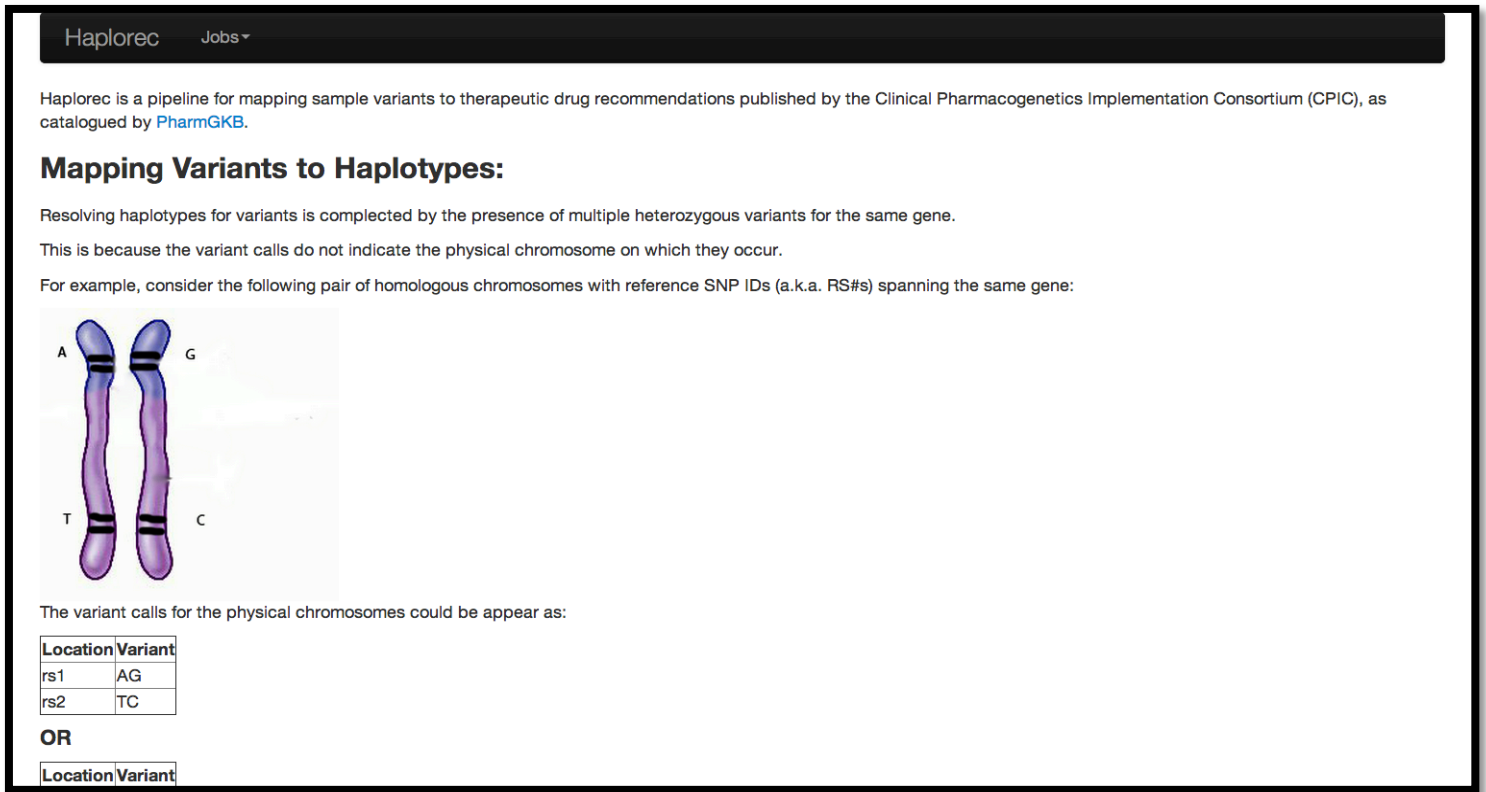


Haplorec User Guide

Home Page

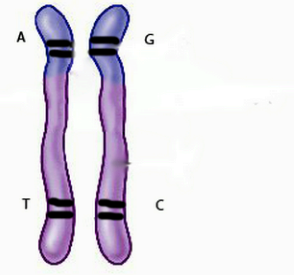
The Home page explains how the drug recommendations and novel haplotypes are generated from the variants that are inputted.



