

## ★ Adolescent idiopathic scoliosis (Kou, 2019)

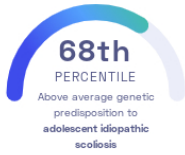
Ikuyo Kou, et al.  
Nature Communications

Spine Development

### STUDY SUMMARY

Discovery of 14 novel genetic loci associated with adolescent *idiopathic* scoliosis, or abnormal curvature of the spine.

#### YOUR RESULT



#### STUDY DESCRIPTION

Adolescent *idiopathic* scoliosis (AIS) is a sideways curvature of the spine (greater than 10 degrees) that appears in children and adolescent ages 10 to 18. AIS is a common disease, affecting 2-3% of adolescents worldwide. In fact, ~30% of individuals with AIS have a family history of scoliosis, suggesting that genetics plays a role in AIS development. To identify genetic regions associated with AIS, this study examined genetic information of 79,211 Japanese individuals. The researchers identified 20 genetic regions significantly associated with AIS, 14 of which are novel. Together, these genetic regions explain ~5% of the heritability of AIS.

#### DID YOU KNOW?

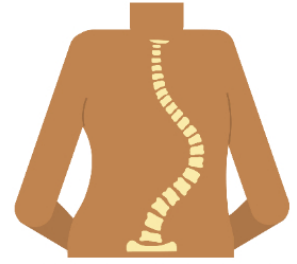
Females are more likely to develop AIS than males. Moreover, females are 8 times more likely to develop worse curvature over time than males. While it is unclear why there is a sex-bias, it is possible that the hormone leptin plays a role. Leptin is involved in telling your brain that you are full after a meal and females have higher leptin levels.

#### YOUR DETAILED RESULTS

To calculate your genetic predisposition to adolescent idiopathic scoliosis we summed up the effects of genetic variants that were linked to adolescent idiopathic scoliosis in the [study that this report is based on](#). These variants can be found in the table below. The variants highlighted in green have **positive effect sizes** and increase your genetic predisposition to adolescent idiopathic scoliosis. The variants highlighted in blue have **negative effect sizes** and decrease your genetic predisposition to adolescent idiopathic scoliosis. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to adolescent idiopathic scoliosis. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for adolescent idiopathic scoliosis to be 2.84**. To determine whether your score is high or low, we compared it to the scores of 5,000 other Nebula Genomics users. We found that your polygenic score for adolescent idiopathic scoliosis is in the **68th percentile**. This means that it is higher than the polygenic scores 68% of people. We consider this to be an **above average genetic predisposition to adolescent idiopathic scoliosis**. However, please note that genetic predispositions do not account for important non-genetic factors like lifestyle. Furthermore, the genetics of most traits has not been fully understood yet and many associations between traits and genetic variants remain unknown. For additional explanations, click on the column titles in the table below and visit our [Nebula Library tutorial](#).

VARIANT <sup>Ⓞ</sup>	YOUR GENOTYPE <sup>Ⓞ</sup>	EFFECT SIZE <sup>Ⓞ</sup>	VARIANT FREQUENCY <sup>Ⓞ</sup>	SIGNIFICANCE <sup>Ⓞ</sup>
rs11190870_T	T / T	0.42 (↑)	66%	$2.01 \times 10^{-82}$
rs9389985_G	A / G	0.19 (↑)	43%	$3.51 \times 10^{-20}$
rs7028900_C	G / C	0.18 (↑)	42%	$2.19 \times 10^{-17}$
rs6047716_C	C / T	0.14 (↑)	47%	$1.45 \times 10^{-11}$
rs141903567_C	NA	0.29 (-)	5%	$9.78 \times 10^{-11}$
rs11205303_C	T / T	0.16 (-)	21%	$1.62 \times 10^{-10}$
rs12029076_G	G / C	0.17 (↑)	78%	$2.17 \times 10^{-10}$
rs1978060_G	G / G	0.15 (↑)	47%	$3.26 \times 10^{-10}$
rs2467146_A	A / G	0.14 (↑)	67%	$5.96 \times 10^{-10}$
rs11787412_A	C / C	0.13 (-)	38%	$1.32 \times 10^{-9}$
rs188915802_T	NA	0.51 (-)	1%	$1.94 \times 10^{-9}$
rs658839_G	A / A	0.13 (-)	51%	$3.15 \times 10^{-9}$
rs2194285_G	A / A	0.17 (-)	11%	$8.69 \times 10^{-9}$
rs160335_G	G / G	0.12 (↑)	51%	$9.10 \times 10^{-9}$
rs482012_T	G / T	0.13 (↑)	72%	$2.30 \times 10^{-8}$
rs17011903_A	C / C	0.18 (-)	10%	$3.56 \times 10^{-8}$
rs397948882_A	A / A	0.18 (↑)	10%	$3.66 \times 10^{-8}$
rs12149832_G	G / A	0.15 (↑)	79%	$4.40 \times 10^{-8}$

N/A indicates variants that could not be imputed using the 1000 genomes project datasets and variants that have a frequency of < 5%. Your genome was sequenced at 30x/100x coverage and is not imputed. However, to calculate percentiles, we need to compare your data with other users imputed data. To make the data comparable, we need to exclude some of the variants from your data.



Scoliosis is a sideways curvature of the spine.