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:

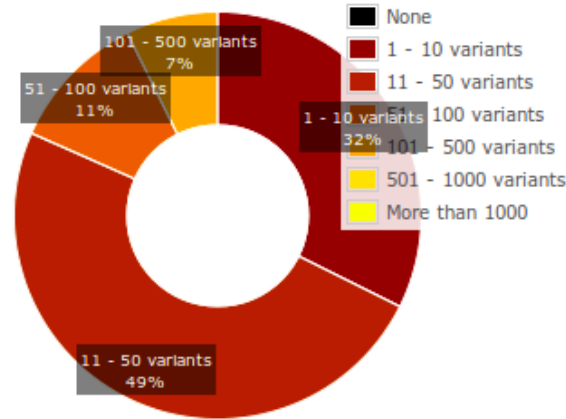http://bipmed.iqm.unicamp.br

Current status

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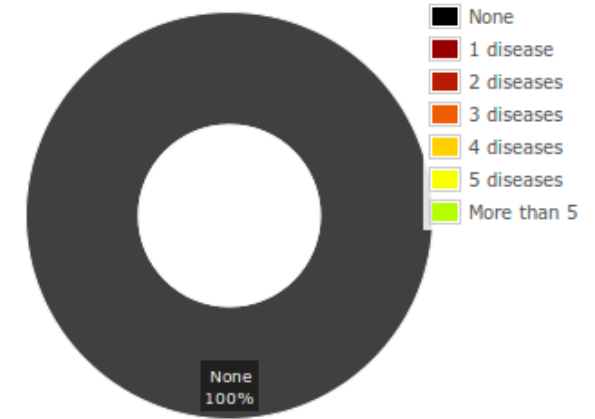- We accept patient data;
- We can control data access;
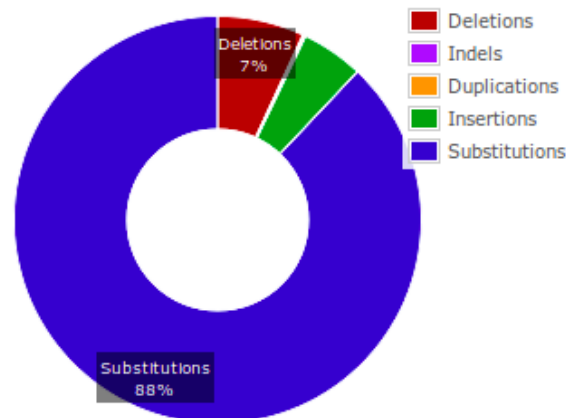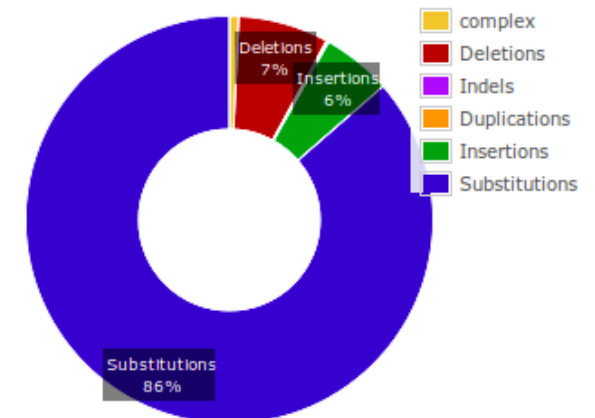
The Beacon

- 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 Beacon

This beacon provides data on individuals from the reference population.

Chromosome:

| 1 |

Position:

| 14463 |

Allele:

| TI |

| Submit |

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.

Beacon Network

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

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

bipmed@bipmed.iqm.unicamp.br