What areas should NHGRI support to facilitate understanding of basic biological questions/processes using genomic assays of function?

How I interpret the question:

- What should NHGRI be doing for me (and clinician/scientists like me)?

- What does the draft epigenome mean to the rank and file physicians trying to engage in precision medicine?

- When a patient comes into the NIH CC, how can I use Encode/Roadmap to better understand dz?

- How can I use Encode/Roadmap in animal models to better understand dz pathogenesis in humans and identify/devise new therapies?

Ease of use of epigenomics data for rank and file clinician/scientists

www.ncbi.nlm.nih.gov/gene/?term=jak3

Gene

| Gene ⇕ | jak3 |

Save search   Advanced

Gene sources
Genomic

Categories
Alternatively spliced
Annotated genes
Protein-coding
Pseudogene

Sequence content
CCDS
Ensembl
RefSeq
RefSeqGene

Status   clear
✓ Current only

Chromosome locations

more...

Clear all

Show additional filters

Display Settings: ⌄ Tabular, 20 per page, Sorted by Relevance

Send to: ⌄

Filters: N

▼ Top
Homo
Mus m
Rattus
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More...

Find rel

Database

Find it

Search

jak3[A

Did you mean jak3 as a gene symbol?
Search Gene for jak3 as a symbol.

Results: 1 to 20 of 234

<< First   < Prev   Page 1   of 12   Next >   Last >>

ⓘ Filters activated: Current only. Clear all to show 241 items.

Name/Gene ID	Description	Location	Aliases	MIM
JAK3 ID: 3718	Janus kinase 3 [*Homo sapiens* (human)]	Chromosome 19, NC_000019.10 (17824782..17848071, complement)	JAK-3_HUMAN, JAKL, L-JAK, LJAK, JAK3	600173
Jak3 ID: 16453	Janus kinase 3 [*Mus musculus* (house mouse)]	Chromosome 8, NC_000074.6 (71676383..71690577)	fae, wil	
Jak3 ID: 25326	Janus kinase 3 [*Rattus norvegicus* (Norway rat)]	Chromosome 16, NC_005115.4 (20107468..20120678)	RATJAK3	
jak3 ID: 561370	Janus kinase 3 (a protein tyrosine kinase, leukocyte) [*Danio rerio* (zebrafish)]	Chromosome 8, NC_007119.6 (13347948..13375414)	sb:eu418	
JAK3 ID: 395845	Janus kinase 3 [*Gallus gallus* (chicken)]	NW_003769825.1 (40253..46881)	JAK	

jak3[A

jak3 [A

Gene expression on your iPhone
ImmGen app

www.ncbi.nlm.nih.gov/epigenomics/?term=jak3

NCBI   Resources ⊙ How To ⊙   Sign in to
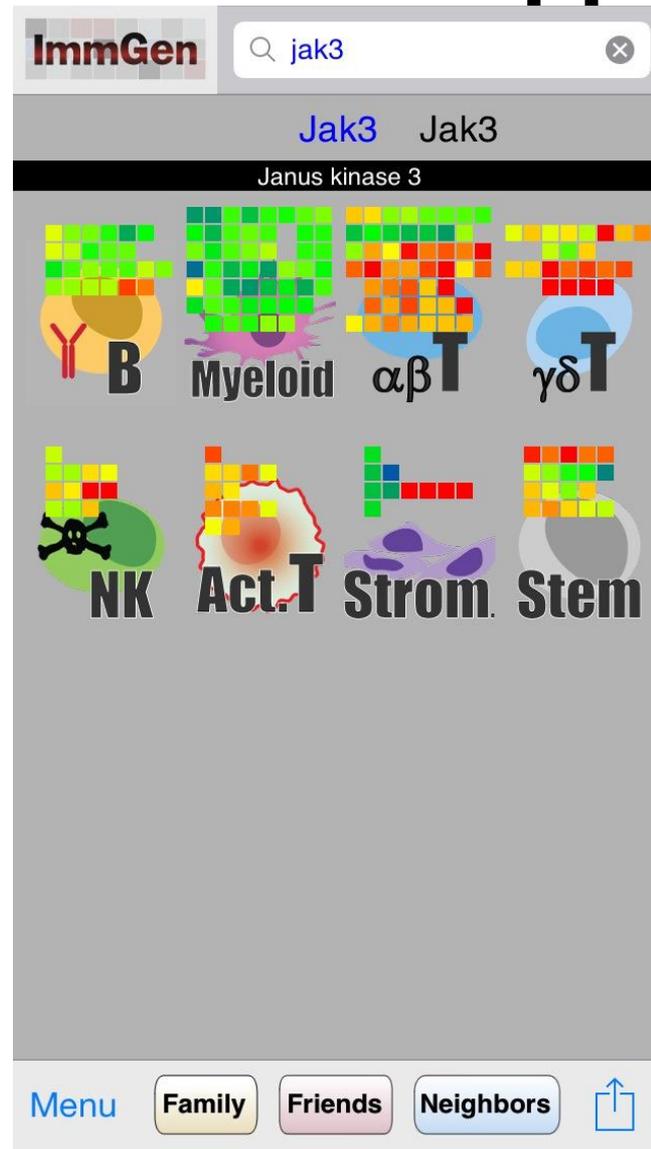
Epigenomics

