

## ☆ Pelvic organ prolapse (Olafsdottir, 2020)

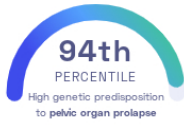
Thorhildur Olafsdottir, et al.  
Nature Communications Biology

Bladder Uterus Pelvis

### STUDY SUMMARY

Identification of 8 genetic variants associated with pelvic organ prolapse.

#### YOUR RESULT



#### STUDY DESCRIPTION

The muscles that support a woman's pelvic organs can weaken, causing organs like the bladder and uterus to drop lower in the *pelvis*. This condition is called pelvic organ prolapse, which results in symptoms like pressure in the pelvic area, bowel problems, and sexual dysfunction. Pelvic organ prolapse is very common, affecting nearly 1 in 3 women at some point during their lifetime. This genome-wide association study examined over 355,000 females of European ancestry to better understand the genetic risk factors for pelvic organ prolapse. The study identified 8 variants associated with pelvic organ prolapse, which collectively explain an estimated 12% of the heritability of the condition. One of the discovered genetic variants appears to affect the *estrogen* signaling pathway, while another variant influences maintenance of connective tissue in the *pelvis*.

#### DID YOU KNOW?

Pelvic organ prolapse is most commonly caused by pregnancy and childbirth. Obesity, old age, and conditions with frequent coughing are also risk factors for developing the condition.

#### YOUR DETAILED RESULTS

To calculate your genetic predisposition to pelvic organ prolapse we summed up the effects of genetic variants that were linked to pelvic organ prolapse 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 pelvic organ prolapse. The variants highlighted in blue have **negative effect sizes** and decrease your genetic predisposition to pelvic organ prolapse. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to pelvic organ prolapse. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for pelvic organ prolapse to be 0.38**. 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 pelvic organ prolapse is in the **94th percentile**. This means that it is higher than the polygenic scores 94% of people. We consider this to be a **high genetic predisposition to pelvic organ prolapse**. 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>
rs3820282_T	C / C	-0.16 (-)	17%	$3.30 \times 10^{-21}$
rs9306894_G	G / G	0.10 (↑)	34%	$3.40 \times 10^{-17}$
rs1430191_T	C / T	0.09 (↑)	48%	$1.00 \times 10^{-9}$
rs3791675_T	C / C	-0.14 (-)	21%	$2.70 \times 10^{-17}$
rs7682992_T	A / A	0.12 (-)	21%	$4.50 \times 10^{-16}$
rs72624976_T	NA	-0.22 (-)	5%	$4.00 \times 10^{-12}$
rs1247943_G	G / A	0.09 (↑)	46%	$2.30 \times 10^{-13}$
rs12325192_T	C / C	-0.12 (-)	18%	$2.80 \times 10^{-12}$

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.