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News Briefs

FDA Approves HDE for Rare Disorder of Placenta

The Food and Drug Administration (FDA) has approved a Humanitarian Device Exemption (HDE) aimed at helping doctors provide treatment during pregnancy for fetuses affected by twin-to-twin transfusion syndrome. This syndrome sometimes occurs when women are pregnant with identical twins. It is a rare disease of the placenta.

As identical twin fetuses develop, there are often blood vessels in their shared placenta that connect the fetuses' circulating blood. In most cases, the blood flows properly through these vessels. However, in TTS the blood flow is uneven, with one fetus receiving too much blood and one receiving too little.

This can cause heart failure in the twin receiving too much blood and life-threatening anemia in the one receiving too little. Many of these babies do not survive delivery or are born with severe handicaps.

HDEs are applications for approval of medical devices being developed for conditions that affect fewer than 4,000 Americans per year. Sponsoring companies must demonstrate the safety and probable benefit of such devices.

In this case, the device, known as Fetoscopy Instrument Sets, is intended to be used for the treatment of TTTS for fetuses between the 16th and 26th month of pregnancy. They are intended to help doctors identify the communicating blood vessels and normalize the blood flow. The devices are distributed by Karl Storz Endoscopy America, Inc., of Culver City, Calif.

University of Washington Scientists Develop Newborn Screening Technique

Scientists at the University of Washington have developed a relatively simple technique for detecting enzyme deficiencies in newborns so that treatment may begin before too much damage has been done. The researchers say the technique may be used for a variety of rare diseases, including Tay-Sachs, Fabry and Gaucher diseases.

Damage from these diseases is permanent, but if enzyme replacement therapy is available and is begun early, the damage may be reduced or minimized.

The scientists reported on their research at a recent meeting of the American Chemical Society. They said their screening method has been used successfully to date in treating seven

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diseases - Krabbe, Pompe, Niemann-Pick, Gaucher, Fabry, Tay-Sachs, and Hurler syndromes - associated with enzyme deficiencies. These diseases are among the group known as "lysosomal storage disorders"; because the enzyme deficiencies occur within the structures known as lysosomes that break down large molecules within cells.

Typically, when one of these enzyme deficiencies is present, the baby may appear healthy at first but begin to show signs of disease over time, as waste material accumulates within the cells. The worst of these diseases can cause devastating effects, including mental retardation, or can be life-threatening.

The screening technique uses a spot of blood drawn from a baby's heel (samples are routinely taken for other tests now) and tandem mass spectrometry. The sample can be screened for several enzyme deficiencies at the same time.

CFC Genes Identified With Help of Patient Organization

A genetic support group of approximately 100 families from the United States and abroad collaborated with researchers at the University of California, San Francisco, on research that has identified three genes associated with a rare condition known as cardio-facio-cutaneous (CFC) syndrome. Their work should make possible a better understanding of this complex developmental disorder. The findings have been reported in the online version of the journal, *Science* (January 26, 2006).

Since 2003, the families of CFC International have collected medical reports, photographs and the DNA of family members diagnosed with the condition. The DNA was collected through a biobank established by the patient organization.

The information and DNA samples allowed Dr. Katherine A. Rauen and her colleagues at the UCSF to identify three genes - known as BRAF, MEK1 and MEK2 - that play a role in the development of this disorder. By studying the factors that cause the disorder to occur, the researchers hope ultimately to develop possible treatment options.

At the present time, there is no therapy for CFC other than treatment aimed at specific conditions associated with the syndrome. CFC is characterized by unusually sparse, brittle, curly hair; skin abnormalities; heart malformations that are present at birth or develop later; growth delays; and/or varying degrees of mental retardation.

Saving Andy Martin's Cells



Andy Martin (left) and Tyler Curiel, MD, MPH (PSurch/Tulane Publications)

In 2004, a medical student at Tulane University in New Orleans named Andrew Martin helped NORD write a report for its Rare Disease Database on a rare form of cancer known as sinonasal undifferentiated carcinoma (SNUC). He wanted to increase awareness of this little-known disease.

Today, that report stands as a

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reminder of the dedication of this young researcher and of his colleagues' heroic efforts to salvage his work in the aftermath of Hurricane Katrina.

Andy Martin - in addition to being a third-year medical student - was an oncology patient. He suffered from the same rare cancer that he was studying - SNUC - which affects the nasal cavity and nearby sinuses. Its first symptoms are a bloody nose, runny nose, bulging eye, or chronic infection. In some cases, SNUC has been associated with prior irradiation for other cancers, but it has also appeared in people who have had no previous irradiation.

Because SNUC is extremely rare, there has been little research on it. Its treatment has no firmly established protocol but relies, instead, on therapies used for other cancers. At Tulane's Cancer Center, Andy Martin had donated tissues from his own tumor for research that, he hoped, might lead to a treatment for SNUC. He and several colleagues were working under the direction of Tyler Curiel, MD, MPH, chief of hematology and medical oncology.

About six months after Andy co-authored the report for NORD, he succumbed to his disease. However, the tissues he had donated still provided hope that others with SNUC might have a brighter future.

Then, in August 2005, Hurricane Katrina roared across the Tulane campus. In the stark aftermath, with destruction all around them, Dr. Curiel and another cancer researcher, Dr. Michael Brumlik, set out to save Andy's cells... and Tulane's SNUC research program. Because there was no power in their building, they decided to move the freezers in which Andy's cells were stored to a nearby building that had working generators.

Pulitzer Prize-winning journalist Amy Dockser Marcus described their efforts as follows in a *Wall Street Journal* article:

"The freezers were huge," weighing as much as three or four kitchen refrigerators. "The two started pushing the first one down the hall, trying to get it into the elevator before the emergency power in their building went out. It was dark in the hallway, with the temperature above 100 degrees. They worked by flashlight."

When they got the first freezer to the other building, it was too big to fit through the door so they carried the vials individually, stuffing them into freezers in other labs. They carried hundreds of boxes of vials, up and down stairways, in the dark aftermath of the storm.

A few hours later, the generators in the second building failed. Again, Drs. Curiel and Brumlik carried boxes of vials through the darkened passageways, this time to an area where there were three liquid nitrogen tanks that, they hoped, would keep the samples at the required temperature for a few weeks without power. Then, National Guardsmen told them they had to leave.

As soon as he was out of New Orleans, Dr. Curiel began to work on getting back in, to save Andy's cells. Others rallied behind him: The University of Texas Southwestern Medical Center donated 800 pounds of dry ice. Trident Aviation Services donated the use of its corporate jet, and Fisher Scientific International provided insulated containers.

Eventually, Drs. Curiel and Brumlik were taken by the Trident jet to New Orleans and by helicopter to the roof of a hospital. At the medical school, they were told by guards that they could stay for four hours.

"Working in the dark," Marcus wrote in her Wall Street Journal article, "they went back to the three liquid nitrogen tanks. Shining their flashlights on the tanks, they popped the top on the first one. A big puff of white vapor hit them in the face. Dr. Curiel reached in and pulled out a rack containing boxes of vials." The very first box was labeled "SNUC".

Andy Martin's cells were placed in donated cooler space at UT Southwestern and Baylor University College of Medicine but recently returned to Tulane. For now, at least, Tulane's research on this very rare cancer has been saved.

"We still face many challenges," said Susan Sarver, RN, who worked with Andy at Tulane, "but we're very grateful for what Dr. Curiel did, and we're determined to keep this research going."

For more information about SNUC research at Tulane, call Susan Sarver, RN, at (504) 988-8840 or write to bounce@tulane.edu.

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