



# Brazilian Initiative on Precision Medicine

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# Database infra-structure


- Based on a solid platform (LOVD v3.x), which catalogs:
  - Variants;
  - Transcripts;
  - Genes;
  - Subjects;
  - Diseases;
- Privacy-aware: users can't access identifiable information;
- Different levels of access to information (open, controlled);
- Isolated computational infra-structure;
- Infra-structure mirrored through partners (USP);

# Visite: bipmed.iqm.unicamp.br

Genes Transcripts Variants Individuals Diseases Screenings Submit Documentation

## View all genes

22165 entries on 222 pages. Showing entries 1 - 100.

100 per page  << First < Prev **1** 2 3 4 5 6 7 8 9 10 11 ... Next > Last >>

Symbol	Gene	Chr	Band	Transcripts	Variants	Unique variants	Last updated
A1BG	alpha-1-B glycoprotein	19	q13.43	1	0	0	2015-11-11
A1BG-AS1	A1BG antisense RNA 1	19	q13.43	0	0	0	2015-11-11
A1CF	APOBEC1 complementation factor	10	q21.1	1	0	0	2015-11-11
A2M	alpha-2-macroglobulin	12	p13.31	1	0	0	2015-11-11
A2M-AS1	A2M antisense RNA 1 (head to head)	12	p13.31	0	0	0	2015-11-11
A2ML1	alpha-2-macroglobulin-like 1	12	p13	0	0	0	2015-11-11
A3GALT2	alpha 1,3-galactosyltransferase 2	1	p35.1	1	0	0	2015-11-10
A4GALT	alpha 1,4-galactosyltransferase	22	q13.2	1	0	0	2015-11-11
A4GNT	alpha-1,4-N-acetylglucosaminyltransferase	3	p14.3	1	11	6	2015-11-11
AAAS	achalasia, adrenocortical insufficiency, alacrimia	12	q13	1	0	0	2015-11-11
AACS	acetoacetyl-CoA synthetase	12	q24.31	1	0	0	2015-11-11
AACSP1	acetoacetyl-CoA synthetase pseudogene 1	5	q35	0	0	0	2015-11-11
AADAC	arylacetamide deacetylase	3	q25.1	1	28	18	2015-11-11
AADACL2	arylacetamide deacetylase-like 2	3	q25.1	1	1	1	2015-11-11
AADACL2-AS1	AADACL2 antisense RNA 1	3	q25.1	1	76	60	2015-11-11

Dynamic searches

# The database

- Current format for assessment of user needs;
- May require changes on the current version;
- Allows for simplified searches to assist on going research;
- Give it a try and let us know what your thoughts are:

<http://bipmed.iqm.unicamp.br>

# Current status

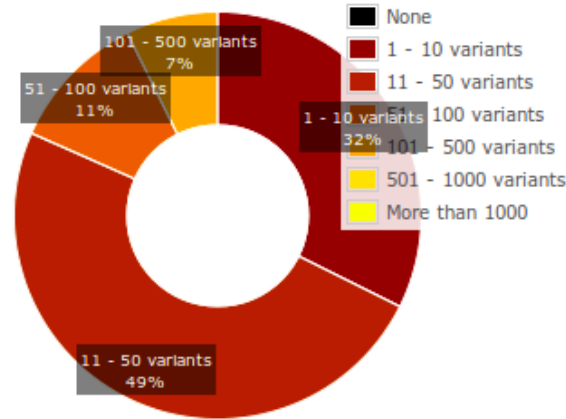
- 29 WES samples;
- Array data waiting to be included;
- Individuals from the reference population;
- We accept patient data;
- We can control data access;

## Current system status

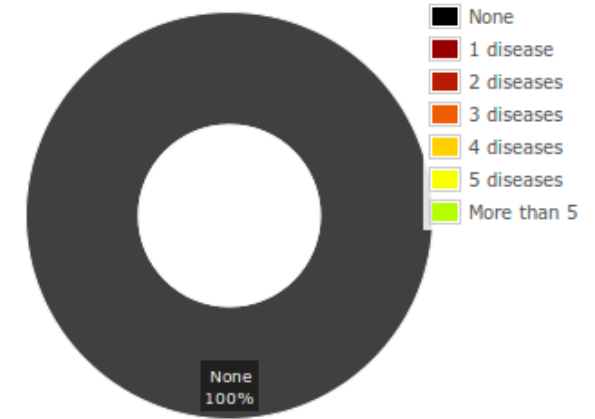
Current time: Thu, 12 Nov 2015 19:34:29 -0200

### Genes (22165)

Public variants per gene

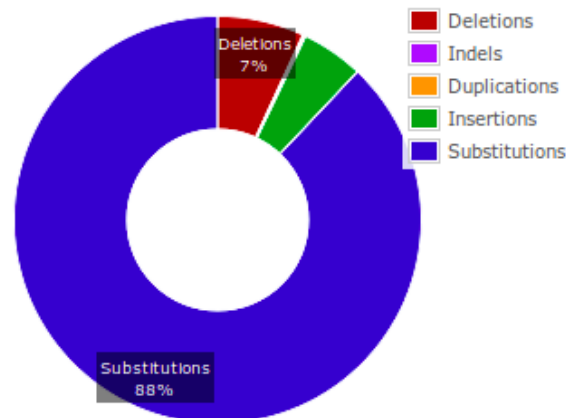


Linked diseases per gene

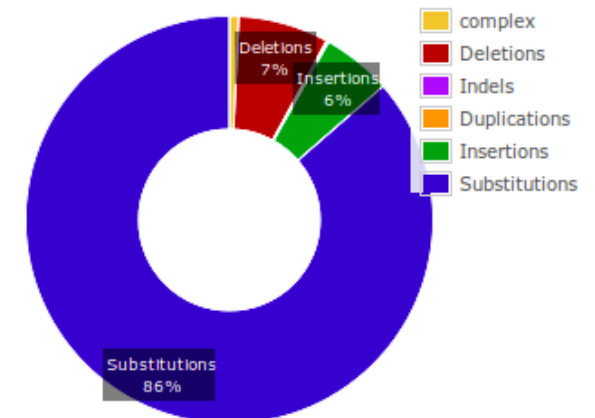


### Variant type (DNA level)

All public variants (10150641)



