

CLARION-LEDGER TUESDAY, AUGUST 31, 2004

A SPECIAL REPORT
LAST PART OF A 10-DAY SERIES

GENETIC DISASTER

A STORY OF FATE, FAITH AND FAMILY



Disease: I return to California to see my aunt for the last time

From 1A
below — ubiquity, which marks defective proteins for destruction. Nucleus can live without VCP, yet the gene has hardly been researched. What has been researched shows disrupting VCP leads to inclusion body formation, something obviously connected to the family disease.

Giles hands me a copy of the report he's writing. It shows that of the families studied, 82 percent had included body sympathetic, 42 percent had. Paper's and 30 percent had early onset of dementia that affected the frontotemporal lobes of the brain. He suspects this mutation could be causing VCP to act in brain ways. By finding pathways affected by this faulty gene, researchers will better understand how the brain, bone and muscles relate to each other at the genetic level, he says.

By getting such insight, researchers may only find answers for the disease that haunts my family. They may also find clues to help solve a range of diseases from Alzheimer's to Parkinson's.

Dr. Virginia Kinross enters and talks about what a devastating disease this is, crippling so many, stripping away body and mind. I flip to the part of the report that lists the family trees for each family, including mine, Family 3. There are many listed, but I'm soon able to figure out the segment that represents Daddy Sam. The darkened upper right corner shows her last inclusion body myopathy. The darkened lower right corner shows her last inclusion body myopathy. The darkened lower right corner shows her last inclusion body myopathy.

I turn to Dr. Kinross. "That means my Dad doesn't have the disease and neither do I," I say. She nods. "That's what children can't possibly get the disease," I say. I hug her and thank her for all she's done to help defeat this disease. For the first time in more than a century, my family has hope.

I call Aunt K.K.'s partner, Marilyn Fitzgerald, and tell her the gene for the monster had been found. There is silence, then a tearful voice. "That's a major, major finding," she says. "That's the biggest thing they've done so far."

She smells K.K.'s scent, to be tested genetically for Huntington's disease, as well as aggregating neurodegenerative movement and emotional diseases. Results came back negative as they had for so many times before. For the first time since K.K. found Moody on her pillow 13 years ago, she is able to prove what she always believed — her illness is

caused by a faulty gene. **BOSTON** — I sat with Dr. Ruth Tanzi, one of the world's most prominent researchers on Alzheimer's.

He says the region Dr. Kinross has been working on is the same region linked to families suffering from Alzheimer's. That same region, he says, also includes those suffering from Lewy's disease and Huntington's disease, which is similar to PK's — the same disease my cousin Margaret has been diagnosed with.

"So here we have three different diseases that are mapping to the same region of the chromosome," he says. "My aunt, who didn't take hormones, developed early onset of dementia at 46, but her first cousin, who did take hormones, didn't get dementia until she was 51." I say, "Perhaps the hormones helped save of the disease."

Recent says epidemiologists originally suggested hormones, but trials have not proven to be effective. "If you had a couple of epidemiologists, they would see there's a correlation between hormone replacement therapy and protection, then there must be something to it," he says. "I think the evidence is more against it right now, but in science it's just a matter of time to be proven."

Afterward, I talk with Giles about the idea of hormones possibly serving as a trigger for the genetic time bomb. "From what we can tell, there is no difference in male and female onset. So some degree, that would rule out hormones," he says. "That then leaves us with many women with dementia than men."

Nature Genetics publishes the research of Dr. Kinross and others in the Age-Related, dementia, and Huntington's section. The work, she has Harvard resume her lab the Kinross Laboratory, and the National Institutes of Health have awarded her a one-year grant of \$184,000 a year. She has a genetic counselor, had to have worked in Europe and has headed her research to the genetic disease of Angelman's syndrome, which only children of speech and mental functioning.

My 11-year-old cousin Ryan loves and his wife are expecting a child in February. He was named in my will. He has a 50-50 chance of having the disease — the same odds his child faces should he come down with the disease. But he thinks time on his child's side. "That's 40 or 50 years down the road," he says. "Hopefully, we'll be able to prove what kind of genetic treatment by that time."



Marilyn Fitzgerald watches Mary Fitzgerald look at K.K. Mary, who has Huntington's, sat on a 1964 striped bench for when she suffered from psychosis in 1964.

had a second test thankfully proved the first one false. "Scared the pants off me," she says. She remembers how devastating her grandfather and mother became before they died and thinks about the monster who may attack her at any time. "It's scary," she says. "It's very scary."

