New genetic test predicts progress of scoliosis

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Nikki Koval, a fifth grader at St. Andrew School in Newtown, was one of the first patients in the Delaware Valley to receive a new genetic test that predicts whether a child diagnosed with mild scoliosis will develop a severe form of the disease.

Scoliosis is an abnormal curvature of the spine, which affects two to three percent of the population, or six million people in the United States, according to the National Scoliosis Foundation, and there is no cure. Only 1 to 4 percent of patients diagnosed with scoliosis experience curve progression severe enough to warrant surgery.

Nikki was diagnosed by her family doctor during a regular checkup at the age of eight. X-rays showed Nikki had a 23-degree spinal curve.

"I didn’t really understand what scoliosis was at first, but I was nervous," Nikki says. "I didn’t want to stop any of my activities because I had it."

She plays on her school soccer and basketball teams and also enjoys swimming, lacrosse, biking and horseback riding. "My parents explained to me that there was nothing to worry about now, but that the doctors would want to watch me to see if my curve got worse."

Her mother, Maureen, took Nikki to Dr. Suken A. Shah, a pediatric orthopedic surgeon and division chief of the Spine and Scoliosis Service at the Nemours/Alfred I. DuPont Hospital for Children in Wilmington, Del.

Dr. Shah told Nikki and her mom about a clinical trial of Scoliscore, a prognostic test he has worked to develop as a consultant with Axial Biotech.

Researchers like Shah reviewed the medical records and DNA samples of more than 10,000 scoliosis patients at 15 high-volume spine centers and discovered they have two kinds of genetic markers — one that may predispose them to developing severe scoliosis and another that may protect them from severe curve progression.

The Scoliscore test is painless and requires no needles — just a quick swab into a tube. A sample of Nikki’s saliva was sent to a laboratory for DNA analysis which yields a numerical score between 1 and 200 that would tell her doctor whether she was at a low (less than 50), intermediate (51-180) or high risk (181 to 200) for developing a severe curve (greater than 40 degrees). Spine surgery is generally recommended for curves of 45 degrees or more. Nikki had a score of 18 indicating she has a 99 percent probability she will not experience progression to a severe curve.

"We all took a deep breath," her mom said. "We were very relieved to say the least."

"Until now, it was extremely difficult to distinguish low-risk patients like Nikki from everyone else and we had to treat everybody like they had the potential to progress," said Shah. "Now with the addition of this DNA test along with other radiographic and clinical information obtained at the time of diagnosis, we can for the first time develop a more customized approach."

Children with mild scoliosis (curves of 25 degrees or less) are generally monitored for possible progression until they stop growing. This typically means x-rays and physical examinations every four to six months over a period of several years.

In some cases, children wear a brace in an attempt to slow or stop possible curve progression.

According to the web site of DePuy Spine Inc, the company marketing the test, children with scoliosis receive an average of 29 spinal x-rays over a nine-year period and seven to 10 percent of initially diagnosed patients are prescribed a brace. Studies by the National Cancer Institute have shown that adolescents repeatedly exposed to diagnostic radiation have a higher incidence of thyroid, breast and blood cancers later in life.

"This test means we may be able to reduce the frequency of x-rays and doctor visits and potentially reduce the need for bracing as a preventive measure in low-risk patients," said Dr. Shah.

Developed by Axial Biotech, the Scoliscore Test is the first commercially available genetic test for a spinal deformity and is intended for Caucasian children between age 9 and 13 diagnosed with mild AIS (10°-25° Cobb Angle).

"This is not a screening test for all kids in elementary school," says Shah. "Nor is it for those with cerebral palsy or other muscular diseases or for children younger than early adolescence."

The test does not predict an individual’s susceptibility to inherit the condition, or the final outcome of a patient’s progression.