



CENTER FOR COMPUTATIONAL BIOLOGY AND BIOINFORMATICS
INDIANA UNIVERSITY SCHOOL OF MEDICINE | DEPARTMENT OF BIOCHEMISTRY AND MOLECULAR BIOLOGY
CUTTING EDGE RESEARCH THROUGH COLLABORATION



INDIANA UNIVERSITY

MutDB: Enhanced Biochemical Analysis of Structural and Functional Features of Human Genetic Variation

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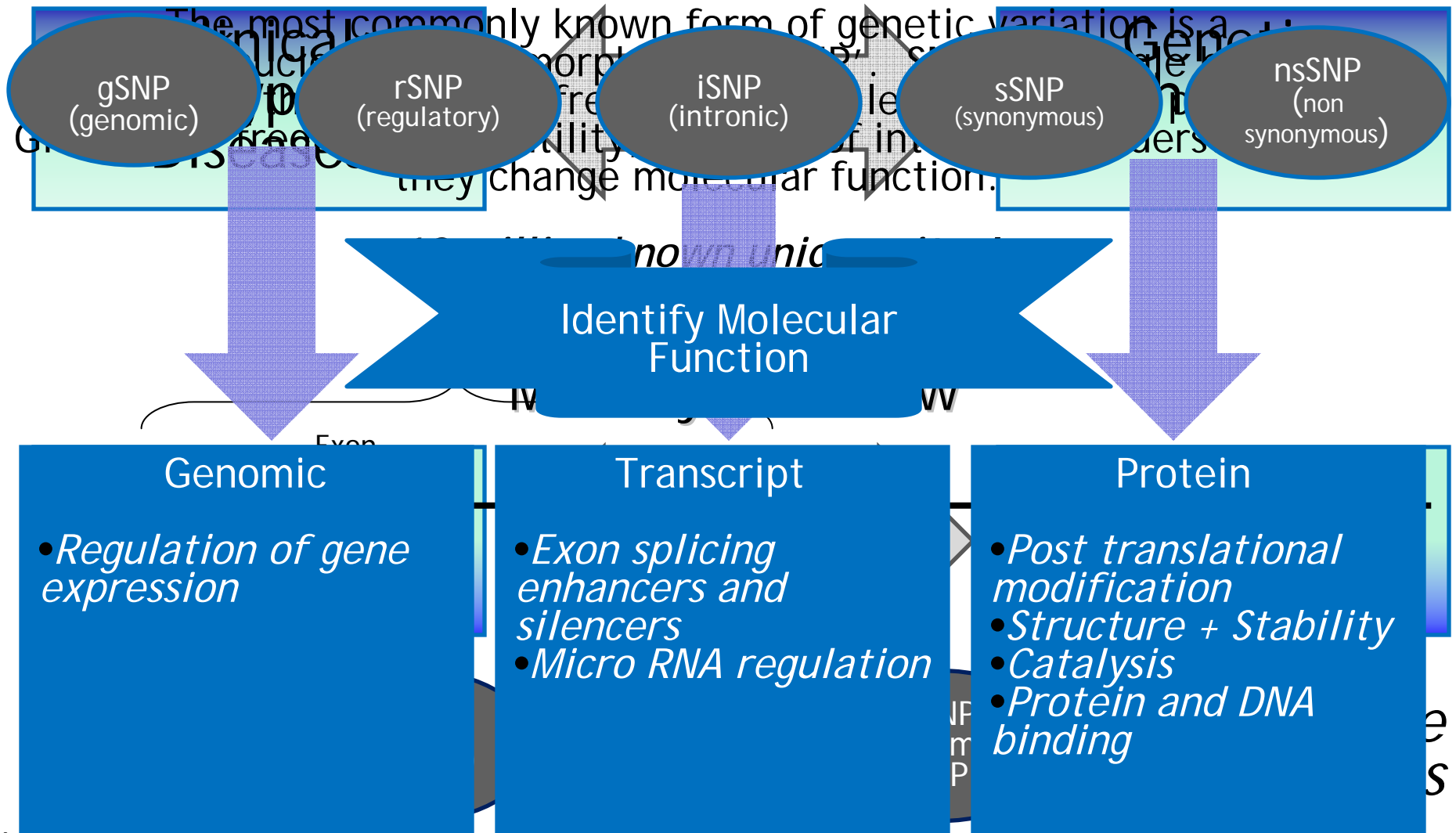
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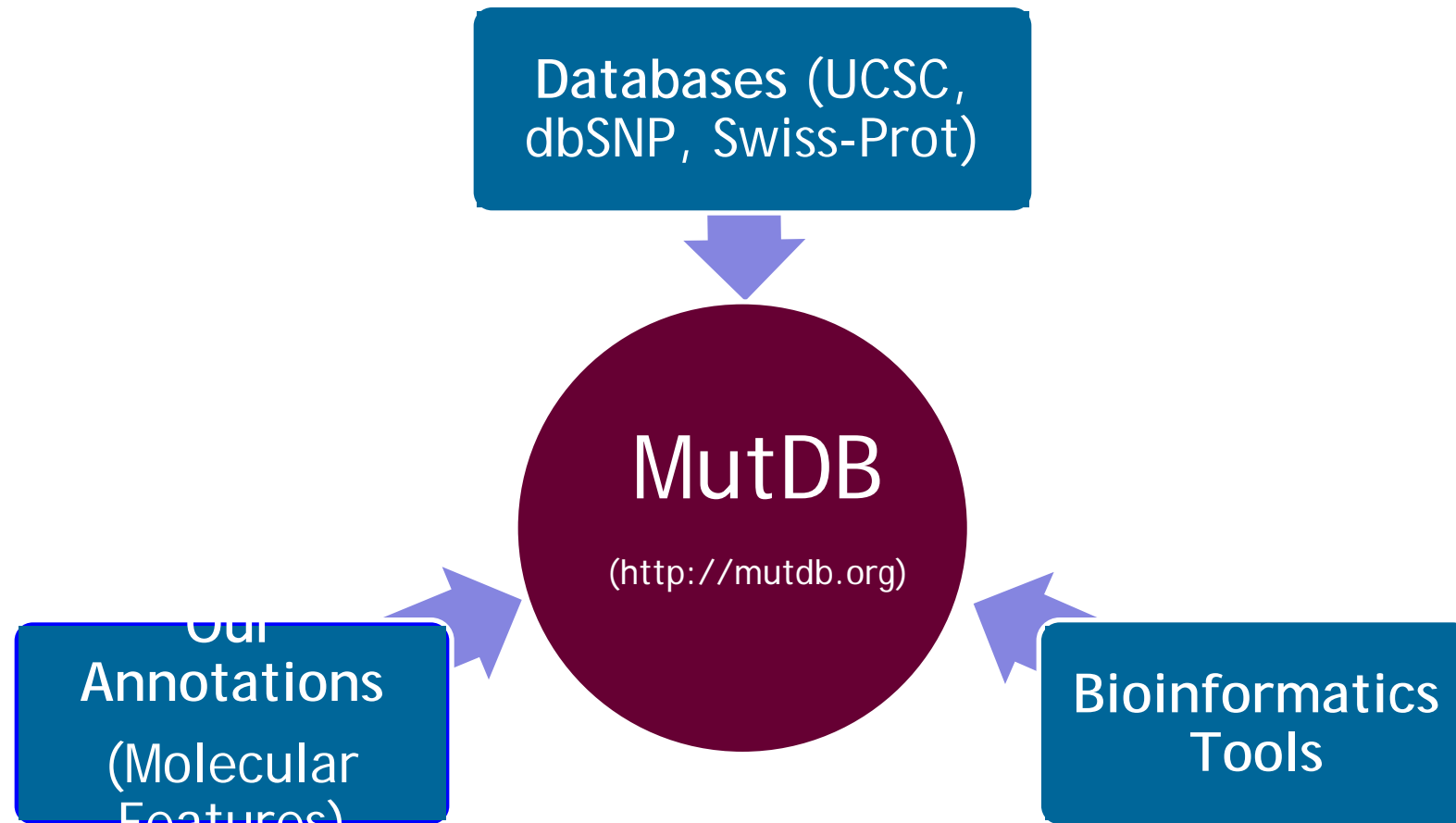
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SNPs Are Important Approaches in Genetic Studies