The screenshot shows the Haplorec Home Page. At the top is a dark navigation bar with 'Haplorec' and 'Jobs' (with a dropdown arrow). Below the bar, the page explains that Haplorec is a pipeline for mapping sample variants to therapeutic drug recommendations published by the Clinical Pharmacogenetics Implementation Consortium (CPIC), as catalogued by [PharmGKB](#).

Mapping Variants to Haplotypes:

Resolving haplotypes for variants is complicated by the presence of multiple heterozygous variants for the same gene. This is because the variant calls do not indicate the physical chromosome on which they occur. For example, consider the following pair of homologous chromosomes with reference SNP IDs (a.k.a. RS#s) spanning the same gene:



The variant calls for the physical chromosomes could appear as:

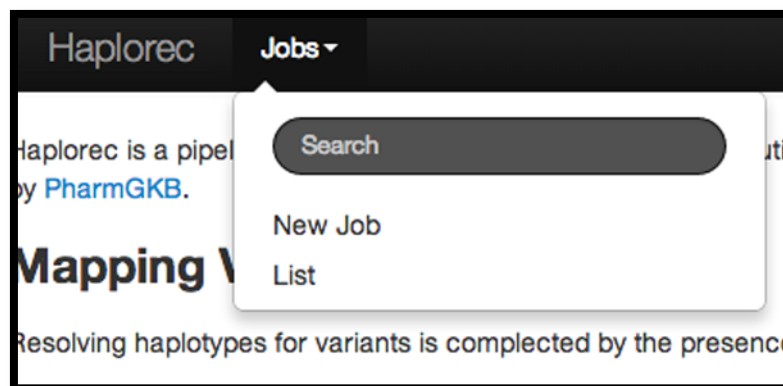
Location	Variant
rs1	A
rs2	T

OR

Location	Variant
rs1	G
rs2	C

The Navigation Bar

There are two buttons, Haplorec and Jobs. Click on **Haplorec** to return the Home Page. Click on **Jobs**, to get the following dropdown menu.



The screenshot shows the 'Jobs' dropdown menu. The menu is open, displaying three options: 'Search' (with a magnifying glass icon), 'New Job' (with a plus icon), and 'List' (with a list icon). The background shows the Haplorec Home Page content, including the title 'Mapping Variants to Haplotypes'.

- **Search**, searches through the list of created jobs and displays all the names of jobs that have that letter combination in them
- **New Job**, takes you to the Create Job page
- **List**, takes you to the list of jobs that have already been created

Create Job Page

In order to create a new job click on the **Jobs** button in the navigation bar and in the dropdown menu click on **New Job**. This takes you to the Create Page.

HaplorecJobs

Create Job

Job Name

Variants

Heterozygous Variant Combinations

Haplotypes

Genotypes

Phenotypes

Genotype Drug Recommendations

Novel Haplotypes

Phenotype Drug Recommendations

DataFile

Create

Scroll over the variant node to display a Sample Input file.

HaplorecJobs

Create Job

Job Name

Variants

Heterozygous Variant Combinations

Haplotypes

Genotypes

Phenotypes

Genotype Drug Recommendations

Novel Haplotypes

Phenotype Drug Recommendations

Sample Input File:

PLATE	EXPERIMENT	CHIP	WELL_POSITION	ASSAY_ID	GENOTYPE_ID	DESCRIPTION	SAMPLE_ID	ENTRY_OPERATOR
RA_CCP_RXN1_TCC-P5-7+SickKidsP12_May2011	1	1	B02	chr1_117098850	C	A.Conservative	1057-101938	Automatic
RA_CCP_RXN1_TCC-P5-7+SickKidsP12_May2011	1	1	B02	chr1_196991682	G	A.Conservative	1057-101938	Automatic
...

