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TSRHC Researchers Link First Gene to Scoliosis

After years of surgeries, braces and clinic visits, Judy Worthington knows firsthand how scoliosis, a condition that causes one's spine to curve into a "C" or "S" shape, affected her life and the lives of three generations of her family.

"All five siblings had scoliosis — my three sisters, my brother and I," said Judy, whose two daughters, son and a granddaughter were also diagnosed with the same condition. Judy's treatment for scoliosis as an adolescent included four surgeries and a one-year stint in a body cast, and as an adult, a curve-correcting spine implant system involving two surgeries.

Judy was treated by Texas Scottish Rite Hospital for Children's second chief of staff, Dr. Brandon Carrell, at the W. B. Carrell Memorial Clinic, and she knew that her family's medical history could be vital to TSRHC's research study to find the genes responsible for scoliosis.

She was right, and with the help of her family and more than 50 other families, TSRHC

"It has been known for many decades that scoliosis tends to be inherited within families, but now we have found the first gene that is clearly related to the development of scoliosis."

— DR. CAROL WISE

was able to identify the first gene associated with scoliosis. Thanks to Judy and her family, hundreds of other blood donors to the study and the research team at TSRHC, there is hope for a life free of scoliosis in the future.

"Scoliosis was just something to live with," said Helen Ray Nicholson, Judy's oldest sister.

"But when we met Dr. Carol Wise, that changed everything."

Through a study as part of the Sarah M. and Charles E. Seay/Martha and Pat Beard Center for Excellence in Spine Research, Dr. Wise, director of molecular genetics at the

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L to R: Judy Worthington with her daughter Kelley Worthington, her sister Helen Ray Nicholson, and her granddaughter Rebecca Headrick, three generations of women reunited with TSRHC's Dr. Carol Wise to celebrate the gene discovery.

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hospital and leader of the 10-year research study, and her team at TSRHC recently announced that they identified the first gene, CHD7, associated with idiopathic scoliosis. Idiopathic scoliosis, or IS, is the most common spinal deformity in children, and previously had no known cause and still no cure. This gene discovery gives the medical community clues to explain what causes the condition and provides tools for future research.

IS poses a significant health burden to children, as it has a prevalence of two to three percent of school-aged youth. Children who are still growing, particularly girls, are at the greatest risk of developing severe spinal curvature. IS often appears in family members, indicating a genetic cause.

"At the hospital, we have found families that are struggling with this condition, and it is occurring over and over again, in generation after generation, and in multiple siblings and other family members," said Dr. Wise.

The research team sought out families with a history of IS to participate in their study, and Judy and her family were among the first to sign up.

"We collected a sample of DNA from family members and analyzed the DNA to tell us where the gene is located," said Dr. Wise. "By looking at DNA from enough families, we can tell where the actual genetic change occurred."

The research team conducted genome-wide scans and follow-up studies of 53 families, totaling 130 individuals with a confirmed IS diagnosis. The findings were published in the May 2007 issue of the *American Journal of Human Genetics*.

"This is the most definitive link between genetics and scoliosis that has been reported so far," said Dr. Wise. "It has been known for many decades that scoliosis tends to



(Above) Gene discovery research team. L to R: Dr. Xiaochong Gao, Samuel Weber, Dr. Carol Wise, Nora Elrnoufy, Shorn Devroy and Dr. Dongping Zhang (Right) Dr. Xiaochong Gao and Dr. Carol Wise in the lab



be inherited within families, but now we have found a gene that is clearly related to the development of scoliosis."

Although more research is needed to make significant strides, according to Dr. Tony Herring, chief of staff at TSRHC, this discovery lays the groundwork for future research that will hopefully identify the specific abnormalities that cause the spine curvature.

"When we understand these mechanisms, we may be able to develop new preventative measures and better treatment methods," said Dr. Herring. The news of the gene discovery has Robin Tuley, Judy's daughter who also has a mild form of scoliosis, very excited.

"I have two girls that are almost grown, and I already worry about scoliosis affecting their children," said Robin. "But as they get closer to finding a solution, it gets easier not to be as concerned."

According to Dr. Wise, researchers and clinicians at TSRHC envision that one day, diagnosis can take place before the curve progresses, and noninvasive treatments can be applied

to correct the spinal curvature before surgery is needed.

"I am so thankful to the research group at TSRHC for their commitment to finding a cure for scoliosis," said Judy. "Maybe one day my great grandchildren and great-great grandchildren won't have to deal with this disease."

"This is a big day for us," said Dr. Wise. "We are so appreciative of the Worthington family and of many other families as well. We owe it to them that we've made this much progress."

But for Dr. Wise and her research team, the search is not over.

"We've made one discovery," she said. "And we have many more genes to discover, and we will discover them." □

Nora Elrnoufy, TSRHC Research department extracts DNA samples for use in genetic research.

