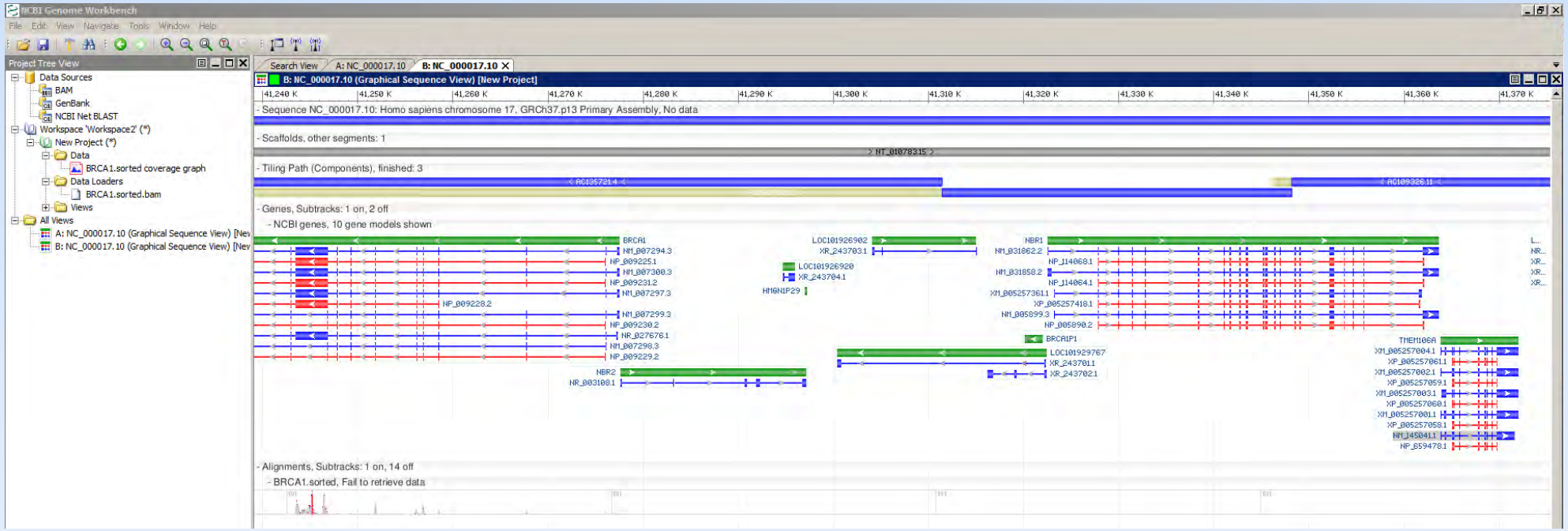
Genomic Variation in the Rising Era of Individual Genome Sequence:

Why we are Enabling Everyone to Share and Use Public Datasets

August 2016



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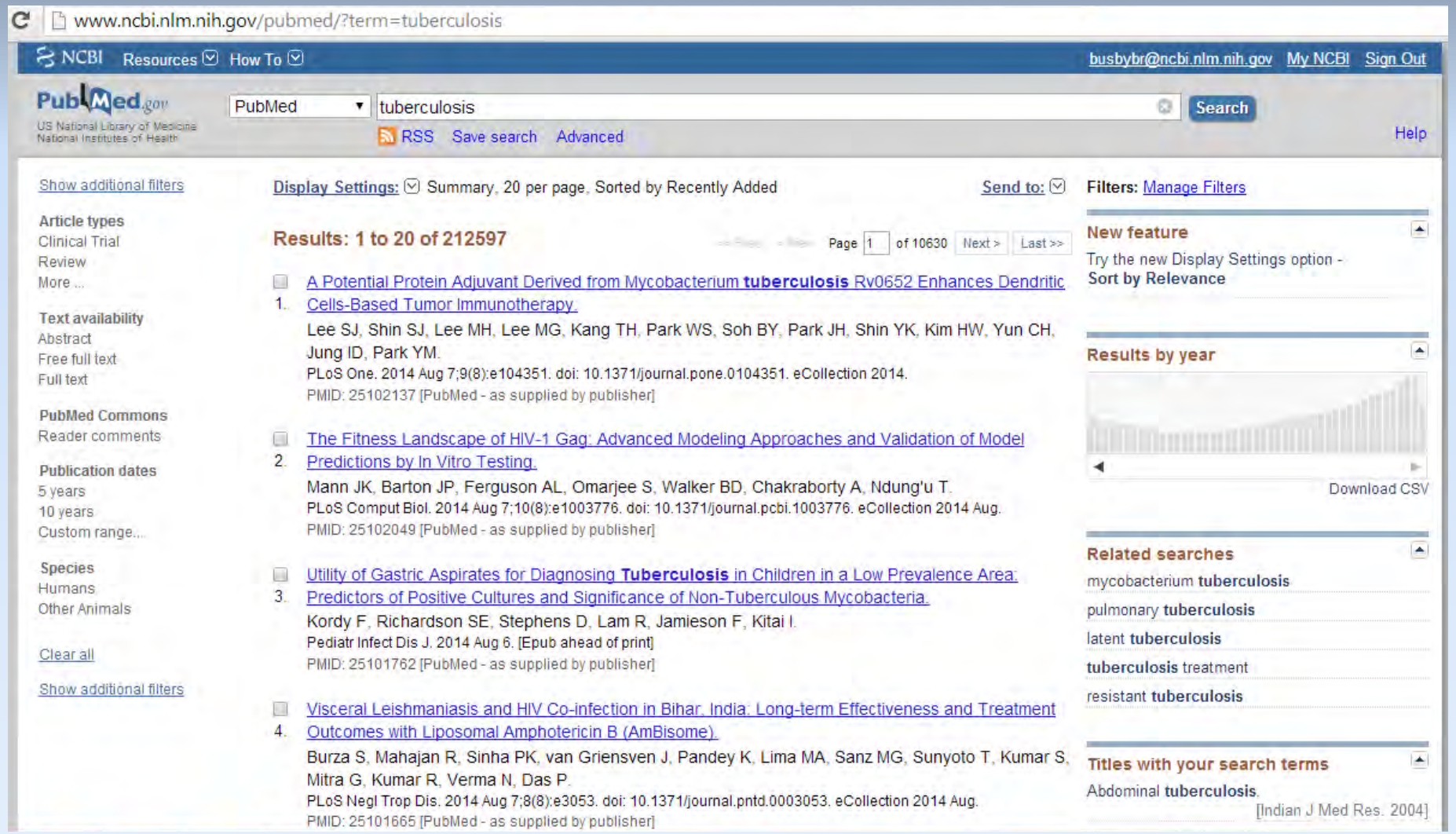
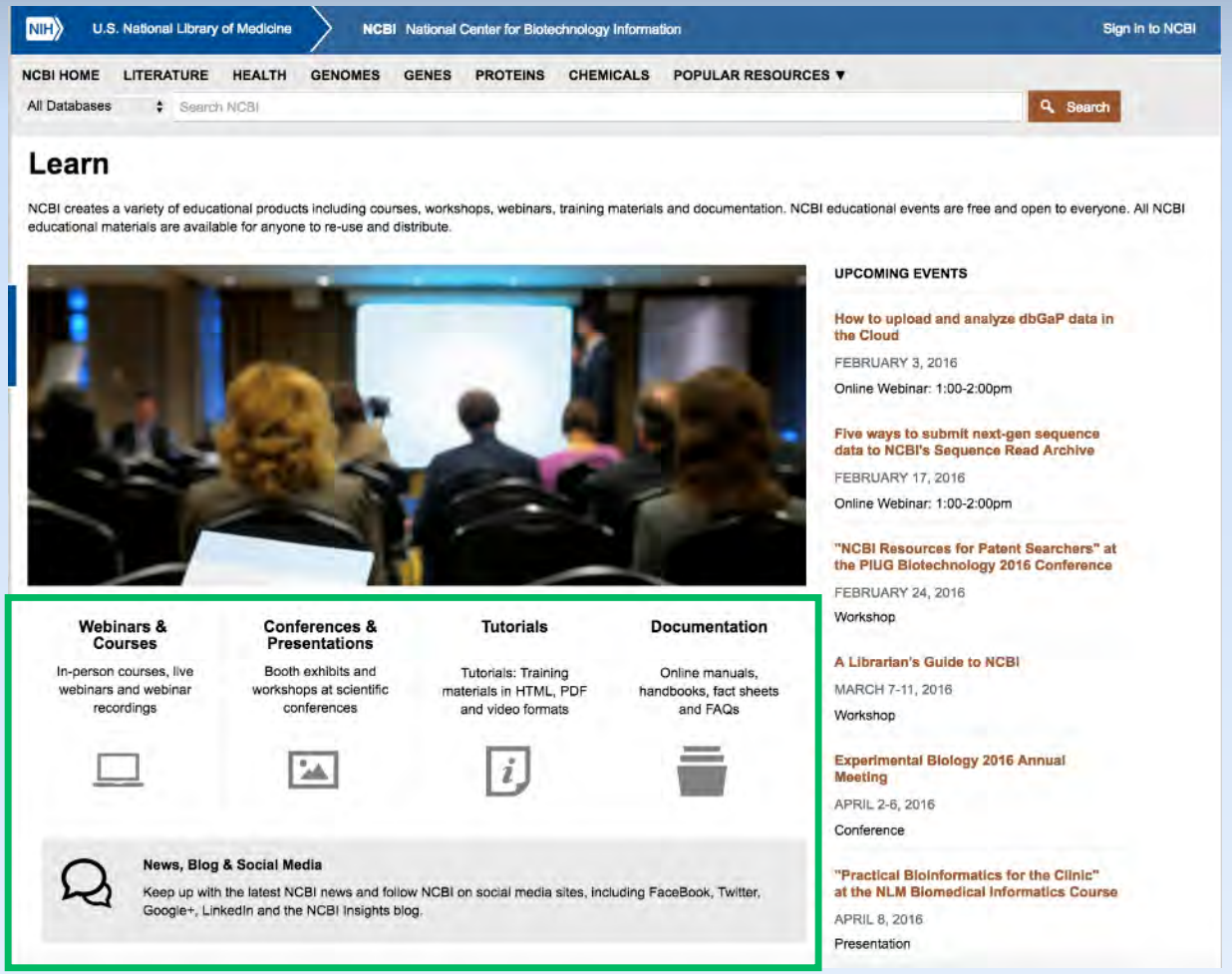
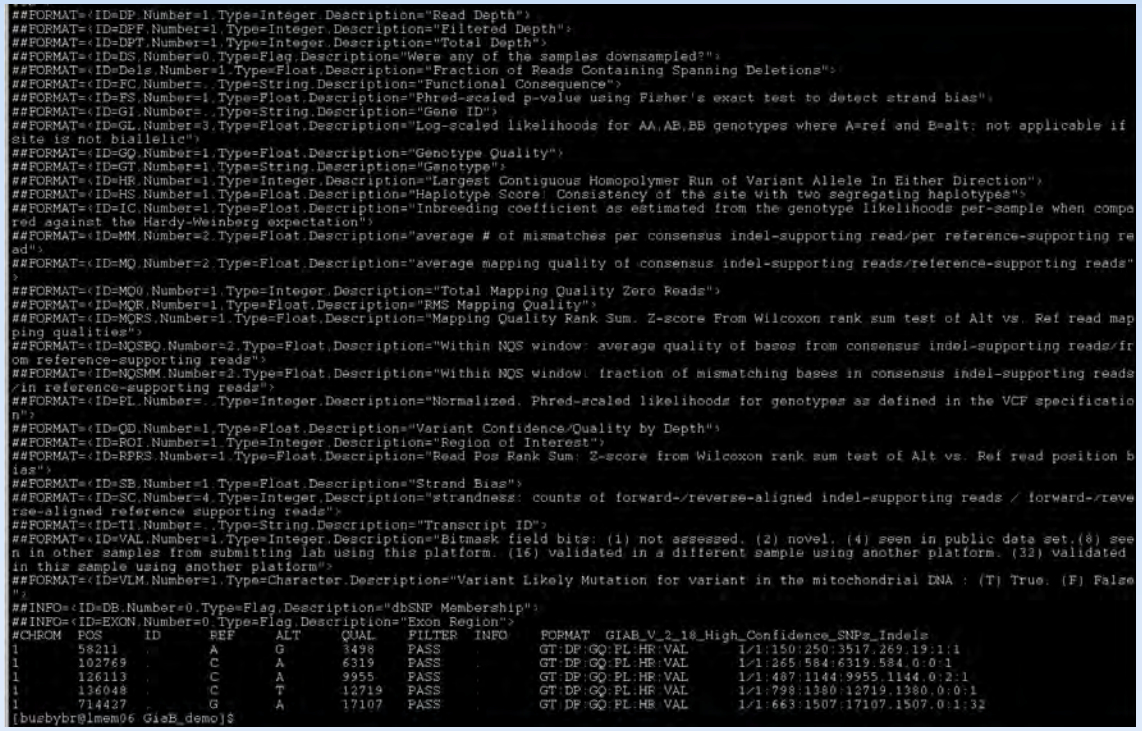
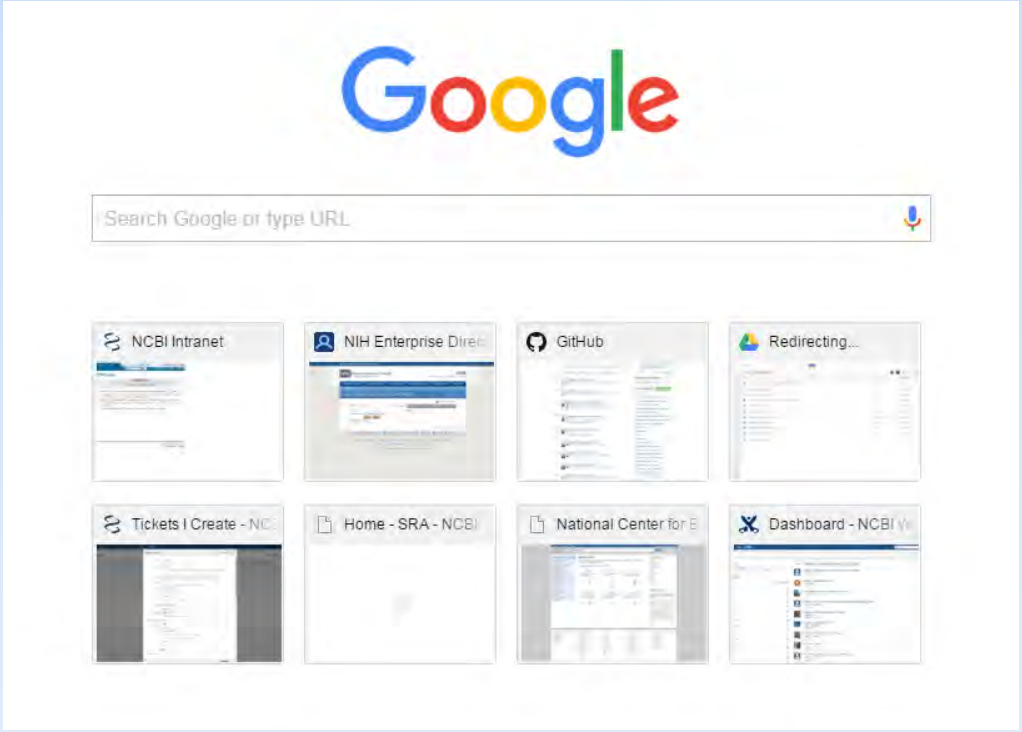


