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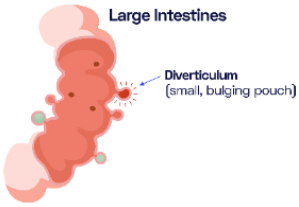
Diverticular disease (Maguire, 2018)

Lillias Maguire, et al.
Nature Genetics

Intestines



The pouches can get inflammed and cause pain.

STUDY SUMMARY

Identification of 42 genetic regions associated with diverticular disease.

YOUR RESULT

20th
PERCENTILE
Below average genetic predisposition to **diverticular disease**

STUDY DESCRIPTION

The colon, also known as the large intestine, squeezes water and nutrients out of the food we eat. Diverticular disease occurs when pressure causes small pouches to form in the colon, which can result in abdominal pain, intestinal bleeding, and diarrhea.
Diverticular disease is very common, affecting around 35% of those under 50 and nearly 60% of individuals over 60, though not everyone experiences symptoms. Genetics is thought to explain over 50% of an individual's risk of developing diverticular disease. To discover variants associated with diverticular disease, researchers examined the genetic information of over 430,000 individuals of European ancestry. The study identified 42 genomic regions, 39 of them novel, that are associated with the risk of diverticular disease. Some of the genes identified in this study are important for the functioning of smooth muscles, the type of muscles that allow intestines to contract. Other discovered genes play a role in transport of minerals in the colon.

DID YOU KNOW?

Fiber intake is recommended for those at risk for diverticular disease because it can help soften stool and reduce pressure in the colon. Fiber-rich foods include beans, rice, oatmeal, and vegetables such as broccoli and carrots.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to diverticular disease we summed up the effects of genetic variants that were linked to diverticular disease 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 diverticular disease. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to diverticular disease. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to diverticular disease. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for diverticular disease to be -0.05**. 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 diverticular disease is in the **20th percentile**. This means that it is higher than the polygenic scores 20% of people. We consider this to be a **below average genetic predisposition to diverticular disease**. 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	YOUR GENOTYPE	EFFECT SIZE	VARIANT FREQUENCY	SIGNIFICANCE
rs6734367_G	T / G	-0.01 (↓)	82%	4.29×10^{-44}
rs4333882_G NEW	A / A	0.01 (-)	19%	4.44×10^{-22}
rs7609897_T	G / T	-0.01 (↓)	22%	2.72×10^{-18}
rs7086249_C NEW	T / T	-0.00 (-)	46%	5.37×10^{-16}
rs1802575_C NEW	G / C	0.01 (↑)	13%	7.71×10^{-16}
rs11667256_T NEW	A / T	-0.00 (↓)	52%	1.24×10^{-14}
rs962369_C NEW	T / C	-0.01 (↓)	31%	2.16×10^{-14}
rs6949391_T NEW	C / C	0.00 (-)	34%	3.74×10^{-14}
rs61823192_T NEW	NA	-0.01 (-)	3%	1.15×10^{-13}
rs9520344_A	T / T	-0.00 (-)	24%	5.23×10^{-12}
rs7098322_T NEW	T / T	-0.01 (↓)	87%	9.94×10^{-12}
rs10472291_A NEW	C / C	0.00 (-)	33%	1.01×10^{-11}
rs582094_T NEW	T / T	-0.00 (↓)	32%	1.55×10^{-11}
rs75434097_A NEW	G / G	0.01 (-)	15%	4.90×10^{-11}
rs2280028_A NEW	G / A	-0.01 (↓)	14%	7.05×10^{-11}
rs9856118_G NEW	A / A	-0.01 (-)	17%	8.80×10^{-11}
rs71472433_C NEW	A / A	0.01 (-)	17%	8.90×10^{-11}
rs2131755_G NEW	A / A	0.00 (-)	41%	1.50×10^{-10}
rs4839715_A NEW	G / A	-0.00 (↓)	37%	1.62×10^{-10}
rs148376933_T NEW	NA	1.16 (-)	< 1%	1.88×10^{-10}
rs1381335_T NEW	C / T	-0.00 (↓)	24%	1.91×10^{-10}
rs61814883_A NEW	G / G	-0.00 (-)	30%	2.05×10^{-10}
rs8074740_A NEW	A / A	0.00 (↑)	32%	2.34×10^{-10}
rs3113037_T NEW	C / T	0.00 (↑)	23%	2.52×10^{-10}
rs12293535_A NEW	G / G	0.00 (-)	28%	6.20×10^{-10}
rs11619840_A	C / A	-0.00 (↓)	19%	1.70×10^{-9}
rs875107_A NEW	C / C	-0.00 (-)	52%	2.33×10^{-9}
rs3823878_A NEW	G / G	0.01 (-)	6%	2.63×10^{-9}
rs10471645_C NEW	C / C	-0.00 (↓)	83%	3.03×10^{-9}
rs1888693_A NEW	G / G	-0.00 (-)	34%	3.58×10^{-9}
rs4871180_T NEW	C / T	0.00 (↑)	25%	4.15×10^{-9}
rs2049865_A NEW	C / A	0.00 (↑)	58%	5.54×10^{-9}
rs1544387_T NEW	G / T	0.00 (↑)	58%	5.74×10^{-9}
rs11934833_G NEW	G / G	-0.00 (↓)	30%	6.21×10^{-9}
rs2784255_C NEW	T / C	-0.00 (↓)	48%	1.06×10^{-8}
rs10120333_G NEW	G / G	0.00 (↑)	53%	1.54×10^{-8}

rs12942267_T NEW	C / C	0.00 (–)	64%	2.55×10^{-8}
rs62126581_A NEW	G / G	-0.00 (–)	17%	3.77×10^{-8}
rs115490395_A NEW	NA	0.02 (–)	1%	4.39×10^{-8}
rs2470653_A NEW	T / T	-0.00 (–)	23%	4.51×10^{-8}
rs10173528_T NEW	T / T	-0.00 (↓)	61%	4.73×10^{-8}
rs72945112_T NEW	C / C	-0.00 (–)	15%	6.30×10^{-6}

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.