STUDI BIOINFORMATIKA VARIASI DAN EKSPRESI GEN YANG BERHUBUNGAN DENGAN *TAKAYASU ARTERITIS* (TA)

SKRIPSI

Disusun Untuk Memenuhi Sebagian Persyaratan
Mencapai Derajat Sarjana Farmasi

Disusun Oleh :

Nabila Aulia Rahma
NIM : 202205046

PROGRAM STUDI FARMASI PROGRAM SARJANA
FAKULTAS ILMU KESEHATAN
UNIVERSITAS MUHAMMADIYAH GOMBONG
GOMBONG
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2026

HALAMAN PERSETUJUAN

STUDI BIOINFORMATIKA VARIASI DAN EKSPRESI GEN YANG BERHUBUNGAN DENGAN *TAKAYASU ARTERITIS* (TA)

Telah disetujui dan dinyatakan Telah memenuhi syarat untuk diujikan

Pada Tanggal 26 Mei 2026

Yang dipersiapkan dan disusun oleh :

Nabila Aulia Rahma

NIM: 202205046

Sususnan Tim Pembimbing

apt. Dwiki Fitri, M.Farm (.....)

Mengetahui

Ketua Program Studi Farmasi Program Sarjana

Universitas Muhammadiyah Gombong

apt. Tri Cahyani Widiastuti, M.Sc

NIDN : 616028901

HALAMAN PENGESAHAN

STUDI BIOINFORMATIKA VARIASI DAN EKSPRESI GEN YANG BERHUBUNGAN DENGAN *TAKAYASU ARTERITIS* (TA)

Yang dipersiapkan dan disusun oleh :

Nabila Aulia Rahma

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Telah dipertahankan di hadapan Tim Penguji

pada tanggal 26 Mei 2026

Susunan Tim Penguji

1. Dr. apt. Muh Husnul Khuluq, M.Farm (.....)
2. apt. Wahyu Rahmatulloh, M.Farm (.....)
3. apt. Dwiki Fitri, M.Farm (.....)

Mengetahui

Ketua Program Studi Farmasi Program Sarjana

Universitas Muhammadiyah Gombong

apt. Tri Cahyani Widiastuti, M.Sc

NIDN : 616028901

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Nabila Aulia Rahma

KATA PENGANTAR

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Gombong, 13 Januari 2026

Nabila Aulia Rahma

NIM. 202205046

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PROGRAM STUDI FARMASI PROGRAM SARJANA
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Skripsi, Mei 2026

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ABSTRAK

STUDI BIOINFORMATIKA VARIASI DAN EKSPRESI GEN YANG BERHUBUNGAN DENGAN *TAKAYASU ARTERITIS* (TA)

Latar Belakang: *Takayasu Arteritis* (TA) merupakan penyakit inflamasi kronis idiopatik yang menyerang pembuluh darah besar. Faktor genetik berperan penting dalam patogenesis penyakit TA, namun penelitian bioinformatika terkait variasi dan ekspresi gen pada TA masih terbatas.
Tujuan Penelitian: Penelitian ini bertujuan untuk mengidentifikasi gen risiko TA berdasarkan data genomik serta menganalisis hubungan variasi dengan ekspresi gen pada jaringan tubuh manusia.
Metode Penelitian: Penelitian ini menggunakan pendekatan bioinformatika dengan data sekunder dari GWAS *Catalog*, *HaploReg* v4.2, GTEx *Portal*, dan *Ensembl Genome Browser*. SNP diseleksi berdasarkan nilai *p-value* < 5×10^{-8}, kemudian dianalisis untuk mengidentifikasi variasi *missense*, pola ekspresi gen, dan distribusi frekuensi alel.
Hasil Penelitian: Sebanyak 166 SNP teridentifikasi berkaitan dengan TA, dengan 68 SNP memenuhi ambang signifikansi genomik. Enam SNP teridentifikasi sebagai variasi *missense*, yaitu rs3130618 (GPANK1), rs1576 (CCHCR1), rs9267532 (LY6G5B), rs10885 (PRRC2A), rs9267547 (LY6G6F), dan rs45479291 (HLA-V), dengan pola ekspresi yang relevan pada jaringan arteri, *whole blood*, paru-paru, dan testis.
Kesimpulan: Keenam gen kandidat yang teridentifikasi berpotensi berkontribusi terhadap patofisiologi TA melalui pola ekspresi gen pada berbagai jaringan tubuh.
Rekomendasi: Gen kandidat perlu dikembangkan sebagai biomarker *Takayasu Arteritis*, namun masih diperlukan penelitian lanjutan dengan populasi yang lebih luas untuk validasi hasil.

Kata Kunci;
Takayasu Arteritis, Bioinformatika, GWAS Catalog

[1] *Mahasiswa, Universitas Muhammadiyah Gombong*
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BACHELOR PHARMACY STUDY PROGRAM
FACULTY OF HEALTH SCIENCE
UNIVERSITAS MUHAMMADIYAH GOMBONG
Thesis, May 2026

Nabila Aulia Rahma[1)], Dwiki Fitri[2)]

ABSTRACT

BIOINFORMATIC STUDY OF GENE VARIATIONS AND EXPRESSION ASSOSIATED WITH *TAKAYASU ARTERITIS* (TA)

Background: *Takayasu Arteritis* (TA) is a chronic idiopathic inflammatory disease affecting the large blood vessels. Genetic factors play an important role in the pathogenesis of TA, however bioinformatics research on gene variations and expression in TA remains limited.
Purpose: This study aimed to identify TA risk genes based on genomic data and to analyse the relationship between genetic variations and gene expression across human tissues.
Method: This study employed a bioinformatics approach using secondary data from the GWAS Catalog, *HaploReg* v4.2, *GTEx Portal*, and *Ensembl Genome Browser*. SNPs were selected based on a p-value threshold of < 5 x 10-8 and further analysed to identify missense variants, gene expression patterns, and allele frequency distributions.
Result: A total of 166 SNPs were identified as associated with TA, of which 68 SNPs met the genome-wide significance threshold. Six SNPs were identified as missense variants, namely rs313618 (GPANK1), rs1576 (CCHCR1), rs9267532 (LY6G5B), rs10885 (PRRC2A), rs9267547 (LY6G6F), and rs45479291 (HLA-V), with relevant expression patterns observed in arterial tissue, whole blood, lung, and testis.
Conclusion: The six identified candidate genes potentially contribute to the pathophysiology of TA through tissue-specific gene expression patterns.
Recommendation: Candidate genes need to be developed as biomarkers for *Takayasu Arteritis*, however further studies with larger populations are still needed to validate the findings.

Keywords;
Takayasu Arteritis, Bioinformatics, GWAS Catalog

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[1] *Student of Universitas Muhammadiyah Gombong*
[2] *Lecturer of Universitas Muhammadiyah Gombong*

DAFTAR ISI

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DAFTAR GAMBAR

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DAFTAR LAMPIRAN

DAFTAR SINGKATAN

TA.	*Takayasu Arteritis*
GWAS.	*Genome Wide Association*
GTEx.	*Genome-Tissue Expression*
SNP.	*Single Nucleotide Polymorphisms*
HaploReg V4.2.	*HaploReg Versi 4.2*
eQTL.	*Expression Quantitative Trait Loct*
cis-eQTL.	*cis-Expression Quantitative Trait Loci*
AFR.	*Africa*
AMR.	*America*
EUR.	*Europe*
SAS.	*South Asia Summit*
EAS.	*East Asia Summit*
RNA.	*Ribonucleic Acid*
DNA.	*Deoxyribonucleic Acid*
NK.	*Natural Killer*
HLA.	*Human Leukocyte Antigen*
MHC.	*Major Histocompatibility Complex*
GPANK1.	*G-Patch Domain Ankyrin Repeats 1*
CCHCR1.	*Coiled-Coil alpha-Helical Rod protein 1*
PRRC2A.	*Proline Rich Coiled-Coil 2A*
LY6G6F.	*Lymphocyte antigen 6 family member G6F*
LY6G5B.	*Lymphocyte antigen 6 family member G5B*
HLA-V.	*Human Leukocyte Antigen V*
NES.	*Normalized Effect Size*
CC.	*Cytosine Cytosine*
TT.	*Thymine Thymine*
AA.	*Adenine Adenine*
GG.	*Guanine Guanine*
CA.	*Cytosine Adenine*
GA.	*Guanine Adenine*
GC.	*Guanine Cytosine*

CT.	*Cytosine Thymine*
mRNA.	*messenger Ribonucleic Acid*
DbSNP func annot.	*Database of Single Nucleotide Polymorphisms Functional Annotation*
Ref.	*Reference Allele*
Alt.	*Alternative Allele*
OFD1.	*Oral Facial Digital Syndrome 1*
PCM1.	*Pericentriolar Materials 1*
Lcn.	*Lipocalin*
TNF.	*Tumor Necrosis Factor*
ITGAM.	*Intergrin Subunit Alpha M*
GCA.	*Giant Cell Arteritis*
ADAM3	*A Disintegrin And Metalloprotease 3*
NMOSD.	*Neuromyelitis Optica Spectrum Disorder*
MS.	*Multiple Sclerosis*
TAP1.	*Transporter Associated with Antigen Processing 1*
TAP2.	*Transporter Associated with Antigen Processing 2*
LD.	*Linkage Disequilibrium*

BAB I
PENDAHULUAN

1.1 Latar Belakang

Pengembangan teknologi yang pesat saat ini sangat membantu dalam mengidentifikasi kerentanan genomik, terutama dalam mendeteksi variasi gen. Variasi gen yang berperan penting pada terjadinya kesalahan urutan pada struktur DNA yang tersusun atas basa nukleotida A, T, G, dan C (Astuti *et al*., 2023). *Takayasu Arteritis* (TA) merupakan suatu penyakit genetik yang disebabkan inflamasi kronis yang progresif dan penyebab aslinya belum diketahui secara pasti (idiopatik). Penyakit TA ini menyebabkan penyempitan pada pembuluh darah, oklusi (pembuluh darah tertutup total, sehingga aliran darah sempit), serta aneurisma (dinding pembuluh darah menjadi lemah dan menonjol) (Satriyanto, 2020). TA juga dikenal sebagai *pulseless disease*, *aortic arch syndrome*, dan *occlusive thromboaortopathy* (Wira Pratama Yasa *et al*., 2022).

Prevalensi TA di dunia mencapai 2,6 juta kasus per tahun yang banyak ditemukan di Jepang, Asia Tenggara, India, dan Amerika Selatan (Febrina, 2015). TA sering dialami oleh wanita muda berusia < 40 tahun, dengan rasio wanita terhadap pria sebesar 5-12:1 (Liu *et al*., 2022). Tingkat kejadian TA di Amerika Serikat dilaporkan sebesar 0,9 kasus per 1 juta penduduk, sementara di Eropa berkisar antara 0,4-1,5 kasus per 1 juta penduduk (Chair *et al*., 2018). Data mengenai TA di Benua Afrika masih terbatas. Akan tetapi, terdapat suatu studi terbesar di Afrika Selatan tepatnya di Rumah Sakit Groote Schuur, Provinsi Western Cape, dimana berdasarkan penelitian tersebut diketahui terdapat sebanyak 272 pasien yang mengalami TA (Botha, 2023). Angka kejadian TA di Indonesia juga masih jarang terjadi, namun di Pasuruan, Jawa Timur ditemukan pasien wanita berusia 18 tahun yang didiagnosis menderita TA (Lusida *et al*., 2020).

