

★ Mosaic loss of chromosome Y (Thompson, 2019)

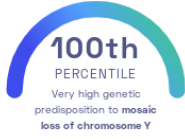
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Blood

STUDY SUMMARY

Discovery of 166 genetic variants associated with a mosaic loss of *chromosome Y*.

YOUR RESULT



STUDY DESCRIPTION

Our DNA is packed into 23 pairs of *chromosomes* including two 'sex *chromosomes*'. Females have two X *chromosomes*, while males have one Y and one X *chromosome*. In males, loss of the Y *chromosome* in some cells of the body, particularly white blood cells, has been previously associated with multiple medical conditions including various cancers, autoimmune disease, diabetes, and cardiovascular disease. To determine genetic factors that may predispose a male to loss of *chromosome Y*, this study examined the genomes of over 750,000 males of European and Japanese ancestry. A detectable loss of the Y *chromosome* was present in approximately 20% of male study participants. The study identified 166 variants, 137 of which are novel, that together explain more than 30% of the heritability of Y *chromosome* loss. While the loss of *chromosome Y* can occur only in males, the study also found an association between the discovered variants and breast cancer risk in females and well as delayed menopause onset.

DID YOU KNOW?

Smoking has previously been identified as one of the biggest non-genetic risk factors for loss of chromosome Y.

YOUR DETAILED RESULTS

To calculate your genetic predisposition to mosaic loss of chromosome Y we summed up the effects of genetic variants that were linked to mosaic loss of chromosome Y 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 mosaic loss of chromosome Y. The variants highlighted in blue have **negative effect sizes** and decrease your genetic predisposition to mosaic loss of chromosome Y. Variants that are not highlighted are not found in your genome and do not affect your genetic predisposition to mosaic loss of chromosome Y. By adding up the effect sizes of the highlighted variants **we calculated your polygenic score for mosaic loss of chromosome Y to be 12.75**. 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 mosaic loss of chromosome Y is in the **100th percentile**. This means that it is higher than the polygenic scores 100% of people. We consider this to be a **very high genetic predisposition to mosaic loss of chromosome Y**. 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 [Ⓞ]
rs17758695_C	C / C	0.56 (↑)	97%	5.20 x 10 ⁻¹⁴⁹
rs2887399_G	G / G	0.22 (↑)	80%	4.30 x 10 ⁻¹³⁶
rs381500_C	C / A	0.15 (↑)	55%	1.00 x 10 ⁻⁹²
rs1849209_T	G / T	0.17 (↑)	77%	2.10 x 10 ⁻⁹²
rs1052053_G	A / A	0.15 (-)	38%	3.60 x 10 ⁻⁹¹
rs11082396_C	T / C	0.21 (↑)	13%	1.60 x 10 ⁻⁸¹
rs6089050_C	C / C	0.16 (↑)	79%	6.20 x 10 ⁻⁷⁶
rs78378222_G	NA	0.57 (-)	1%	1.40 x 10 ⁻⁷⁰
rs9400267_T	T / C	0.12 (↑)	48%	3.90 x 10 ⁻⁶⁷
rs2280548_T	G / G	0.12 (-)	40%	1.50 x 10 ⁻⁶⁷
rs72698720_C	C / C	0.17 (↑)	86%	9.70 x 10 ⁻⁶⁴
rs13062095_C	T / C	0.11 (↑)	34%	1.30 x 10 ⁻⁴⁷
rs73721669_C	C / C	0.15 (↑)	88%	9.70 x 10 ⁻⁴⁴
rs16982394_A	C / C	0.10 (-)	27%	5.90 x 10 ⁻⁴⁰
rs118035610_C	C / C	0.23 (↑)	96%	5.50 x 10 ⁻³⁹
rs7129527_G	A / A	0.10 (-)	41%	4.30 x 10 ⁻³⁸
rs10456506_T	T / T	0.13 (↑)	87%	6.60 x 10 ⁻³⁶
rs56116444_G	T / T	0.17 (-)	8%	9.30 x 10 ⁻³⁶
rs3743503_G	T / G	0.13 (↑)	13%	2.50 x 10 ⁻³⁵
rs111725880_C	C / C	0.14 (↑)	89%	6.50 x 10 ⁻³⁵
rs609018_G	T / T	0.09 (-)	44%	8.90 x 10 ⁻³⁵
rs7271671_C	C / C	0.20 (↑)	93%	1.10 x 10 ⁻³¹
rs72899729_G	NA	0.37 (-)	1%	1.20 x 10 ⁻³¹
rs10048745_A	G / G	0.10 (-)	26%	4.40 x 10 ⁻³¹
rs2979469_C	G / G	0.09 (-)	74%	2.80 x 10 ⁻³⁰
rs62618742_C	C / C	0.25 (↑)	97%	7.00 x 10 ⁻³⁰
rs4714550_C	A / C	0.10 (↑)	76%	1.70 x 10 ⁻²⁸
rs73176930_A	A / A	0.10 (↑)	80%	1.30 x 10 ⁻²⁶
rs35355140_C	C / C	0.14 (↑)	93%	8.80 x 10 ⁻²⁵
rs12695310_G	G / T	0.07 (↑)	47%	3.40 x 10 ⁻²⁴
rs35979828_T	C / C	0.13 (-)	7%	2.80 x 10 ⁻²²
rs10849448_G	A / G	0.09 (↑)	75%	3.70 x 10 ⁻²²
rs17255991_C	C / C	0.11 (↑)	89%	2.20 x 10 ⁻²¹
rs2293661_G	A / A	0.07 (-)	45%	2.30 x 10 ⁻²¹
rs11769630_A	T / T	0.13 (-)	8%	3.90 x 10 ⁻²¹
rs4981287_A	G / A	0.08 (↑)	24%	2.50 x 10 ⁻²⁰
rs11679328_T	C / C	0.14 (-)	6%	4.80 x 10 ⁻²⁰
rs758275723_T	T / T	0.26 (↑)	97%	1.10 x 10 ⁻¹⁹
rs9844706_G	A / A	0.10 (-)	11%	1.60 x 10 ⁻¹⁹