DataFile

Create

Click on the variant Node to add a file.

HaplorecJobs

Create Job

Job Name

Variables

one

1234 copy 2.txt2013-07-02 12:07 PM

chromosome_1.png2013-07-10 11:49 AM

DNA SEQUENCING...FORM- Updated.pdf2013-06-11 2:06 PM

drugrec.png2013-07-09 3:38 PM

G6PD.png2013-07-10 10:57 AM

Grails_files2013-06-17 11:00 AM

Grails.html2013-06-17 11:00 AM

matrix.png2013-07-10 11:54 AM

pgx_variants.txt2013-07-02 12:04 PM

PLATE EXPERIMENT CHIP WELL2013-06-27 9:06 AM

scons-2.3.0.tar.gz2013-06-04 10:54 AM

seq_order_form.pdf2013-06-07 11:45 AM

variants.txt2013-05-21 11:16 AM

Cancel

Open

Phenotypes

Phenotype Drug Recommendations

Novel Haplotypes

Sample Input File:

PLATE	EXPERIMENT	CHIP	WELL_POSITION	ASSAY_ID	GENOTYPE_ID	DESCRIPTION	SAMPLE_ID	ENTRY_OPERATOR
RA_CCP_RXN1_TCC-P5-7+SickKidsP12_May2011	1	1	B02	chr1_117098850	C	A.Conservative	1057-101938	Automatic
RA_CCP_RXN1_TCC-P5-7+SickKidsP12_May2011	1	1	B02	chr1_196991682	G	A.Conservative	1057-101938	Automatic
...

Data File

Create

Make sure you name the job with a unique name before creating it.
It will redirect you to the create page if you try to create a job with no name or with a name that already exists.

Job Name

jobThree

Variables

Then click the create button that appears at the bottom of the page.

This starts the process of creating the job. The create page will display the progress:

- Nodes with the loading spinner are currently being created
- Nodes that have no loading spinner are complete
- Nodes in red have failed

HaplorecJobs

Create Job

Job Name

jobThree

Variants

Heterozygous Variant Combinations

Sample Input File:

PLATE	EXPERIMENT	CHIP	WELL_POSITION	ASSAY_ID	GENOTYPE_ID	DESCRIPTION	SAMPLE_ID	ENTRY_OPERATOR
RA_CCP_RXN1_TCC-P5-7+SickKidsP12_May2011	1	1	B02	chr1_117098850	C	A.Conservative	1057-101938	Automatic
RA_CCP_RXN1_TCC-P5-7+SickKidsP12_May2011	1	1	B02	chr1_196991682	G	A.Conservative	1057-101938	Automatic
...

DataFile

1234 copy 2.txt

Choose FileDelete

Create

Once the Job is done being created, it redirects you to the Show job page.

Note:
The job will continue to be created if you leave the create page while it is in the process of being created. The progress will also be displayed on the job’s show page.

Also if a node fails, the job stops being created and you will be redirected to the show page.