Gejala TA secara klinis dibedakan menjadi dua fase utama, yaitu fase awal dan fase stenosis. Fase awal ditandai oleh gejala umum yang tidak

spesifik seperti demam, rasa lelah, dan nyeri kepala, sedangkan gejala untuk fase stenosis ditandai oleh hipertensi, klaudikasio, bradikardi, *bruit*, serta angina (Rajlawot *et al*., 2022). Faktor risiko penyakit kardiovaskular sendiri terdapat pada pasien dengan hipertensi arteri, hiperlipidemia, serta kebiasaan merokok (Laurent *et al*., 2021). Penyebab pasti TA hingga kini belum diketahui, namun terdapat bukti ilmiah yang menunjukkan bahwa faktor genetik berperan penting dalam patogenesis penyakit ini. Sejumlah studi genomik, khususnya yang menggunakan pendekatan *Genome-Wide Association Study* (GWAS), telah berhasil mengidentifikasi sejumlah gen yang berpotensi dalam penyakit TA. Penelitian tersebut menunjukkan bahwa alel HLA-B*52:01 muncul sebagai penanda genetik paling kuat pada berbagai populasi (Ortiz-Fernández, 2021a).

Pemanfaatan basis data genomik menjadi salah satu pendekatan utama dalam mengidentifikasi hubungan antara gen dengan mekanisme suatu penyakit. Terdapat berbagai basis data yang memuat informasi mengenai keterkaitan antara gen dengan proses patogenesis suatu penyakit, salah satunya adalah katalog GWAS. Basis data ini berisi variasi genetik yang menghubungkan keseluruhan genom dengan fenotip yang dipetakan melalui *cis-expression quantitative trait loci* (cis-eQTL), yang berperan dalam memengaruhi karakteristik fenotip alami (Medi Sushanti *et al*., 2023) Katalog GWAS juga berfungsi untuk mengidentifikasi varian yang berasosisasi dengan penyakit umum pada populasi, yang secara akumulatif dapat berkontribusi terhadap mekanisme patogenesis penyakit seperti TA. Basis data lain yang digunakan yaitu *HaploReg* versi (4.2), GTEx *Portal*, dan *Ensembl genome browser 115*.

Al-Qur’an merupakan kitab suci umat islam yang diturunkan Allah SWT kepada Nabi Muhammad SAW, juga mengandung isyarat yang dapat dikaitkan dengan ilmu genetika. Salah satu ayat Al-Qur’an, menafsirkan terkait silsilah manusia yaitu berasal dari Nabi Adam AS dan Hawa. Hal ini dibuktikan dengan kesamaan ciri fisik manusia di bumi sebagai keturunan

mereka, serta proses penciptaan manusia dari setetes air mani yang kemudian berkembang dan menyebar ke seluruh penjuru dunia (Fadly *et al*., 2023). Ayat yang dikaitkan dengan ilmu genetika tersebut terdapat dalam surah An-Nisa' ayat 1, yang berbunyi :

يَا أَيُّهَا النَّاسُ اتَّقُوا رَبَّكُمُ الَّذِي خَلَقَكُم مِّن نَّفْسٍ وَاحِدَةٍ وَخَلَقَ مِنْهَا زَوْجَهَا وَبَثَّ مِنْهُمَا رِجَالًا كَثِيرًا وَنِسَاءً ۚ وَاتَّقُوا اللَّهَ الَّذِي تَسَاءَلُونَ بِهِ وَالْأَرْحَامَ ۚ إِنَّ اللَّهَ كَانَ عَلَيْكُمْ رَقِيبًا

Artinya :

"Wahai manusia, bertakwalah kepada Tuhanmu yang telah menciptakanmu dari diri yang satu (Adam) dan dia menciptakan darinya pasangan (Hawa). Dari keduanya Allah memperkembangbiakkan laki-laki dan perempuan yang banyak. Bertakwalah kepada Allah yang dengan nama-Nya kamu saling meminta dan (peliharalah) hubungan kekeluargaan. Sesungguhnya Allah selalu menjaga dan mengawasimu".

Berdasarkan penjelasan diatas, penelitian bioinformatika mengenai variasi dan ekspresi gen yang berkaitan dengan TA masih sangat terbatas, teutama penelitian yang memanfaatkan data dari katalog GWAS. Studi lanjutan berbasis data genomik diperlukan untuk menelusuri variasi dan ekspresi gen secara lebih spesifik. Pendekatan ini dinilai efisien dari segi waktu dan biaya serta berpotensi mengungkap gen risiko baru yang dapat memperdalam pemahaman terkait mekanisme molekuler TA.

1.2 Rumusan Masalah

Perumusan masalah berikut dapat dibuat berdasarkan latar belakang yang telah diuraikan, yaitu :

1.2.1 Apa saja gen yang berpotensi berperan dalam risiko terjadinya penyakit *Takayasu Arteritis* (TA) berdasarkan hasil penelitian pada basis data katalog GWAS dengan nilai *p-value* < 5 x 10^{-8}?

1.2.2 Apakah terdapat hubungan antara variasi gen dengan ekspresi gen yang teridentifikasi melalui basis data GTEx *Portal*?

1.3 Tujuan Penelitian

1.3.1 Tujuan Umum

Penelitian ini bertujuan untuk mengidentifikasi variasi gen yang berkaitan dengan TA serta menganalisis hubungan antara variasi dan ekspresi gen melalui pemanfaatan basis data dengan Metode Bioinformatika.

1.3.2 Tujuan Khusus

1.3.2.1 Mengetahui gen berisiko pada penyakit TA yang ditemukan dalam basis data dengan pendekatan bioinformatika.

1.3.2.2 Mengetahui hubungan antara variasi gen dengan tingkat ekspresi gen yang ditemukan pada jaringan tertentu.

1.4 Manfaat Penelitian

1.4.1 Manfaat Bagi Pengembangan Ilmu

Bagi pengembangan ilmu, penelitian ini berpotensi memperkaya pengetahuan ilmiah dibidang bioinformatika, terutama dalam pemanfaatan data genetik untuk mengidentifikasi gen-gen yang berperan dalam penyakit TA.

1.4.2 Manfaat Bagi Praktisi

Bagi praktisi, hasil penelitian ini dapat memberikan wawasan baru mengenai gen-gen yang berkontribusi terhadap risiko TA, sehingga dapat dijadikan referensi maupun literatur pertimbangan dalam penemuan variasi gen menggunakan pendekatan bioinformatika.

1.4.3 Manfaat Bagi Masyarakat

Penelitian ini diharapkan mampu meningkatkan pemahaman masyarakat mengenai peran faktor genetik pada TA.

1.4.4 Manfaat Bagi Peneliti

Kegiatan ini menjadi sarana pengembangan kompetensi dalam analisis data bioinformatika serta memberikan pengalaman dalam mengintegrasikan variasi genetik untuk mengidentifikasi hubungan antara variasi dan ekspresi gen pada TA.

1.5 Keaslian Penelitian

Keaslian penelitian ini disusun dengan merujuk pada sejumlah studi terdahulu yang memiliki keterkaitan topik dengan penelitian yang dilakukan. Meskipun demikian, penelitian-penelitian tersebut menunjukkan perbedaan pada aspek karakteristik subjek, jumlah dan penempatan variabel penelitian, serta pendekatan analisis yang digunakan.

Tabel 1. 1 Keaslian Penelitian

Nama, Peneliti, Tahun	Judul Penelitian	Metode Penelitian	Hasil Penelitian	Perbedaan dan Persamaan Penelitian
(Fitri *et al*., 2024)	Integrasi basis data genomik dan bioinformatika untuk mengidentifikasi varian seluruh genom pada penyakit genom pada penyakit *myasthenia gravis* di berbagai benua.	Pendekatan bioinformatika	Hasil penelitian ini yaitu ditemukan dua varian utama (rs2071591 dan rs231770) yang memengaruhi aktivitas gen LTA di jaringan otak dan gen CTLA4 di jaringan testis.	Persamaan : metode yang digunakan Perbedaan ; jenis penyakit

Nama, Peneliti, Tahun	Judul Penelitian	Metode Penelitian	Hasil Penelitian	Perbedaan dan Persamaan Penelitian
(Huang *et al*., 2018)	Analisis Bioinformatika Mengidentifikasi Tiga Gen yang Berpotensi Penting dan Terekspresi Berbeda pada Sel Mononuklear Darah Perifer Pasien dengan Arteritis Takayasu	Pendekatan bioinformatika	Ditemukan 932 gen terekspresi berbeda, terdiri dari 372 meningkat dan 560 menurun. Analisis jaringan protein menunjukkan empat gen utama (RHOA, FOS, EGR1, dan GNB1) sebagai gen sentral yang berpotensi berperan dalam patogenesis TA.	Persamaan : jenis penyakit Perbedaan : Metode yang digunakan - Menggunakan analisis ekspresi gen *Whole Human Genome Microarray* - Data ekspresi gen diunduh dari basis data *Gene Expression Omnibus* (GEO)
(Terao, 2018)	Determinasi Genetik dan Epistasis LILRA3 dan HLA-B*52 pada Arteritis Takayasu	Pendekatan bioinformatika	Hasil penelitian ini yaitu ditemukan empat lokus genetik baru yang terkait dengan *Takayasu Arteritis* (PTK2B, LILRA3/LILR	Persamaan : - Jenis penyakit - Menggunakan katalog GWAS Perbedaan : - Menggunakan nilai *p-value* ($5,0 \times 10^{-6}$

Nama, Peneliti, Tahun	Judul Penelitian	Metode Penelitian	Hasil Penelitian	Perbedaan dan Persamaan Penelitian
			B2, DUSP22, dan KLHL33) serta dua lokus tambahan (FCGR3A dan 21q.22) yang sebelumnya tidak dilaporkan pada populasi non Eropa.	$< p < 5{,}0 \times 10^{-5}$) - Tidak menggunakan gen *missense*

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LAMPIRAN

Lampiran 1. Lembar Bimbingan Proposal

	UNIVERSITAS MUHAMMADIYAH GOMBONG	Nomor	: PDN-SKP/12/005
		Revisi ke	: 02
		Tgl. Terbit	18 Agustus 2020
		Halaman	

LEMBAR BIMBINGAN PROPOSAL SKRIPSI

Nama mahasiswa : Nabila Aulia Rahma
NIM : 2022050196
Nama Pembimbing : apt. Dwiki Elki, M. Farm.