For more information go to: ncbi.nlm.nih.gov/learn

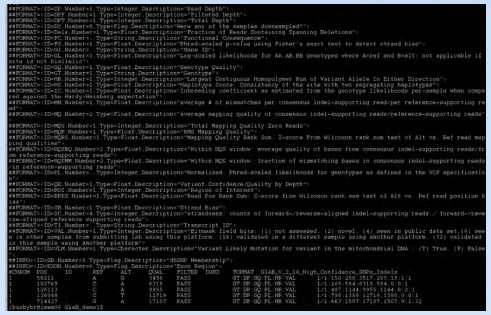


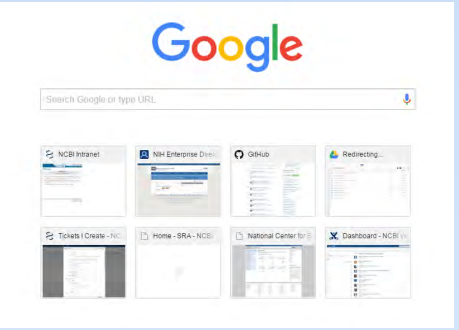
The Two Things You Need to Know to Handle "Big Data"





The Two Things You Need to Know to Handle "Big Data"

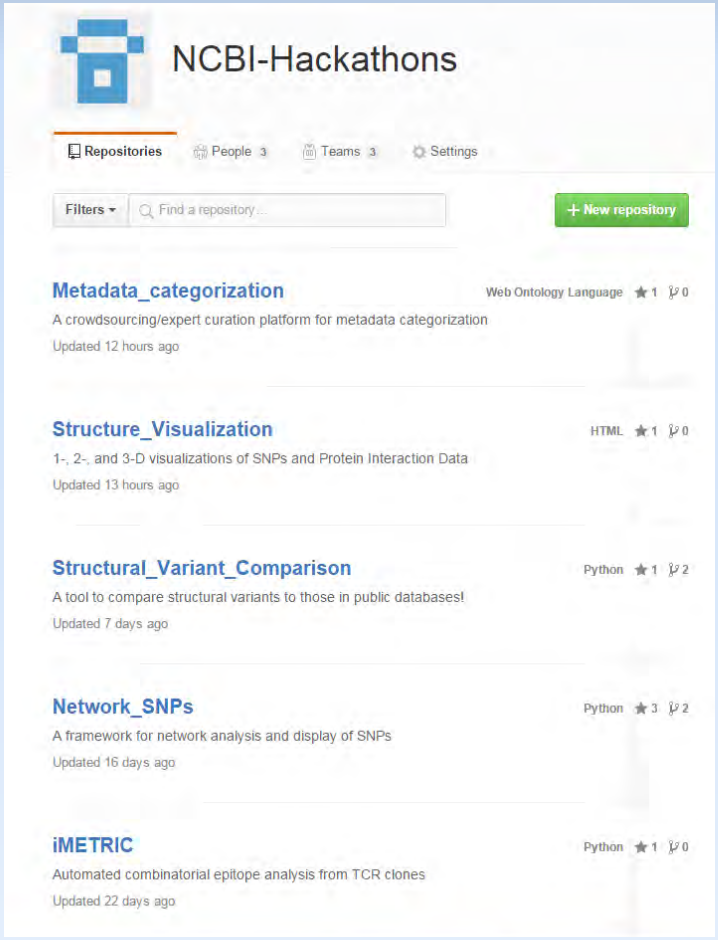




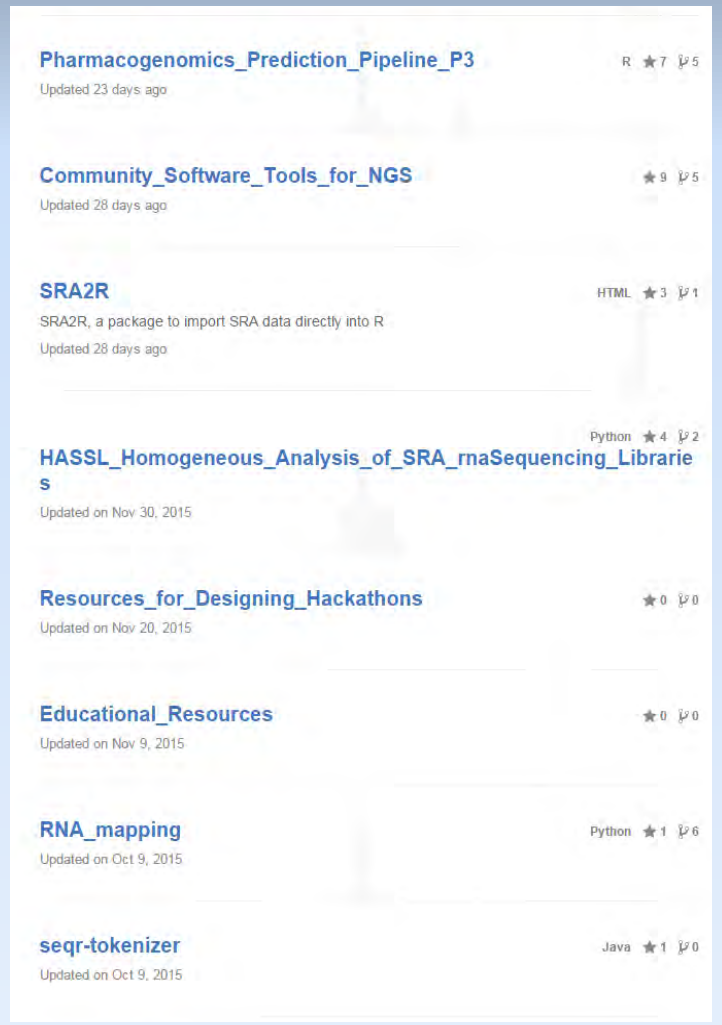
- Software Carpentry
- Stack Overflow
- Biostars and SEQanswers

Oh yeah, and this...