HaplorecJobs

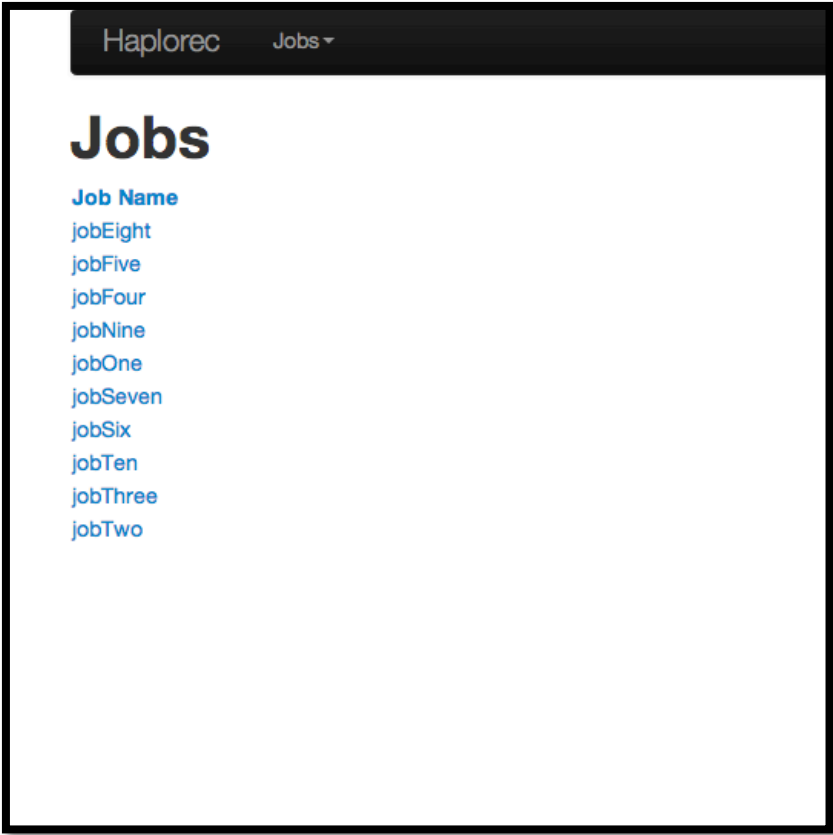
jobNine

Delete Job

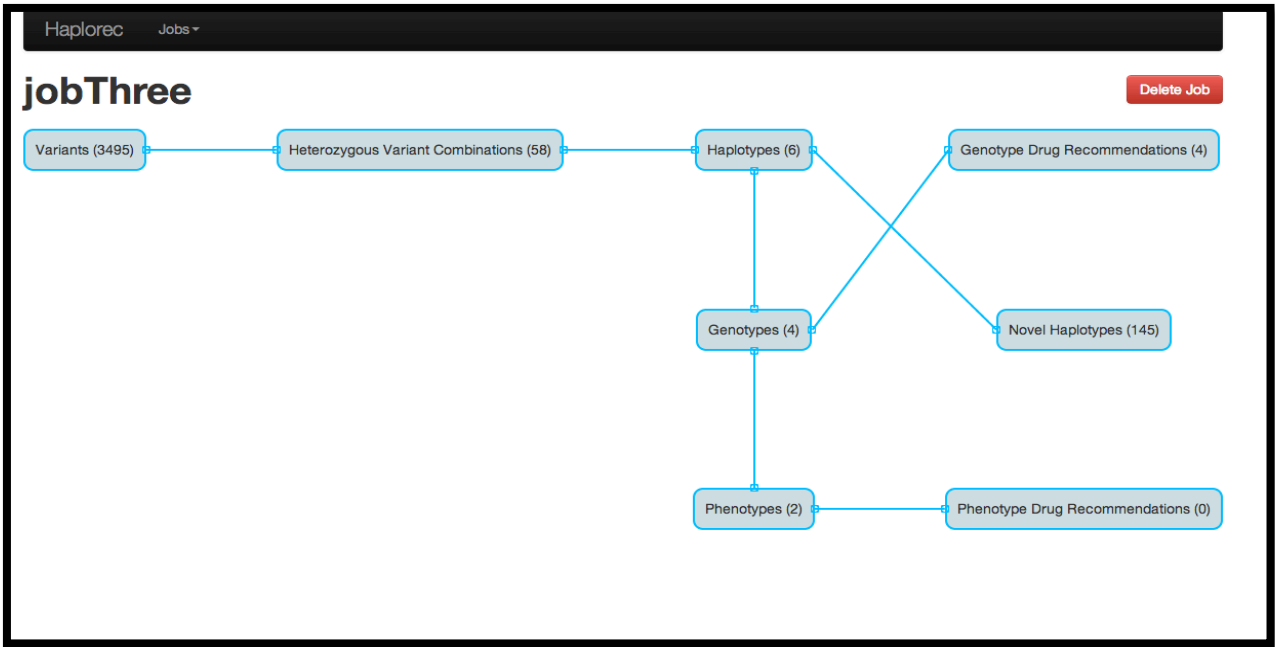
Variants (0)

Job List Page

The List page displays the list of created jobs in Alphabetical order, click on a job to go to the job’s Show Page.



