The Latest in Health

Experimental Drug Shows Promise In Treating Cystic Fibrosis

LOS ANGELES (MCT) – All her life, Lindsay Shipp knew that she was dying. As a baby, she would cry after eating, and salt collected on her forehead. The diagnosis was cystic fibrosis, an incurable genetic disease that, at the time, meant a life expectancy of 18 years.

The disease, which affects 30,000 people in the United States, hinders the movement of salt water in their body. Because of this, the pancreas fails soon after birth, patients cannot properly digest food and their airways fill with mucus, leaving them vulnerable to lung infections and other problems. The current average life expectancy is 37.

"I had this race-against-time mentality where I did everything I could possibly do in a day," said Shipp. "I never let a moment go to waste."

But since she started taking an experimental drug as part of a closely watched clinical trial, her outlook has changed. The drug, called ivacaftor, cleared her lungs and allowed her to add a healthy 15 pounds to her 5-foot, 100-pound frame. On the day in February of last year when a regular check-up revealed her lungs were functioning at 96 percent — bringing her to essentially the same level as a healthy person — Shipp collapsed on a bench outside the hospital and sobbed with relief. "I realized I would have a full life at that moment," she said.

The trial results, published in Thursday's edition of the *New England Journal of Medicine*, showed that ivacaftor reduced the incidence of pulmonary exacerbations — sudden, serious flare-ups that can send patients to the hospital and cause irreversible lung damage — by 55 percent compared to a placebo. The study of 161 patients also found that patients who took the drug saw their lung function improve 10.4 percent and gained nearly 6 pounds over the course of 48 weeks.

"This is exciting work," said Dr. Michael Welsh, a pulmonary physician at the University of Iowa who was not involved in the study. "I think that it is very promising for the people who have this particular mutation and it may turn out to have broader significance."

Ivacaftor affects a genetic mutation in 4 percent to 5 percent of cystic fibrosis patients that prevents a protein called CFTR from allowing enough chloride ions to exit a certain type of cell. Chloride, when coupled with sodium to make salt, helps regulate the flow of water in the body. When that channel doesn't work, the lungs dry up and the once-protective mucus grows thick and immovable, providing an ideal breeding ground for bacteria.

This saltwater imbalance affects the pancreas and digestive tract, so patients are often small and malnourished. The sweat glands don't work properly either, leading to overly salty skin.

The drug helps that chloride gateway to stay open, restoring the saltwater balance in the body. It is the first therapy that fixes the basic problem that causes the disease, not just the symptoms, said study coauthor Dr. Michael Konstan, a pediatric pulmonologist at Case Western Reserve University School of Medicine.

"The goal is if you would give a therapy like this before [the lungs deteriorate], you can prevent lung disease from developing," Konstan said.

The drug can't yet help the vast majority of cystic fibrosis sufferers — around 90 percent of them have a mutation in which the protein, once manufactured inside the cell, never even makes it to the cell surface where it is supposed to act as a chloride channel. Researchers are trying to develop drugs that would help bring that protein to the surface. Once a patient is on such a drug, ivacaftor could then pick up the process from there.

"It's creating such a sense of hope and optimism for all the cystic fibrosis patients because this approach will work," said Robert Beall, president and chief executive of the Bethesda, Md.-based Cystic Fibrosis Foundation, which has spent millions funding such research.

Although therapies to fight the disease seemed to be around the corner when the CFTR gene was discovered in 1989, it took more than two decades to develop a treatment based on the genetic defect.

"This timeline suggests that much of the promise of the Human Genome Project has yet to be fulfilled and that realizing the therapeutic benefits will take persistence and determination," Dr. Pamela Davis of Case Western Reserve University School of Medicine wrote in an editorial accompanying the study. "Society cannot allow support for research and development to be compromised in the current rush to cut the federal budget."

If all goes smoothly, ivacaftor may be approved by the Food and Drug Administration in 2012.

The drug is not an absolute cure. It cannot rid the body of bacterial infections that stick around even after mucus has been cleared from the lungs. But perhaps, if given to patients that were young enough — say, newborns — it could allow them to live an essentially normal life.

"It's like there's someone else's lungs inside of me," Shipp said. "It's awesome — I feel like I can do anything."

Wall Street Fights Autism

BY KIM ROBINSON

On November 8, more than fifty leading philanthropists gathered in the Wall Street boardroom of investment bank DH Blair to hear Jerusalem Mayor Nir Barkat discuss the urgent need to support the creation of the world's first Global Center for Autism Research and Education on Mt. Scopus.

The Global Autism Center is a project of ICare4Autism, the International Center for Autism Research and Education, a New York-based charitable organization founded in 2004 by Dr. Joshua Weinstein to tackle the global autism crisis by promoting breakthrough innovations in research, diagnosis and treatment.

When it opens in 2014, the center's five-acre campus will feature a state-of-the-art on-site research institute; the world's first university-level school of autism studies; an educational center applying the latest research and technology to the special needs of students with autism; and a foundation to support the transformation of autism education and treatment worldwide.

In addition to these program plans, Dr. Joshua Weinstein, founder and CEO of ICare4Autism, announced an important new initiative: the world's first Comprehensive Autism Workforce Development Center. The Workforce Development Center will provide preparation for workforce entry; vocational training and employment services in more than a dozen fields, ranging from semi-skilled to the high-tech sector; specialized support to persons with Asperger's syndrome; and ongoing job coaching and mentoring.

Dr. Eric Hollander, the chairman of the advisory committee for ICare4Autism, explained why the Global Autism Center is so important now. "The Global Autism Center will bring together the large-scale data and collaborative networks urgently needed to drive breakthrough genetic and epigenetic autism research," he stated. "These networks do not yet exist, and no other organization is building them. It will create the world's first integrated platform for cross-disciplinary collaboration between biomedical researchers and educators. The creation of such a platform is critical to global efforts to foster early detection and effective intervention.

"And finally, having the Global Autism Center in Jerusalem will have a powerful and transformative impact on the Israeli autism community, bringing the world's leading researchers into close collaboration with their Israeli colleagues and rapidly bringing their most important advances to Israelis with autism."

Mayor Barkat said, "The Global Autism Center is of vital importance for the city of Jerusalem and the world. We are pleased and honored to help bring the world's leading scientific and educational experts together to spark the innovations that will transform the lives of millions of people with autism and their families."

Said host J. Morton Davis, DH Blair founder and chairman, "I am proud to support the Global Autism Center and grateful to ICare4Autism for bringing this center of excellence to Jerusalem, where it will transform the lives of so many people."

Event co-chair John Catsimatidis, owner and CEO of Red Apple Group and Gristedes Foods, applauded the Global Autism Center as "an example of international collaboration that is worthy of our support because it will help to permanently change the world for the better."

For information about ICare4Autism's Global Autism Center in Jerusalem, please contact krobinson@Icare4autism.org.

Jerusalem Mayor Nir Barkat discusses the ICare4Autism Global Autism Center at the ICare4Autism breakfast. L-R: Dr. Joshua Weinstein, founder and CEO of ICare4Autism; Mayor Barkat, speaking; host J. Morton Davis, DH Blair founder and chairman.