred, for those with a high risk for recurrence or for cancers on the face or other areas in which maximal tissue preservation is critical for cosmetic or functional purposes.

The procedure removes the visible portion of

the tumor along with another thin layer of skin, which is examined microscopically for evidence of cancer. If cancer cells are found, another layer of tissue is removed from the area. The process continues until there is no evidence of remaining cancer. “The technique ensures we get all the cancer while minimizing removal of healthy tissue and scarring,” says Dr. Whalen.

The surgery is typically performed as an outpatient procedure using local anesthesia. Most cases require removing three or fewer layers and take less than four hours.

**For more information about Mohs surgery at the UConn Health Center, call the Dermatology Department at 860-679-4600. To speak with Dr. Whalen, call 860-679-1718.**

# GENETIC SCREENING YIELDS DIAGNOSIS AND TREATMENT STRATEGIES



Robert Greenstein, M.D.

Dramatic advances in molecular biology and genetics technology make it possible to examine the DNA of the general population for mutations that cause disease. Information provided by this type of genetic screening can provide information crucial to early detection and treatment of hereditary diseases.

“We can now screen the adult population for a variety of adult-onset diseases,” says Robert Greenstein, M.D., professor of genetics and director of the Division of Human Genetics at the UConn Health Center. “We can identify people susceptible to a particular disease like breast or colon cancer or hemochromatosis before they have symptoms and complications.

This technology also may be used for cardiomyopathies and certain types of cardiac arrhythmias, Parkinson’s disease and certain types of diabetes.

“We have long understood there was a hereditary component to many diseases, and we are beginning to take advantage of new DNA technology to identify those individuals at increased risk for developing disease when they are pre-symptomatic. This information may then be incorporated into their medical care.”

**For more information about genetic screening tests, call the Human Genetics Clinic at 860-523-6464.**

## NEW SCREENING TECHNIQUES INCREASE EARLY DETECTION OF BIRTH DEFECTS

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“The UConn Health Center is the only facility in the state that can combine the results of both the first and second trimester screenings,” says Dr. Benn. “Because the analysis involves seven different pieces of information, which are not completely independent of each other, the mathematical calculations are extremely complex. The complexity currently puts the analysis out of reach of most small laboratories.”

“Because of the high accuracy of these screening tests when used together, normal results can provide considerable reassurance to women and their partners about the health of their developing baby,” says Winston Campbell, M.D., director of the Division of Maternal-Fetal Medicine at the UConn Health Center.

While the combined first and second trimester screening results are the gold standard in terms of accuracy, good results also can be obtained with the first trimester tests alone. This screening includes the blood test and the nuchal translucency ultrasound. “It does not provide the definitive diagnosis achieved through amniocentesis, but it’s helpful to many women because it can be performed early in pregnancy and allows many low-risk women to avoid the 0.5 percent chance of miscarriage associated

with amniocentesis,” says Dr. Benn. Another diagnostic test, chorionic villi sampling, can be done in the first trimester but it carries a higher risk, about 1 percent chance for miscarriage.

“The American College of Obstetricians and Gynecologists has recommended non-invasive first trimester screening tests be offered to women at risk for a genetic disorder,” says Dr. Campbell.

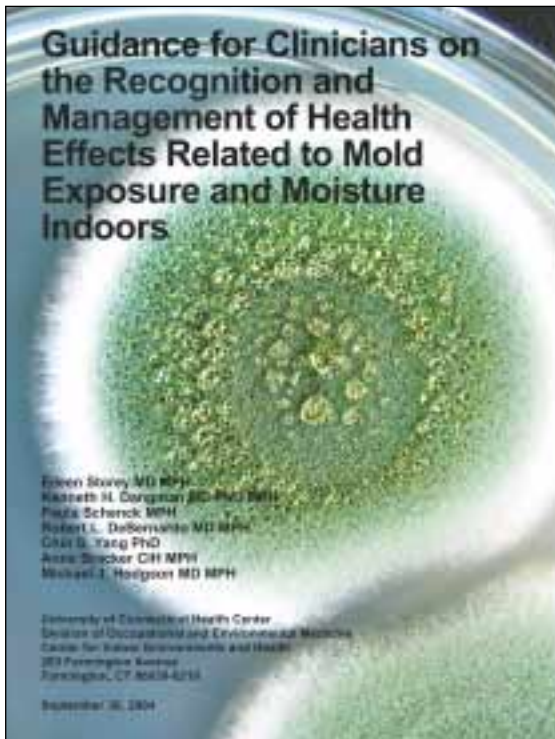
First trimester screening generates either a screen-negative or a screen-positive result. A screen-negative result is obtained when there is a normal ultrasound examination and the blood test shows concentrations of hCG and PAPP-A within expected ranges. The result indicates there is not enough risk for a birth defect for the woman to consider additional immediate follow-up testing.