rs291699_T	C / C	0.11 (-)	9%	2.90 x 10 ⁻¹⁸
rs116483731_G	G / G	0.35 (↑)	99%	9.20 x 10 ⁻¹⁸
rs36084354_A	G / A	0.10 (↑)	9%	1.70 x 10 ⁻¹⁷
rs13063578_A	A / A	0.06 (↑)	40%	2.10 x 10 ⁻¹⁷
rs7781977_C	C / T	0.07 (↑)	69%	2.50 x 10 ⁻¹⁷
rs621940_C	C / C	0.09 (↑)	84%	8.30 x 10 ⁻¹⁷
rs74911261_A	NA	0.18 (-)	3%	1.20 x 10 ⁻¹⁶
rs186430430_C	NA	0.70 (-)	< 1%	1.50 x 10 ⁻¹⁶
rs1805419_G	G / G	0.07 (↑)	72%	2.30 x 10 ⁻¹⁶
rs3811444_C	C / C	0.06 (↑)	67%	2.90 x 10 ⁻¹⁶
rs74845559_C	C / C	0.09 (↑)	86%	4.30 x 10 ⁻¹⁶
rs9939347_G	G / G	0.06 (↑)	30%	5.10 x 10 ⁻¹⁶
rs4669037_T	T / C	0.06 (↑)	32%	5.50 x 10 ⁻¹⁶
rs11062924_T	G / T	0.08 (↑)	17%	1.40 x 10 ⁻¹⁶
rs710422_G	G / A	0.07 (↑)	22%	3.00 x 10 ⁻¹⁶
rs141777833_C	NA	0.33 (-)	1%	3.70 x 10 ⁻¹⁶
rs72966755_G	C / G	0.06 (↑)	46%	6.70 x 10 ⁻¹⁶
rs670180_A	A / A	0.05 (↑)	57%	1.50 x 10 ⁻¹⁴
rs7172615_G	G / G	0.07 (↑)	76%	2.00 x 10 ⁻¹⁴
rs3756364_G	G / A	0.08 (↑)	87%	3.60 x 10 ⁻¹⁴
rs57565337_T	NA	0.22 (-)	3%	4.40 x 10 ⁻¹⁴
rs2852780_A	A / G	0.08 (↑)	13%	8.40 x 10 ⁻¹⁴
rs200123937_G	TGT / TGT	0.07 (-)	76%	1.30 x 10 ⁻¹³
rs76428106_C	NA	0.25 (-)	1%	1.60 x 10 ⁻¹³
rs73111522_A	NA	0.14 (-)	4%	2.50 x 10 ⁻¹³
rs2407797_A	A / A	0.06 (↑)	72%	3.00 x 10 ⁻¹³
rs2396902_G	G / G	0.06 (↑)	57%	4.10 x 10 ⁻¹³
rs1569419_T	T / C	0.06 (↑)	23%	1.10 x 10 ⁻¹²
rs117145034_A	NA	0.22 (-)	2%	1.60 x 10 ⁻¹²
rs6479226_T	T / T	0.05 (↑)	46%	2.50 x 10 ⁻¹²
rs1049612_A	A / A	0.05 (↑)	61%	2.70 x 10 ⁻¹²
rs34890930_T	C / C	0.06 (-)	73%	3.30 x 10 ⁻¹²
rs9262570_T	NA	0.17 (-)	3%	5.00 x 10 ⁻¹²
rs12549737_T	T / T	0.07 (↑)	85%	7.10 x 10 ⁻¹²
rs392020_T	C / C	0.04 (-)	25%	1.20 x 10 ⁻¹¹
rs6731121_G	G / T	0.05 (↑)	39%	1.40 x 10 ⁻¹¹
rs11177383_C	A / A	0.05 (-)	31%	1.50 x 10 ⁻¹¹
rs3761704_G	A / G	0.07 (↑)	14%	1.90 x 10 ⁻¹¹
rs230533_G	G / G	0.05 (↑)	65%	1.90 x 10 ⁻¹¹
rs739846_A	G / G	0.09 (-)	8%	1.90 x 10 ⁻¹¹
rs6924733_G	T / T	0.05 (-)	49%	2.30 x 10 ⁻¹¹
rs13141_G	G / G	0.22 (↑)	99%	3.10 x 10 ⁻¹¹
rs76428668_G	T / T	0.06 (-)	25%	4.00 x 10 ⁻¹¹
rs760622583_T	/	0.07 (-)	16%	5.70 x 10 ⁻¹¹
rs1531842_T	T / T	0.05 (↑)	31%	6.00 x 10 ⁻¹¹
rs218264_A	A / A	0.06 (↑)	75%	6.20 x 10 ⁻¹¹
rs7223609_T	C / C	0.07 (-)	19%	6.90 x 10 ⁻¹¹
rs11642909_A	G / A	0.05 (↑)	68%	8.30 x 10 ⁻¹¹
rs56795609_A	A / A	0.06 (↑)	83%	8.80 x 10 ⁻¹¹
rs1926785_A	G / G	0.07 (-)	16%	1.10 x 10 ⁻¹⁰
rs34087210_G	C / C	0.05 (-)	58%	1.40 x 10 ⁻¹⁰
rs5757613_A	A / A	0.05 (↑)	34%	1.50 x 10 ⁻¹⁰
rs10305667_C	C / T	0.05 (↑)	76%	1.70 x 10 ⁻¹⁰
rs551319785_C	C / C	0.08 (↑)	14%	2.40 x 10 ⁻¹⁰
rs17064495_C	NA	0.14 (-)	3%	2.40 x 10 ⁻¹⁰
rs2527884_G	G / A	0.05 (↑)	51%	2.50 x 10 ⁻¹⁰
rs2688887_T	C / C	0.05 (-)	46%	3.70 x 10 ⁻¹⁰
rs60808706_G	G / G	0.10 (↑)	95%	3.70 x 10 ⁻¹⁰
rs113736796_G	NA	0.11 (-)	4%	3.80 x 10 ⁻¹⁰
rs5914348_T	T / T	0.04 (↑)	74%	3.90 x 10 ⁻¹⁰
rs980381_T	A / A	0.06 (-)	17%	5.20 x 10 ⁻¹⁰