Unique public variants (27758)



# The Beacon

- Genetic mutation sharing platform;
- Proposed by the Global Alliance for Genomics and Health;
- Answers questions of the type:
  - ‘do you have information about this mutation?’
- Responds:
  - ‘yes or no’
- Meant to be accessed from computer programs.

# Beacon Interface

- Access it through:  
<http://bipmed.iqm.unicamp.br/beacon>
- Needs: Chromosome, Position, Allele;
- Returns: yes/no;
- Computer programs can ask questions too;



**BIPMed** Brazilian Initiative on **PRECISION MEDICINE**

## BIPMed Beacon

This beacon provides data on individuals from the reference population.

**Chromosome:**

**Position:**

**Allele:**

# Programmatic Access to the BIPMed Beacon

<http://bipmed.iqm.unicamp.br/cgi-bin/beacon/query?chromosome=1&position=14463&alternateBases=T&format=text>

- Beacon will respond: true, false;
- Computer programs can ask several questions like this;
- Resulting information can be used to feed analyses;



# Beacon Network

<https://www.beacon-network.org>

- 52 beacons around the world:
  - EMBL/EBI;
  - HapMap;
  - Broad Institute;
  - Ontario Institute for Cancer Research;
  - Wellcome Trust Sanger Institute
- BIPMed is the first Brazilian and first Latin-American beacon.

A global search engine for genetic mutations.

GRCh37 ▾ e.g. 1 : 100,000 A>C Search

Example: BRCA2 V...

### What's a Beacon?

Beacon is a genetic mutation sharing platform developed by the Global Alliance for Genomics and Health. A beacon is web service that any institution can implement to share genetic data. A beacon answers questions of the form "Do you have information about the following mutation?" and responds with one of "Yes" or "No", among potentially more information.

A site offering this service is called a "beacon". This open web service is designed both to be technically simple while providing data generators options for distributing data through proportional safeguards.

### The Beacon Network

The Beacon Network is a search engine across the world's public beacons. It enables global discovery of genetic mutations, federated

### Recent News

- [Beacon Project Cracks the Door for Genomic Data Sharing](#)  
BioIT World, August 14, 2015
- [Beacons for Data Sharing](#)  
BioTechniques, August 12, 2015
- [Lighting beacons for data access in genomic research](#)  
EMBL-EBI, June 15, 2015
- [10 Breakthrough Technologies: Internet of DNA](#)  
MIT Technology Review, February 18, 2015
- [Geneticists Begin Tests of an Internet for DNA](#)  
MIT Technology Review, December 17, 2014

Global Alliance for Genomics & Health | Terms of Use | Email Us | Mailing List | @BeaconOfBeacons

# Beacon Network

- Asks questions to several beacons;
- Reports the answer to the user;
- Joint effort to share knowledge on genomic variation;
- Answers are subsettable;

Advanced Options










GRCh37 ▾ 13 : 32954208 T>A Search

Response	All None
<input checked="" type="checkbox"/> Found	4
<input checked="" type="checkbox"/> Not Found	23
<input checked="" type="checkbox"/> Error	2

Organization	All None
<input checked="" type="checkbox"/> AMPLab, University o...	
<input checked="" type="checkbox"/> BioReference Labora...	
<input checked="" type="checkbox"/> DNASTack	only
<input checked="" type="checkbox"/> SolveBio	
<input checked="" type="checkbox"/> University of Californi...	

Show All

	1000 Genomes - Thousand Genomes YRI ... DNASTack	<span>Not Found</span> <span>More info...</span>
	AMPLab - 1000 Genomes Project AMPLab, University of California	<span>Not Found</span>
	Biobase - HGMD University of California, Santa Cruz	<span>Not Found</span>
	BioReference BioReference Laboratories	<span>Not Found</span>
	SolveBio - 1000G/1.1.0-2015-01-08/1000g SolveBio	<span>Not Found</span>
	SolveBio - CIVIC/1.0.0-2015-04-07/Variants SolveBio	<span>Not Found</span>
	SolveBio - ClinVar/3.6.0-2015-07-13/ClinVar SolveBio	<span>Found</span>
	SolveBio - ClinVar/3.6.0-2015-07-13/Variants SolveBio	<span>Found</span>
	SolveBio - COSMIC/1.1.0-COSMIC71/Som... SolveBio	<span>Not Found</span>

# Safety Mechanism

- To avoid abuse (e.g., computer program asking questions several times, hoping to identify individuals), safety measures are implemented;
- For every request, the beacon contacts our Bottleneck Server;
- The server adds 150msec per extra request made from same IP;
- After every second without request, timer is reduced by 10msec;
- If timer gets to 10s, response gets delayed by this timer;
- If timer gets to 20s, beacon blocks the IP, until timer gets to lower than 20s;

Try and Let Us Know!!

<http://bipmed.iqm.unicamp.br>

<https://bipmed.iqm.unicamp.br/beacon>

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