



Searching for SNPs or Specific SNP Data Using a Specific ID

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Locating SNPs using Affymetrix IDs

How do I use Affymetrix IDs to find dbSNP refSNP (rs) numbers? The Affymetrix ID is: SNP_A-1508104

There are two methods you can use to search for the refSNP number using the Affymetrix ID:

Method #1:

1. Go to the [dbSNP home page](#) and select [Entrez SNP](#) from the dbSNP search options located just above the announcement section.
2. When you get to the Entrez SNP page, click on the Preview/Index tab located at the top of the page just below the search box.
3. Once on the Preview/Index page, click on the arrow next to the text box that reads "All Fields" to release a drop down menu.
4. Scroll down the drop-down menu and select "Submitter SNP ID"
5. In "Preview/Index" tab, choose "Submitter SNP ID" in drop down field, and place your Affymetrix ID in the empty text box to the right, and click the "preview" button.

Method #2:

1. Go back to the [dbSNP home page](#), and go to the "Search by IDs on All Assemblies" section, and Click the arrow next to the text box that says "Reference Cluster ID(rs#)". This will open a drop-down menu.
2. Select "Submitter SNP ID" (your Affymetrix ID is the SNP ID the Submitter gave to it, thus "Submitter SNP ID").
3. Place your Affymetrix ID in the text box to the left of the one that now reads "Submitter SNP ID", and press the "Search" button.

Your specific Affymetrix SNP ID did not turn up a match, so try the following:

1. Go back to the "Search by IDs on All Assemblies" section in the [dbSNP home page](#), and click the arrow next to the text box that says "Reference Cluster ID(rs#)" to re-open the drop-down menu.
2. Select "Submitter SNP ID"
3. Place the following truncated search term in the text box to the left: SNP_A-15*
4. Press the search button.

The [result page](#) for this search does at least indicate that dbSNP does contain SNPs with similar ID numbers (i.e. SNP_A-1507972http://www.ncbi.nlm.nih.gov/SNP/snp_viewTable.cgi?handle=AFFY). (7/27/07)

I inquired about Affymetrix number "SNP_A-1508104", and just heard back from Affymetrix. Apparently, SNP_A-1508104 is on their "10K 2.0 array", for which dbSNP does not yet have information — this would explain why we could not find this Affymetrix number in dbSNP.

Affymetrix tech support confirmed that SNP_A-1508104 does indeed correspond to rs1374108. (I think Affymetrix downloads dbSNP data and then uses the information in making their chips. That is why they know the corresponding refSNP[rs] number). Affymetrix recently submitted the sequences from four of their chip products to dbSNP (see below):

Mapping250K_Nsp

Mapping250K_Sty

Mapping50K_Hind240

Mapping50K_Xba240.

To access dbSNP data for the SNPs on these chips, users can query dbSNP by using the Affymetrix ID number. You can find more information at the [Affymetrix site](#). (8/03/07)

Locating SNPs using Celera IDs

I followed the instructions in the FAQs and can find rs IDs for Celera IDs starting with hCV, but not for Celera IDs starting with hDV.

Celera did not submit all of their variations to dbSNP. We have most (but not all) of Celera's human (hCV*) and mouse (mCV*) variations, but we don't have any that starts with hDV*. (02/22/08)

How do I use Celera ID numbers to find dbSNP refSNP (rs) numbers?

1. Go to the [SNP home page](#)
2. In the "Search by IDs" section of the home page, type the Celera ID number in the text box, and select "Celera SNP ID" from the drop down menu to the right of the text box.
3. Click on the "Search" button. (3/3/06)

How do I search for rs numbers that correspond to a list of hCV (Celera) SNP numbers? When I tried to "Search by ID", I got an ss number but not an rs number.

The reason why your list of hCV numbers has associated submitted SNP (ss) numbers but not refSNP (rs) numbers is that these particular variations have not yet been mapped and clustered.

rs numbers have not been assigned for these ss numbers in the current build. They will be automatically assigned during the next dbSNP build mapping and clustering process, and will be available when the new build is released. Information about the dbSNP rs numbering and clustering process is available in the [dbSNP handbook](#).

To receive notification of new build releases, subscribe to [dbSNP-announce](#). (2/16/06).

When I run a Batch query using Celera/ABI SNP IDs, the Chromosome report I get back doesn't contain the original query IDs, and is sorted by rs number, so I've lost correlation between the IDs. What can I do?

