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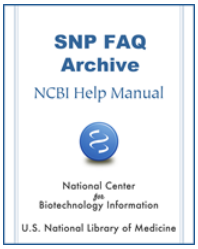

Classes of Genetic Variation Included in dbSNP

Created: July 23, 2005; Updated: February 18, 2014.

Can I submit Structural (copy number) variations to dbSNP?

NCBI is in the process of creating a database to house CNV/structural Variation (STV) — we hope to see submissions for this database accepted in the next few months, and the database itself is expected to launch in about a year.(**07/11/08**)

If dbSNP is a database containing Single Nucleotide Polymorphisms (SNP), it doesn't accept mutations, right?

Think of dbSNP as a "dbVariation" since it contains not only Single Nucleotide Polymorphism (SNP), but also indels, STRs, MNPs, etc. Biologists have also been using the term "Polymorphism" to refer to common variations and the term "mutation" to refer to rare allele variation. dbSNP includes both polymorphisms and mutations. Please note, however, that the use of the word "mutation" is being phased out in dbSNP, and will be replaced by the term "Clinical/LSDB variation".

Starting in the Spring of 2008, dbSNP began accepting submissions of human Clinical/LSDB variations as well as annotations to existing human variations (including phenotype) on the Human Variation: Search, Annotate, Submit site (for single submissions) as well as on the Human Variation: Annotate and Submit Batch Data site (for multiple submissions). As of this date, there are a total of 1266 records in dbSNP that were submitted as Clinical/LSDB variations (select "Clinical/LSDB variation" in the Entrez SNP limits page and click "GO" with out entering a search term in the Entrez search box), and 1134 records submitted as Clinical/LSDB variations that also have OMIM links (select "Clinical/LSDB variation" and "OMIM" in the Entrez SNP limits page and click "GO" with out entering a search term in the Entrez search box).

Those SNPs with clinical association(s) will have a red "VarView" (Variation Viewer) link in the "allele" section at the upper right of the refSNP cluster report. Clicking the link will take you to the Variation Viewer Report for the gene in which the SNP is found.

We expect that the number of Clinical/LSDB variation records in dbSNP will grow rapidly as more users discover dbSNP's resources for submitting them. (**07/09/08**)

rs28937569 and rs28937568 are listed as disease-related mutations in OMIM. Can a mutation be listed as a variation in dbSNP?

Please Note: the use of the word "mutation" is being phased out in dbSNP, and will be replaced by the term "Clinical/LSDB variation".

rs28937569 and rs28937568 were submitted by OMIMSNP using an automated program that extracted OMIM variations occurring in the coding regions of OMIM records. Only those variations with reference sequences available and where the reference allele was confirmed were added to dbSNP.

Originally, the great majority of data in dbSNP was collected and defined as variations simply using sets of co-aligned genomic or DNA sequences. Because this process typically had little to no focus on disease

condition, only about 250 records in dbSNP were successfully associated with phenotype-causing variations or a clinical outcome in OMIM.

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Does dbSNP include all known single nucleotide variations without taking function (e.g. disease-associated mutations) into account?

dbSNP accepts all kinds of variations regardless their functional associations. (**5/29/07**)

R978C has been described by two journal articles as a mutation, but you have it listed as a polymorphism with ID rs28942108. Is R978C a mutation or a polymorphisim?

dbSNP accepts nucleotide variations submissions, including both mutations and polymorphisms. These submissions might include variation frequencies, and sample populations. Not only does dbSNP accept single nucleotide variations(trueSNP), it also accepts micro-satellites(STR), indels (insertion deletion variations) as well as multiple bases substitutions(MNP), and is planning to accept large structural variations as well. Please note that dbSNP even has accepted a small subset of somatic mutation submissions. The dbSNP Handbook includes a table describing all the variations accepted by, and included in, dbSNP.

The rs28942108 refSNP cluster contains a single submitted SNP (ss) submitted by OMIMSNP. As of this date, no frequency information has been submitted for this SNP. An OMIM link for this refSNP(rs28942108) is located at the far right side of the "NCBI Resource Links" section of the rs28942108 cluster report (**4/26/07**)

Why is rs8111802 classified as a SNP rather than "mixed" when its exemplar submission is a DIP record?

The SNP development group thinks that it is best if we do not cluster or merge SNPs of different variation classes even when they map to the exact same contig location. This affects about 50K of the current refSNP (rs) numbers, including rs8111802. We will split these current SNP clusters by SNP class and work out all related details soon.

(**10/6/06**)

What classes of genetic variation are included in dbSNP?

dbSNP accepts several classes of genetic variation:

- Single Nucleotide Polymorphism (SNP)
- Deletion/Insertion Polymorphism (DIP)
- Microsatellite or Short Tandem Repeat (STR)
- Multi-Nucleotide Polymorphism (MNP)

dbSNP also accepts reports of “No-variation”; that is, dbSNP accepts reports of segments of assayed sequence that show no variation in the sample.

As dbSNP uses the term "SNP" in the much looser sense of "minor genetic variation", there is no requirement or assumption about minimum allele frequencies for the polymorphisms accepted by dbSNP. Therefore, dbSNP includes both disease causing clinical mutations as well as neutral polymorphisms.(**04/05/06**)

How do I find out the kind of polymorphisims contained in dbSNP? I want to see what kind I have in randomly cloned human sequences I have.

dbSNP has several variation classes, which are defined in Table 3 of the dbSNP handbook.

You can search for examples of these variations within dbSNP by doing the following:

1. Go to Entrez SNP
2. Click on the grey “Limit” tab located near the top of the page just below the text box used for searches.
3. Once on the “Limits” page, select the appropriate organism, SNP (variation) class, and any other “limits” that are appropriate, and press the ”Go” button at the top of the page. (**08/13/07**)

Does dbSNP have any human mitochondrial DNA data?

As of February 2005, dbSNP has not received any human mitochondrial DNA submissions. You may want to resend your request to info@ncbi.nlm.nih.gov in case other resources like GenBank have received such submissions. **(2/14/05)**