Tanggal Bimbingan	Topik/Materi Bimbingan	Paraf Mahasiswa	Paraf Pembimbing
26/9 - 2025	Bimbingan Judul		
29/9 - 2025	Bimbingan Judul, pengajuan judul		
13/10 - 2025	Bimbingan Bab I		
29/10 - 2025	ACC Bab I		
22/11 - 2025	Bimbingan bab II & III, ACC Bab II & III, Bimbingan PPT seminar		

Gombong, 22 Bulan 12 Tahun 2025

Mengetahui,

Koordinator Skripsi

apt. Wahyu Rahmatulloh, M.Farm
NIDN : 0617039602

Ka. Prodi S1 Farmasi

Dr. apt. Naclaz Zukhruf WK, M.Pharm.Sci
NIDN : 0618109202

Lampiran 2. Lembar Bimbingan Hasil

UNIVERSITAS MUHAMMADIYAH GOMBONG
FAKULTAS ILMU KESEHATAN
PROGRAM STUDI FARMASI PROGRAM SARJANA
Jl. Yos Sudarso No. 461 Gombong, Kebumen 54412 Telp./Fax. (0287) 472433, 473750
Website: www.unimugo.ac.id E-mail: s1farmasi@unimugo.ac.id

LEMBAR BIMBINGAN HASIL SKRIPSI

Nama Mahasiswa : NABILA AULIA RAHMA
NIM : 202205046
Nama Pembimbing : apt. Dwiki Fitri, M.Farm

Tanggal	Topik/Materi Bimbingan	Paraf Mahasiswa	Paraf Pembimbing
09 april 2026	Bimbingan bab 4 (hasil)		
22 april 2026	Revisi bab 4, bimbingan bab 5		
29 april 2026	Acc Bab 4, revisi Bab 5		
08 Mei 2026	revisi Bab 5 (pembahasan)		
09 Mei 2026	revisi Bab 5, Bimbingan abstrak		
12 Mei 2026	Acc Bab 5, revisi abstrak		
13 Mei 2026	Acc abstrak.		

Gombong, 12 Bulan Mei Tahun 2026

Mengetahui,
Koordinator Skripsi

apt. Wahyu Rahmatulloh, M.Farm

Ka. Prodi S1 Farmasi

apt. Tri Cahyani Widiastuti, M.Sc

DOKUMEN TERVERIFIKASI SISTEM SIMPUS
No : LB-H/FAR/2026/5046-FWSRM
Nama : NABILA AULIA RAHMA
NIM : 202205046
Tgl : 01 April 2026
Scan QR untuk verifikasi keaslian dokumen.
Tidak sah tanpa tanda tangan basah koordinator skripsi dan ka prodi s1 farmasi

SIMPUS Dokumen ini dicetak melalui Aplikasi SIMPUS Farmasi UM Gombong

Lampiran 3. Lembar Bimbingan Abstrak

UNIVERSITAS MUHAMMADIYAH GOMBONG
FAKULTAS ILMU KESEHATAN
PROGRAM STUDI FARMASI PROGRAM SARJANA
Jl. Yos Sudarso No. 461 Gombong, Kebumen 54412 Telp./Fax. (0287) 472433, 473750
Website: www.unimugo.ac.id E-mail: sl farmasi@unimugo.ac.id

LEMBAR REVISI ABSTRAK SKRIPSI

Nama Mahasiswa	:	NABILA AULIA RAHMA
NIM	:	202205046
Judul Skripsi	:	STUDI BIOINFORMATIKA VARIASI DAN EKSPRESI GEN YANG
Tanggal Ujian	:	Selasa, 26/05/2026 07.30–09.00
Tempat Ujian	:	Ruang G302
Batas Akhir Revisi	:	26 Juni 2026

NO	REVISI	PARAF PEMBIMBING
1.	Revisi abstrak bahasa inggris	
2.	ACC abstrak.	
3.		
4.		
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9.		
10.		

Gombong, 19 bulan Juni tahun 2026

Koordinator Skripsi,

apt. Wahyu Rahmatulloh, M.Farm

Pembimbing Abstrak,

Khamim Murtopo., M.Pd.

DOKUMEN TERVERIFIKASI SISTEM SIMPUS
No : LRA/FAR/2026/5046-YJDBZX
Nama : NABILA AULIA RAHMA
NIM : 202205046
Tgl : 15 Juni 2026
Scan QR untuk verifikasi keaslian dokumen.
Tidak sah tanpa tanda tangan basah koordinator skripsi dan pembimbing abstrak

SIMPUS Dokumen ini dicetak melalui Aplikasi SIMPUS Farmasi UM Gombong

Lampiran 4. Surat Pernyataan Tidak Melakukan Etik Penelitian

UNIVERSITAS MUHAMMADIYAH GOMBONG
FAKULTAS ILMU KESEHATAN
PROGRAM STUDI FARMASI PROGRAM SARJANA
Jl. Yos Sudarso No. 461 Gombong, Kebumen 54412 Telp./Fax. (0287) 472433, 473750
Website: www.unimugo.ac.id E-mail: s1farmasi@unimugo.ac.id

SURAT PERNYATAAN
TIDAK MELAKUKAN ETIK PENELITIAN

Nomor : ETK/FAR/2026/50460670
Tanggal : Gombong, 8 Mei 2026

Assalamu'alaikum wr wb

Saya yang bertanda tangan dibawah ini:

Nama	:	NABILA AULIA RAHMA
NIM	:	202205046
Prodi	:	Farmasi Program Sarjana
Judul skripsi	:	STUDI BIOINFORMATIKA VARIASI DAN EKSPRESI GEN YANG BERHUBUNGAN DENGAN TAKAYASU ARTERITIS (TA)

Menerangkan bahwa penelitian yang saya lakukan tidak bersinggungan dengan subyek yang berkaitan dengan manusia ataupun hewan. Demikian surat pernyataan ini saya buat dengan sebenar-benarnya dan untuk dipergunakan sebagai salah satu persyaratan ujian hasil penelitian (skripsi). Terimakasih.

Wassalamu'alaikum wr wb

Peneliti,

NABILA AULIA RAHMA
NIM. 202205046

Mengetahui,
Dosen Pembimbing Utama

apt. Dwiki Fitri, M.Farm
NUPTK. 4435777678230072

DOKUMEN TERVERIFIKASI SISTEM SIMPUS
No : ETK/FAR/2026/50460670
Nama : NABILA AULIA RAHMA
NIM : 202205046
Tgl : 8 Mei 2026
Scan QR untuk verifikasi keaslian
Tidak sah tanpa tanda tangan basah peneliti dan dosen pembimbing

SIMPUS Dokumen ini dicetak melalui
Aplikasi SIMPUS Farmasi UM Gombong

Lampiran 5. Surat Pernyataan Bebas Plagiarisme

UNIVERSITAS MUHAMMADIYAH GOMBONG
PERPUSTAKAAN
Jl. Yos Sudarso No. 461, Telp./Fax. (0287) 472433 GOMBONG, 54412
Website : https://library.unimugo.ac.id/
E-mail : lib.unimugo@gmail.com

SURAT PERNYATAAN CEK SIMILARITY/PLAGIASI

Yang bertanda tangan di bawah ini Pustakawan Universitas Muhammadiyah Gombong. Menyatakan bahwa karya tulis di bawah ini **sudah lolos** uji cek similarity/plagiasi:

Judul : Studi Bioinformatika Variasi dan Ekspresi Gen yang Berhubungan dengan Takayasu Arteritis
Nama : Nabila Aulia Rahma
NIM : 202205046
Program Studi : Farmasi Program Sarjana
Hasil Cek : 8 %

Gombong, 11 Mei 2026

Mengetahui,
Pustakawan Universitas Muhammadiyah Gombong

Dwi Sundari Yati

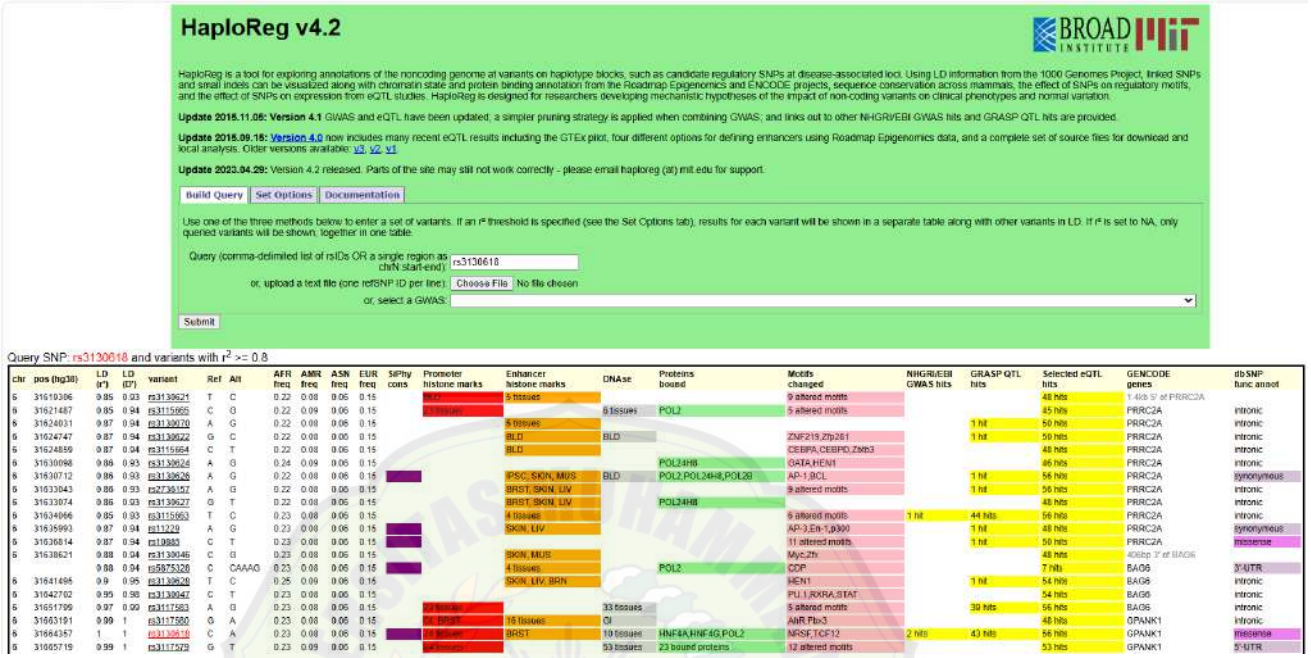
Lampiran 6. Hasil Analisi SNP yang ditemukan pada Basis Data Katalog GWAS sebanyak 166