Only the FLATFILE report lists the submitter's ID (i.e. in your case, the hCV IDs) of all submitted SNPs. hCV IDs, however, are not reported for SNPs that are submitted by an entity other than Celera.

dbSNP does not have control over the assignment of specific rs numbers to hCV IDs. Not every rs number has a corresponding hCV ID, and Celera may change or drop the rs number based on their internal processing. Likewise, dbSNP's mapping and clustering process may change, merge, or drop rs numbers. I suggest that you convert your processing from hCV to dbSNP rs numbers, since rs numbers are the standard identifier for SNP.

There is a table of hCV IDs and their corresponding rs and ss numbers (for dbSNP build 127) located in in dbSNP's FTP site in the [misc. folder](#) of the human_9606 directory. The file name is hCV_dbSNP_ID.txt.gz. The column definitions are located in the hCV_dbSNP_ID readme file located in the [same directory](#) and folder. (2/13/07)

Locating SNPs using HGVS Recommended Nomenclature

Where can I search for variations using HGVS nomenclature?

There are two places you can use to search for variations using HGVS nomenclature:

Go to the "Search by IDs on All Assemblies" section of the [dbSNP home page](#), and select "HGVS names" from the dropdown menu.

OR

Use the [Human Variation: Search, Annotate, Submit](#) site

(04/17/08)

I am trying to find an rs number for a SNP called CD14-159C/T. Can you tell me what the rs number is or how I can find it?

You can use the [Human Variation: Search, Annotate, Submit](#) site to find the rs number you need, but you'll need the accession number for the reference sequence that codes for CD14 to do so. (04/09/08)

Will the HGVS based variation search you are testing also be available through eutils?

Yes, it should also be available through eutils once we have finished developing the resource (9/27/07)

Can you tell me how to find the rs number for a SNP when we have just position [TNFa (-308G/A)] to search with?

Use the [Human Variation: Search, Annotate, Submit](#) site, which performs a position search using names based on HGVS recommended nomenclature (e.g. NM_000594.2:c.-308G>A) that you can try, but in order to do so, you'll need to have a accession for the reference sequence (a mRNA, or protein, or a genomic sequence in GenBank). (09/07: 07/31/08)

Locating SNPs using Seattle SNPs IDs

How I match SNP names from the Seattle SNPs database (e.g. a Seattle SNPs named UI1498 in the IL10 gene) to dbSNP rs numbers in dbSNP?

Seattle SNPs names were submitted to dbSNP using the gene name concatenated with the position of the variation. For example, a couple of SNPs submitted for gene [CPSF4](#) were submitted by Seattle SNPs as CPSF4-000171 and CPSF4-000205.

1. To find the Seattle SNPs names you need to use to search for dbSNP IDs, go to the Seattle SNPs page for the gene of interest (our example, [CPSF4](#)). Once you're on this page, scroll down to the "Gene Specific Links" section of the page, and select the "SNP Context" link to see a list of names used by Seattle SNPs for SNPs in this gene (please note: As we have no control over Seattle SNPs web page, the link mentioned above may change in the future from this description).
2. Go to the "Search by IDs" section of the [dbSNP homepage](#).
3. Enter the Seattle SNPs names (ie.CPSF4-000171) into the text box.
4. Click on the box located immediately to the right of the text box to activate a drop-down menu. Select "Submitter SNP ID" from the menu.

- Click on the “Search” box to get the corresponding refSNP (rs) number for that submitter ID. (3/7/07)

Locating SNPs using STS IDs

What do I enter in the "Between Markers STS Search" field in the dbSNP search window?

Enter the name or ID numbers of an STS (i.e. SHGC-156091 and D1S3135) in dbSTS. (1/7/05)

How do I find the refSNP (rs) numbers for Y chromosome markers published by the Y-chromosome consortium (e.g. what is the corresponding rs number for STS DYS190)?

- Enter "DYS190" into [Entrez cross-database search](#), and click the “Go” button.
- You will find that there is a single hit (AF337053), which is located in the NCBI Nucleotide database. Click on the Nucleotide Icon to go to the Nucleotide result page.
- Click on the AF337053 link to get to the Sequence View page for this accession number. You will find the relationship between DYS190 and the human Y chromosome in the “features” section of this page:
/organism="Homo sapiens"
/mol_type="genomic DNA"
/db_xref="taxon: 9606"
/chromosome="Y"
/clone="3-11; DYS190"
- Click on the word “Links” at the top right corner of the sequence view page; clicking on it will release a dropdown menu. Select “Mapviewer” to see that AF337053 is located on the q11 region of Y.
- Click on the Y chromosome link to go to Mapviewer. Click on the “Maps & Options” button to add the "Variation" option to your Mapviewer display. Two SNPs are within 2000 bp of AF337053. (2/14/06)

Every time I search for STS markers in dbSNP, dbSNP returns a screen that says that no SNPs were found in my region of interest. What am I doing wrong?