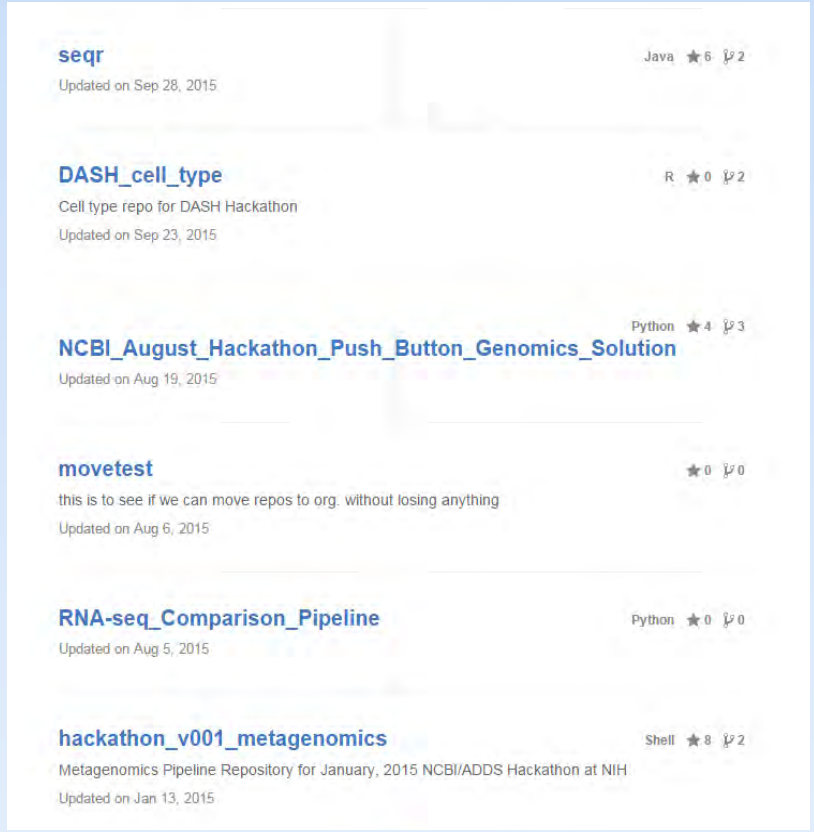
github.com/NCBI-hackathons



github.com/NCBI-hackathons



github.com/NCBI-hackathons



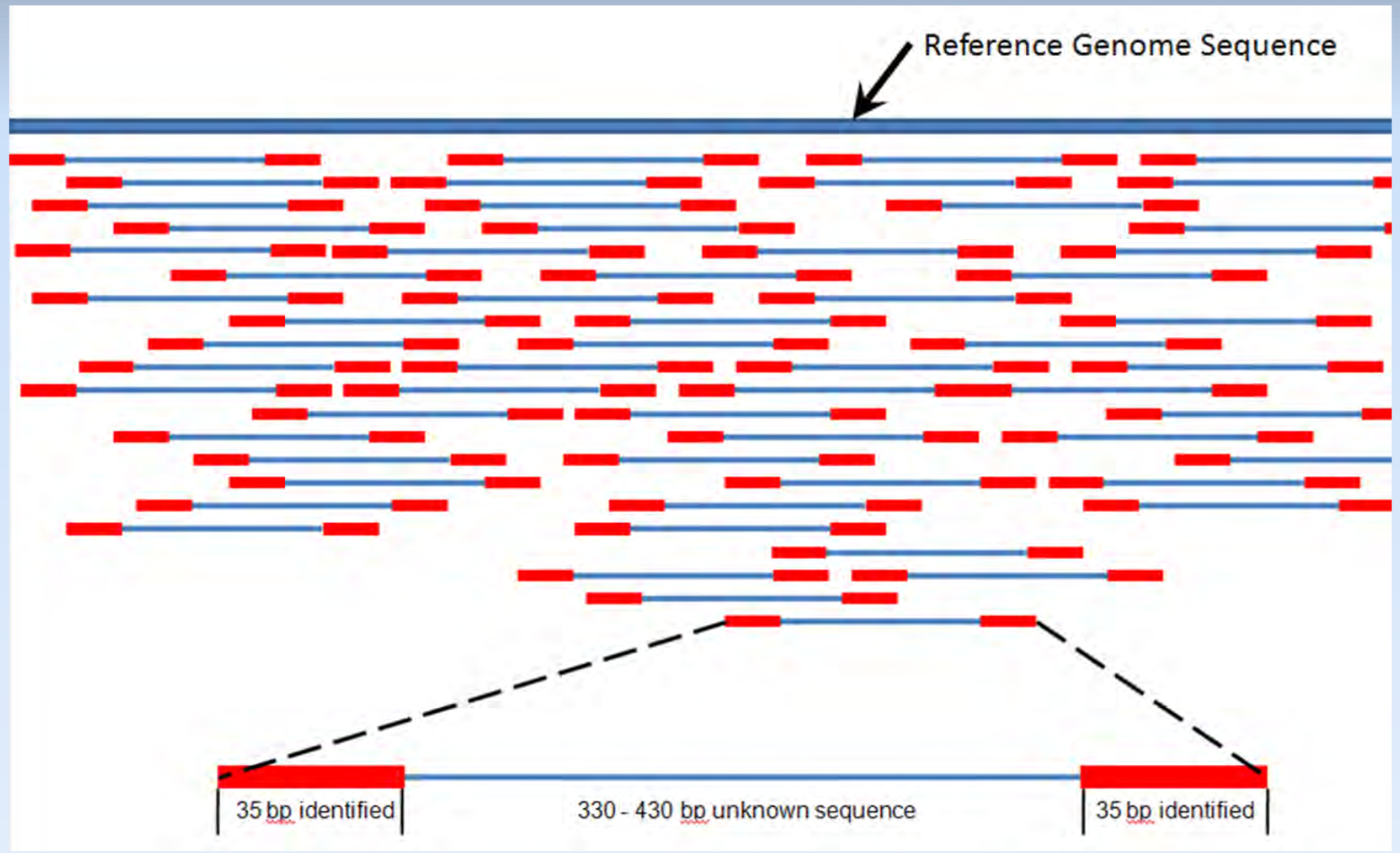
The Five Things You Need to Know About "Big Data"

examples from genomics

- Check data quality
  - Current largest source of noise is library prep
- Setting parameters (or not)
- Remember multiple etiologies
  - "If it looks like a duck"
- Multiple testing correction
  - Not Bonferroni
- Validation can come in many forms (as long as its independent)

Review of terminology and concepts

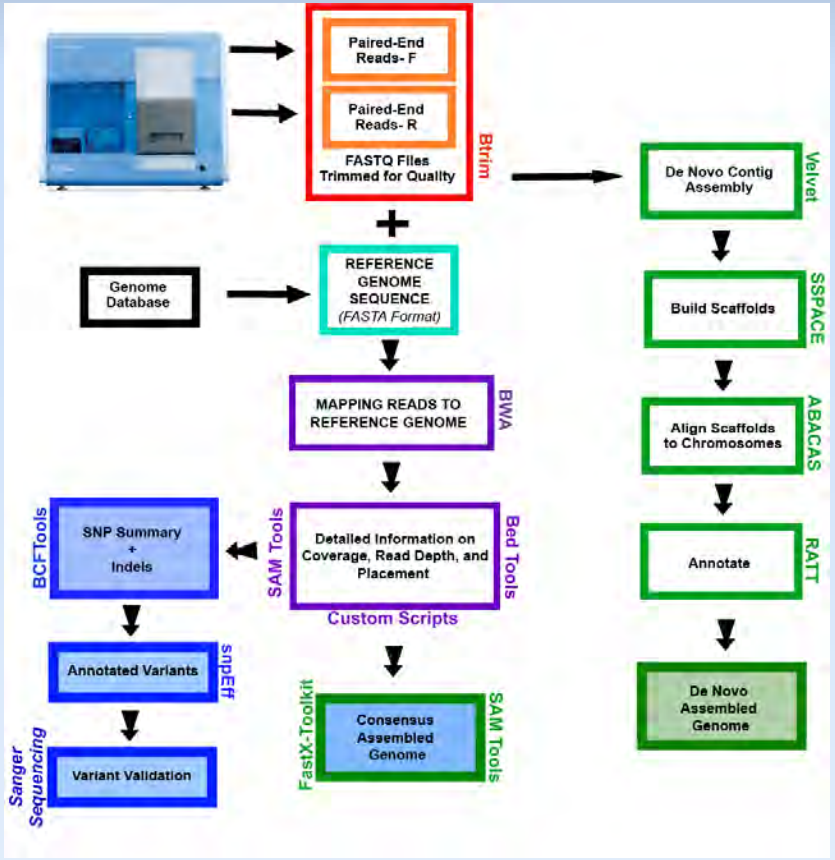
Next Generation Sequencing



Graphic Credit:
Spencer Martin, UBC

Review of terminology and concepts

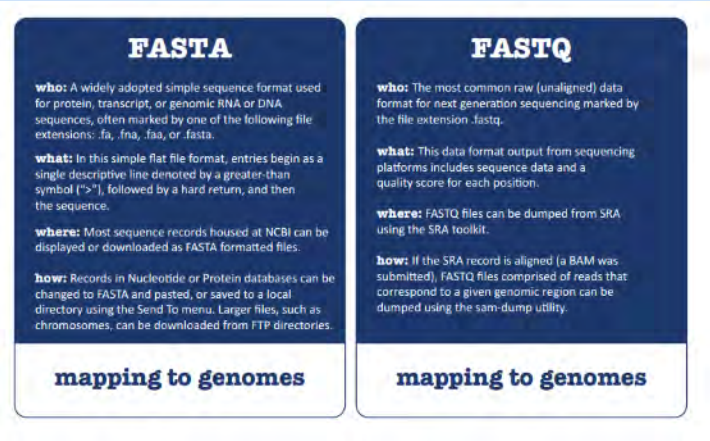
How Genomes are Mapped and Assembled

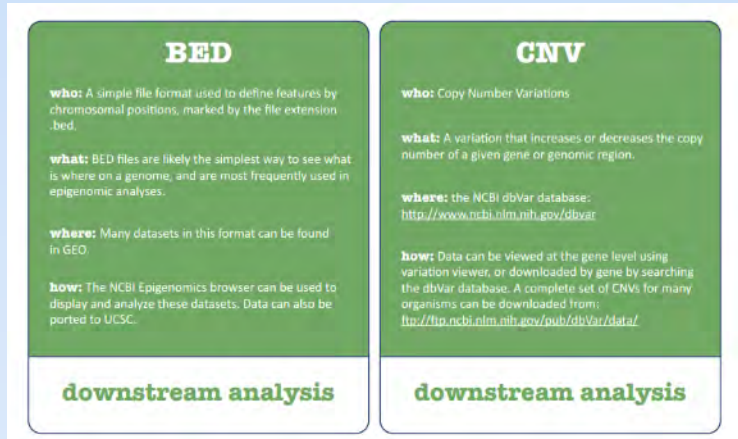


Review of terminology and concepts

How Genomes are Mapped and Assembled

http://1.usa.gov/1J1xmYs





NCBI NGS Online Workshop – Available on the NCBI YouTube Channel!

