

## ★ Mosaic loss of chromosome Y (Terao, 2019)

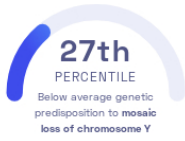
Chikashi Terao, et al.  
Nature Communications

Blood Sex

### STUDY SUMMARY

Identification of 46 genetic variants associated with mosaic loss of chromosome Y.

### YOUR RESULT



### STUDY DESCRIPTION

Cells in the human body store genetic information in 23 pairs of chromosomes. Mosaic loss of chromosome Y (mLOY) is a male-specific condition characterized by the loss of chromosome Y by some cells. mLOY has been linked to various medical conditions, including cancer, and is most commonly observed in the white blood cells of ageing men. This genome-wide association study analyzed the genetic data of 95,380 Japanese men to better understand the genetic factors that contribute to the loss of the *Y chromosome*. The study discovered 46 genetic variants of which 35 are novel, and collectively these variants explain an estimated 9% of the heritability of mLOY. Many of the identified genetic variants are near genes that are active in blood *stem cells*.

### DID YOU KNOW?



Studies have shown that smoking may significantly increase the risk of mosaic 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 **-0.08**. 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 **27th percentile**. This means that it is higher than the polygenic scores 27% of people. We consider this to be a **below average 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 <sup>Ⓞ</sup>	YOUR GENOTYPE <sup>Ⓞ</sup>	EFFECT SIZE <sup>Ⓞ</sup>	VARIANT FREQUENCY <sup>Ⓞ</sup>	SIGNIFICANCE <sup>Ⓞ</sup>
rs56116444_G	T / T	-0.09 (-)	31%	9.20 x 10 <sup>-81</sup>
rs80277818_G	A / G	-0.07 (↓)	26%	7.80 x 10 <sup>-55</sup>
rs2842873_T	C / C	-0.06 (-)	37%	1.50 x 10 <sup>-36</sup>
rs72698721_A	G / G	0.06 (-)	32%	4.20 x 10 <sup>-33</sup>
rs59543286_C <span>NEW</span>	A / A	0.06 (-)	23%	8.00 x 10 <sup>-32</sup>
rs4709819_A	G / A	0.05 (↑)	41%	1.30 x 10 <sup>-30</sup>
rs227079_A	C / A	-0.04 (↓)	48%	5.90 x 10 <sup>-26</sup>
rs4721217_T	C / C	-0.04 (-)	42%	6.80 x 10 <sup>-23</sup>
rs2237896_A <span>NEW</span>	G / G	0.04 (-)	40%	1.70 x 10 <sup>-22</sup>
rs78997619_T	C / C	0.08 (-)	6%	2.50 x 10 <sup>-21</sup>
rs56727837_T <span>NEW</span>	G / G	0.06 (-)	11%	9.70 x 10 <sup>-20</sup>
rs34324_A <span>NEW</span>	A / A	-0.04 (↓)	43%	3.10 x 10 <sup>-19</sup>
rs11769630_A <span>NEW</span>	T / T	-0.06 (-)	13%	3.70 x 10 <sup>-19</sup>
rs4681200_G	G / G	0.07 (↑)	92%	3.80 x 10 <sup>-19</sup>
rs4683900_T	C / T	0.04 (↑)	37%	7.90 x 10 <sup>-19</sup>
rs10849448_G <span>NEW</span>	A / G	-0.05 (↓)	81%	1.50 x 10 <sup>-18</sup>
rs728739_G	A / G	0.08 (↑)	6%	3.80 x 10 <sup>-18</sup>
rs871134_T <span>NEW</span>	C / T	-0.03 (↓)	49%	1.40 x 10 <sup>-14</sup>
rs35356140_A <span>NEW</span>	C / C	0.04 (-)	14%	2.30 x 10 <sup>-13</sup>
rs138423884_G <span>NEW</span>	NA	0.08 (-)	4%	1.30 x 10 <sup>-12</sup>
rs10948011_A <span>NEW</span>	G / G	0.04 (-)	23%	4.00 x 10 <sup>-12</sup>
rs9921295_G <span>NEW</span>	T / T	0.03 (-)	33%	4.00 x 10 <sup>-12</sup>
rs17049722_T <span>NEW</span>	C / C	0.04 (-)	17%	4.40 x 10 <sup>-12</sup>
rs11251_T	G / G	0.03 (-)	54%	3.40 x 10 <sup>-11</sup>
rs77406149_G <span>NEW</span>	A / A	0.04 (-)	20%	2.50 x 10 <sup>-10</sup>
rs2646425_T <span>NEW</span>	C / C	0.03 (-)	31%	4.30 x 10 <sup>-10</sup>
rs2811487_A <span>NEW</span>	G / G	0.03 (-)	36%	6.80 x 10 <sup>-10</sup>
rs1859259_T <span>NEW</span>	T / T	-0.03 (↓)	58%	1.10 x 10 <sup>-9</sup>
rs12668837_T <span>NEW</span>	C / C	-0.03 (-)	46%	1.80 x 10 <sup>-9</sup>
rs2853677_A <span>NEW</span>	G / A	0.03 (↑)	70%	2.10 x 10 <sup>-9</sup>
rs9299129_G <span>NEW</span>	A / A	0.03 (-)	23%	2.20 x 10 <sup>-9</sup>
rs527504_A <span>NEW</span>	G / A	-0.03 (↓)	19%	4.10 x 10 <sup>-9</sup>
rs77874075_G	T / G	-0.03 (↓)	22%	4.60 x 10 <sup>-9</sup>
rs117587217_C <span>NEW</span>	NA	-0.10 (-)	2%	9.70 x 10 <sup>-9</sup>
rs201753350_T	NA	-0.15 (-)	1%	1.00 x 10 <sup>-8</sup>
rs12225799_G <span>NEW</span>	C / G	-0.04 (↓)	13%	1.10 x 10 <sup>-8</sup>
rs79058858_T <span>NEW</span>	NA	0.16 (-)	1%	1.40 x 10 <sup>-8</sup>
rs74843651_T <span>NEW</span>	G / G	0.06 (-)	5%	2.20 x 10 <sup>-8</sup>



rs189309686_T 	C / C	0.03 (-)	25%	4.40 x 10 <sup>-8</sup>
rs2804301_A 	G / G	-0.03 (-)	24%	4.40 x 10 <sup>-8</sup>
rs2979469_C	G / G	-0.04 (-)	88%	4.90 x 10 <sup>-8</sup>

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.