I tested the STS search and it works for the query: between STS “RH98513” and “SHGC-74072”. For your query to work, the STS markers you use must exist in dbSTS and must be on the same chromosome. [Check your markers](#) against dbSTS to make sure that they are valid.

Locating SNPs using PubMed ID

If I know the PubMed number of a SNP, how do I get the rs ID of this SNP?

Query Pubmed using the ID (pmid). Once you get the result, you can either select SNP Link from Display, or you can or click on the blue word Links at the top right side of the result and select SNP, if it is shown. The SNP option is not shown if there is no SNP associated with the publication.

[Here's an example](#) of a publication with a SNP association.

Locating SNPs using CGAP IDs

How do I search for a SNP that is associated with a CGAP identifier?

dbSNP does not contain CGAP identifiers for one of two reasons: either SNPs with CGAP identifiers have not been submitted to dbSNP, or the SNPs were submitted using a different identifier.

If you know the identifier that a particular submitter has used in their submission to SNP, you can search for SNPs of interest using this identifier:

- Go to the [dbSNP homepage](#)

2. Go to the Search by IDs section, and enter the SNP identifier in the text box.
3. Select "Submitter SNP ID" from the drop-down menu to the right of the text box and click on the "search" button located below the text box.

If you have the sequence, you could also try a [BLAST](#) search. (3/14/06)

Locating SNPs using BAC Clone IDs

Why does the "[ACC]" term used in SNP searches return the following error message: "The following term was not found: AC019340[ACC]"?

We only index sequence accession numbers (NT and NM) that have SNPs mapped directly to them, and do not project them onto BAC clone sequences (ie. AC000120). (3/6/07)

Locating Specific Data Using RefSNP(rs) or Submitted SNP(ss) ID

Finding an RS Number Using an SS Number and Vice Versa

Is there a way to find a SNPs using an ss number? How would I use ss159830799 to find the rs number for that SNP?

Go to the [dbSNP home page](#), and enter ss159830799 in the text box located in the "Search by ID on all Assemblies" section located just below the announcement section near the top of the page, and select "NCBI Assay ID(ss#)" from the drop down menu to the right of the text box where you entered ss159830799. Click the "Search" button. (08/04/09)

How do I find the ss numbers for an rs number?

Go to the [dbSNP home page](#), and enter your rs number (in this example I will use rs3751143) in the text box located in the "Search by ID on all Assemblies" section located just below the announcement section near the top of the page, and select "Reference cluster ID(rs#)" (this should be the default setting) from the drop down menu to the right of the text box where you entered rs3751143. Click the "Search" button, and you will get a [refSNP cluster report](#) for that rs number. Scroll down the cluster report until you get to the submitter records section, where you will find all the ss numbers for this rs number. (08/04/09)

Finding Submitter (Local) ID used for a Particular SNP

I need data that shows the conversion between Baylor IDs and rs IDs, but the Batch Query limit is 30,000 SNP IDs, which means I'd have to run 56 batch queries. Is there an easier way?

You can download the [SubSNP](#) table from the FTP site and use that to lookup Baylor IDs using dbSNP rs IDs.

The [column definitions](#) for the SubSNP table can be found using dbSNP's [data dictionary](#) (07/08/08)

If I have a refSNP ID number, how do I find the trivial or local name used by the submitter in published manuscripts for that variation?

Use the "Search by IDs" query module that is located on the dbSNP homepage. It will allow you to select SNPs based on dbSNP record identifiers, including: the reference SNP (refSNP) cluster ID numbers (rs#), submitted SNP Accession numbers (ss#), and Submitter SNP (trivial/local) IDs for the same variations.

To use Search by IDs:

1. Enter the ID in the input text box and select the ID type from the drop down menu located next to the input text box. Now click "Search".
2. The result page will show the reference SNP (refSNP) cluster ID numbers (rs#), submitted SNP Accession numbers (ss#), and Submitter SNP (trivial/local) ID for the ID you submitted. (11/17/05)

Finding Submitter Information

How do I find the submitter and assay information for rs3918289, and how do I cite it in a journal article I am writing?