Some Supplemental Material for Those New to Bioinformatics and Genomics

The opinions expressed here are those of the author.
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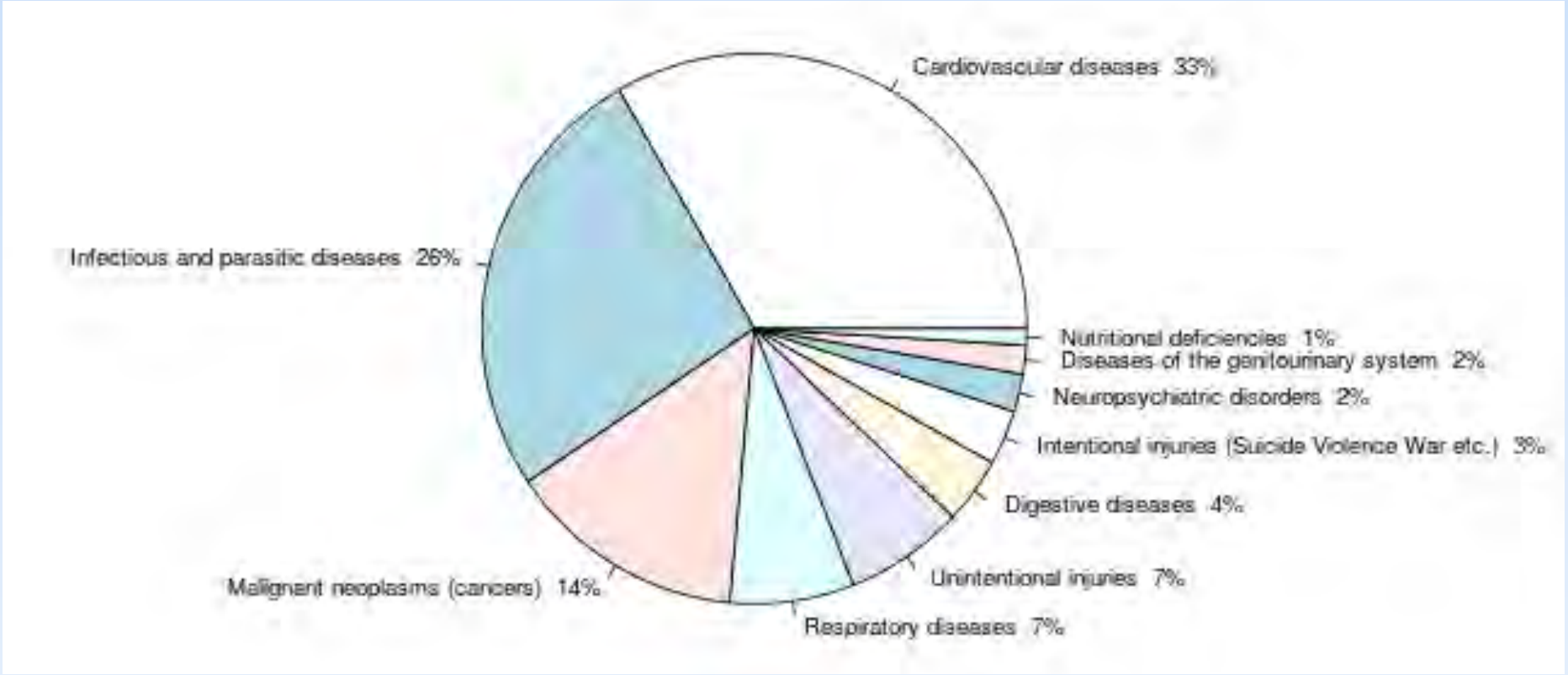
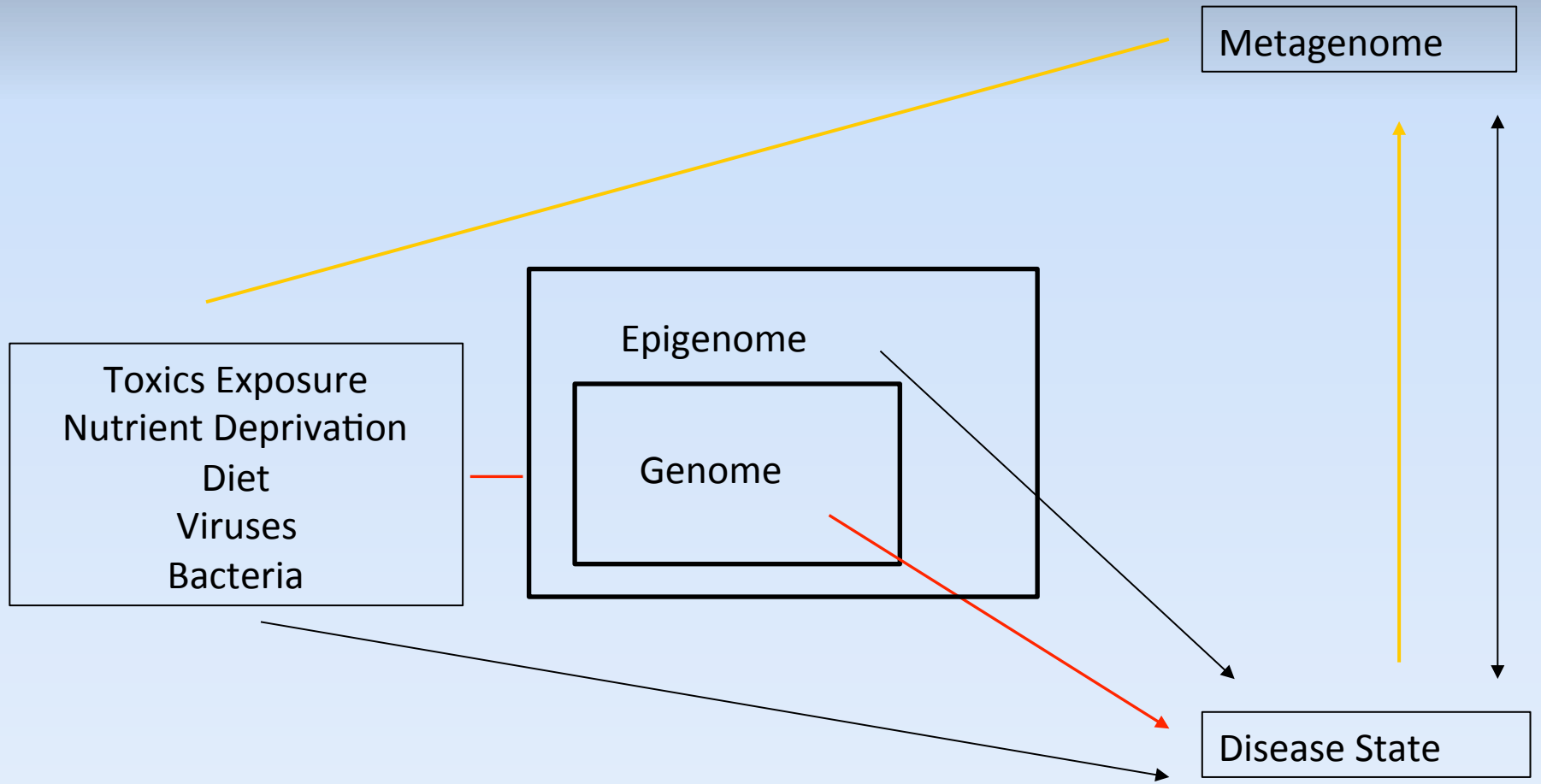
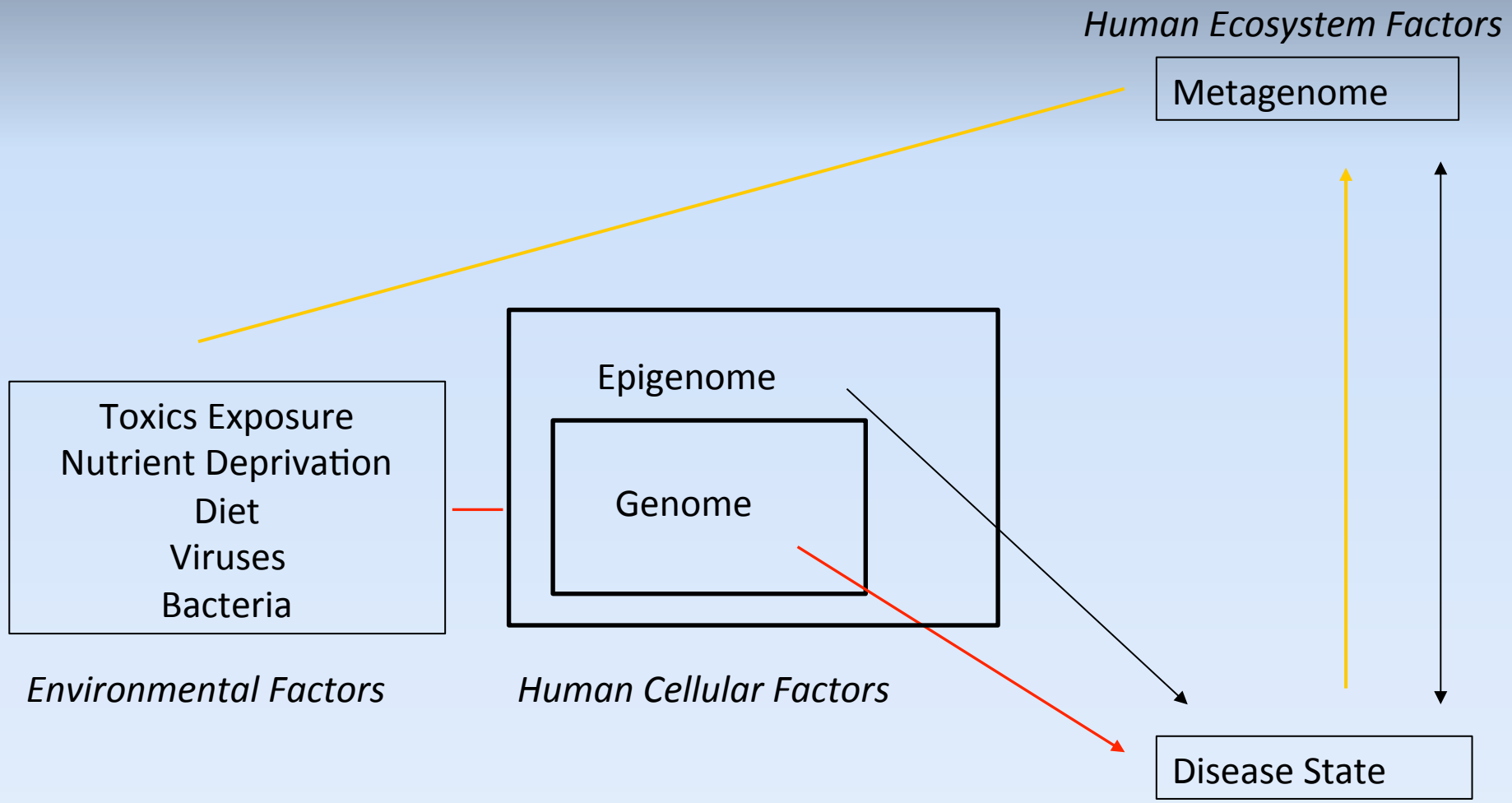


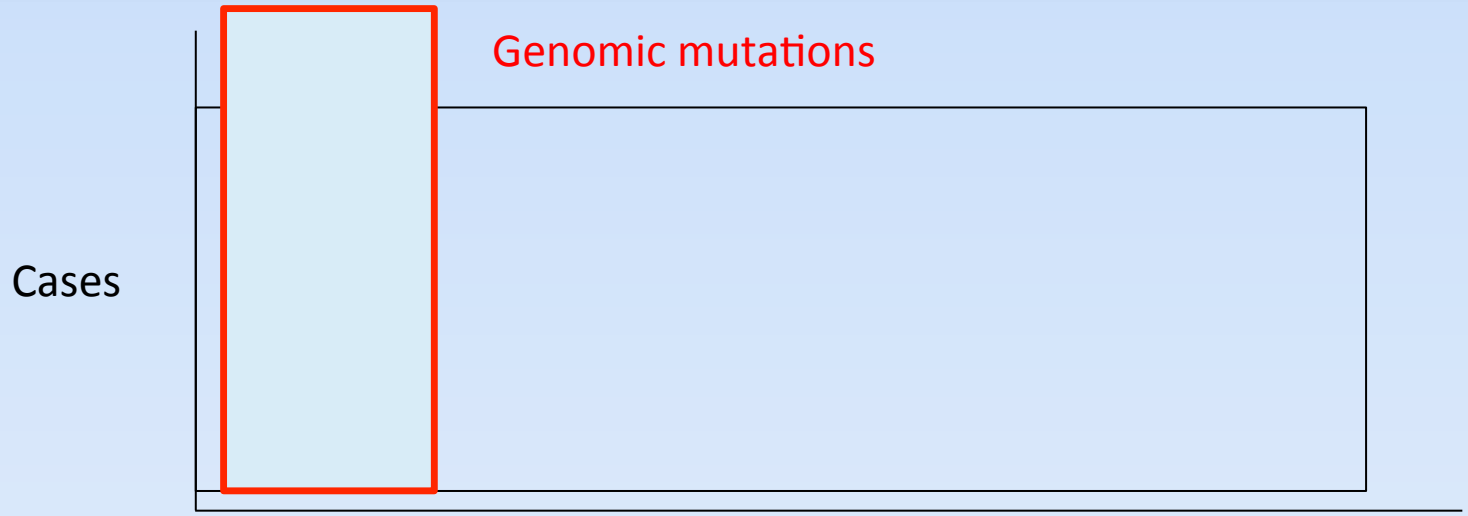
Biological Information Flows and Disease States