Show Job Page



Click on a node to display its list.

HaplorecJobs

jobThree

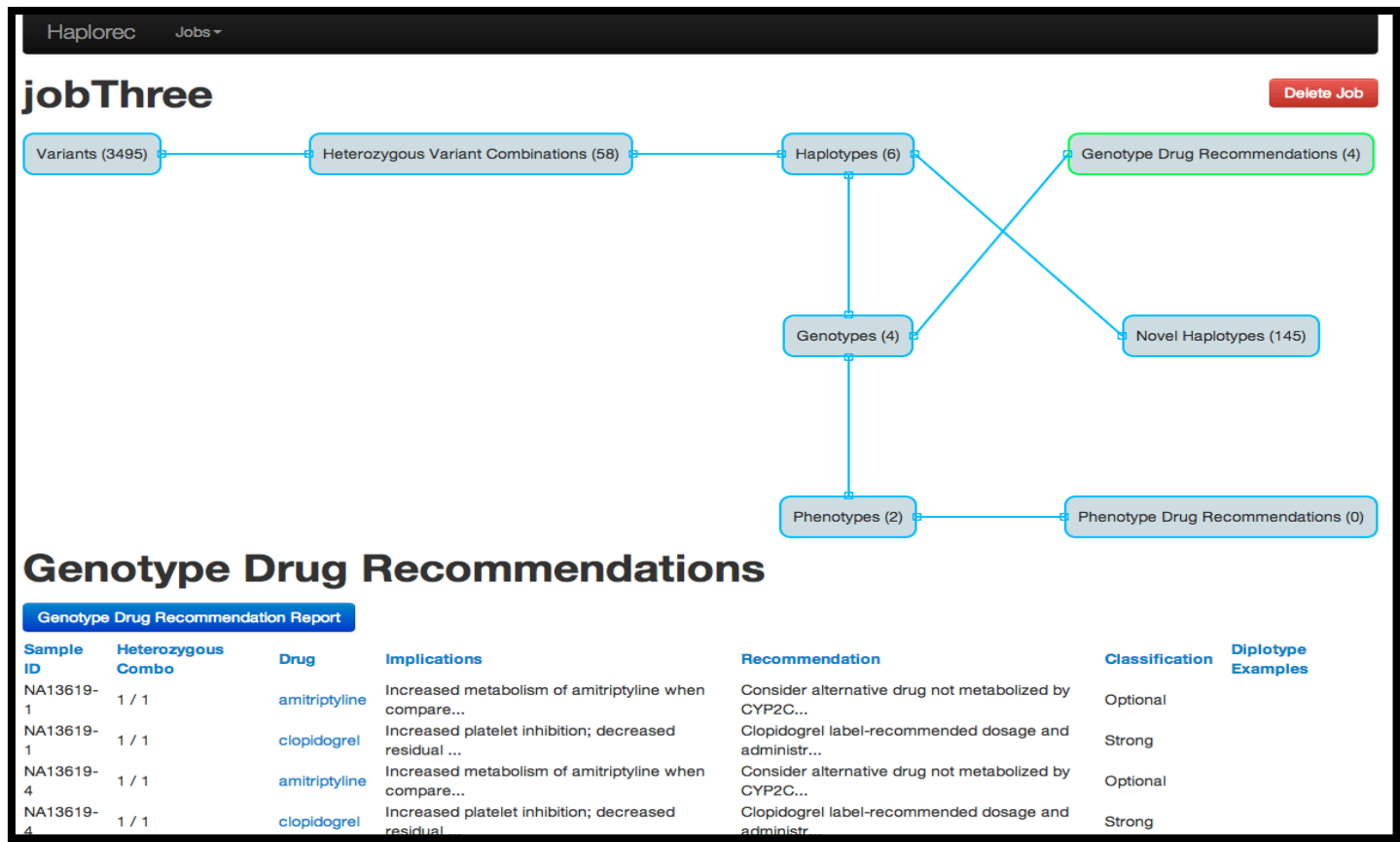
Delete Job

Nodes

Nodes

1234567891011121314151617181920212223242526272829303132333435363738394041424344454647484950515253545556575859606162636465666768697071727374757677787980818283848586878889909192939495969798991001011021031041051061071081091101111121131141151161171181191201211221231241251261271281291301311321331341351361371381391401411421431441451461471481491501511521531541551561571581591601611621631641651661671681691701711721731741751761771781791801811821831841851861871881891901911921931941951961971981992002012022032042052062072082092102112122132142152162172182192202212222232242252262272282292302312322332342352362372382392402412422432442452462472482492502512522532542552562572582592602612622632642652662672682692702712722732742752762772782792802812822832842852862872882892902912922932942952962972982993003013023033043053063073083093103113123133143153163173183193203213223233243253263273283293303313323333343353363373383393403413423433443453463473483493503513523533543553563573583593603613623633643653663673683693703713723733743753763773783793803813823833843853863873883893903913923933943953963973983994004014024034044054064074084094104114124134144154164174184194204214224234244254264274284294304314324334344354364374384394404414424434444454464474484494504514524534544554564574584594604614624634644654664674684694704714724734744754764774784794804814824834844854864874884894904914924934944954964974984995005015025035045055065075085095105115125135145155165175185195205215225235245255265275285295305315325335345355365375385395405415425435445455465475485495505515525535545555565575585595605615625635645655665675685695705715725735745755765775785795805815825835845855865875885895905915925935945955965975985996006016026036046056066076086096106116126136146156166176186196206216226236246256266276286296306316326336346356366376386396406416426436446456466476486496506516526536546556566576586596606616626636646656666676686696706716726736746756766776786796806816826836846856866876886896906916926936946956966976986997007017027037047057067077087097107117127137147157167177187197207217227237247257267277287297307317327337347357367377387397407417427437447457467477487497507517527537547557567577587597607617627637647657667677687697707717727737747757767777787797807817827837847857867877887897907917927937947957967977987998008018028038048058068078088098108118128138148158168178188198208218228238248258268278288298308318328338348358368378388398408418428438448458468478488498508518528538548558568578588598608618628638648658668678688698708718728738748758768778788798808818828838848858868878888898908918928938948958968978988999009019029039049059069079089099109119129139149159169179189199209219229239249259269279289299309319329339349359369379389399409419429439449459469479489499509519529539549559569579589599609619629639649659669679689699709719729739749759769779789799809819829839849859869879889899909919929939949959969979989991000100110021003100410051006100710081009101010111012101310141015101610171018101910201021102210231024102510261027102810291030103110321033103410351036103710381039104010411042104310441045104610471048104910501051105210531054105510561057105810591060106110621063106410651066106