A screen-positive result indicates that some consideration should be given to follow-up testing to help determine whether an abnormality is actually present. Women who receive a screen-positive result may consider chorionic villi sampling or, during the second trimester, additional serum screening tests, another ultrasound evaluation or amniocentesis.

**For more information about first trimester screening tests, call the Maternal-Fetal Medicine Ultrasound Unit at 860-679-4363 or the Human Genetics Clinic at 860-523-6464.**

**Because of the high accuracy of these screening tests when used together, normal results can provide considerable reassurance to women and their partners about the health of their developing baby.**

# HEALTH CENTER PUBLISHES MANUAL FOR PHYSICIANS ON BUILDING-RELATED ILLNESSES



From sinusitis to asthma and pneumonitis, serious illness as a result of poor indoor air quality has been well documented in recent years. Less clearly established are the specific causes of building-related illnesses and the measures required to prevent or ameliorate them.

For physicians and other health care providers faced with a rapidly growing number of patients troubled by indoor contaminants, there is a new manual to guide them, entitled *Guidance for Clinicians on the Recognition and Management of Health Effects Related to Mold Exposure and Moisture Indoors*,

published by the Center for Indoor Environments and Health at the UConn Health Center with a grant from the Environmental Protection Agency.

“It’s a manual about illnesses related to mold and indoor moisture for primary care physicians and other health care providers,” says Eileen Storey, M.D., M.P.H., director of the Division of Occupational and Environmental Medicine and one of the authors of the book. “We provide an approach to use with patients. We help physicians and other providers identify patients and the

illnesses or complaints that may be related to mold or other indoor contaminants. We give providers assessment tools for those patients. We tell them how to counsel their worried well patients and guide them to resources their patients can use to reduce moisture and mold in their homes.”

“We know that exposure to mold and other contaminants in indoor environments may adversely affect a person’s overall health and well-being,” says Paula Schenck, M.P.H., another author of the book. “We want providers to ask their patients about environmental issues that might be related to their illnesses and be able to provide counseling about them.”

**“We help physicians and other providers identify patients and the illnesses or complaints that may be related to mold or other indoor contaminants.”**

“It’s like tobacco. Thirty years ago, doctors did not think of tobacco as an issue for them in their practice. Now it’s standard health care practice for physicians to ask their patients about tobacco use and provide counseling for it. We hope this book will do the same thing for indoor air quality,” says Dr. Storey.

For information about *Guidance for Clinicians on the Recognition and Management of Health Effects Related to Mold Exposure and Moisture Indoors*, go online to the Center for Indoor Environments and Health at [www.oehc.uconn.edu/clinser/indoor.htm](http://www.oehc.uconn.edu/clinser/indoor.htm). To speak with Dr. Storey, call 860-679-2386.

## SYMPTOMS OF PULMONARY ARTERIAL HYPERTENSION MAKE DIAGNOSIS DIFFICULT *continued from page 1*

ventricle is less efficient at pumping blood through the pulmonary vasculature. As a result, patients develop exertional dyspnea, and simple tasks like climbing stairs become a challenge.

“There is no cure, but treatment can improve symptoms, exercise capacity, quality of life and survival,” says Dr. Foley.

**For more information about the Pulmonary Hypertension Program, please call 860-679-3343.**

### SYMPTOMS OF PULMONARY ARTERIAL HYPERTENSION

- breathlessness
- racing pulse
- dizziness
- fatigue
- fainting spells
- swelling in ankles or legs
- chest pain
- bluish lips and skin
- palpitations



Remarkable Care Through  
Research and Education