Defining Terms



Rethinking Disease

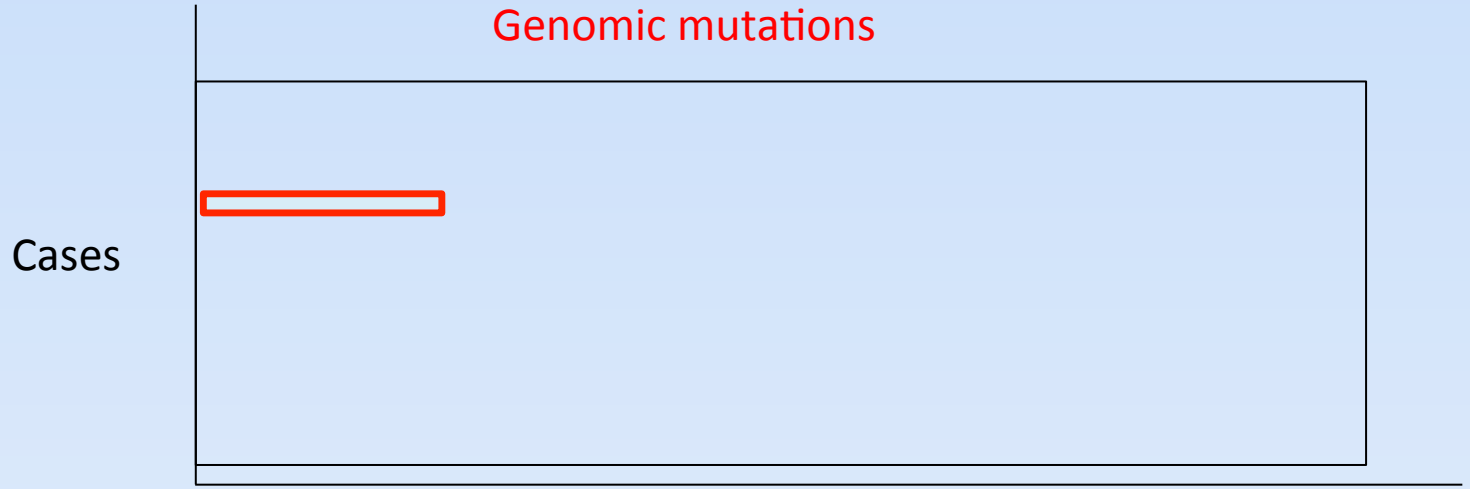


"Etiology == Genomic Cause"

A completely hypothetical example to Illustrate a point

~~1 disease~~
~~1 genetic~~
~~cause~~

Rethinking Disease

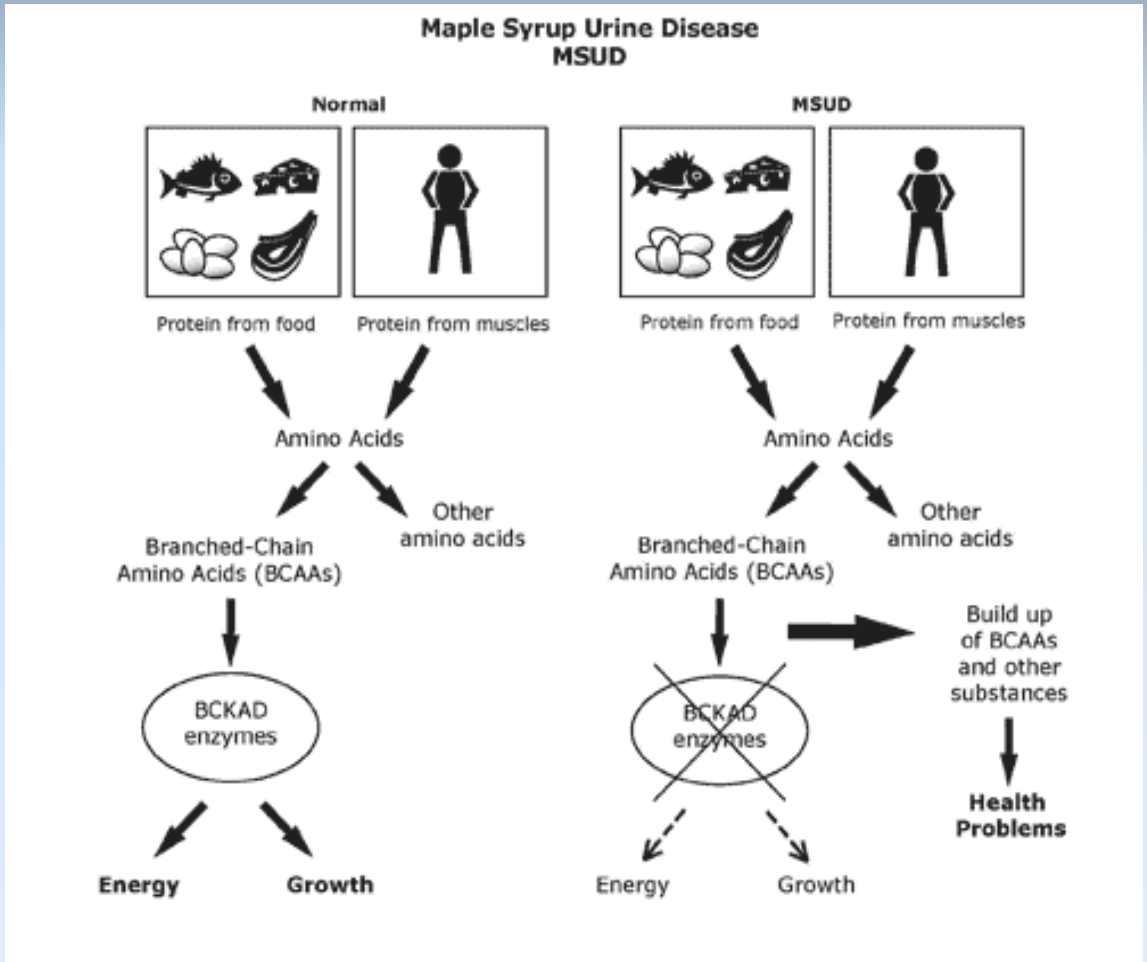


"Etiology == Genomic Cause"

A completely hypothetical example to Illustrate a point

~~1 disease~~
~~1 genetic~~
~~cause~~

Rare but Treatable Variants!



http://www.nature.com/news/rare-diseases-genomics-plain-and-simple-1.10125

Gene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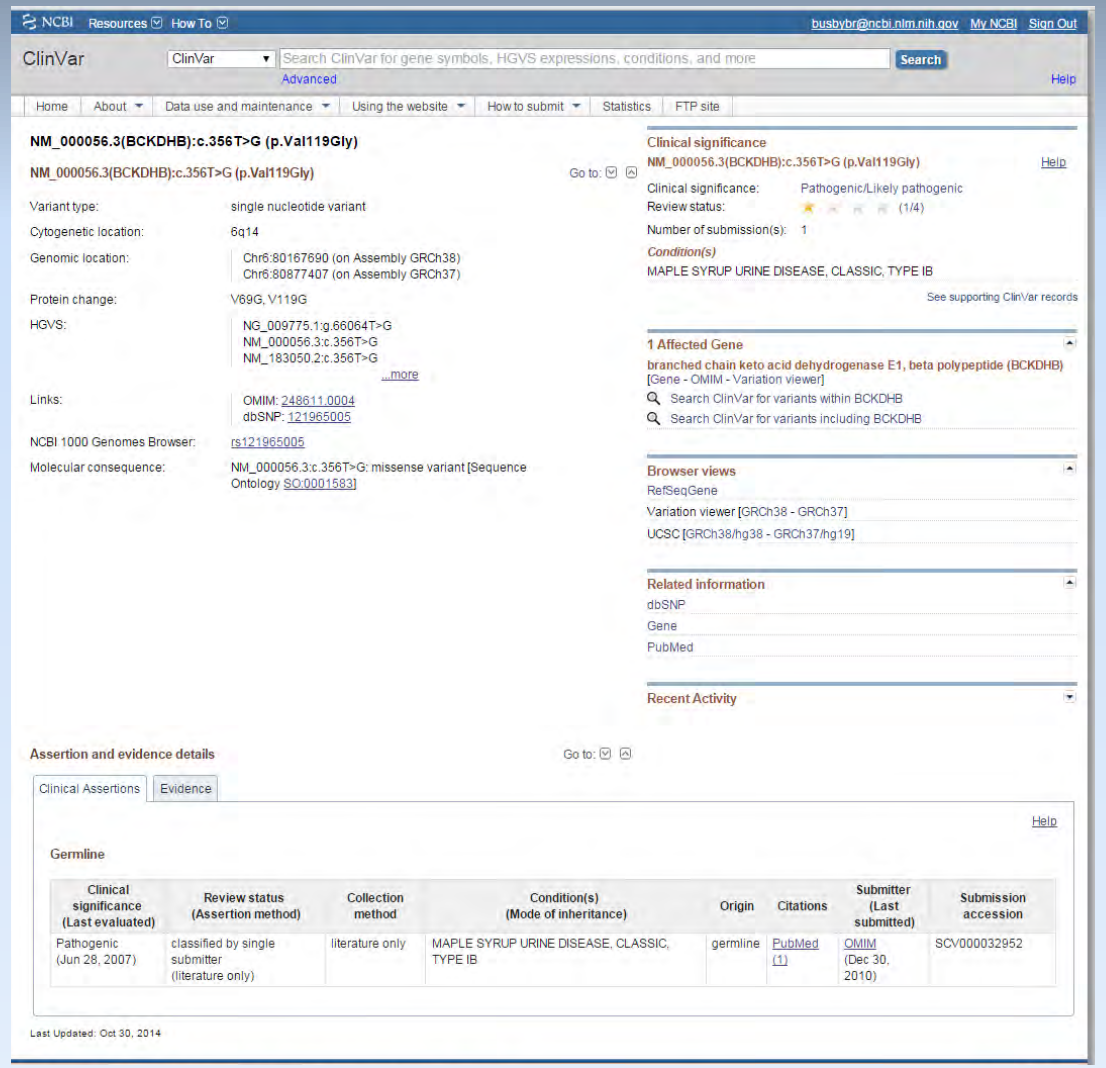
BCKDHB branched chain keto acid dehydrogenase E1, beta polypeptide [Homo sapiens (human)]
Gene ID: 594, updated on 2-Nov-2014
Summary
Official Symbol BCKDHB provided by HGNC
Official Full Name branched chain keto acid dehydrogenase E1, beta polypeptide provided by HGNC
Primary source HGNC:HGNC:987
Locus tag RP1-279A18.1
See related Ensembl:ENSG00000083123; HPRD:02011; MIM:248611; Vega:OTTHUMG00000016430
Gene type protein coding
RefSeq status REVIEWED
Organism Homo sapiens
Lineage Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as E1B; dJ279A18.1
Summary Branched-chain keto acid dehydrogenase is a multienzyme complex associated with the inner membrane of mitochondria, and functions in the catabolism of branched-chain amino acids. The complex consists of multiple copies of 3 components: branched-chain alpha-keto acid decarboxylase (E1), lipoamide acyltransferase (E2) and lipoamide dehydrogenase (E3). This gene encodes the E1 beta subunit, and mutations therein have been associated with maple syrup urine disease (MSUD), type 1B, a disease characterized by a maple syrup odor to the urine in addition to mental and physical retardation, and feeding problems. Alternative splicing at this locus results in transcript variants with different 3' non-coding regions, but encoding the same isoform. [provided by RefSeq, Jul 2008]
Genomic context
Location: 6q14.1
See BCKDHB in Epigenomics, MapViewer
Table of contents
Summary
Genomic context
Genomic regions, transcripts, and products
Bibliography
Phenotypes
Variation
Pathways from BioSystems
Interactions
General gene information
Markers, Clone Names, Homology, Gene Ontology
General protein information
NCBI Reference Sequences (RefSeq)
Related sequences
Additional links
Locus-specific Databases
Related information
Order cDNA clone
3D structures
BioAssay
BioAssay by Target (List)

