NHLBI Exome Variant Server

- Search for SLC6A3 on the NHLBI and click on “display snp summary”. Click the “MAF (%)” option (Labeled [1] on the next slide). Sort by “GVS Function” [2]. Change the number of entries shown to “all” [3].

- 1) **There are no coding non-synonymous SNPs with a frequency greater than 5% in either population.**

- 2) **The most common coding SNP is rs6347 with a minor allele frequency of 47.9%**

- Bonus question: You can still do an association analysis but you would probably want to focus on covering the whole gene instead of picking specific SNPs

Gene Name: OPRM1 (Gene ID: 4988) (+)

Chromosome 6: 154407642 - 154440594

Population: AfricanAmerican, EuropeanAmerican

GWAS Catalog: OPRM1
KEGG Pathway: OPRM1
Sanger COSMIC: OPRM1
PPI STRING 9.0: OPRM1
OMIM: OPRM1

Variation Color Code:

- splice or nonsense or frameshift
- missense
- coding-synonymous
- coding
- utr
- codingComplex

Download Option:

File Format: Text
Zip Format: gzip
download

Add or Remove Columns (Description of Columns)

[1]

☑ dbSNP rs ID	☑ Alleles	☑ EA Allele Count	☑ AA Allele Count	☑ Allele Count	☑ EA Genotype Count	☑ AA Genotype Count
☑ Genotype Count	☑ MAF (%)	☑ Sample Read Depth	☑ Genes	☑ Gene Accession #	☑ GVS Function	☑ cDNA Change
☑ cDNA Size	☑ Protein Change	☑ Conservation (GERP)	☐ Conservation (phastCons)	☑ Grantham Score	☑ PolyPhen Prediction	☐ Clinical Link
☐ NCBI 37 Allele	☐ Chimp Allele	☐ Filter Status	☐ Illumina HumanExome Chip	☐ GWAS Hits	☐ EA Est. Age (kyrs)	☐ AA Est. Age (kyrs)

[2] **Sort Variants by** Variant Pos **Select Population** All **Select Transcript** Union of Transcripts

If "Select Transcript" above is set to "Union of Transcripts", and if multiple transcripts of a gene are involved in a variant and the function annotations for the variant are the same, only one representative tra file if one chooses to download the data.

[3] Show 10 entries

Variant Pos	rs ID	Alleles	EA Allele #	AA Allele #	All Allele #	EA Genotype #	AA Genotype #	All Genotype #	MAF (%) (EA/AA/All)
6:154410919	rs201598368	G>C	C=0/G=8334	C=1/G=3829	C=1/G=12163	CC=0/CG=0/GG=4167	CC=0/CG=1/GG=1914	CC=0/CG=1/GG=6081	0.0/0.0261/0.0082
6:154410932	rs374691791	A>C	C=1/A=8369	C=0/A=3886	C=1/A=12255	CC=0/CA=1/AA=4184	CC=0/CA=0/AA=1943	CC=0/CA=1/AA=6127	0.0119/0.0/0.0082
6:154410940	rs368117860	T>A	A=0/T=8380	A=1/T=3913	A=1/T=12293	AA=0/AT=0/TT=4190	AA=0/AT=1/TT=1956	AA=0/AT=1/TT=6146	0.0/0.0255/0.0081
6:154410958	rs371723278	T>C	C=1/T=8439	C=0/T=3978	C=1/T=12417	CC=0/CT=1/TT=4219	CC=0/CT=0/TT=1989	CC=0/CT=1/TT=6208	0.0118/0.0/0.0081
6:154410963	rs373741336	A>G	G=1/A=8465	G=0/A=4028	G=1/A=12493	GG=0/GA=1/AA=4232	GG=0/GA=0/AA=2014	GG=0/GA=1/AA=6246	0.0118/0.0/0.008
6:154410963	rs373741336	A>G	G=1/A=8465	G=0/A=4028	G=1/A=12493	GG=0/GA=1/AA=4232	GG=0/GA=0/AA=2014	GG=0/GA=1/AA=6246	0.0118/0.0/0.008
6:154411018	rs368512118	T>C	C=1/T=8489	C=0/T=4102	C=1/T=12591	CC=0/CT=1/TT=4244	CC=0/CT=0/TT=2051	CC=0/CT=1/TT=6295	0.0118/0.0/0.0079
6:154411054	rs145700569	G>T	T=1/G=8471	T=0/G=4114	T=1/G=12585	TT=0/TG=1/GG=4235	TT=0/TG=0/GG=2057	TT=0/TG=1/GG=6292	0.0118/0.0/0.0079