To find the submitter for rs3918289:

1. Go to the [dbSNP home page](#) .
2. Key in the rs number in the "Search by IDs on all Assemblies" section in the empty text box. Make sure the box to the left of the text box reads "Reference cluster ID(rs#)".
3. Push the "search" button located just below both boxes. You will get the [refSNP cluster report](#) for your rs number.
4. Go to the "submitter records" section of the report, and click on the NCBI Assay ID link for the only submitted SNP in the cluster, which is also the refSNP (ss5112514). You will get the [submitted SNPdetail page](#) for this SNP.
5. Click on the Handle Link "IPCDG" located near the top of the page to get information on the [submitter](#).

To find assay information for rs3918289:

1. Go to the [dbSNP home page](#)
2. Key in the rs number in the "Search by IDs on all Assemblies" section in the empty text box. Make sure the box to the left of the text box reads "Reference cluster ID(rs#)"
3. Push the "search" button located just below both boxes. You will get the [refSNP cluster report](#) for your rs number.
4. Go to the "submitter records" section of the report, and click on the NCBI Assay ID link for the only submitted SNP in the cluster, which is also the refSNP (ss5112514). You will get the [submitted SNPdetail page](#) for this SNP.
5. Look down the Submitted SNP detail page a little until you find the "Assay" section, and click on the method link "PROTOCOL-PCR" to get [detailed information](#) on the assay method.

For citation information, consult the "Citing dbSNP" section of this archive.(01/07/08)

Finding Lists of Chromosome Positions

How do I use dbSNP refSNP (rs) numbers to get a list of chromosome positions?

1. Go to the [dbSNP home page](#), and scroll down to the "Batch" section.
2. In the "upload list" sub-section, select "Reference SNP ID (rs)".
3. On the data request form, enter the email address where you want your data sent, select the organism of interest from the "organism" dropdown menu, and in the "Select Result Format" section choose "CHROMOSOME RPT" from the dropdown menu (3/1/06).

Finding Primers and Assay Conditions

How do I find the submitter and assay information for rs3918289, and how do I cite it in a journal article I am writing?

To find the submitter for rs3918289:

6. Go to the [dbSNP home page](#).
7. Key in the rs number in the "Search by IDs on all Assemblies" section in the empty text box. Make sure the box to the left of the text box reads "Reference cluster ID(rs#)".
8. Push the "search" button located just below both boxes. You will get the [refSNP cluster report](#) for your rs number.
9. Go to the "submitter records" section of the report, and click on the NCBI Assay ID link for the only submitted SNP in the cluster, which is also the refSNP (ss5112514). You will get the [submitted SNPdetail page](#) for this SNP.
10. Click on the Handle Link "IPCDG" located near the top of the page to get information on the [submitter](#).
To find assay information for rs3918289:

6. Go to the [dbSNP home page](#)
7. Key in the rs number in the "Search by IDs on all Assemblies" section in the empty text box. Make sure the box to the left of the text box reads "Reference cluster ID(rs#)"
8. Push the "search" button located just below both boxes. You will get the [refSNP cluster report](#) for your rs number.
9. Go to the "submitter records" section of the report, and click on the NCBI Assay ID link for the only submitted SNP in the cluster, which is also the refSNP (ss5112514). You will get the [submitted SNPdetail page](#) for this SNP.
10. Look down the Submitted SNP detail page a little until you find the "Assay" section, and click on the method link "PROTOCOL-PCR" to get [detailed information](#) on the assay method.

For citation information, consult the "[Citing dbSNP](#)" section of this archive.(01/07/08)

I have been searching dbSNP using a SNP accession number (rs2979099) to find information on primers and assay conditions. I can't find them. Can you help me?

Go to the dbSNP homepage and select Search by IDs, then enter the prefix "ss" next to the accession number (2979099) in the search box. You will then be taken to the [Submitted SNP\(ss\) Details](#) page for that accession number. To view the details of the experiment, locate the ASSAY section, and click on the link that follows the method "PROTOCOL_1".

Searching for Missing or Replaced refSNPs

The "rsCurrent" field in RsMergeArch contains 129,000 rs numbers not in the ASN.1 flat files or in a dbSNP web search. Is there a list of expired rs numbers?