710681069107010711072107310741075107610771078107910801081108210831084108510861087108810891090109110921093109410951096109710981099110011011102110311041105110611071108110911101111111211131114111511161117111811191120112111221123112411251126112711281129113011311132113311341135113611371138113911401141114211431144114511461147114811491150115111521153115411551156115711581159116011611162116311641165116611671168116911701171117211731174117511761177117811791180118111821183118411851186118711881189119011911192119311941195119611971198119912001201120212031204120512061207120812091210121112121213121412151216121712181219122012211222122312241225122612271228122912301231123212331234123512361237123812391240124112421243124412451246124712481249125012511252125312541255125612571258125912601261126212631264126512661267126812691270127112721273127412751276127712781279128012811282128312841285128612871288128912901291129212931294129512961297129812991300130113021303130413051306130713081309131013111312131313141315131613171318131913201321132213231324132513261327132813291330133113321333133413351336133713381339134013411342134313441345134613471348134913501351135213531354135513561357135813591360136113621363136413651366136713681369137013711372137313741375137613771378137913801381138213831384138513861387138813891390139113921393139413951396139713981399140014011402140314041405140614071408140914101411141214131414141514161417141814191420142114221423142414251426142714281429143014311432143314341435143614371438143914401441144214431444144514461447144814491450145114521453145414551456145714581459146014611462146314641465146614671468146914701471147214731474147514761477147814791480148114821483148414851486148714881489149014911492149314941495149614971498149915001501150215031504150515061507150815091510151115121513151415151516151715181519152015211522152315241525152615271528152915301531153215331534153515361537153815391540154115421543154415451546154715481549155015511552155315541555155615571558155915601561156215631564156515661567156815691570157115721573157415751576157715781579158015811582158315841585158615871588158915901591159215931594159515961597159815991600160116021603160416051606160716081609161016111612161316141615161616171618161916201621162216231624162516261627162816291630163116321633163416351636163716381639164016411642164316441645164616471648164916501651165216531654165516561657165816591660166116621663166416651666166716681669167016711672167316741675167616771678167916801681168216831684168516861687168816891690169116921693169416951696169716981699170017011702170317041705170617071708170917101711171217131714171517161717171817191720172117221723172417251726172717281729173017311732173317341735173617371738173917401741174217431744174517461747174817491750175117521753175417551756175717581759176017611762176317641765176617671768176917701771177217731774177517761777177817791780178117821783178417851786178717881789179017911792179317941795179617971798179918001801180218031804180518061807180818091810181118121813181418151816181718181819182018211822182318241825182618271828182918301831183218331834183518361837183818391840184118421843184418451846184718481849185018511852185318541855185618571858185918601861186218631864186518661867186818691870187118721873187418751876187718781879188018811882188318841885188618871888188918901891189218931