ClinVar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NM_000056.3(BCKDHB):c.356T>G (p.Val119Gly)
NM_000056.3(BCKDHB):c.356T>G (p.Val119Gly)
Go to:
Variant type: single nucleotide variant
Cytogenetic location: 6q14
Genomic location: Chr6:80167690 (on Assembly GRCh38)
Chr6:80877407 (on Assembly GRCh37)
Protein change: V69G, V119G
HGVS: NG_009775.1:g.66064T>G
NM_000056.3:c.356T>G
NM_183050.2:c.356T>G
...more
Links: OMIM: 248611.0004
dbSNP: 121965005
NCBI 1000 Genomes Browser: rs121965005
Molecular consequence: NM_000056.3:c.356T>G: missense variant [Sequence Ontology SO:0001583]
Clinical significance
NM_000056.3(BCKDHB):c.356T>G (p.Val119Gly)
Help
Clinical significance: Pathogenic/Likely pathogenic
Review status: (1/4)
Number of submission(s): 1
Condition(s)
MAPLE SYRUP URINE DISEASE, CLASSIC, TYPE IB
See supporting ClinVar records
1 Affected Gene
branched chain keto acid dehydrogenase E1, beta polypeptide (BCKDHB)
[Gene - OMIM - Variation viewer]
Search ClinVar for variants within BCKDHB
Search ClinVar for variants including BCKDHB
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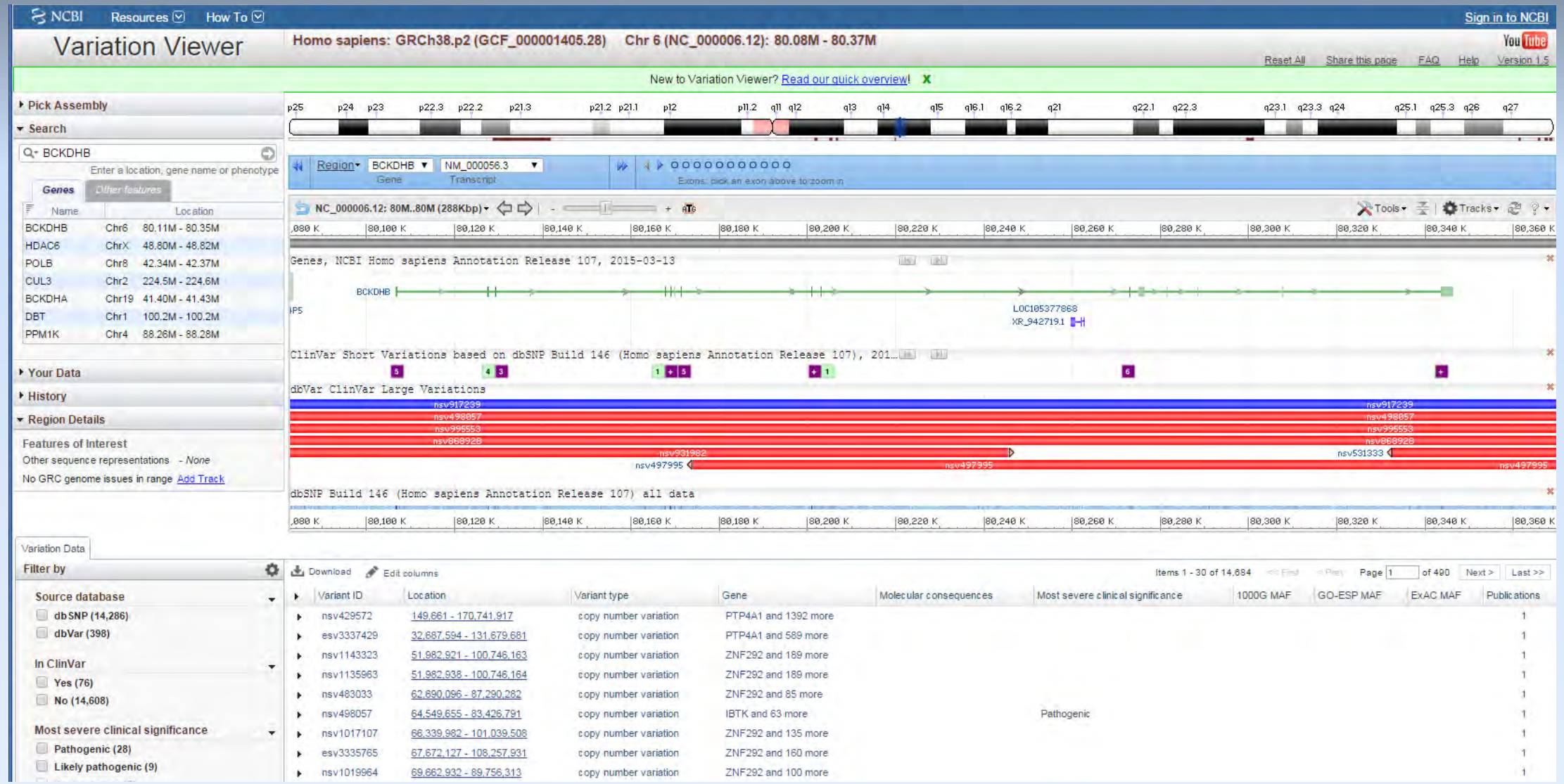
Review status (Assertion method)
Collection method
Condition(s) (Mode of inheritance)
Origin
Citations
Submitter (Last submitted)
Submission accession
Pathogenic (Jun 28, 2007)
classified by single submitter (literature only)
literature only
MAPLE SYRUP URINE DISEASE, CLASSIC, TYPE IB
germline
PubMed (1)
OMIM (Dec 30, 2010)
SCV000032952
Last Updated: Oct 30, 2014

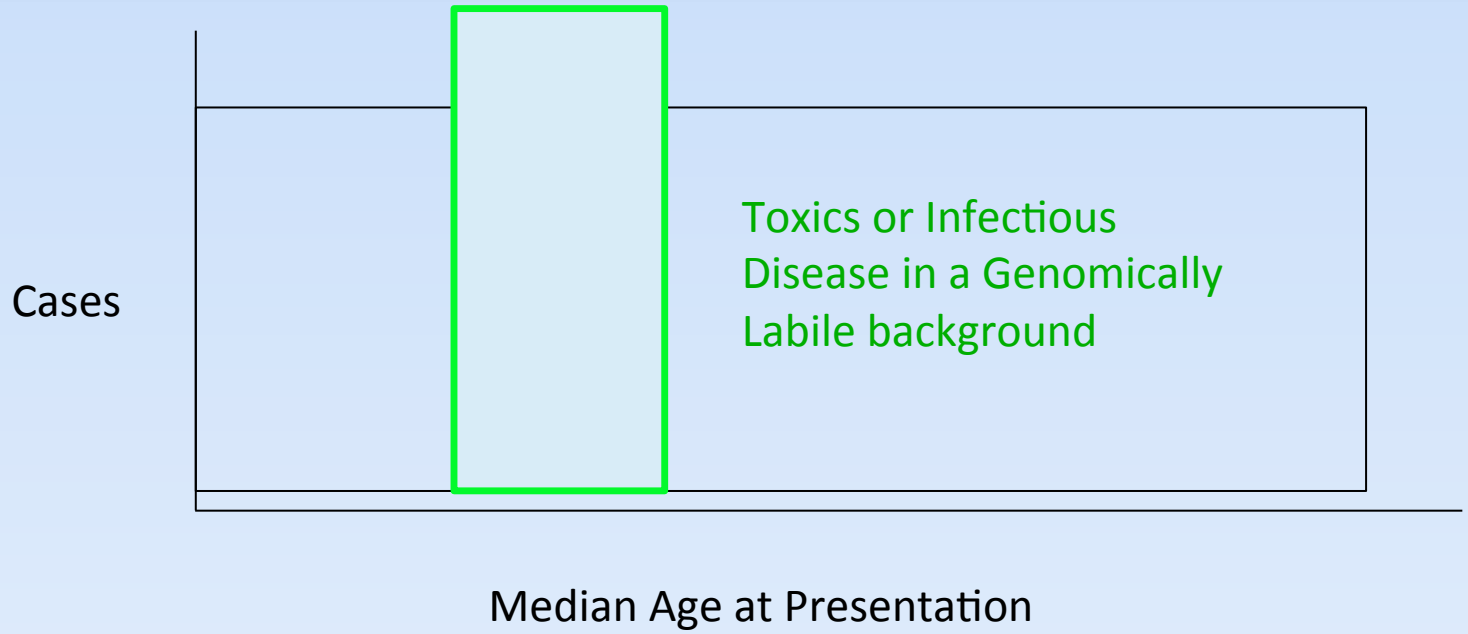
Variation Viewer



Clinical Applications

Gene – Bonus Round!

Rethinking Disease



A completely hypothetical example to
Illustrate a point

Pharmacogenomics

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Thomson Reuters and NuMedii Launch Ground-Breaking Initiative to Identify Drugs for Repurposing

Companies leverage content, Big Data analytics and expertise to improve success of drug discovery

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PHILADELPHIA, Nov. 4, 2013 /PRNewswire/ -- The IP & Science business of Thomson Reuters, the world's leading provider of intelligent information for businesses and professionals, today announced a strategic initiative with NuMedii, a biotech company dedicated to revolutionizing drug discovery, to identify new treatments for areas of unmet medical need. The partnership, which pairs NuMedii's technology with the authoritative, hand-curated drug and disease information and systems biology expertise of Thomson Reuters, will create an unprecedented repository of content and methodologies to systematically identify new applications for existing drug compounds.

NuMedii will use Thomson Reuters MetaCore and Integrity content, as well as Thomson Reuters Life Sciences Professional Services expertise in systems biology, in conjunction with its unique Big Data technology and databases, to find FDA-approved drugs or discontinued development compounds that are appropriate for repurposing. The companies will leverage this comprehensive collection of high-quality data, knowledge and predictive technologies to identify therapeutic candidates with the greatest probability for clinical success, in turn fulfilling NuMedii's mission to discover and de-risk drugs that target new pathologies.

"NuMedii is blending various innovative technologies and life science data to create a next generation drug discovery and development engine," said Gini Deshpande, Ph.D., founder and chief executive officer of NuMedii. "This unique partnership

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A Drug Repositioning Approach Identifies Tricyclic Antidepressants as Inhibitors of Small Cell Lung Cancer and Other Neuroendocrine Tumors

Nadine S. Jahchan[1,2], Joel T. Dudley[1], Pawel K. Mazur[1,2], Natasha Flores[1,2], Dian Yang[1,2], Alec Palmerton[1,2], Anne-Flore Zmoos[1,2], Dedeepya Vaka[1,2], Kim Q.T. Tran[1,2], Margaret Zhou[1,2], Karolina Krasinska[3], Jonathan W. Riess[4], Joel W. Neal[5], Purvesh Khatri[1,2], Kwon S. Park[1,2], Atul J. Butte[1,2] and Julien Sage[1,2]

Author Affiliations

Author Notes

Corresponding Authors:
Julien Sage, Stanford University, 265 Campus Drive, SIM1 G2078, Stanford, CA 94305-5457. Phone: 650-724-9246; Fax: 650-736-0195; E-mail: julsage@stanford.edu; and Atul J. Butte, Stanford University, 251 Campus Drive West, MSOB X1C63, Stanford, CA 94305-5479. Phone: 650-725-1337; Fax: 650-723-7070;abutte@stanford.edu

J.T. Dudley and P.K. Mazur contributed equally to this work.

