Almost alone

Jefferson County man is one of the few with Erdheim-Chester Disease

By Peggy Scott
For the Leader

A year ago, Ken Els of Hillsboro had no idea he was a member of an exclusive group – people who have been diagnosed with Erdheim-Chester Disease, a mysterious and rare ailment that causes fibrous growths around vital organs.

He's one of just 350 people diagnosed worldwide with the disease since it was first identified in 1930.

The disease is so rare it isn't included in standard medical school textbooks and few doctors have ever heard of it.

But after more than 100 X-rays, four CT scans and an MRI of Els' heart and brain, doctors at Washington University School of Medicine figured out what was causing his unusual symptoms.

For Els, 67, the mystery started in winter 2010, when he was diagnosed with a touch of pneumonia. He went back to his family doctor to get a scan of his lungs to make sure the pneumonia was gone.

The radiologist said indeed the infection was gone, but the scan showed something unusual around his heart and his kidneys.

Another CT scan was scheduled, which showed fibrous growths around his kidneys, adrenal glands and arteries to his heart. In addition, doctors found bone lesions on his legs and spine.

Those discoveries led to a full-body scan, which turned up more lesions and raised more questions. In the meantime, Els suffered intermittent pain and aching in his joints.

Els said his local doctor decided to send him to see a specialist. Like an episode of the television show "House," the Washington University medical team came together at Barnes Hospital to try to figure out what was going on.

"They did more tests. At one time, there were five doctors in my room," said Els, who was hospitalized at Barnes-Jewish Hospital in St. Louis while doctors tried to find the source of his pain.

Eureka!

Among the team was Dr. Lesley Davila, a rheumatology fellow at Washington University in St. Louis. She was on her way home for the weekend when she got a call asked for a consultation on Els' case.

"I remember being a little annoyed that they called at 5 p.m. on a Friday," she said.

But her annoyance was quickly replaced with curiosity as she read Els' medical history and looked at his test results and scans.

As a rheumatologist, Davila treats patients who have arthritis and lupus. She also is studying people with a condition called retroperitoneal fibrosis, a disease that includes fibrous tissue growing in the abdomen.

Gordon Bess photo

Ken Els with some of the research he has collected about Erdheim-Chester disease.

"He had all these weird findings, and they thought he might have retroperitoneal fibrosis," Davila said.

That Friday night, a number of doctors pored over Els' test results, Davila said. At one point, a radiologist pointed out a strange finding on a CT scan of Els' belly.

"The limits of the scan went to the mid-thigh," Davila said. "In the bone were some strange findings. The radiologist said we should consider something called Erdheim-Chester Disease. I had never heard of it."

She spent the weekend reading everything she could find about ECD. "The more I read about the disease, he had almost everything. Wow, this really fits."

What is it?

Els and his doctors began a crash course to learn as much as possible about the rare condition.

The disease was first described in 1930 by Austrian pathologist Jakob Erdheim and American pathologist William Chester. In the 80 years since, 350 cases have been reported worldwide.

Currently, there are 69 living patients reported worldwide; most are in the United States. Els is one of four ECD patients reported in Missouri.

The disease is tough to diagnose, Davila said, in part because it can affect many different organs in the body. As a result, patients don't exhibit the same symptoms.

"It's fairly complex," she said. "Usually, people get aches and pains in their arms and legs. But that could be from thousands of different things. Some will get a rash. Some get these little yellow patches on each side of the nose. But those can occur for other reasons."

The disease can attack the heart, the lungs or the eyes.

Davila said diagnosing the disease is just the first step. Doctors are struggling to determine what causes it and how to treat it.

"It's really unclear what's causing it, what's driving it," she said. "Because it's so rare, there's not really that much research going on."

The St. Louis team suggested Els go to MD Anderson in Houston, Texas, a medical facility that has treated several cases of ECD. He discussed his case with doctors there, but didn't stay.

"But I decided to come back to Barnes," Els said. "We'll figure this out. I didn't get it overnight. I won't get better overnight."

In fact, Els thinks he's had symptoms over many years that may have been linked to ECD.

Four years ago, when Els broke his knee, his doctor saw lesions on the bone usually associated with cancer. No cancer was found. The broken knee healed, and Els was thankful that he was cancer-free.

However, bone lesions may have been a sign of ECD.

Two years ago, Els had a mild heart attack that his cardiologist said seemed a little unusual. A month after the attack, Els suffered congestive heart failure.

"I got a couple of stints out of the deal," he said. But now he knows he has fibrous ECD tumors around his heart that may have played a role in the "unusual" heart findings.

In 1985, he was diagnosed with myasthenia gravis, an autoimmune disease that leads to muscle weakness and fatigue. He was diagnosed after sudden onset of double vision.

Over time, his vision improved, but now Els wonders if that, too, could have been a symptom of ECD.

Finding help

Els said about 60 percent of people who have the disease die within 2 1/2 years of being diagnosed.

"But some live for decades," he said. "There is higher risk if the brain or heart or lungs are involved."

Els said his heart is involved.

"But they don't know what to expect," he said.

Davila said answers are hard to find in large part because the pool of patients is so small.

Els and his doctors discovered the ECD Global Alliance, a group of ECD patients and caregivers around the world connected via the Internet.

The Global Alliance recently secured a $50,000 grant for the first ECD study and chose a doctor in Italy to lead the research.

Els said the alliance has pledged to help educate doctors and patients in hopes of identifying additional ECD patients for the study.

The goal is to determine a cause and a treatment. In addition, he said, the study should help raise awareness.

Information is mostly anecdotal. People post treatments that seem to be working, and those that aren't.

As research is conducted, Davila said diagnosis of ECD may become somewhat more common.

In addition, she said, medical imaging has improved over the past decade.

"We are picking up on things even though they are not causing symptoms," she said.

But, Davila said, the disease remains rare. She said Els may be the only patient with ECD she encounters in her professional career, but she's not ruling out the possibility of others.

Els and his wife of 45 years, Kathy, have three daughters and seven grandchildren.

Currently, Els' symptoms allow him to live a pretty normal life.

"I'm lucky because I'm still able to get out and go," he said. "I'm really fortunate and thankful for that."

At present, doctors are stymied about treatment options.

Els said he has taken interferon, a drug that boosts immunity, and is considering another drug that is typically used to fight cancer. But he said he wants to pursue a cautious approach to battle his ECD since he still has a good quality of life.

For information, visit the ECD Global Alliance Web site erdheim-chester.org.