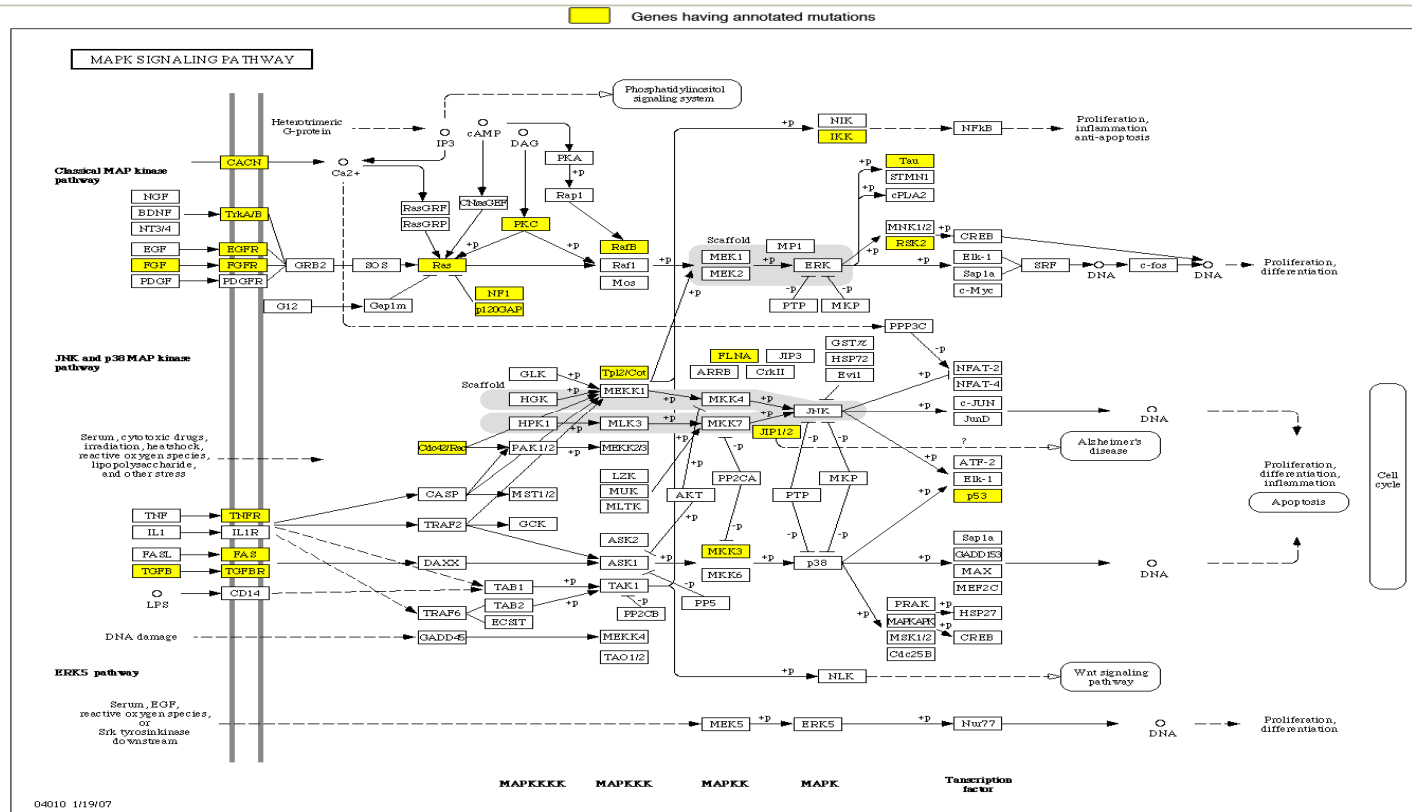


What is MutDB ?