This Article

Published OnlineFirst September 26, 2013; doi: 10.1158/2159-8290.CD-13-0183

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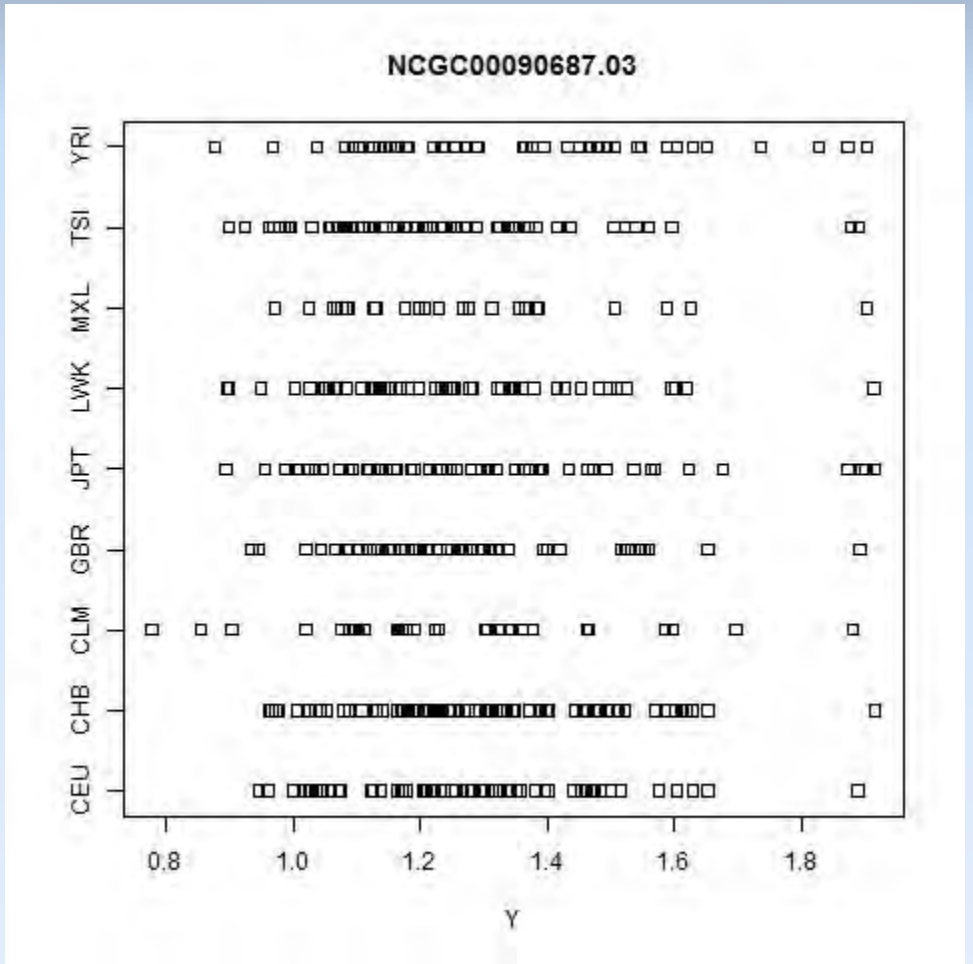
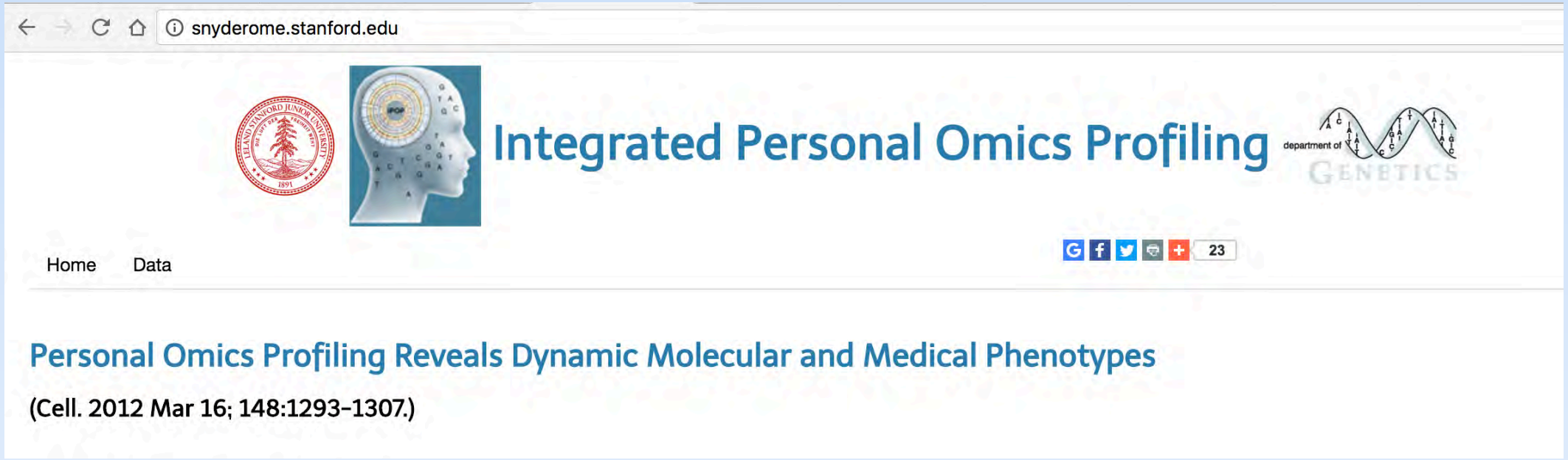
Toxicogenomics



Bacterial and Viral Infections may Trigger Disease Incidence



Bacterial and Viral Infections may Trigger Disease Incidence

Fig. S8

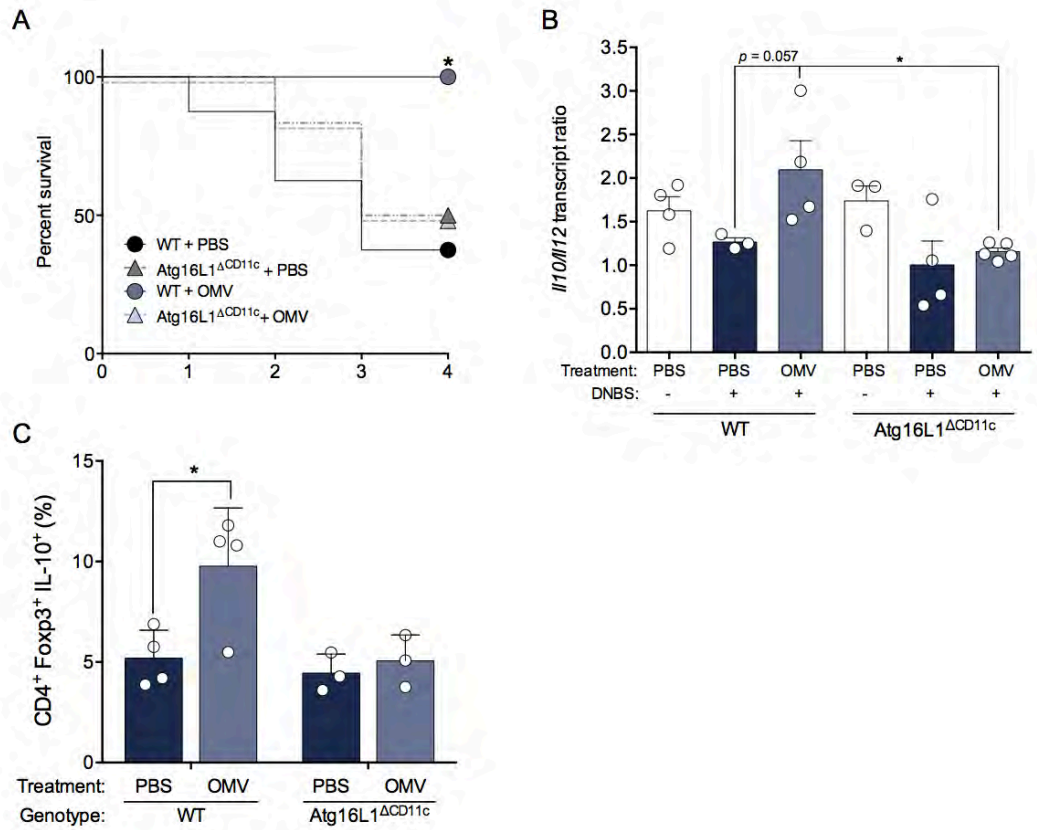
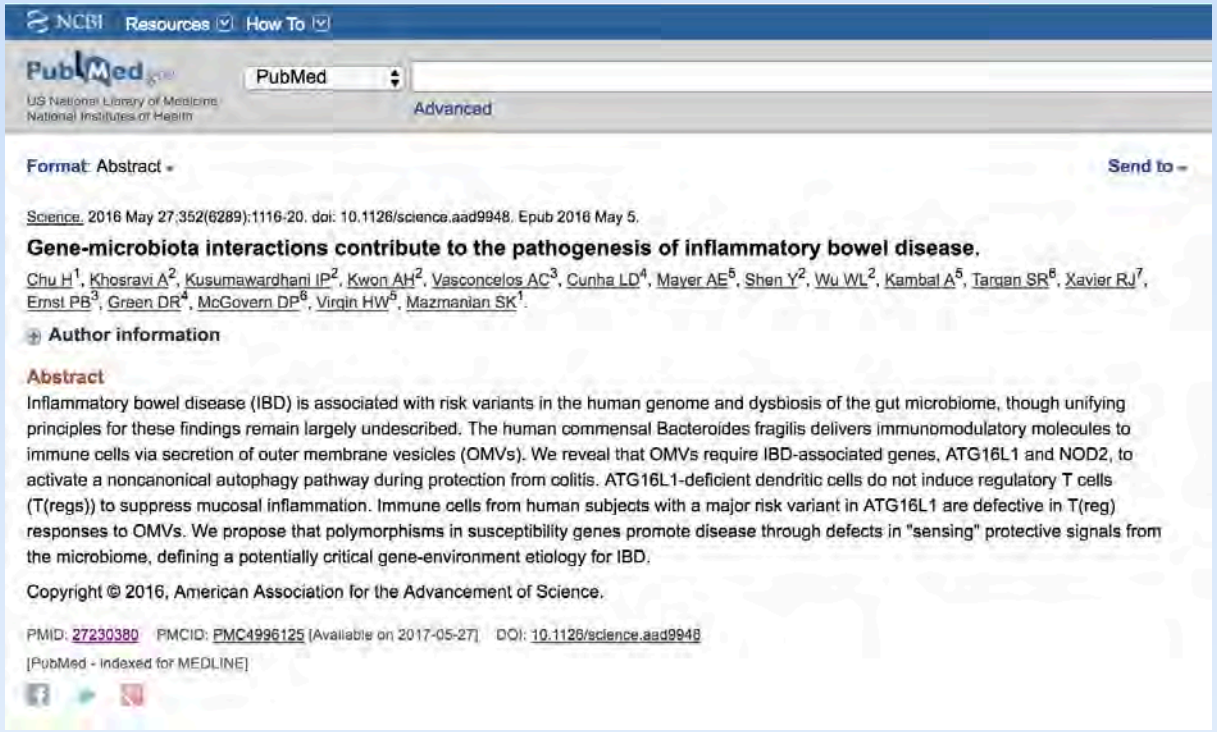


Fig. S8. *B. fragilis* OMV-mediated protection from colitis requires ATG16L1 in DCs.



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Science. 2016 May 27;352(6289):1116-20. doi: 10.1126/science.aad9948. Epub 2016 May 5.

Gene-microbiota interactions contribute to the pathogenesis of inflammatory bowel disease.

Chu H[1], Khosravi A[2], Kusumawardhani IP[2], Kwon AH[2], Vasconcelos AC[3], Cunha LD[4], Mayer AE[5], Shen Y[2], Wu WL[2], Kambal A[5], Targan SR[6], Xavier RJ[7], Ernst PB[3], Green DR[4], McGovern DP[6], Virgin HW[5], Mazmanian SK[1].