Let me explain how we track both merged and deleted (an entirely different process from merging) refSNP (rs) numbers, by using a hypothetical example where a "chain merge" (multiple rs numbers merge into each other) occurs:

For example, let us say rs "A" merged into rs "B", and later, rs "B" merged into rs "C". As a result of the first merge, the entry for rs "A" in rsCurrent

is updated to rs "B"; after the second merge, rsCurrent is then updated to rs "C". Now, if rs "C"'s submitters withdraw all the member ss numbers within the refSNP cluster rs "C", then rs "C" will get an entry in the SNPHistory table (the SNPHistory table ONLY contains SNPs that have "become history" — that is, SNPs that have been completely deleted). Please see ftp file for SNPHistory.bcp (located in the [snp/database/organism_data/species_of_interest](#) directory). To find the column names for the SNPHistory table, download the human_9606_table.sql, which is located in the [human organism_schema](#) directory.

Getting back to RsMergeArch: since "withdrawing rs "C" is not a merge action, the table RsMergeArch is not updated. RsMergeArch is used to track "rs merge" actions only. I can see that this might be confusing, so when time allows, we will add the following explanation to the RsMergeArch table definition, to make the RsMergeArch.rsCurrent meaning clearer:

RsMergeArch is used to track each rs merge event.

If an rs number in RsMergeArch.rsCurrent is withdrawn from dbSNP by submitter request, then an the rs number of the same value as that in rsCurrent will be entered into the SNPHistory table (which contains deleted rs numbers only).

Please note: "rsCurrent" in RsMergeArch does not mean the "current rs number" in the current dbSNP build".
(08/12/08)

Where can I get a list of all deleted rs clusters? The merge history is available (RsMergeArch), but I don't see the equivalent for deleted rs numbers.

You will find a list of deleted SNPs in the SNPHistory table, which is located in the same FTP directory where the RsMergeArch table is located.

The SNPHistory table does not track history of a SNP — it only stores SNPs that have been deleted (i.e. "they're 'history"). Currently, the [dbSNP Data Dictionary](#) does not contain a description of this table or its columns. I will try to ass them as time permits.

To find the column names for the SNPHistory table, download the human_9606_table.sql in the [human organism_schema directory](#) (06/11/08)

A publication gives rs2857713 a SNP, but when I looked for it in dbSNP, it took me some time to find that it changed to rs2229094. Can't you inform users when refSNP numbers have changed?

dbSNP does provide notice when refSNPs have merged. Currently, there are three different entry points in dbSNP that will lead you to the partner numbers of a merge:

1. You can retrieve a list of merged rs numbers from [Entrez SNP](#). Just type "mergedrs" (without the quotation marks) in the text box at the top of the page and click the "go" button. Each entry in the returned list will include the old rs numbers that has merged, and the new rs number it has merged into (with a link to the refSNPpage for the new rs number). You can limit the output to merged rs numbers within a certain species by clicking on the "Limits" tab and then selecting the organism you wish from the organism selection box.
2. If you enter an old rs number (in this case **rs2857713**) into the "Search for IDs" search on the dbSNP home page, the response page will state that the SNP has been merged, and will provide the new rs number (in this case **rs2229094**) and a link to the refSNP page for that new rs number.
3. The [RsMergeArch table](#) houses the merged SNPs, and is available on the dbSNP ftp site. A full description of the table can be found in the [dbSNP Data Dictionary](#), and the column definitions are located in the dbSNP_main_table.sql.gz, which can be found in the [shared_schema](#) directory of the dbSNP FTP site.

(9/07:11/07:11/08)

I had heard that RS numbers are not stable. For example, rs17216163 is now rs717620, and rs17231380 is now rs5186. I assume that you don't ever reuse the "retired" numbers? Why did you make some numbers obsolete?

The examples you cite are instances where multiple rs numbers were assigned at the same genomic location, and the higher rs number was merged into a lower rs number (this is the dbSNP merge rule for rs numbers). Such a merge can happen when submissions differ in the length and quality of flanking sequence. We only

merge rs numbers that have an identical set of mappings to the genome and have the same type of alleles (e.g. both must be the same variation type and share one allele in common). We would not merge a SNP and an indel (insertion/deletion) into a single rs number (different variation classes) since they represent to different types of mutational "events".

The location of the rs number remains valid and we never reuse rs numbers.