894189518961897189818991900190119021903190419051906190719081909191019111912191319141915191619171918191919201921192219231924192519261927192819291930193119321933193419351936193719381939194019411942194319441945194619471948194919501951195219531954195519561957195819591960196119621963196419651966196719681969197019711972197319741975197619771978197919801981198219831984198519861987198819891990199119921993199419951996199719981999200020012002200320042005200620072008200920102011201220132014201520162017201820192020202120222023202420252026202720282029203020312032203320342035203620372038203920402041204220432044204520462047204820492050205120522053205420552056205720582059206020612062206320642065206620672068206920702071207220732074207520762077207820792080208120822083208420852086208720882089209020912092209320942095209620972098209921002101210221032104210521062107210821092110211121122113211421152116211721182119212021212122212321242125212621272128212921302131213221332134213521362137213821392140214121422143214421452146214721482149215021512152215321542155215621572158215921602161216221632164216521662167216821692170217121722173217421752176217721782179218021812182218321842185218621872188218921902191219221932194219521962197219821992200220122022203220422052206220722082209221022112212221322142215221622172218221922202221222222232224222522262227222822292230223122322233223422352236223722382239224022412242224322442245224622472248224922502251225222532254225522562257225822592260226122622263226422652266226722682269227022712272227322742275227622772278227922802281228222832284228522862287228822892290229122922293229422952296229722982299230023012302230323042305230623072308230923102311231223132314231523162317231823192320232123222323232423252326232723282329233023312332233323342335233623372338233923402341234223432344234523462347234823492350235123522353235423552356235723582359236023612362236323642365236623672368236923702371237223732374237523762377237823792380238123822383238423852386238723882389239023912392239323942395239623972398239924002401240224032404240524062407240824092410241124122413241424152416241724182419242024212422242324242425242624272428242924302431243224332434243524362437243824392440244124422443244424452446244724482449245024512452245324542455245624572458245924602461246224632464246524662467246824692470247124722473247424752476247724782479248024812482248324842485248624872488248924902491249224932494249524962497249824992500250125022503250425052506250725082509251025112512251325142515251625172518251925202521252225232524252525262527252825292530253125322533253425352536253725382539254025412542254325442545254625472548254925502551255225532554255525562557255825592560256125622563256425652566256725682569257025712572257325742575257625772578257925802581258225832584258525862587258825892590259125922593259425952596259725982599260026012602260326042605260626072608260926102611261226132614261526162617261826192620262126222623262426252626262726282629263026312632263326342635263626372638263926402641264226432644264526462647264826492650265126522653265426552656265726582659266026612662266326642665266626672668266926702671267226732674267526762677267826792680268126822683268426852686268726882689269026912692269326942695269626972698269927002701270227032704270527062707270827092710271127122713271427152716271727182719