Months, I continue my search for my family's past. When I ask researcher Giles about the ancestry connected with the disease, he says none of the 10 families has been able to trace its ancestry across the ocean, although all have links to Western Europe. "We have now found three families in mainland Europe with mutations in VCP."

Those families, he says, live in Poland and Germany, a country that matches up with the ancestry of my Illinois cousin Roma, because we're both of ancestry would also match up with mine.

Dr. Kinross and her researchers became the first to find a disease linked to VCP. "It's going to open up a whole industry of research connected to VCP because it's an important mechanism being disrupted," she says.

She wishes as Mary Hilton, who has been one of my aunt since 1994, looks K.K., counting her and seeing how getting her side to which, Marilyn thinks back to 1964, when she and her mother visited K.K. in Huntington. When Marilyn visited, K.K. begged to return to California — and Marilyn quickly agreed.

She points to the wheelchair. "I was sitting in this, she would be taking care of me," she says. "I don't think you can say that about my people."



J.D. Schaefer/The Clarion-Ledger

EPISODE
Dad wonders the news he's dodged the family disease. That news makes him realize for the first time that the constant ache he feels in his head has nothing to do with the disease that robbed him of his father and is now stealing his sister.

A few months later, he goes to a chiropractor. After a couple of visits, he tells me treatments are helping. He puts my grandfather on his cell phone to talk with me. "Hello, darling," Mom says. "What are you doing?" "I'm writing about the family disease," I tell her. "I'm writing about the family disease."

Still unable to understand, she tells me my grandfather on his cell phone to talk with me. "Hello, darling," Mom says. "What are you doing?" "I'm writing about the family disease," I tell her. "I'm writing about the family disease."

Each day, my 16-year-old grand mother shares stories of my childhood with Dad. But she can no longer remember where he lives, no longer remember her family's traits, no longer remember her great-grandchild's names.

Perhaps the only blessing is she can no longer remember her daughter is dying of the family disease. "I tell her."

LOS MOLINOS, Calif. — My aunt steps to one side on her wheelchair, drool dripping from her mouth to a towel on her chest. K.K. doesn't know I'm here and not know when I leave. The monster is almost done with her, ready to move on to a new victim. This week passed, my aunt's fever shot up to nearly 103 degrees, but it finally returned again. "I was so worried," Marilyn says. "She's never had that kind of fever."

ABOUT THE TEAM
Since 1989, Jerry Mitchell, a 45-year-old neurologist, reports for The Clarion-Ledger, has focused attention on an orphaned illness from the South's past. Now he focuses on a sister that continues to attack his family, a rare genetic disease that has taken the lives of so many Mitchell grew up in Texas.

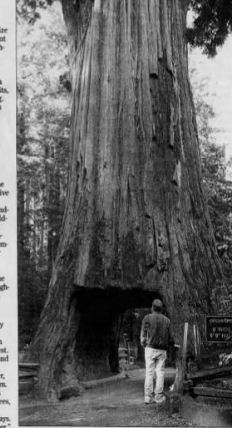
Since joining The Clarion-Ledger in 1984, J.D. Schaefer has created images that depict the lives and challenges of Mississippians. He passed his own after seeing the rapid deterioration of his mother, who is suffering from dementia. The 49-year-old photographer grew up in Indiana.

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stood in front of the Chandler Tree, which is 2 1/2 feet wide and 315 feet tall. It's one of the many redwoods in Northern California.

asked to lead on Pacific Coast Highway 1 in Northern California. As a redwood forest in Leggett, I longed to see the Chandler Tree, a tree so huge curly-oak seeders drive their cars through the hole carved out of the trunk.

I found made this 115-foot mammoth, walking through time. Hippocratic practices in medicine in Greece when this redwood was a sapling 21 centuries ago, developing an ethical oath physicians still swear today.

I can't help but wonder if the monster that attacked my family for generations has lived on this Earth as long as this tree. More importantly, I can't help but wonder how many more years it will take to rid the Earth of this monster. The early light of day dawns through the limbs, and I find myself praying for a cure for the family disease. A cure for K.K. and my cousin Margaret, a cure for my cousin Donna, who is now bedridden. I