We have discussed the issue of supporting query by merged rs numbers more robustly in dbSNP, Entrez and our web based services. That way a retired rs number can be found easily and used as a proxy for the current "live" number. Please note that merging is only used to reduce redundancy in the catalog of rs numbers so each position has a unique identifier.

In the first example you cite, prior to their merge, both rs17216163 and rs717620 would have been the "address" for the same nucleotide. Now only rs717620 is used in annotation, and rs17216163 is retained in our merge history tables. With extended annotation, users would be able to query by the full set of retired rs numbers.

Currently, there are three different entry points in dbSNP that will lead you to the partner numbers of a merge:

1. You can retrieve a list of merged rs numbers from [Entrez SNP](#). Just type "mergedrs" (without the quotation marks) in the text box at the top of the page and click the "go" button. Each entry in the returned list will include the old rs numbers that has merged, and the new rs number it has merged into (with a link to the refSNP page for the new rs number). You can limit the output to merged rs numbers within a certain species by clicking on the "Limits" tab and then selecting the organism you wish from the organism selection box.
2. If you enter a merged old rs number into the "Search for IDs" search on the dbSNP home page, the response page will state that the SNP has been merged, and will provide the new rs number and a link to the refSNP page for that new rs number.
3. The [RsMergeArch table](#) houses the merged SNPs, and is available on the dbSNP ftp site. A full description of the table can be found in the [dbSNP Data Dictionary](#), and the column definitions are located in the dbSNP_main_table.sql.gz, which can be found in the [shared_schema](#) directory of the dbSNP FTP site.

(11/07:05/08:11/08)

Why were SNPs rs12722468 and rs12722469 deleted?

These SNPs were removed in July 2004 by [IMGT/HLA](#), the curator for HLA. HLA allele E*0102 was withdrawn because it was shown to be identical (see the [IMGT/HLA database](#)) to E*0101 despite the allele differences presented before July 2004. (10/5/07)

I extracted rs7452972 from SNP genotype bulk data I downloaded from the HAPMAP Jan '07 data release, but I can't find rs7452972 in dbSNP.

I see that HapMap has [genotype data](#) for rs7453972 available online. dbSNP also has genotype data for this refSNP, but it looks like rs7452972 has [merged](#) into rs4357147. To see the genotype information you will have to search using the current refSNP (rs) number ([rs4357147](#)). (5/4/07)

Why has rs947894 been replaced by replaced by rs1695?

rs947894 was merged into rs1695. refSNP (rs) numbers merge when we find that two SNPs have exactly the same mapping properties. When two refSNP numbers merge, all previous member submitted SNP (ss) numbers cluster together under the refSNP with the smallest number.

Such clustering of submitted SNPs provides additional available data. For example, you can get extra information on the SNP that might not have been accessible if one submitted SNP in the cluster has frequency/genotype data and another submitted SNP has a pubmed link.

You can always find out if a SNP has been merged by going to the refSNP Cluster Report for the reSNP in question, in this case, [rs947894](#). An Entrez SNP query, however, will indicate “no items found” if the queried refSNP has merged into another refSNP, but will indicate that another SNP merged into the queried refSNP if such an event has occurred. (08/22/07)

Locating SNPs using IDs Published in the Literature

I've tried without success to find human SNPs from a publication in dbSNP using the IDs mentioned in the article as well as the author's name. Is there anything else I can try?

Yes, you can now submit previously published human variations described in a publication not authored by you to a new resource called [Human Variation: Annotate and Submit Batch Data](#). You must, however, be able to describe the variations using [HGVS nomenclature](#). You will see that the submission template for this resource has a field for the inclusion of the PubMed ID number, so that the original reference for the variation will be linked to the submission. You can also submit a single variation from the literature using the [Human Variation: Search, Annotate, Submit](#) site. **Please Note:** these online submission resources accept human data submissions only. (06/11/08)

I have a SNP that has no rs number, but does have a ID (human TNF alpha -308G>T), I've searched in dbSNP, but can't find an rs number for it.

Unless the submitter used the exact name "human TNF alpha -308G>T" in their submission, it won't show up as a result in your dbSNP search. It is also possible that the authors who mentioned "human TNF alpha -308G>T" in their publication never submitted this SNP to dbSNP.