272027212722272327242725272627272728272927302731273227332734273527362737273827392740274127422743274427452746274727482749275027512752275327542755275627572758275927602761276227632764276527662767276827692770277127722773277427752776277727782779278027812782278327842785278627872788278927902791279227932794279527962797279827992800280128022803280428052806280728082809281028112812281328142815281628172818281928202821282228232824282528262827282828292830283128322833283428352836283728382839284028412842284328442845284628472848284928502851285228532854285528562857285828592860286128622863286428652866286728682869287028712872287328742875287628772878287928802881288228832884288528862887288828892890289128922893289428952896289728982899290029012902290329042905290629072908290929102911291229132914291529162917291829192920292129222923292429252926292729282929293029312932293329342935293629372938293929402941294229432944294529462947294829492950295129522953295429552956295729582959296029612962296329642965296629672968296929702971297229732974297529762977297829792980298129822983298429852986298729882989299029912992299329942995299629972998299930003001300230033004300530063007300830093010301130123013301430153016301730183019302030213022302330243025302630273028302930303031303230333034303530363037303830393040304130423043304430453046304730483049305030513052305330543055305630573058305930603061306230633064306530663067306830693070307130723073307430753076307730783079308030813082308330843085308630873088308930903091309230933094309530963097309830993100310131023103310431053106310731083109311031113112311331143115311631173118311931203121312231233124312531263127312831293130313131323133313431353136313731383139314031413142314331443145314631473148314931503151315231533154315531563157315831593160316131623163316431653166316731683169317031713172317331743175317631773178317931803181318231833184318531863187318831893190319131923193319431953196319731983199320032013202320332043205320632073208320932103211321232133214321532163217321832193220322132223223322343223532236322373223832239322403224132242322433224432245322463224732248322493225032251322523225332254322553225632257322583225932260322613226232263322643226532266322673226832269322703227132272322733227432275322763227732278322793228032281322823228332284322853228632287322883228932290322913229232293322943229532296322973229832299323003230132302323033230432305323063230732308323093231032311323123231332314323153231632317323183231932320323213232232323323234323235323236323237323238323239323

Genotype Drug Recommendation, Novel Haplotypes, Phenotype Drug Recommendation have a report button, which when clicked, downloads a text file of the report. Also, click on a drug's name to get more information about the drug.



Novel Haplotypes’s list is displayed in a table.

Novel Haplotypes

Novel Haplotype Report

CYP2C19

CYP2C19

Haplotype	151A>G	19239G>A	87259A>G	87275G>A	rs11188072	rs11568732	rs118203756	rs118203757	rs118203759	rs12248!
*5B	A	G	A	G	C	T	G	G	C	C
*6	A	G	A	G	C	T	G	G	C	C
*7	A	G	A	G	C	T	G	G	C	C
*8	A	G	A	G	C	T	G	G	C	C
*9	A	G	A	G	C	T	G	G	C	C
NA11322-4, chrA (1/1)										C
NA11322-4, chrB (1/1)										C

If you want to look at another gene, select it from the dropdown list.
To scroll through the matrix make sure the mouse is on the lower right quadrant of the table.