No	SNPs	*p-value*	No	SNPs	*p-value*
1	rs74712413	1×10^{-5}	34	rs2852306	5×10^{-6}
2	rs74712413	1×10^{-5}	35	rs9615754	4×10^{-6}
3	rs13260543	9×10^{-6}	36	rs9615754	4×10^{-6}
4	rs12419742	9×10^{-6}	37	rs410852	4×10^{-6}
5	rs3847061	9×10^{-6}	38	rs7956657	4×10^{-6}
6	rs1250566	9×10^{-6}	39	chr6:33036435	4×10^{-6}
7	rs11675428	8×10^{-6}	40	rs1868971	3×10^{-6}
8	rs12511310	8×10^{-6}	41	rs10816540	3×10^{-6}
9	rs9380294	7×10^{-6}	42	rs375244	3×10^{-6}
10	rs5875188	7×10^{-6}	43	rs12596533	3×10^{-6}
11	rs2660739	7×10^{-6}	44	rs56167332	2×10^{-6}
12	rs76139923	7×10^{-6}	45	rs10470752	2×10^{-6}
13	rs1877209	7×10^{-6}	46	rs3812560	2×10^{-6}
14	rs76139923	7×10^{-6}	47	rs1113601	2×10^{-6}
15	rs115155040	6×10^{-6}	48	rs2322599	2×10^{-6}
16	rs113569359	6×10^{-6}	49	rs8111	1×10^{-6}
17	rs2242944	6×10^{-6}	50	rs4463283	1×10^{-6}
18	rs115155040	6×10^{-6}	51	rs34629529	1×10^{-6}
19	rs10144053	6×10^{-6}	52	rs34629529	1×10^{-6}
20	rs7005183	6×10^{-6}	53	rs7743661	1×10^{-6}
21	rs12571685	6×10^{-6}	54	rs9380141	1×10^{-6}
22	rs136301	6×10^{-6}	55	rs7843437	1×10^{-6}
23	rs136301	6×10^{-6}	56	rs12608210	1×10^{-6}
24	rs7956657	6×10^{-6}	57	rs12321966	1×10^{-6}
25	rs3817971	5×10^{-6}	58	rs4713281	1×10^{-6}
26	rs4624865	5×10^{-6}	59	rs6560480	9×10^{-7}
27	rs9917659	5×10^{-6}	60	rs1051957	9×10^{-7}
28	rs1462717	5×10^{-6}	61	rs2836883	8×10^{-7}
29	rs176237	5×10^{-6}	62	rs41267090	8×10^{-7}
30	rs4981676	5×10^{-6}	63	rs1376741	7×10^{-7}
31	rs28381348	5×10^{-6}	64	rs35409540	7×10^{-7}
32	rs6904014	5×10^{-6}	65	rs3869115	6×10^{-7}
33	rs12492780	5×10^{-6}	66	rs142314071	6×10^{-7}

No	SNPs	*p-value*	No	SNPs	*p-value*
67	rs142314071	6×10^{-7}	102	rs2069837	7×10^{-9}
68	rs77422524	5×10^{-7}	103	rs113452171	4×10^{-9}
69	chr6:26264671	5×10^{-7}	104	rs58693904	4×10^{-9}
70	rs1376741	4×10^{-7}	105	rs17133698	3×10^{-9}
71	chr21:40462378:D	4×10^{-7}	106	rs4379175	3×10^{-9}
72	rs10921544	3×10^{-7}	107	rs76457959	3×10^{-9}
73	rs2457837	3×10^{-7}	108	rs28749167	2×10^{-9}
74	rs7603494	2×10^{-7}	109	rs12111032	1×10^{-9}
75	rs1429365	2×10^{-7}	110	rs28834970	1×10^{-9}
76	rs12047961	2×10^{-7}	111	rs1116221	1×10^{-9}
77	rs9271539	2×10^{-7}	112	chr6:31570670	1×10^{-9}
78	rs2069837	2×10^{-7}	113	rs9267547	5×10^{-10}
79	rs2736173	2×10^{-7}	114	rs4817988	3×10^{-10}
80	rs9540128	1×10^{-7}	115	rs707936	2×10^{-10}
81	rs7911604	1×10^{-7}	116	rs73426382	2×10^{-10}
82	rs1611209	1×10^{-7}	117	rs1383258	2×10^{-10}
83	HLA-DQB1*0502	1×10^{-7}	118	rs4410768	2×10^{-10}
84	rs28399987	8×10^{-8}	119	rs12526858	2×10^{-10}
85	rs1610644	8×10^{-8}	120	rs12110437	2×10^{-10}
86	rs9273902	7×10^{-8}	121	HLAB*5201	2×10^{-10}
87	rs7038415	5×10^{-8}	122	rs4817984	1×10^{-10}
88	rs34109750	5×10^{-8}	123	rs11229	8×10^{-11}
89	rs28752871	5×10^{-8}	124	rs707915	6×10^{-11}
90	rs11666543	4×10^{-8}	125	rs3130628	5×10^{-11}
91	HLA-DQB1*0502	4×10^{-8}	126	rs3130626	5×10^{-11}
92	rs103294	3×10^{-8}	127	rs1062070	5×10^{-11}
93	rs4817984	3×10^{-8}	128	rs3130349	5×10^{-11}
94	rs9540128	2×10^{-8}	129	rs3130070	4×10^{-11}
95	rs12515766	2×10^{-8}	130	rs2736157	4×10^{-11}
96	rs6959179	2×10^{-8}	131	rs10885	4×10^{-11}
97	rs9266745	2×10^{-8}	132	rs2517985	3×10^{-11}
98	rs12980600	2×10^{-8}	133	rs9267535	3×10^{-11}
99	rs1264697	1×10^{-8}	134	rs3134940	3×10^{-11}
100	rs2523721	8×10^{-9}	135	rs1265078	3×10^{-11}
101	rs1713450	7×10^{-9}	136	rs3130627	3 x 10-11

No	SNPs	*p-value*	No	SNPs	*p-value*
137	rs1576	3×10^{-11}	152	rs12524487	6×10^{-16}
138	rs9267532	3×10^{-11}	153	rs45479291	2×10^{-16}
139	rs62392216	3×10^{-11}	154	HLAB*1302	2×10^{-16}
140	rs10573	3×10^{-11}	155	rs61444472	2×10^{-17}
141	rs9271066	2×10^{-11}	156	rs17193507	2×10^{-18}
142	rs3130618	2×10^{-11}	157	rs12524487	7×10^{-20}
143	rs62392216	1×10^{-11}	158	HLAB*5201	3×10^{-20}
144	HLAB*1501	1×10^{-11}	159	HLA-B*1302	3×10^{-25}
145	rs1265067	8×10^{-12}	160	rs17193507	7×10^{-30}
146	rs2099684	7×10^{-12}	161	rs9262437	5×10^{-31}
147	rs10919543	6×10^{-12}	162	rs117298325	4×10^{-34}
148	rs2836883	1×10^{-12}	163	HLA-B*5201	4×10^{-36}
149	rs2322599	3×10^{-13}	164	rs12524487	6×10^{-38}
150	rs12524487	3×10^{-15}	165	rs2844678	2×10^{-42}
151	HLA-B*1501	1×10^{-15}	166	rs2844678	6×10^{-47}

Lampiran 7. Hasil Analisis Basis Data HaploReg_Versi 4.2



HaploReg v4.2

BROAD INSTITUTE

HaploReg is a tool for exploring annotations of the noncoding genome at variants on haplotype blocks, such as candidate regulatory SNPs at disease-associated loci. Using LD information from the 1000 Genomes Project, linked SNPs and small indels can be visualized along with chromatin state and protein binding annotation from the Roadmap Epigenomics and ENCODE projects, sequence conservation across mammals, the effect of SNPs on regulatory motifs, and the effect of SNPs on expression from eQTL studies. HaploReg is designed for researchers developing mechanistic hypotheses of the impact of non-coding variants on clinical phenotypes and normal variation.

Update 2015.11.05: Version 4.1 GWAS and eQTL have been updated; a simpler pruning strategy is applied when combining GWAS; and links out to other NHGRI/EBI GWAS hits and GRASP QTL hits are provided.

Update 2015.09.15: Version 4.0 now includes many recent eQTL results including the GTEx pilot, four different options for defining enhancers using Roadmap Epigenomics data, and a complete set of source files for download and local analysis. Older versions available: v3, v2, v1

Update 2023.04.29: Version 4.2 released. Parts of the site may still not work correctly - please email haploreg (at) mit edu for support.

Build Query | Set Options | Documentation

Use one of the three methods below to enter a set of variants. If an r^2 threshold is specified (see the Set Options tab), results for each variant will be shown in a separate table along with other variants in LD. If r^2 is set to NA, only queried variants will be shown, together in one table.

Query (comma-delimited list of rsIDs OR a single region as chrN:start-end): rs3130618

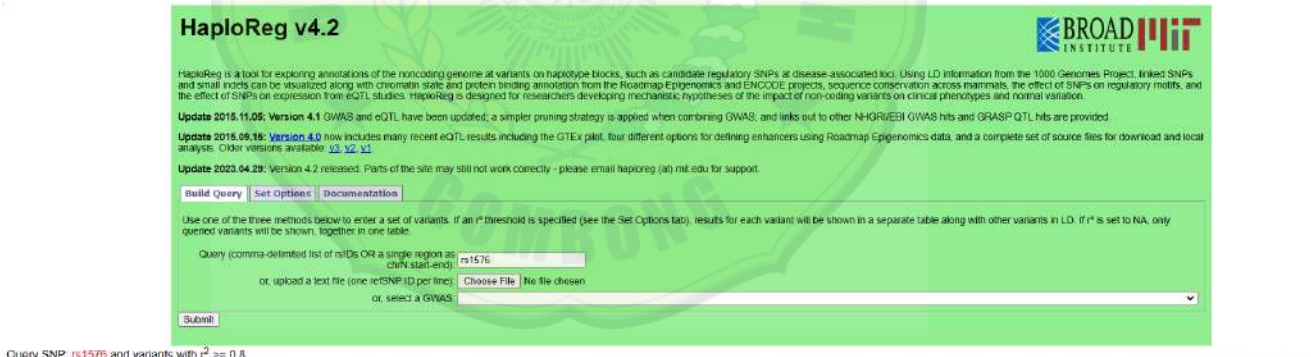
or, upload a text file (one rsID/SNP ID per line): Choose File | No file chosen

or, select a GWAS:

Submit

Query SNP: rs3130618 and variants with r^2 >= 0.8

chr	pos (hg38)	LD (r^2)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNAse	Proteins bound	Motifs changed	NHGRI/EBI GWAS hits	GRASP QTL hits	Selected eQTL hits	GENCODE genes	dbSNP func annot
6	31610306	0.85	0.93	rs3130621	T	C	0.22	0.08	0.05	0.15		BLD	5 tissues			9 altered motifs			48 hits	1.4kb 5' of PRRC2A	
6	31621487	0.85	0.94	rs3115665	C	G	0.22	0.09	0.05	0.15		21 tissues		6 tissues	POL2	5 altered motifs			45 hits	PRRC2A	intronic
6	31624031	0.87	0.94	rs3130670	A	G	0.22	0.08	0.05	0.15			5 tissues					1 hit	50 hits	PRRC2A	intronic
6	31624747	0.87	0.94	rs3130622	G	C	0.22	0.08	0.05	0.15			BLD	BLD		ZNF219,Zfp161		1 hit	50 hits	PRRC2A	intronic
6	31624859	0.87	0.94	rs3115664	C	T	0.22	0.08	0.05	0.15			BLD			CEBPA,CEBPD,Zfx3			48 hits	PRRC2A	intronic
6	31630098	0.86	0.93	rs3130624	A	G	0.24	0.09	0.05	0.15					POL2,HNF4G	GATA,HEN1			46 hits	PRRC2A	intronic
6	31630712	0.86	0.93	rs3130626	A	G	0.22	0.08	0.05	0.15			IPSC, SKIN, MUS	BLD	POL2,POL2AHB,POL2B	AP-1,BCL		1 hit	56 hits	PRRC2A	synonymous
6	31633043	0.86	0.93	rs2736157	A	G	0.22	0.08	0.05	0.15			BRST, SKIN, LIV			9 altered motifs		1 hit	56 hits	PRRC2A	intronic
6	31633074	0.86	0.93	rs3130627	G	T	0.22	0.08	0.05	0.15			BRST, SKIN, LIV		POL2AHB				48 hits	PRRC2A	intronic
6	31634066	0.85	0.93	rs3115663	T	C	0.23	0.08	0.05	0.15			4 tissues			6 altered motifs	1 hit	44 hits	56 hits	PRRC2A	intronic
6	31635993	0.87	0.94	rs11229	A	G	0.23	0.08	0.05	0.15			SKIN, LIV			AP-3,En-1,p300		1 hit	48 hits	PRRC2A	synonymous
6	31636814	0.87	0.94	rs19885	C	T	0.23	0.08	0.05	0.15						11 altered motifs		1 hit	50 hits	PRRC2A	missense
6	31638621	0.88	0.94	rs3130646	C	G	0.23	0.08	0.05	0.15			SKIN, MUS			Myc,Zfx			48 hits	406bp 3' of BAG6	
		0.88	0.94	rs5875326	C	CAAAG	0.23	0.08	0.05	0.15			4 tissues		POL2	CDP			7 hits	BAG6	3'-UTR
6	31641496	0.9	0.95	rs3130629	T	C	0.25	0.09	0.05	0.15			SKIN, LIV, BRN			HEN1		1 hit	54 hits	BAG6	intronic
6	31642702	0.95	0.98	rs3130647	C	T	0.23	0.08	0.05	0.15						PU.1,RXRA,STAT			54 hits	BAG6	intronic
6	31651799	0.97	0.99	rs3117583	A	G	0.23	0.08	0.05	0.15		22 tissues		33 tissues		5 altered motifs		38 hits	56 hits	BAG6	intronic
6	31663191	0.99	1	rs3117580	G	A	0.23	0.08	0.05	0.15		23 tissues	16 tissues	GI		AhR,Pbx3			48 hits	GPANK1	intronic
6	31664357	1	1	rs3130636	C	A	0.23	0.08	0.05	0.15		23 tissues	BRST	10 tissues	HNF4A,HNF4G,POL2	NRSF,TCF12	2 hits	43 hits	56 hits	GPANK1	missense
6	31665719	0.99	1	rs3117579	G	T	0.23	0.09	0.05	0.15		24 tissues		53 tissues	23 bound proteins	12 altered motifs			53 hits	GPANK1	5'-UTR



HaploReg v4.2

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Query (comma-delimited list of rsIDs OR a single region as chrN:start-end): rs1576

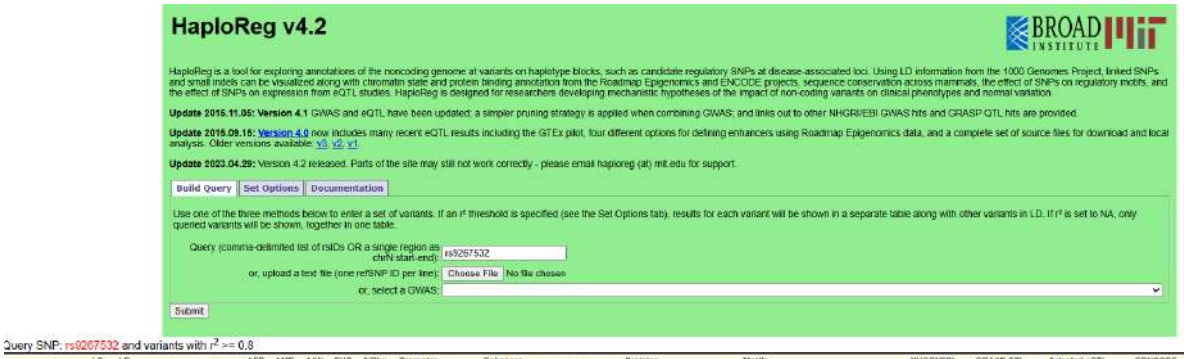
or, upload a text file (one rsID/SNP ID per line): Choose File | No file chosen

or, select a GWAS:

Submit

Query SNP: rs1576 and variants with r^2 >= 0.8

chr	pos (hg38)	LD (r^2)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNAse	Proteins bound	Motifs changed	NHGRI/EBI GWAS hits	GRASP QTL hits	Selected eQTL hits	GENCODE genes	dbSNP func annot
6	31139540	0.99	1	rs1265992	C	T	0.25	0.30	0.13	0.33		ESC,IPSC	17 tissues	ESDR		9 altered motifs			66 hits	PSORS1C1	intronic
6	31140494	0.99	1	rs199773024	C	CTCT	0.25	0.29	0.09	0.33			ESC, IPSC, BRST			E2A,Nkx2,Pbx2			29 hits	481bp 3' of PSORS1C1	
6	31142033	1	1	rs1265987	G	A,C,T	0.25	0.29	0.09	0.33			9 tissues	SKIN					74 hits	405bp 3' of CCHCR1	
6	31142169	0.89	1	rs2523834	T	A	0.22	0.28	0.08	0.30			9 tissues	SKIN		PU.1			51 hits	269bp 3' of CCHCR1	
6	31142170	0.89	1	rs2523833	C	T	0.22	0.28	0.08	0.30			9 tissues	SKIN		Irf,PU.1			51 hits	268bp 3' of CCHCR1	
6	31142614	1	1	rs1576	G	C	0.25	0.29	0.09	0.33			13 tissues	7 tissues		4 altered motifs		1 hit	53 hits	CCHCR1	missense
6	31144331	1	1	rs1265979	G	T	0.24	0.29	0.09	0.33		GI	ESDR, FAT, GI			E2A,Myf			53 hits	CCHCR1	intronic
6	31144825	1	1	rs1265978	G	C	0.24	0.29	0.09	0.33		GI	IPSC, SKIN, GI	4 tissues		4 altered motifs		2 hits	54 hits	CCHCR1	intronic
6	31145306	1	1	rs1265976	G	A	0.24	0.29	0.09	0.33		GI	IPSC, SKIN	GI		6 altered motifs		2 hits	53 hits	CCHCR1	intronic
6	31146163	0.99	0.99	rs746846	T	C	0.24	0.28	0.09	0.33			GI			GR			51 hits	CCHCR1	intronic
6	31146405	0.99	0.99	rs746647	A	G	0.24	0.29	0.09	0.33			GI			NRSF,Fax-4		5 hits	52 hits	CCHCR1	intronic
6	31147923	0.98	0.99	rs1265969	A	G	0.24	0.29	0.09	0.33						5 altered motifs			51 hits	CCHCR1	intronic
6	31148365	0.99	0.99	rs1265967	G	A	0.24	0.29	0.09	0.33			BLD, GI			Rora			51 hits	CCHCR1	intronic
6	31148433	0.82	0.99	rs130671	G	A	0.20	0.28	0.06	0.29			BLD			6 altered motifs			46 hits	CCHCR1	synonymous
6	31149411	0.99	0.99	rs1265114	C	T	0.24	0.29	0.09	0.33								14 hits	53 hits	CCHCR1	intronic
6	31150242	0.99	0.99	rs1265112	T	C	0.24	0.29	0.09	0.33			IPSC, LIV, GI			Egr-1,Znf143	1 hit	7 hits	53 hits	CCHCR1	intronic
6	31151165	0.98	0.99	rs2517985	T	C	0.24	0.29	0.09	0.33			ESDR			Gli3		2 hits	51 hits	CCHCR1	intronic
6	31151603	0.99	0.99	rs1265111	G	A	0.24	0.29	0.09	0.33			ESDR, IPSC			AP-2,EBFINSM1			51 hits	CCHCR1	intronic



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Query (comma-delimited list of rsIDs OR a single region as chrN:start-end): rs9267532

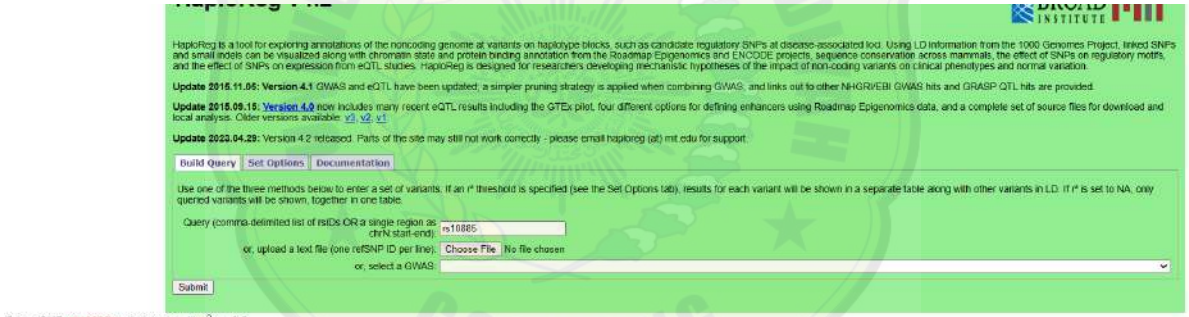
or, upload a text file (one refSNP ID per line): Choose File No file chosen

or, select a GWAS:

Submit

Query SNP: rs9267532 and variants with r^2 >= 0.8

chr	pos (hg38)	LD (r^2)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNAse	Proteins bound	Motifs changed	NHGRI/EBI GWAS hits	GRASP QTL hits	Selected eQTL hits	GENCODE genes	dbSNP func annot
6	31640165	0.84	0.92	rs9267525	T	C	0.08	0.05	0.06	0.07			18 tissues	MUS		AP-1			9 hits	BAG6	intronic
6	31656793	0.94	1	rs9267529	A	C	0.19	0.06	0.06	0.07		24 tissues	GI	8 tissues	POL2,ELF1	Foxa,GCNF,HDAC2		1 hit	17 hits	CSNK2B	intronic
6	31671352	0.94	1	rs2142234	G	A	0.19	0.06	0.06	0.07			SKIN			GR1,GR1b		4 hits	17 hits	LY6G5B	intronic
6	31672202	1	1	rs9267532	C	T	0.19	0.05	0.06	0.07						BCL		2 hits	16 hits	LY6G5B	missense
6	31676863	0.92	1	rs13295	A	G	0.19	0.06	0.06	0.07			FAT, BLD, LNG	BLD		EWSR1-FLI1,Pou2f2,Zbtb33		1 hit	17 hits	LY6G5C	3-UTR
6	31678145	0.92	1	rs9267533	C	T	0.19	0.06	0.06	0.07		SKIN	BLD, THYM	BLD		CHOP::C/EBPalpha,Mat,NF-E2		1 hit	17 hits	LY6G5C	intronic
6	31678880	0.92	1	rs9267534	T	C	0.19	0.06	0.06	0.07		SKIN	BLD					1 hit	17 hits	LY6G5C	intronic
6	31681372	0.92	1	rs9267535	C	G	0.19	0.06	0.06	0.07		21 tissues	BRN, GI, LIV	24 tissues	4 bound proteins	6 altered motifs			10 hits	LY6G5C	
6	31687891	0.92	1	rs10873	G	A	0.16	0.06	0.06	0.07					CTCF			1 hit	15 hits	ABHD16A	synonymous
6	31698829	0.92	1	rs9267540	C	T	0.19	0.06	0.06	0.07			BLD			HEN1,Pax-5			10 hits	ABHD16A	intronic
6	31699116	0.92	1	rs9267541	C	G	0.19	0.06	0.06	0.07			BLD, LIV			Bcl6b,p53			10 hits	ABHD16A	intronic
6	31700771	0.92	1	rs9267543	T	A	0.19	0.06	0.06	0.07			7 tissues		FOXA1,FOXA2	6 altered motifs			10 hits	ABHD16A	intronic
6	31704102	0.9	0.98	rs9267545	G	A	0.19	0.06	0.06	0.07		6 tissues	18 tissues	4 tissues	CEBPB,JUND	15 altered motifs			17 hits	LY6G6E	
6	31720423	0.8	0.9	rs376519	C	T	0.19	0.07	0.06	0.07		SKIN, FAT, DST	5 tissues	7 tissues		6 altered motifs		5 hits	12 hits	C6orf25	intronic
6	31732880	0.8	0.9	rs2736184	G	T	0.20	0.08	0.06	0.07			9 tissues			IK-1			6 hits	CLIC1	intronic
6	31749010	0.8	0.9	rs150793	A	G	0.20	0.07	0.06	0.07			BLD, BONE	GI	FOXA1,JUND,GATA3	Brachyury,Evi-1			14 hits	MSH5	intronic



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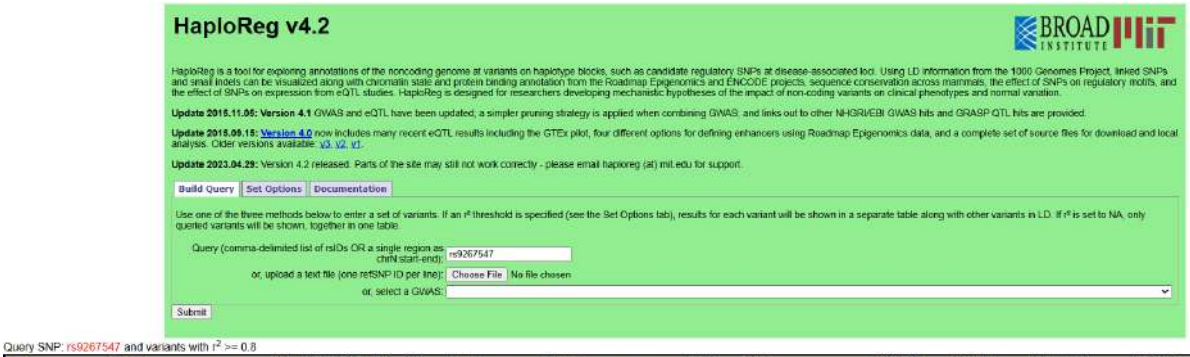
or, upload a text file (one refSNP ID per line): Choose File No file chosen

or, select a GWAS:

Submit

Query SNP: rs10885 and variants with r^2 >= 0.8

chr	pos (hg38)	LD (r^2)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNAse	Proteins bound	Motifs changed	NHGRI/EBI GWAS hits	GRASP QTL hits	Selected eQTL hits	GENCODE genes	dbSNP func annot
6	31614248	0.87	0.98	rs3132451	G	C	0.22	0.09	0.06	0.16		BLD, LIV	10 tissues			7 altered motifs			49 hits	926bp 5' of AIF1	
6	31619366	0.98	0.99	rs3130621	T	C	0.22	0.08	0.06	0.15		BLD	5 tissues			9 altered motifs			48 hits	1.4kb 5' of PRRC2A	
6	31621487	0.98	1	rs3115905	C	G	0.22	0.09	0.06	0.15		24 tissues		6 tissues	POL2	5 altered motifs			45 hits	PRRC2A	intronic
6	31624031	1	1	rs3130670	A	G	0.22	0.08	0.06	0.15			5 tissues					1 hit	50 hits	PRRC2A	intronic
6	31624747	1	1	rs3130622	G	C	0.22	0.08	0.06	0.15			BLD	BLD		ZNF219,Zfp281		1 hit	50 hits	PRRC2A	intronic
6	31624859	1	1	rs3115904	C	T	0.22	0.08	0.06	0.15			BLD			CEBPA,CEBPD,Zbtb3			48 hits	PRRC2A	intronic
6	31629923	0.92	0.98	rs3130623	C	T	0.24	0.08	0.06	0.15					POL24H8				35 hits	PRRC2A	intronic
6	31630098	0.99	1	rs3130624	A	G	0.24	0.09	0.06	0.15					POL24H8	GATA,HEN1			46 hits	PRRC2A	intronic
6	31639752	0.99	1	rs3130626	A	G	0.22	0.08	0.06	0.15			IPSC, SKIN, MUS	BLD	POL2,POL24H8,POL2B	AP-1,BCL		1 hit	56 hits	PRRC2A	synonymous
6	31633043	0.99	1	rs2736157	A	G	0.22	0.08	0.06	0.15			BRST, SKIN, LIV			0 altered motifs		1 hit	56 hits	PRRC2A	intronic
6	31633674	0.99	1	rs3130627	G	T	0.22	0.08	0.06	0.15			BRST, SKIN, LIV		POL24H8				48 hits	PRRC2A	intronic
6	31634066	0.98	0.99	rs3115903	T	C	0.23	0.08	0.06	0.15			4 tissues			6 altered motifs	1 hit	44 hits	56 hits	PRRC2A	intronic
6	31635983	1	1	rs11229	A	G	0.23	0.08	0.09	0.15			SKIN, LIV			AP-3,Ets-1,p300		1 hit	48 hits	PRRC2A	synonymous
6	31636814	1	1	rs10885	C	T	0.23	0.08	0.09	0.15						11 altered motifs		1 hit	50 hits	PRRC2A	missense
6	31638621	0.99	1	rs3130646	C	G	0.23	0.08	0.09	0.15			SKIN, MUS			Myc,Zfx			48 hits	408bp 3' of BAG6	
6	31638960	0.87	1	rs199900659	AAAAAC	A	0.23	0.08	0.09	0.13			SKIN, MUS			18 altered motifs			66 hits	127bp 3' of BAG6	
		0.99	1	rs5875126	C	CAAAG	0.23	0.08	0.09	0.15			4 tissues		POL2	CDP			7 hits	BAG6	3-UTR
6	31641485	0.95	0.98	rs3130628	T	C	0.25	0.09	0.09	0.15			SKIN, LIV, BRN			HEN1		1 hit	54 hits	BAG6	intronic
6	31642762	0.92	0.98	rs3130647	C	T	0.23	0.08	0.06	0.15						PU.1,RXRA,STAT			54 hits	BAG6	intronic
6	31651769	0.9	0.95	rs3117583	A	G	0.23	0.08	0.06	0.15		23 tissues		33 tissues		5 altered motifs		39 hits	56 hits	BAG6	intronic
6	31653191	0.88	0.94	rs3117580	G	A	0.23	0.08	0.06	0.15		24 tissues	18 tissues	GI		AhR,Pbx3			48 hits	GPANK1	intronic
6	31664357	0.87	0.94	rs3130618	C	A	0.23	0.08	0.06	0.15		24 tissues	BRST	10 tissues	HNF4A,HNF4G,POL2	NRSF,TCF12	2 hits	43 hits	50 hits	GPANK1	missense
6	31665719	0.88	0.94	rs3117579	G	T	0.23	0.09	0.06	0.15		24 tissues		53 tissues	23 bound proteins	12 altered motifs			53 hits	GPANK1	5'-UTR



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Query (comma-delimited list of rsIDs OR a single region as chrN:start-end): rs9267547

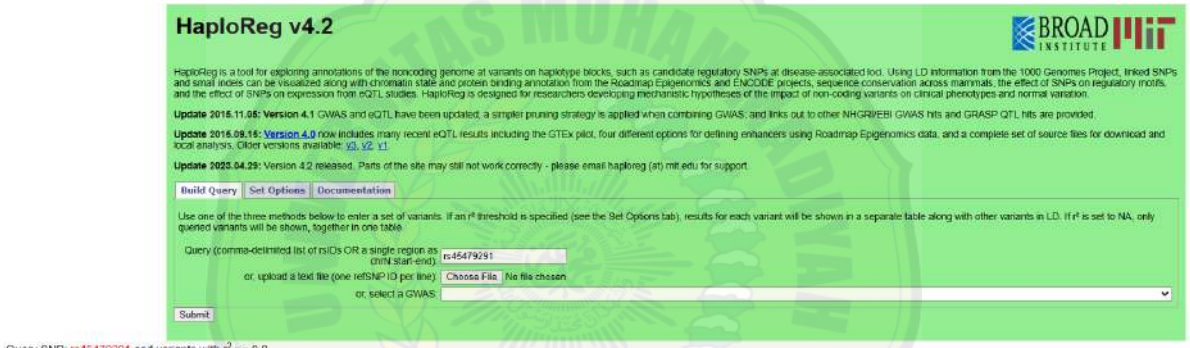
or, upload a text file (one rsID per line): Choose File | No file chosen

or, select a GWAS:

Submit

Query SNP: rs9267547 and variants with r^2 >= 0.8

chr	pos (hg38)	LD (r²)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNAse	Proteins bound	Motifs changed	NHGRI/EBI GWAS hits	GRASP QTL hits	Selected eQTL hits	GENCODE genes	dbSNP func annot
6	31705659	1	1	rs9267546	G	A	0.30	0.12	0.08	0.09			BLD			Hand1		13 hits	12 hits	LY6G6E	
6	31707724	1	1	rs9267547	G	A	0.30	0.12	0.09	0.09		SKIN	BLD					2 hits	11 hits	LY6G6F	missense
6	31708208	1	1	rs9267549	C	T	0.30	0.12	0.08	0.09						AhR::Arnt::Ahrr::p300			0 hits	LY6G6F	intronic



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Update 2023.04.29: Version 4.2 released. Parts of the site may still not work correctly - please email haploreg (at) mit.edu for support.

