



Submitting Information for Existing SNPs

Created: June 6, 2005; Updated: June 15, 2010.

Submitting Variations that already exist in dbSNP

Is it possible to deposit nonsynonymous coding region variant information from a re-sequencing study of a gene using 560 samples from the Coriell repository?

It doesn't matter if a variation is novel or recorded, you can submit the variation again as your own data. If the variation already exists, we encourage you to submit the variation and its discovery data anyway, as it will serve as a kind of validation for the existing SNP.

The easiest way to submit is to use the Excel submission template, which is located in the [submission file](#) on the dbSNP FTP site or follow the submission examples, which are located in the same [file](#).

In your case, you should submit using the SNPASSAY form (to record the variation), and the SNPINDUSE form or the SNPPOPUSE form (to record the genotypes of the variation).

If you are going to submit using the SNPINDUSE form, you will need to submit population information and the information regarding individuals in that population. If the individuals and the population are already in dbSNP, you can just use their IDs.

If you think submitting the individual information is too overwhelming, you can submit the genotypes using the SNPPOPUSE form. In contrast to the SNPINDUSE form, the SNPPOPUSE form allows you submit a summary of your genotypes over the entire population instead submitting individual genotypes. (07/24/08)

Submitting SNPs to dbSNP that were Published by Others

Human minor histocompatibility antigen HA-2 does not appear in dbSNP under the MYO1G gene, despite its SNP having been published 7 years ago. Could you please update dbSNP to include this?

In general, dbSNP does not mine publications for input, and the variations in dbSNP are submitted by submitters directly to dbSNP. If you would like the SNP mentioned in this publication to be included in dbSNP, go to the [Human Variation: Annotate and Submit Batch Data](#) site. Here you can submit variations described in a publication not authored by you, but you must be able to describe the variations using [HGVS nomenclature](#). You will see that the submission template 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 (03/05/08)

Is it possible for me to submit human SNPs that are not available in dbSNP, but are described in research articles I've read?

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 do not take submissions from other species (06/11/08)

I have detected a SNP that has been previously reported, but I can't find it in dbSNP. I want to publish the SNP and therefore need a refSNP number for it. Do I mention the previous report of this SNP when I submit the data for it?

You can submit the SNP so long as you detected it independently and reference both your publication and the previous publication in the submission to dbSNP. To reference both publications in a single submission, make two publication (PUB) sections and put two citation fields in the SNPASSAY section that contain the publication titles. (2/2/05)

A number of SNPs we typed and validated were published by others, but do not appear in dbSNP. Can we submit information on these variants even though we did not originally identify them and have no electronic record of who did?

Yes, you can submit the data. (9/12/05)

If I submit SNPs that already have refSNP (rs) numbers, do I need to submit the 3' and 5' flanking sequences as well?

You do not need to submit the SNPs if they already exist in dbSNP with assigned ss or rs numbers. (6/5/06)

Annotating existing SNPs

Will dbSNP allow me to annotate an existing human SNP record that I did not originally submit?

Yes, you can now annotate existing human SNP records using two new resources. One is called the [Human Variation: Search, Annotate, Submit](#) site, where you can submit a single variation or annotation to a single variation, and the other is the [Human Variation: Annotate and Submit Batch Data](#) site, where you can submit a number of annotations or variations simultaneously.

Both of these resources allow you to submit phenotypic data for an existing SNP in the “Comments” section of each respective submission form. You can also submit a “Linkout” URL that will point to this variant on your organization’s website, as well as OMIM IDs, Allelic Variant IDs, and other associated PubMed IDs (in addition to the primary PubMed ID for the variant’s original publication). **Please Note:** these online submission resources do not take annotations or submissions for SNPs of other species. (07/10/08)

I have some phenotypic data I'd like to annotate to an existing human SNP. If I can do this, where do I submit this data?

Yes, you can now annotate existing SNP records using two new resources. One is called the [Human Variation: Search, annotate, Submit](#) site, where you can submit a single variation or annotation to a single variation, and the other is the [Human Variation: Annotate and Submit Batch Data](#) site, where you can submit a number of annotations or variations simultaneously.

Both of these resources allow you to submit phenotypic data for an existing SNP in the “Comments” section of each respective submission form. You can also submit a “Linkout” URL that will point to this variant on your organization’s website, as well as OMIM IDs, Allelic Variant IDs, and other associated PubMed IDs (in addition to the primary PubMed ID for the variant’s original publication). **Please Note:** these online submission resources do not take annotations or submissions for SNPs of other species.(07/10/08)

Submitting Frequency Data for SNPs already in dbSNP

We want to submit allele frequency information for SNPs already housed in dbSNP. How do we submit these data?

You can submit allele information for existing SNPs by submitting a SNPPOPUSE file, which contains HANDLE, BATCH, and METHOD headers, as well as a series of ALLELEFREQ entries. Each ALLELEFREQ entry requires a refSNP ID, alleles with frequencies, and direction of the probe relative to the refSNP sequence.

Can a user submit frequency data for SNPs that are currently in dbSNP?

Yes, a user can submit frequency data for SNPs already in dbSNP. Please [follow the instructions](#) on the web for formatting.

Submitting Genotype Data for SNPs already in dbSNP

I want to submit the genotypic data for the refSNPs used in a population. What section of the excel submission template do I need to fill out?

Fill out the SNPINDUSE section. Remember to use the refSNP (rs) or submitted SNP (ss) numbers when reporting genotype in this section (6/5/06)