- MutDB portal for mutation annotation of SNPs/Mutations helps identify the primary molecular cause of disease

SNPs / Mutations - Gene Level



D
C
G
E
N
V

Source ID	AA Position (help?)	WT -> MT (help?)	Sequence	Phenotype	Structure	PubMed	SIFT Score (help?)
SWISS-PROT_0379	57	K -> R	...K... ...K... ...K...	in prostate cancer	None	8823308	0.00*
SWISS-PROT_04681	57	L -> Q	...L... ...L... ...L...	in prostate cancer	None		0.00*
SWISS-PROT_00971	64	Q -> R	...Q... ...Q... ...Q...	in prostate cancer	None		0.02*
SWISS-PROT_00772	11	S -> T	...S... ...S... ...S...	in prostate cancer	None		0.00*
SWISS-PROT_00772	10	S -> T	...S... ...S... ...S...	in prostate cancer	None		0.00*

- MutDB enables visualizing 188 KEGG human pathways
- Genes / Proteins with mutations are highlighted dynamically using SOAP based web services



SNPs / Mutations - Protein Level

- Protein Data Bank (PDB) - greater than 4

- Annotate

- Search

search the tool

- 10 are of

- Mutation data

MutDB Position: SWISS:VAR_009773 [SNP Browser](#) [Home](#) [About](#)


MOONEY LAB [Home](#) | [Research](#) | [Publications](#) | [People](#) | [Employment](#) | [Entrepreneurship](#) | [Fun](#) | [Contact](#)

SNP Selection Tool (UCSC Build 17, DBSNP Build 122) [MutDB](#) [SNP Home](#) [Logout](#) [Help](#)

TOTAL SNPS	CHROMOSOME	NUMBER OF CURRENTLY SELECTED SNP
4583	chr1	0

Page (1 of 46) Records [0 - 100]

Get Sequences				Excel	Haplotype Struct:	CEU	CHB	JPI	YRI					
<input type="button" value="Update Image"/>	<input type="button" value="Clear"/>	<input type="button" value="Show All"/>	<input type="button" value="Show Selected"/>	<input type="button" value="Show Unselected"/>	<input type="button" value="Select All"/>	Session Name:			<input type="button" value="Save"/>					
SNP Info				UCSC		dbSNP		HapMap Freq		Scores				
rsid	observed	validation	hapmap	Location	strand	position	avHet	avHetSE	CEU	CHB	JPT	YRI	SIFT score	conser
<input type="checkbox"/> rs241231	A / G	X	X	unknown	-	4491481	0.192132	0.24321	0.192	0.034	0.067	0.108
<input type="checkbox"/> rs12567893	C / G	X	X	unknown	+	4490944	0.244898	0.249948		0.119	0.125	0.22
<input type="checkbox"/> rs12755100	G / A	X	X	unknown	+	4490939	0.0198	0.0975088		0.011	0	0.017
<input type="checkbox"/> rs241232	C / T	X	X	unknown	-	4490828	0.428917	0.17461		0.208	0.388	0.326
<input type="checkbox"/> rs34995066				unknown	+	4490633	0	0				
<input type="checkbox"/> rs241233	A / G	X	X	unknown	-	4490409	0.481403	0.0946183	0.292	0.311	0.5	0.2
<input type="checkbox"/> rs12410112	T / A	X	X	unknown	+	4490320	0	0	0	0	0	0
<input type="checkbox"/> rs241234				unknown	-	4489270	0	0				
<input type="checkbox"/> rs34879999				unknown	+	4489269	0	0				
<input type="checkbox"/> rs34846714				unknown	+	4489249	0	0				
<input type="checkbox"/> rs34865748				unknown	+	4489224	0	0				
<input type="checkbox"/> rs241235		X		unknown	-	4489121	0	0				
<input type="checkbox"/> rs241236		X		unknown	-	4489023	0	0				
<input type="checkbox"/> rs4654444				unknown	+	4488251	0	0				
<input type="checkbox"/> rs11807073		X		unknown	+	4487985	0	0				
<input type="checkbox"/> rs241237	G / A	X	X	unknown	-	4487828	0.473211	0.117426	0.467	0.222	0.322	0.475
<input type="checkbox"/> rs17348762		X		unknown	+	4487739	0.138353	0.223685				
<input type="checkbox"/> rs35174098				unknown	+	4487637	0	0				
<input type="checkbox"/> rs10915551	C / T	X	X	unknown	+	4487454	0.072888	0.176441	0.018	0	0.045	0.085
<input type="checkbox"/> rs241238	A / G	X	X	unknown	+	4487378	0.347567	0.230175	0.433	0.044	0.045	0.183
<input type="checkbox"/> rs241239	A / G	X	X	unknown	+	4487036	0.178465	0.239547	0.258	0	0	0.009