rs11965885_G	G / G	0.05 (↑)	42%	5.30 × 10 ⁻¹⁰
rs769022853_T	/	0.05 (-)	58%	8.50 × 10 ⁻¹⁰
rs2291119_C	C / C	0.05 (↑)	71%	1.00 × 10 ⁻⁹
rs2382230_T	A / T	0.05 (↑)	67%	1.10 × 10 ⁻⁹
rs4671127_C	C / G	0.05 (↑)	66%	1.70 × 10 ⁻⁹
rs7035770_A	C / C	0.04 (-)	46%	2.00 × 10 ⁻⁹
rs57026767_C	C / T	0.06 (↑)	16%	2.50 × 10 ⁻⁹
rs561123020_A	NA	0.17 (-)	1%	3.40 × 10 ⁻⁹
rs2701539_G	T / T	0.09 (-)	6%	3.50 × 10 ⁻⁹
rs60781426_A	G / G	0.07 (-)	12%	3.50 × 10 ⁻⁹
rs2908986_A	G / G	0.05 (-)	29%	4.30 × 10 ⁻⁹
rs6939093_A	NA	0.11 (-)	4%	4.90 × 10 ⁻⁹
rs7410534_T	C / C	0.09 (-)	7%	6.50 × 10 ⁻⁹
rs458069_G	G / G	0.05 (↑)	70%	6.80 × 10 ⁻⁹
rs11258419_G	A / A	0.04 (-)	60%	7.60 × 10 ⁻⁹
rs4288784_G	C / C	0.09 (-)	8%	8.50 × 10 ⁻⁹
rs173913_C	T / T	0.03 (-)	38%	8.80 × 10 ⁻⁹
rs56345976_G	G / A	0.04 (↑)	40%	9.30 × 10 ⁻⁹
rs9943081_C	C / C	0.06 (↑)	85%	9.60 × 10 ⁻⁹
rs478093_G	G / G	0.05 (↑)	68%	1.20 × 10 ⁻⁸
rs66700428_A	AA / AA	0.06 (-)	85%	1.20 × 10 ⁻⁸
rs560646371_G	GCTGCAGGAG / GCTGCAGGAG	0.60 (-)	> 99%	1.40 × 10 ⁻⁸
rs80036149_A	A / A	0.07 (↑)	89%	1.40 × 10 ⁻⁸
rs10831321_G	A / A	0.05 (-)	30%	1.50 × 10 ⁻⁸
rs6020413_T	T / T	0.05 (↑)	79%	1.50 × 10 ⁻⁸
rs2651932_G	A / G	0.08 (↑)	9%	1.90 × 10 ⁻⁸
rs13286011_C	C / C	0.12 (↑)	97%	2.90 × 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.