Build Query | Set Options | Documentation

Use one of the three methods below to enter a set of variants. If an r^2 threshold is specified (see the Set Options tab), results for each variant will be shown in a separate table along with other variants in LD. If r^2 is set to NA, only queried variants will be shown, together in one table.

Query (comma-delimited list of rsIDs OR a single region as chrN:start-end): rs45479291

or, upload a text file (one rsID per line): Choose File | No file chosen

or, select a GWAS:

Submit

Query SNP: rs45479291 and variants with r^2 >= 0.8

chr	pos (hg38)	LD (r²)	LD (D')	variant	Ref	Alt	AFR freq	AMR freq	ASN freq	EUR freq	SiPhy cons	Promoter histone marks	Enhancer histone marks	DNAse	Proteins bound	Motifs changed	NHGRI/EBI GWAS hits	GRASP QTL hits	Selected eQTL hits	GENCODE genes	dbSNP func annot
6	29602585	0.82	0.9	rs3998894	T	C	0.01	0.03	0.14	0.06					EGR1	15 altered motifs			38 hits	12kb 5' of ZFP57	
6	29602883	0.82	0.9	rs4607427	G	A	0.00	0.03	0.15	0.06						BCL,Hic1,YY1			38 hits	12kb 5' of ZFP57	
6	29603495	0.82	0.9	rs2076897	A	G	0.00	0.03	0.14	0.06						4 altered motifs			38 hits	12kb 5' of ZFP57	
6	29603525	0.82	0.9	rs2076896	G	A	0.00	0.03	0.14	0.06						YY1			38 hits	12kb 5' of ZFP57	
6	29603858	0.82	0.9	rs62391757	G	T	0.00	0.03	0.14	0.06						20 altered motifs			38 hits	13kb 5' of ZFP57	
6	29604992	0.82	0.9	rs62391758	C	T	0.00	0.03	0.14	0.06			ESC, IPSC, BLD	BLD		4 altered motifs			38 hits	14kb 5' of ZFP57	
6	29605140	0.82	0.9	rs149213917	C	CA	0.00	0.03	0.14	0.06			ESC, IPSC, BLD			EWSR1-FLI1,RBP-Jkappa			6 hits	14kb 5' of ZFP57	
6	29605680	0.82	0.9	rs56155978	C	T	0.00	0.03	0.14	0.06			ESC, IPSC			E2A,TBX5,ZEB1			38 hits	15kb 5' of ZFP57	
6	29697206	0.82	0.9	rs62391770	C	T	0.00	0.03	0.14	0.06			IPSC			Nanog			38 hits	16kb 5' of ZFP57	
6	29702221	0.82	0.9	rs56269224	C	G	0.00	0.03	0.14	0.06						5 altered motifs			38 hits	21kb 5' of HLA-F	
6	29705042	0.82	0.9	rs17184661	C	T	0.00	0.03	0.15	0.06						NERF1a			38 hits	18kb 5' of HLA-F	
6	29705929	0.82	0.9	rs62391785	C	T	0.00	0.03	0.14	0.06		ESC	4 tissues	48 tissues	4 bound proteins	9 altered motifs			38 hits	17kb 5' of HLA-F	
6	29706511	0.82	0.9	rs62391786	C	T	0.00	0.03	0.14	0.06			4 tissues						38 hits	16kb 5' of HLA-F	
6	29706969	0.82	0.9	rs116285516	T	C	0.00	0.03	0.14	0.06		BLD, THYM	5 tissues			6 altered motifs			40 hits	13kb 5' of HLA-F	
6	29710392	0.82	0.9	rs115651465	T	C	0.00	0.03	0.14	0.06		BLD	BLD, THYM, SPLN	BLD		SIX5,STAT,ZNF263			40 hits	12kb 5' of HLA-F	
6	29712937	0.86	0.93	rs11468601	C	T	0.00	0.03	0.14	0.06			BLD, BRN	BLD					40 hits	9.9kb 5' of HLA-F	
		0.86	0.93	rs17184695	G	A	0.00	0.03	0.14	0.06			BLD			4 altered motifs				9.5kb 5' of HLA-F	
		0.86	0.93	rs17184702	A	G	0.00	0.03	0.14	0.06			BLD			Irf				9.4kb 5' of HLA-F	
6	29723226	0.91	0.95	rs56944823	C	T	0.00	0.03	0.14	0.06		27 tissues	BRN, HRT	33 tissues	6 bound proteins	6 altered motifs			40 hits	HLA-F	
6	29725949	0.91	0.95	rs114325231	G	T	0.00	0.03	0.14	0.06			11 tissues	BLD,BLD,BLD	POL24H8	DBP,NF-kappaB			40 hits	HLA-F	intronic
6	29726066	0.91	0.95	rs62391891	A	G	0.00	0.03	0.14	0.06			BLD, HRT, SPLN		POL24H8	6 altered motifs			40 hits	HLA-F	intronic
6	29728031	0.91	0.95	rs56672647	A	G	0.00	0.03	0.14	0.06			BLD, SKIN, SPLN	BLD,BLD,BLD	POL24H8	CTCF,Rad21,Smad3			46 hits	HLA-F	intronic
6	29729643	0.91	0.95	rs62391819	A	G	0.00	0.03	0.14	0.06			BLD			10 altered motifs			38 hits	HLA-F	intronic
6	29732727	0.95	0.98	rs62391821	C	T	0.01	0.04	0.13	0.06						TBX5			38 hits	HLA-F	intronic
6	29734797	0.95	0.98	rs62391822	T	C	0.00	0.03	0.14	0.06			BRST, BLD, SKIN	7 tissues	PAX5C20	BCL,NF-kappaB			38 hits	HLA-F	intronic
6	29739746	0.9	0.97	rs62391833	T	C	0.00	0.03	0.14	0.06			4 tissues			4 altered motifs			38 hits	HLA-F-AS1	intronic
6	29740266	0.9	0.97	rs55661776	C	G	0.00	0.03	0.14	0.06			LNG, SKIN			BATF,Irf,VDR			38 hits	HLA-F-AS1	intronic
6	29740745	0.95	0.98	rs1413366	C	T	0.00	0.03	0.14	0.06			LNG, SKIN			ERalpha-a			38 hits	HLA-F-AS1	intronic
6	29743603	0.95	0.98	rs55762521	C	T	0.01	0.03	0.14	0.06			SKIN			5 altered motifs			38 hits	HLA-F-AS1	intronic
6	29746701	0.95	0.98	rs62391835	G	T	0.00	0.03	0.14	0.06		54 tissues	15 tissues	7 tissues		E2F,Sin3Ak-20,Znf143			38 hits	651bp 5' of HLA-F-AS1	
6	29751178	0.95	0.98	rs55684048	G	A	0.00	0.03	0.14	0.06		SKIN	SKIN, HRT, SPLN			PEBP,RFX5,Zfp410			38 hits	7958bp 3' of Y_RNA	
6	29755583	0.95	0.98	rs62391842	T	C	0.00	0.03	0.14	0.06		IPSC	6 tissues	ESC,IPSC	BCL3				38 hits	5.1kb 3' of Y_RNA	
6	29756737	0.95	0.98	rs62391843	A	T	0.00	0.03	0.14	0.06			ESC, IPSC			SEF-1			38 hits	6.3kb 3' of Y_RNA	
6	29760844	0.95	0.98	rs62391844	T	C	0.00	0.03	0.14	0.06						22 altered motifs			38 hits	10kb 3' of Y_RNA	
6	29764189	0.95	0.98	rs56287651	T	C	0.00	0.03	0.14	0.06			13 tissues	BRST,VAS		20 altered motifs			38 hits	14kb 3' of Y_RNA	
6	29770265	0.95	0.98	rs3998770	T	C	0.00	0.03	0.15	0.06			SKIN	SKIN		Pou5f1			38 hits	20kb 3' of Y_RNA	
6	29771223	0.95	0.98	rs3888722	C	G	0.00	0.03	0.15	0.06			IPSC, SKIN						40 hits	21kb 3' of Y_RNA	
6	29772247	0.95	0.98	rs62391850	G	A	0.00	0.03	0.15	0.06						4 altered motifs			38 hits	22kb 3' of Y_RNA	
6	29772771	0.95	0.98	rs62391851	G	A	0.00	0.03	0.15	0.06			ESDR			CEBPB,VDR			38 hits	23kb 3' of Y_RNA	
6	29773758	0.95	0.98	rs56362172	T	C	0.00	0.03	0.15	0.06						5 altered motifs			38 hits	23kb 3' of Y_RNA	
6	29775065	0.95	0.98	rs62394668	T	C	0.00	0.03	0.15	0.06						Ev-1,GR1,GR1b			38 hits	25kb 3' of Y_RNA	
6	29776358	0.95	0.98	rs146501834	A	C	0.00	0.03	0.15	0.06						9 altered motifs			8 hits	26kb 3' of Y_RNA	
6	29777454	0.93	0.98	rs2394670	A	G	0.00	0.03	0.15	0.06			ESC	SKIN,SKIN		6 altered motifs			38 hits	27kb 3' of Y_RNA	
6	29780732	0.95	0.98	rs139619383	C	T	0.00	0.03	0.15	0.06						AIRE,RREB-1			46 hits	30kb 3' of Y_RNA	
6	29785806	1	1	rs58290866	A	G	0.00	0.03	0.15	0.06									38 hits	35kb 3' of Y_RNA	
6	29786054	1	1	rs67382831	C	A	0.00	0.03	0.15	0.06			SKIN		USF1	13 altered motifs			38 hits	36kb 3' of Y_RNA	
6	29786591	0.91	1	rs116487225	T	G	0.00	0.04	0.15	0.06				IPSC		14 altered motifs			44 hits	36kb 3' of Y_RNA	
6	29789023	0.89	1	rs2394172	C	T	0.01	0.05	0.15	0.06						Zfp187			23 hits	38kb 5' of HLA-G	
6	29792252	1	1	rs45479291	G	C	0.00	0.03	0.15	0.06		21 tissues	BRN, HRT	10 tissues	POL2,YY1,RFX5	6 altered motifs			38 hits	35kb 5' of HLA-G	missense
6	29792759	1	1	rs45458963	G	A	0.00	0.03	0.15	0.06		21 tissues	ESDR, BRN, HRT	10 tissues		NRSF,RREB-1			38 hits	34kb 5' of HLA-G	intronic
6	29794401	1	1	rs19895757	C	T	0.00	0.03	0.15	0.06			SKIN			Hoxb13			40 hits	33kb 5' of HLA-G	intronic
6	29794862	1	1	rs55791844	A	T	0.00	0.03	0.15	0.06			FAT, SKIN	SKIN		5 altered motifs			38 hits	32kb 5' of HLA-G	intronic

Lampiran 8. Hasil Analisis Basis Data GTEx Portal

Significant Single-Tissue eQTLs for GPANK1 (ENSG00000204438.11) in all tissues

Data Source: GTEx Analysis Release V10 (dbGaP Accession phs000424.v10.p2)

