

# Gene associated with adolescent idiopathic scoliosis identified

12 May 2013



This is a girl with adolescent idiopathic scoliosis (left) and posterior-anterior standing x-ray of spine (right).  
Credit: RIKEN

Researchers from the RIKEN Center for Integrative Medical Sciences in Japan have identified the first gene to be associated with adolescent idiopathic scoliosis (also called AIS) across Asian and Caucasian populations. The gene is involved in the growth and development of the spine during childhood.

Their study is published today in the journal *Nature Genetics*.

AIS is the most common pediatric [skeletal disease](#), affecting approximately 2% of school-age children. The causes of scoliosis remain largely unknown and brace treatment and surgery are the only treatment options. However, many clinical and genetic studies suggest a contribution of genetic factors.

To understand the causes and development of scoliosis, Dr Ikuyo Kou, Dr Shiro Ikegawa and their team have tried to identify genes that are associated with a susceptibility to develop the condition.

By studying the genome of 1,819 Japanese individuals suffering from scoliosis and comparing it to 25,939 Japanese individuals, the team identified a gene associated with a susceptibility to develop scoliosis on [chromosome 6](#). The association was replicated in Han Chinese and Caucasian populations.

The researchers show that the [susceptibility gene](#), GPR126, is highly expressed in cartilage and that suppression of this gene leads to delayed growth and bone [tissue formation](#) in the developing spine. GPR126 is also known to play a role in human height and trunk length.

"Our finding suggest the interesting possibility that GPR126 may affect both AIS susceptibility and height through abnormal spinal development and growth," explain the authors.

"Further functional studies are necessary to elucidate how alterations in GPR126 increase the risk of AIS in humans," they conclude.

**More information:** "Genetic variants in GPR126 are associated with adolescent idiopathic scoliosis." Kou et al. *Nature Genetics* 2013, [DOI: 10.1038/ng.2639](#)

Provided by RIKEN

APA citation: Gene associated with adolescent idiopathic scoliosis identified (2013, May 12) retrieved 11 June 2021 from <https://medicalxpress.com/news/2013-05-gene-adolescent-idiopathic-scoliosis.html>

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