Author information

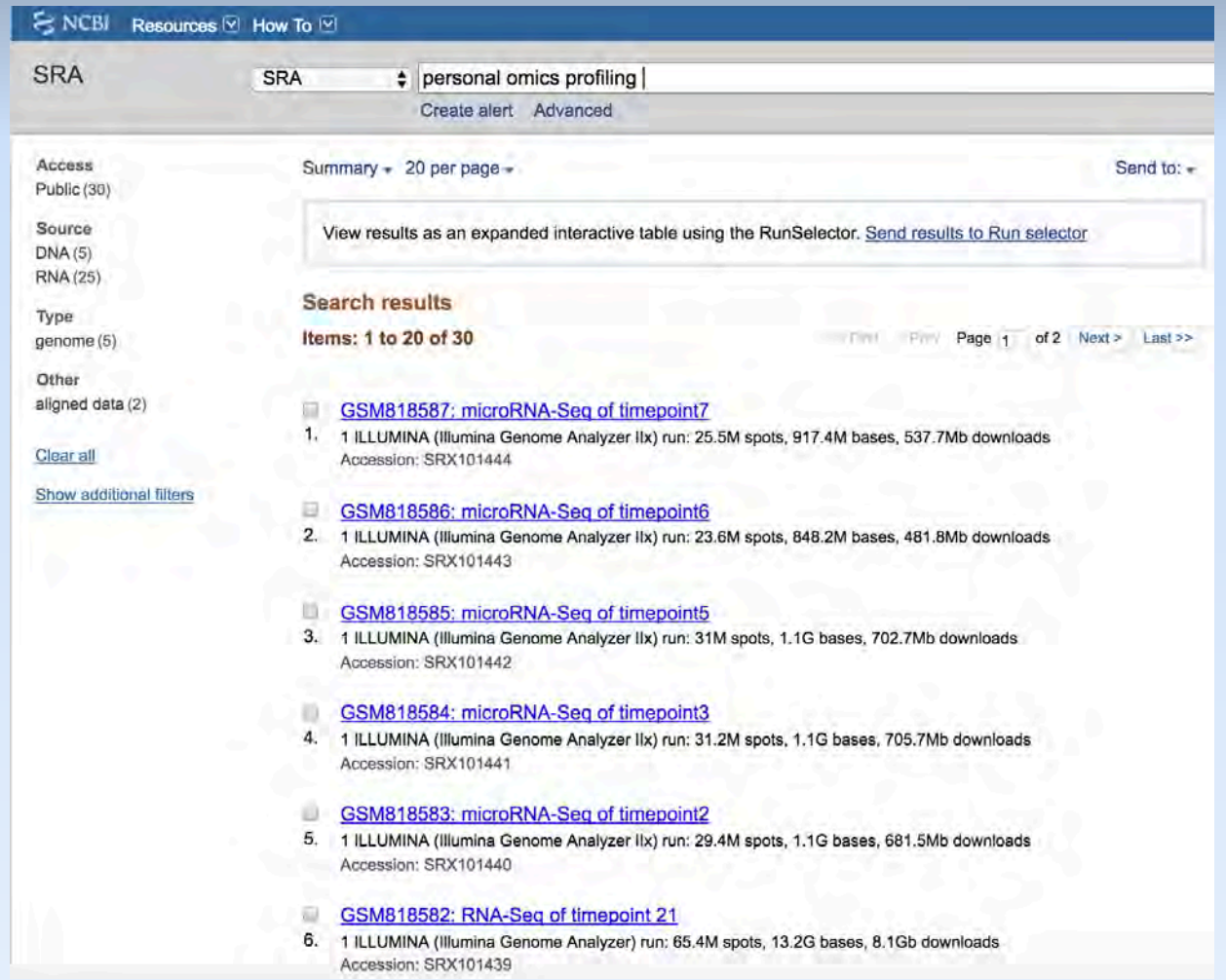
Abstract

Inflammatory bowel disease (IBD) is associated with risk variants in the human genome and dysbiosis of the gut microbiome, though unifying principles for these findings remain largely undescribed. The human commensal Bacteroides fragilis delivers immunomodulatory molecules to immune cells via secretion of outer membrane vesicles (OMVs). We reveal that OMVs require IBD-associated genes, ATG16L1 and NOD2, to activate a noncanonical autophagy pathway during protection from colitis. ATG16L1-deficient dendritic cells do not induce regulatory T cells (T(regs)) to suppress mucosal inflammation. Immune cells from human subjects with a major risk variant in ATG16L1 are defective in T(reg) responses to OMVs. We propose that polymorphisms in susceptibility genes promote disease through defects in "sensing" protective signals from the microbiome, defining a potentially critical gene-environment etiology for IBD.

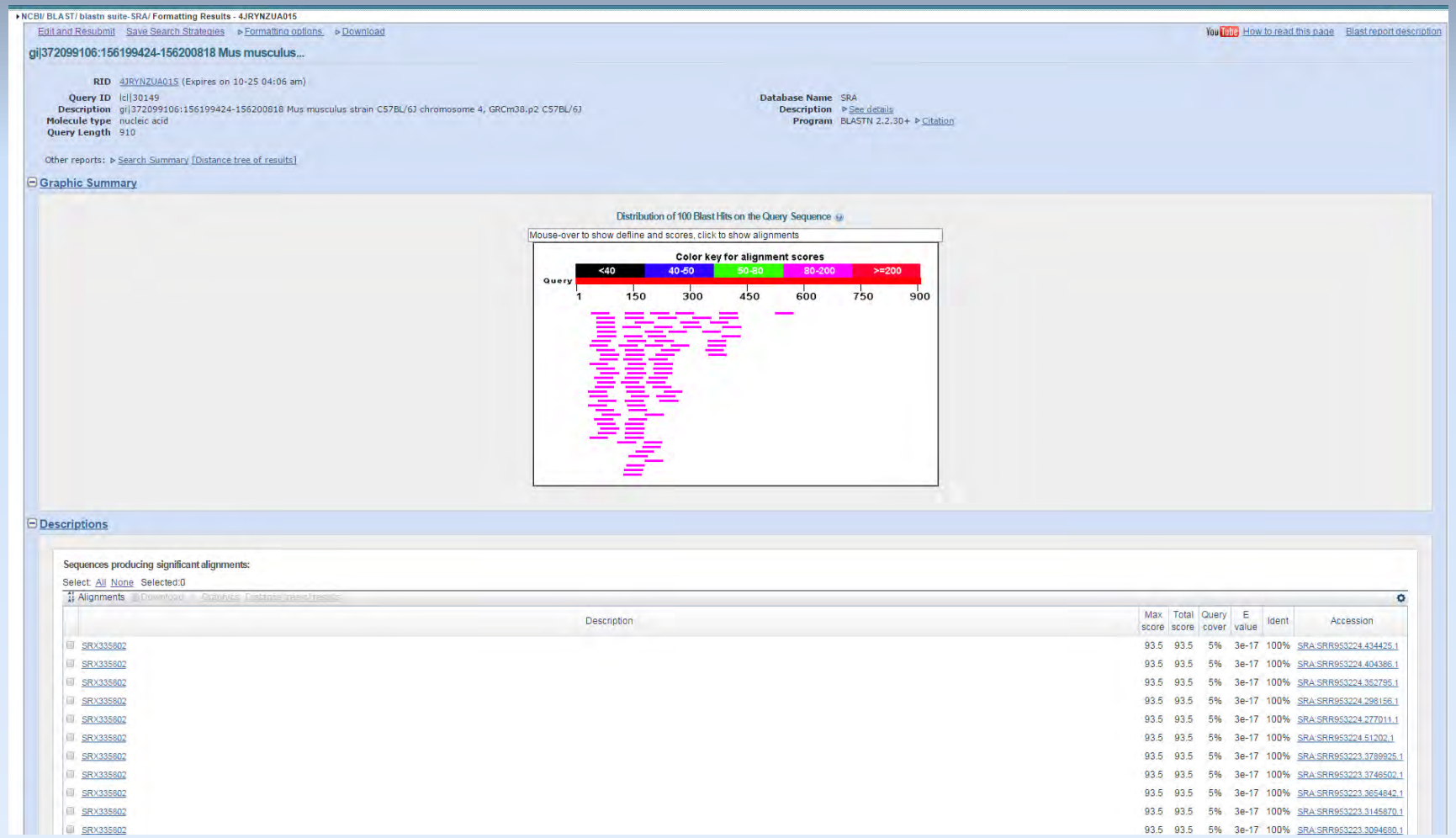
PMID: 27230380 PMCID: PMC4996125 [Available on 2017-05-27] DOI: 10.1126/science.aad9948

[PubMed - indexed for MEDLINE]

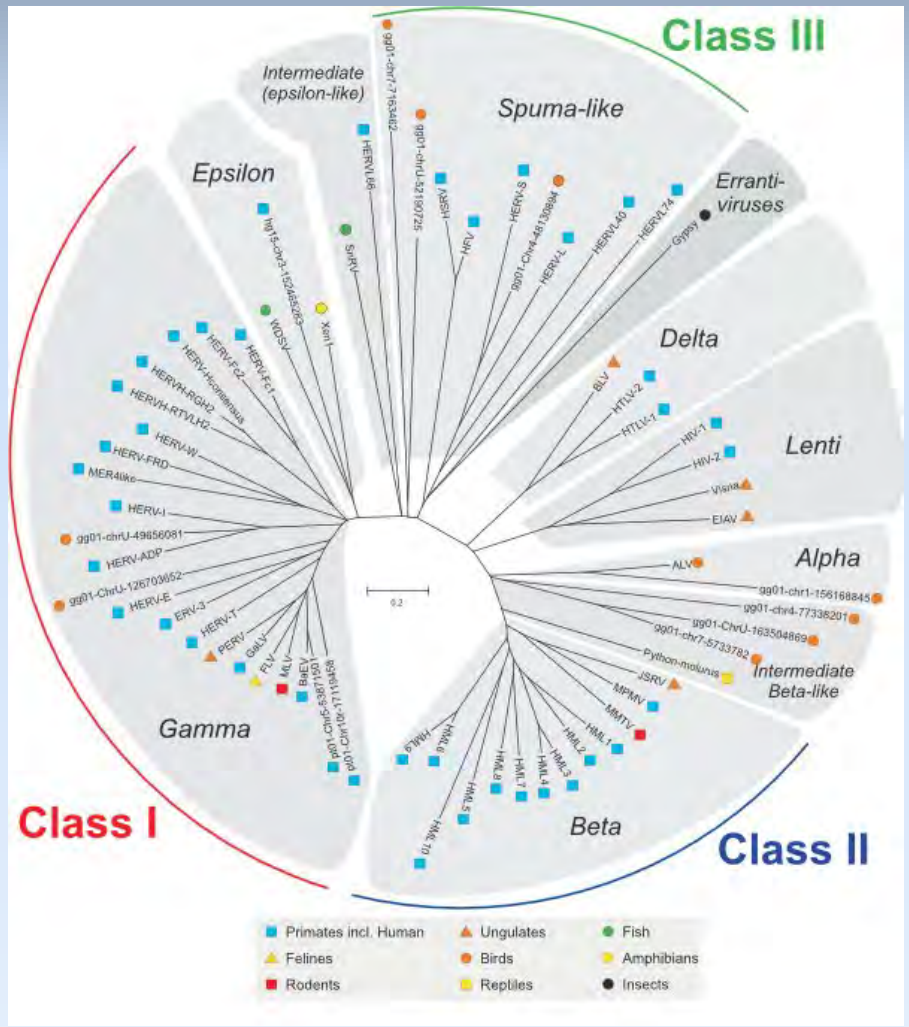
Where is the data? NCBI SRA (and dbGaP)



Blasting into SRA



Endogenous Retroviruses



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