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Systemic lupus erythematosus (Chen, 2020)

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Human Molecular Genetics

Kidneys | Inflammation | Autoimmunity

A typical sign of lupus is a butterfly-shaped rash across the cheeks and bridge of the nose.

STUDY SUMMARY

Development of a genetic risk score to predict the risk of systemic lupus erythematosus (SLE).

YOUR RESULT

STUDY DESCRIPTION

Systemic lupus erythematosus (SLE) is an autoimmune disorder characterized by widespread inflammation and tissue damage. More than 200,000 new SLE cases are diagnosed in the United States every year. About one half of SLE patients experience kidney disease, which is the one of the most common causes of death in SLE patients. In this study, researchers created a genetic risk score for SLE using previously published genetic variants significantly associated with SLE in European and Chinese patients.

DID YOU KNOW?

Women are nine times more likely to develop lupus than men. This is because in addition to genetic factors, environmental factors likely play a role in the development of lupus.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to SLE we summed up the effects of genetic variants that were linked to SLE 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 SLE. The variants highlighted in blue have **negative effects sizes** and decrease your genetic predisposition to SLE. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to SLE. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for SLE to be 5.82**. 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 SLE is in the **43rd percentile**. This means that it is higher than the polygenic scores 43% of people. We consider this to be an **average genetic predisposition to SLE**. 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
rs2187668_T	C / C	0.83 (-)	8%	7.89×10^{-93}
rs11889341_T	C / C	0.55 (-)	20%	1.17×10^{-64}
rs17849502_T	NA	0.94 (-)	3%	1.71×10^{-55}
rs2070197_C	T / T	0.63 (-)	10%	2.57×10^{-45}
rs34572943_A	G / G	0.54 (-)	9%	8.27×10^{-45}
rs4728142_A	G / A	0.36 (↑)	38%	1.02×10^{-27}
rs36014129_A	G / G	0.41 (-)	5%	4.55×10^{-21}
rs7708392_C	G / G	0.24 (-)	43%	1.21×10^{-18}
rs6932056_C	NA	0.60 (-)	3%	7.02×10^{-17}
rs2736340_T	C / T	0.30 (↑)	25%	2.07×10^{-16}
rs2431697_T	T / C	0.37 (↑)	58%	3.37×10^{-14}
rs7444_C	T / C	0.24 (↑)	32%	2.30×10^{-13}
rs2476601_A	G / G	0.30 (-)	8%	6.38×10^{-13}
rs2304256_C	C / C	0.22 (↑)	76%	2.25×10^{-12}
rs2205960_T	G / G	0.38 (-)	18%	2.57×10^{-12}
rs6568431_A	C / C	0.18 (-)	40%	5.07×10^{-12}
rs1801274_G	A / G	0.15 (↑)	44%	6.17×10^{-11}
rs7726414_T	C / C	0.37 (-)	13%	4.39×10^{-10}
rs10028805_G	G / G	0.18 (↑)	56%	5.35×10^{-10}
rs2732552_C	T / C	0.20 (↑)	51%	7.78×10^{-10}
rs1418190_T	C / T	0.21 (↑)	64%	7.23×10^{-9}
rs12802200_C	C / C	0.21 (↑)	80%	1.60×10^{-8}
rs9267992_G	G / A	0.23 (↑)	11%	1.72×10^{-8}
rs6740462_A	A / A	0.10 (↑)	77%	2.14×10^{-8}
rs3768792_G	A / A	0.22 (-)	78%	3.33×10^{-8}
rs10774625_A	A / G	0.12 (↑)	34%	1.04×10^{-7}
rs9652601_G	G / G	0.19 (↑)	66%	3.07×10^{-7}
rs2280381_T	T / T	0.16 (↑)	62%	8.88×10^{-7}
rs564799_C	T / T	0.13 (-)	65%	9.11×10^{-7}
rs7941765_C	T / C	0.13 (↑)	65%	1.18×10^{-6}
rs4948496_C	T / C	0.16 (↑)	45%	1.27×10^{-6}
rs1059312_G	A / A	0.16 (-)	42%	2.24×10^{-6}
rs2941509_T	C / C	0.30 (-)	10%	2.73×10^{-6}
rs6445972_T	T / T	0.21 (↑)	80%	2.90×10^{-6}
rs2327832_G	A / G	0.20 (↑)	16%	3.23×10^{-6}
rs2111485_G	A / G	0.14 (↑)	47%	3.89×10^{-6}
rs3093030_T	T / T	0.15 (↑)	32%	5.81×10^{-6}
rs223881_T	C / C	0.14 (-)	38%	6.01×10^{-6}
rs2934498_G	A / G	0.22 (↑)	35%	7.20×10^{-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.