



## NIDA - Individual Genotype Dataset Description

### Overview

The data set consists of three files. The data file is plain text with tab separated columns. The first row of the data file consists of column headers.

### SNP Information

The file SNP\_INFO.TXT has the following information for a total of 44,454 SNPs.

<i>Column Name</i>	<i>Description</i>
<b>SNP_ID</b>	Perlegen internal SNP identifier.
<b>Refsnp_rsID</b>	The RefSnp rsID (from NCBI) when available. Can be null.
<b>RefSnp_ssID</b>	The RefSnp ssID for SNPs Perlegen submitted to dbSNP. Can be null.
<b>Allele 1</b>	The nucleotide code for Perlegen's reference allele.
<b>Allele 2</b>	The nucleotide code for Perlegen's alternate allele.
<b>Chromosome</b>	Chromosome number of the NCBI Build 35 contig on which the best alignment was found. X is used for the X chromosome, Y for the Y chromosome and U for sequences not assigned to any chromosome in Build 35. May be null if SNP could not be placed on any contig.
<b>Sex-linked</b>	A, autosomal; P, pseudoautosomal (on X or Y, in the pseudoautosomal region); S, sex linked (on X or Y, not in the pseudoautosomal region); U, unassigned (or unknown pseudoautosomal status for X and Y).
<b>Accession ID</b>	The accession number from NCBI Build 35 of the contig to which the SNP aligns; may be null.
<b>Contig Position</b>	Nucleotide position on the NCBI build 35 contig of the 'N' in the assayed sequence; may be null. For SNPs this is always a single position, but in the case of a deletion-insertion polymorphism (DIP), the mapping may be a range. See the table below describing the full range of position notations.
<b>Strand</b>	+ or -, based on the strand for Allele 1 on NCBI Build 35; may be null.
<b>Assayed sequence</b>	The 29mer we assayed for this SNP, with an 'N' representing the SNP or deletion-insertion polymorphism (DIP) at the middle base.



<b>Genes</b>	A comma-separated list of NCBI gene IDs for which the SNP is contained within a footprint extending +/- 10 kb of the largest transcribed sequence.
<b>Selected candidate genes</b>	A comma-separated list of NCBI gene IDs that provided the basis for selecting the SNP. The physical footprint of the gene was extended by use of linkage equilibrium data.
<b>Flags</b>	A comma delimited list indicating selection categories and other information, as described in the table below.

Position notations:

<i>Contig Position</i>	<i>Definition</i>
<b>12345</b>	The N in the assayed sequence aligns to the single base 12345 on the genomic contig.
<b>12345^12346</b>	The N in the assayed sequence maps to the genomic contig between bases 12345 and 12346 (i.e. the N is an insertion with respect to the reference assembly)
<b>12345..12346</b>	The N in the assayed sequence maps to the range of genomic bases beginning with 12345 and ending with 12346.

Flags:

<i>Flags</i>	<i>Definition</i>
<b>CAND_GENE</b>	The SNP was selected because of its association with a candidate gene.
<b>DIP</b>	The variation is a deletion-insertion polymorphism (DIP) rather than a SNP.
<b>CDS</b>	The SNP falls within +/- 10bp of the CDS portion of an exon in a candidate gene.
<b>LD</b>	The SNP was selected because it was found to be in linkage disequilibrium with SNPs in the footprint of a candidate gene for European Americans.
<b>LD_EXONIC</b>	The SNP was selected because it was found to be in linkage disequilibrium with SNPs in an exon of a candidate gene for European Americans.
<b>NIDA_CHOSEN</b>	The SNP was specifically indicated by NIDA for assay.
<b>POOLED</b>	The SNP was assayed in the previous pooled study.
<b>PRLGN_CHOSEN</b>	The SNP was selected by Perlegen.
<b>PROMOTER</b>	The SNP was selected because it was found in the promoter region defined for a candidate gene.
<b>STRAT</b>	The SNP is a Perlegen population stratification analysis SNP.
<b>UTR</b>	The SNP is within +/- 10bp of the UTR portion of an exon in a candidate gene.



<b>WK</b>	The SNP is known to genotype well on the Perlegen platform and is polymorphic in some tested population.
<b>WK_FREQ</b>	The SNP is known to genotype well on the Perlegen platform and is polymorphic in some tested population with MAF >4% in European Americans.