Copy CSV Search: rs3130618 Show 10 entries

Gencode Id	Gene Symbol	Variant Id	SNP	P-Value	NES	Tissue	Actions
ENSG00000204438.11	GPANK1	chr6_31664357_C_A_b38	rs3130618 dbSNP	3.9e-8	-0.18	Nerve - Tibial	eQTL violin plot
ENSG00000204438.11	GPANK1	chr6_31664357_C_A_b38	rs3130618 dbSNP	2.6e-7	-0.17	Artery - Tibial	eQTL violin plot
ENSG00000204438.11	GPANK1	chr6_31664357_C_A_b38	rs3130618 dbSNP	0.000020	-0.14	Esophagus - Muscularis	eQTL violin plot
ENSG00000204438.11	GPANK1	chr6_31664357_C_A_b38	rs3130618 dbSNP	0.000021	-0.15	Thyroid	eQTL violin plot
ENSG00000204438.11	GPANK1	chr6_31664357_C_A_b38	rs3130618 dbSNP	0.00021	-0.15	Artery - Aorta	eQTL violin plot

Significant Single-Tissue eQTLs for CCHCR1 (ENSG00000204536.15) in all tissues

Data Source: GTEx Analysis Release V10 (dbGaP Accession phs000424.v10.p2)

Copy CSV Search: rs1576 Show 10 entries

Gencode Id	Gene Symbol	Variant Id	SNP	P-Value	NES	Tissue	Actions
ENSG00000204536.15	CCHCR1	chr6_31142614_G_C_b38	rs1576 dbSNP	5.3e-88	0.55	Adipose - Subcutaneous	eQTL violin plot
ENSG00000204536.15	CCHCR1	chr6_31142614_G_C_b38	rs1576 dbSNP	5.0e-81	0.51	Nerve - Tibial	eQTL violin plot
ENSG00000204536.15	CCHCR1	chr6_31142614_G_C_b38	rs1576 dbSNP	1.5e-77	0.44	Colon - Transverse	eQTL violin plot
ENSG00000204536.15	CCHCR1	chr6_31142614_G_C_b38	rs1576 dbSNP	1.7e-75	0.50	Thyroid	eQTL violin plot
ENSG00000204536.15	CCHCR1	chr6_31142614_G_C_b38	rs1576 dbSNP	1.1e-74	0.47	Adipose - Visceral (Omentum)	eQTL violin plot
ENSG00000204536.15	CCHCR1	chr6_31142614_G_C_b38	rs1576 dbSNP	2.2e-72	0.54	Lung	eQTL violin plot
ENSG00000204536.15	CCHCR1	chr6_31142614_G_C_b38	rs1576 dbSNP	6.6e-65	0.44	Breast - Mammary Tissue	eQTL violin plot
ENSG00000204536.15	CCHCR1	chr6_31142614_G_C_b38	rs1576 dbSNP	9.2e-59	0.39	Whole Blood	eQTL violin plot

Significant Single-Tissue eQTLs for LY6G5B (ENSG00000240053.8) in all tissues

Data Source: GTEx Analysis Release V10 (dbGaP Accession phs000424.v10.p2)

Copy CSV Search: rs9267532 Show 10 entries

Gencode Id	Gene Symbol	Variant Id	SNP	P-Value	NES	Tissue	Actions
ENSG00000240053.8	LY6G5B	chr6_31672202_C_T_b38	rs9267532 dbSNP	1.8e-10	-0.23	Muscle - Skeletal	eQTL violin plot
ENSG00000240053.8	LY6G5B	chr6_31672202_C_T_b38	rs9267532 dbSNP	5.5e-9	-0.21	Thyroid	eQTL violin plot
ENSG00000240053.8	LY6G5B	chr6_31672202_C_T_b38	rs9267532 dbSNP	1.1e-7	-0.15	Skin - Sun Exposed (Lower leg)	eQTL violin plot
ENSG00000240053.8	LY6G5B	chr6_31672202_C_T_b38	rs9267532 dbSNP	1.2e-7	-0.19	Nerve - Tibial	eQTL violin plot
ENSG00000240053.8	LY6G5B	chr6_31672202_C_T_b38	rs9267532 dbSNP	2.4e-7	-0.17	Adipose - Subcutaneous	eQTL violin plot
ENSG00000240053.8	LY6G5B	chr6_31672202_C_T_b38	rs9267532 dbSNP	2.4e-7	-0.25	Cells - Cultured fibroblasts	eQTL violin plot
ENSG00000240053.8	LY6G5B	chr6_31672202_C_T_b38	rs9267532 dbSNP	4.4e-7	-0.26	Colon - Transverse	eQTL violin plot
ENSG00000240053.8	LY6G5B	chr6_31672202_C_T_b38	rs9267532 dbSNP	6.1e-7	-0.21	Esophagus - Mucosa	eQTL violin plot
ENSG00000240053.8	LY6G5B	chr6_31672202_C_T_b38	rs9267532 dbSNP	0.0000036	-0.13	Whole Blood	eQTL violin plot

Significant Single-Tissue eQTLs for PRRC2A (ENSG00000204469.13) in all tissues

Data Source: GTEx Analysis Release V10 (dbGaP Accession phs000424.v10.p2)

Copy CSV Search: rs10885 Show 10 entries

Gencode Id	Gene Symbol	Variant Id	SNP	P-Value	NES	Tissue	Actions
ENSG00000204469.13	PRRC2A	chr6_31636814_C_T_b38	rs10885 dbSNP	0.00013	-0.053	Whole Blood	eQTL violin plot
ENSG00000204469.13	PRRC2A	chr6_31636814_C_T_b38	rs10885 dbSNP	0.00033	-0.11	Muscle - Skeletal	eQTL violin plot

Significant Single-Tissue eQTLs for LY6G6F (ENSG00000204424.10) in all tissues

Data Source: GTEx Analysis Release V10 (dbGaP Accession phs000424.v10.p2)

Copy CSV Search: rs9267547 Show 10 entries

Gencode Id	Gene Symbol	Variant Id	SNP	P-Value	NES	Tissue	Actions
ENSG00000204424.10	LY6G6F	chr6_31707724_G_A_b38	rs9267547 dbSNP	2.2e-7	-0.30	Testis	eQTL violin plot

Significant Single-Tissue eQTLs for HLA-V (ENSG00000181126.13) in all tissues

Data Source: GTEx Analysis Release V10 (dbGaP Accession phs000424.v10.p2)

Copy CSV Search: rs45479291 Show 10 entries

Gencode Id	Gene Symbol	Variant Id	SNP	P-Value	NES	Tissue	Actions
ENSG00000181126.13	HLA-V	chr6_29792252_G_C_b38	rs45479291 dbSNP	1.7e-9	0.54	Lung	eQTL violin plot
ENSG00000181126.13	HLA-V	chr6_29792252_G_C_b38	rs45479291 dbSNP	0.00017	-0.70	Spleen	eQTL violin plot

Lampiran 9. Hasil Analisis Basis Data *Ensembl Genome Browser*

rs9267532 SNP

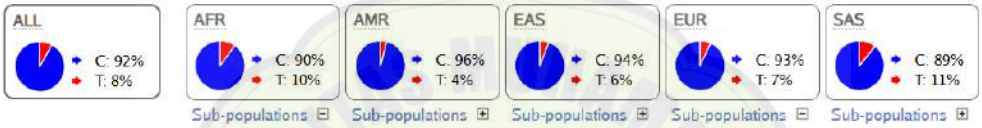
This variant maps to 7 locations

Select a location: None selected Go

Alleles	C/T
Location	This variant maps to 7 genomic locations. Please select a location in the box above.
Evidence status	
Synonyms	This variant has 4 synonyms - Show
Genotyping chips	This variant has assays on 13 chips - Show
Original source	Variants (including SNPs and indels) imported from dbSNP (release 156) | View in dbSNP
About this variant	This variant has 2504 sample genotypes, is associated with 21 phenotypes and is mentioned in 5 citations.

Population genetics

1000 Genomes Project Phase 3 allele frequencies



rs10885 SNP

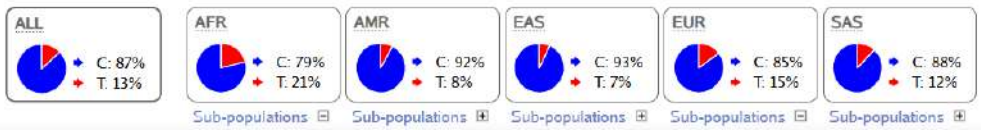
This variant maps to 7 locations

Select a location: None selected Go

Alleles	C/T
Location	This variant maps to 7 genomic locations. Please select a location in the box above.
Evidence status	
Clinical significance	
HGVS names	This variant has 4 HGVS names - Show
Synonyms	This variant has 11 synonyms - Show
Genotyping chips	This variant has assays on 6 chips - Show
Original source	Variants (including SNPs and indels) imported from dbSNP (release 156) | View in dbSNP
About this variant	This variant has 2504 sample genotypes, is associated with 22 phenotypes and is mentioned in 11 citations.

Population genetics

1000 Genomes Project Phase 3 allele frequencies



rs9267547 SNP

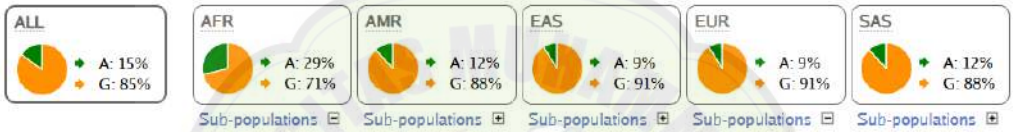
This variant maps to 7 locations

Select a location: None selected Go

Alleles	G/A
Location	This variant maps to 7 genomic locations. Please select a location in the box above.
Evidence status	
Synonyms	This variant has 5 synonyms - Show
Genotyping chips	This variant has assays on 6 chips - Show
Original source	Variants (including SNPs and indels) imported from dbSNP (release 156) | View in dbSNP
About this variant	This variant has 2504 sample genotypes, is associated with 7 phenotypes and is mentioned in 3 citations.

Population genetics

1000 Genomes Project Phase 3 allele frequencies



rs45479291 SNP

This variant maps to 8 locations

Select a location: None selected Go

Alleles	G/C | MAF: 0.09 (C)
Location	This variant maps to 8 genomic locations. Please select a location in the box above.
Evidence status	
Synonyms	Archive dbSNP rs62394673, rs1176604604, rs1160588856
Genotyping chips	This variant has assays on: Illumina_ExomeChip, Illumina_HumanOmni5, HumanCoreExome-12
Original source	Variants (including SNPs and indels) imported from dbSNP (release 156) | View in dbSNP
About this variant	This variant has predicted consequences for 1 regulatory feature, has 2504 sample genotypes, is associated with 8 phenotypes and is mentioned in 1 citation.

Population genetics

1000 Genomes Project Phase 3 allele frequencies