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Systemic sclerosis (López-Isac, 2019)

Elena López-Isac, et al.
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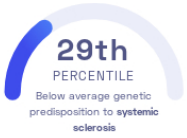
Vasculature | Skin | Autoimmunity

STUDY SUMMARY

Discovery of 13 novel genetic variants associated with the development of systemic sclerosis.

YOUR RESULT



STUDY DESCRIPTION

Normally, the immune system works to protect the body against foreign invaders such as bacteria and viruses. Autoimmune diseases occur when the immune system mistakenly attacks the body's own cells and organs. One of the most debilitating autoimmune diseases is systemic sclerosis, which causes scarring of the skin and internal organs. Over time, this scarring slowly limits the organs' ability to function, and is particularly detrimental to the lungs and the blood vessels. This study attempted to identify genetic variants that contribute to a person's risk of developing of systemic sclerosis. By examining the genetic data of nearly 27,000 individuals of European ancestry, 27 genetic variants that significantly correlate with the risk of systemic sclerosis were identified. Of these, 13 are newly discovered. One identified variant was located in DDX6, a gene previously shown to play a role in the development of blood vessels.

DID YOU KNOW?

Systemic sclerosis can cause skin hardening and joint stiffness, making movement extremely painful. To maintain flexibility, regular exercise is recommended.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to systemic sclerosis we summed up the effects of genetic variants that were linked to systemic sclerosis 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 systemic sclerosis. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to systemic sclerosis. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to systemic sclerosis. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for systemic sclerosis to be -0.15**. 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 systemic sclerosis is in the **29th percentile**. This means that it is higher than the polygenic scores 29% of people. We consider this to be a **below average genetic predisposition to systemic sclerosis**. 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
rs3821236_A	G / G	0.27 (–)	20%	1.94×10^{-23}
rs36073657_T	C / C	0.34 (–)	10%	3.10×10^{-21}
rs2736340_T NEW	C / T	0.22 (↑)	24%	3.33×10^{-21}
rs4853458_A	G / G	0.30 (–)	23%	4.86×10^{-18}
rs11117420_C NEW	G / C	-0.21 (↓)	19%	3.82×10^{-16}
rs1378942_C	A / A	0.17 (–)	39%	1.84×10^{-14}
rs12155080_G	G / C	-0.16 (↓)	37%	2.87×10^{-13}
rs3792783_G	A / A	0.18 (–)	16%	2.42×10^{-12}
rs2056626_G	T / T	-0.21 (–)	39%	1.31×10^{-11}
rs11217020_A NEW	G / G	-0.17 (–)	20%	2.08×10^{-11}
rs11724804_A NEW	A / A	0.16 (↑)	44%	5.31×10^{-11}
rs4076852_G	G / A	0.15 (↑)	26%	1.04×10^{-10}
rs9884090_A NEW	G / G	-0.19 (–)	16%	1.89×10^{-10}
rs589446_T	T / T	-0.15 (↓)	35%	1.95×10^{-10}
rs2651804_T NEW	C / C	-0.20 (–)	17%	2.54×10^{-10}
rs3790566_T	C / C	0.15 (–)	24%	3.84×10^{-10}
rs2305743_A	G / G	-0.19 (–)	20%	4.64×10^{-10}
rs633724_T	C / C	0.12 (–)	35%	2.84×10^{-9}
rs883770_T NEW	C / T	0.12 (↑)	50%	4.79×10^{-9}
rs1857066_A NEW	A / A	-0.14 (↓)	25%	5.02×10^{-9}
rs16832798_C NEW	T / C	0.17 (↑)	14%	5.20×10^{-9}
rs230534_T NEW	T / C	0.14 (↑)	34%	5.38×10^{-9}
rs7355798_T	C / C	0.13 (–)	24%	1.24×10^{-8}
rs1005714_G NEW	C / C	-0.16 (–)	20%	1.87×10^{-8}
rs6598008_A NEW	A / A	-0.22 (↓)	44%	1.97×10^{-8}
rs685985_T NEW	A / A	-0.14 (–)	47%	3.82×10^{-8}
rs2022449_T NEW	G / T	0.14 (↑)	23%	6.28×10^{-8}