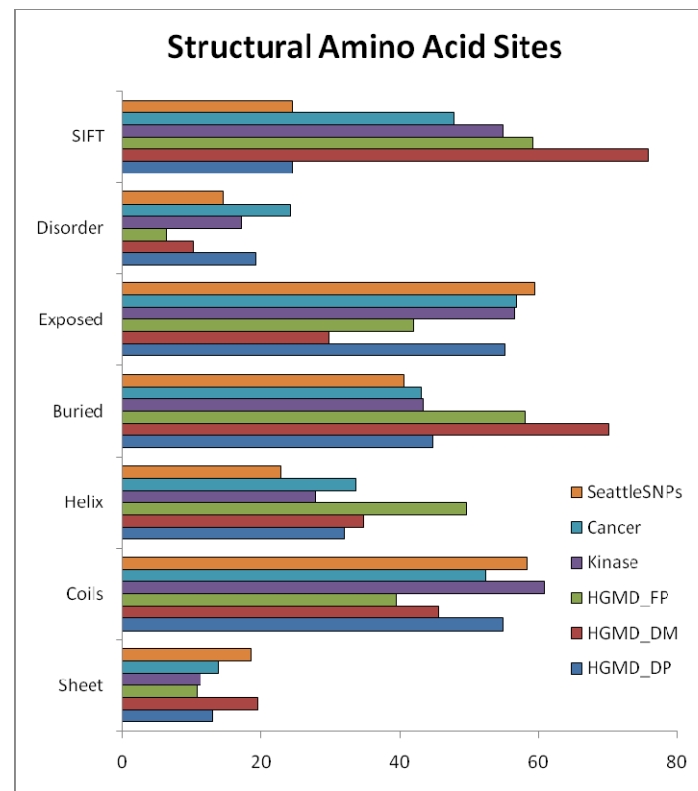
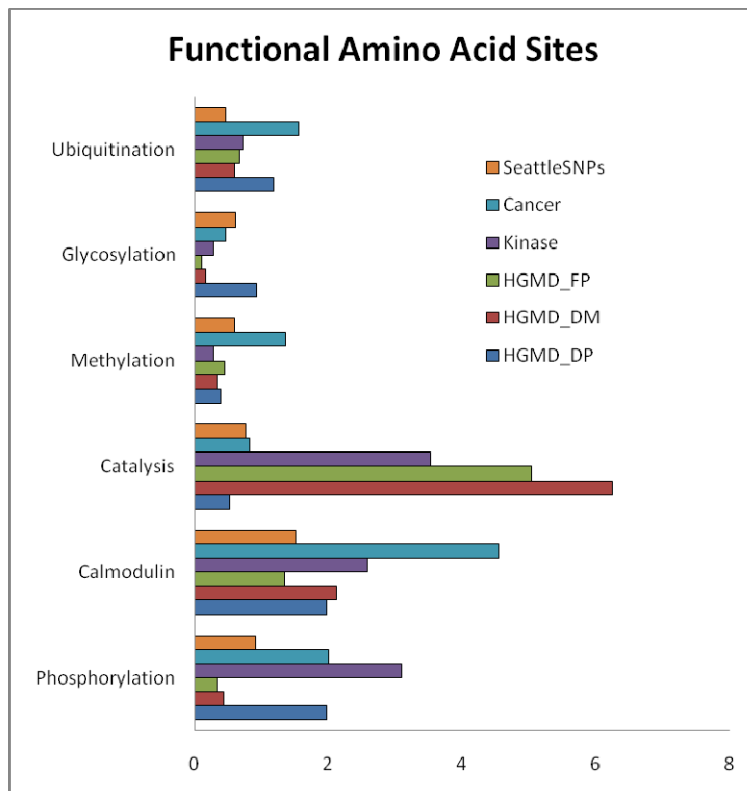


Running
 Structure: SWISS:VAR_009773
 AA:Protein:701
 PDBId: 2Z482:5A3000:1026511
 Phenotype: (in CAS and prosta can
 rev)
 MutDB:rsInfo



Future

- We have recently found evidence that bioinformatic methods can predict the molecular mechanism of disease mutations (see poster). We are currently developing a web portal for predicting disease mechanism.



DM-> Disease Mutations

DP-> Disease Polymorphism

FP-> Functional Polymorphism