Submission to dbSNP is voluntary, but we do try to encourage people to submit SNPs to dbSNP prior to publication so that they can cite their assigned dbSNP IDs (ss or rs) in their manuscript. Check the publication to see if the authors included a dbSNP ID for "human TNF alpha -308G>T" in the manuscript; if they have not, try BLASTing the DNA sequence of human TNF alpha -308G>T (if you have it) against dbSNP to see if there are any matching rs numbers. If there are no matches, and you wish to see this variation in dbSNP, you can submit it yourself using the [Human Variation: Annotate and Submit Batch Data](#) page (multiple submissions) of the [Human Variation: Search, Annotate, Submit](#) site (single submission). The [Human Variation: Annotate and Submit Batch Data Quick Start](#) explains how it is possible to submit published SNPs not already housed in dbSNP. (04/30/08)

CELL. 2004; 119: 591-602 reported a MDM2 SNP309 (A/G polymorphism located in intron 1 of MDM2 gene, but I cannot find it in dbSNP.

If this is the [reference](#) you mentioned, I do not see a dbSNP accession listed. dbSNP has 358 SNPs located in the [MDM2 gene region](#), but I do not see the specific A/G SNP in intron 1 you mentioned.

Although dbSNP currently has 17million+ human SNPs (including 7,529,694 SNPs located in genes), it is still possible that this particular SNP in the MDM2 gene was never submitted to dbSNP. dbSNP is populated by data submitted by users like yourself, and usually, they submit SNP data prior to the release of their publications. If you wish to see this variation in dbSNP, you can submit it yourself using the [Variation Batch Submission](#) page. The [Variation Batch Submission Quick Start](#) explains how it is possible to submit published SNPs not already housed in dbSNP(04/24/08)

I am trying to find an rs number for a SNP called CD14-159C/T. Can you tell me what the rs number is or how I can find it?

You can use the [Human Variation: Search, Annotate, Submit](#) site to find the rs number you need, but you'll need the accession number for the reference sequence that codes for CD14 to do so. (04/09/08)

How do I find a refSNP (rs) number for a SNP using a reference number found in the literature?

The dbSNP [GeneView](#) (SNP linked to Gene) page may be the best way to look for a refSNP (rs) number rs that corresponds to published gene position variations. You can reach the GeneView page by clicking on the pink "GeneView" button located under the refSNP (rs) number of interest in the results page of a Entrez SNP search. Using the GeneView page may not always work if the published gene model is different than the model used in dbSNP, but in your case there is strong evidence that the SNP in position 45 is rs2241766, which is shown on the GeneView page as amino acid 15, codon position 3 ($3 \times 15 = 45$). rs2441766 is located at contig position 93066042.

Looking for the other two variations is a little more difficult. First, click on the radio button labeled "in gene region", which is located in the long, grey, rectangular box found above the **gene model** (contig mRNA transcript) section of the GeneView page. Now click on the "refresh" button located at the far right end of the grey box where you found the "in gene region" button. You will now see all SNPs near ADIPOQ (the default is cSNPs). The SNP in position 713 is likely rs3774261. I determined this by subtracting 45 from 713 (the difference in the position of the two variations) and then adding this number to the contig position of the SNP at position 45. $(713-45) + 93066042 = 93066709$. rs3774261 is located at contig position 93066709. (6/1/06)

Locating SNPs using IDs Published in OMIM Articles

I'm frustrated trying to find the refSNP (rs) number for a SNP listed in an OMIM article (as well as in other publications) only as "c1565t"

The staff at Johns Hopkins manually curates all the OMIM records — it is a slow and tedious process, but they continue to update OMIM records with links from the allelic variants mentioned in OMIM articles to existing dbSNP Reference SNP (rs) numbers. This process, however, is made difficult since variations are reported on different sequences and in many different formats. To improve this situation, NCBI and other institutions are beginning to adopt, and are encouraging others to adopt, a single standard report format for mutations — the [Human Genome Variation Society \(HGVS\) nomenclature recommendations](#), as well as a stable reference sequence standard called [RefSeqGene](#).

dbSNP has begun providing support for submitting variants as well as for variant searches using HGVS nomenclature, and is beginning to add clinical variants to dbSNP:

- Use NCBI's "[Human Variation: Annotate and Submit Batch Data](#)" to submit variations using HGVS nomenclature
- Use NCBI's "[Human Variation: Search, Annotate, Submit](#)" site to search for variations using HGVS nomenclature and to submit single annotations or submissions.
- The [clinical variants found in dbSNP](#) using Entrez SNP (there are only a handful now — click on the "VarView" link)

(06/19/08)