| Epigenomics ⇕ | jak3 | ⊗ | **Search** |

Save search   Advanced

⚠ The following term was not found in Epigenomics: jak3.

ⓘ No items found.

Genome view for human JAK3
See genome view for: mouse Jak3
See Also: jak3 in the Gene database.
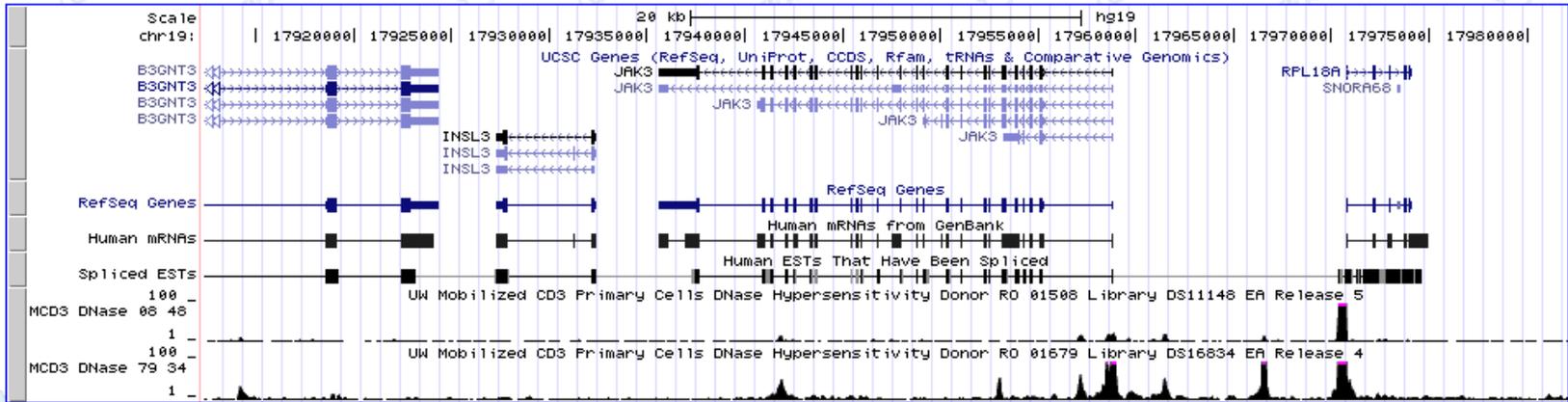
Search details

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(jak3[All Fields])
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Search   See m

Ease of use of epigenomics data for rank and file clinician/scientists

- Integration and visualization of gene expression, epigenomics, 3D chromatin changes (ideally on cell phone app)

- Optimal integration of cross-species epigenomes

- Most effective use will require more user-friendly, highly intuitive modes of accessing datasets

- Facilitate use of epigenomic therapies?

  – Empiric vs targeted use

  – Restoration of "epigenetic homeostasis" – e.g. lupus

- Whose problem? Maybe not NHGRI?

- NCBI/NLM? Dz-specific Institutes? All/joint effort?

What technological breakthroughs would really change how biological questions can be addressed?

- Identification of rare variants/mutations of regulatory elements
- Mendelian disorders of the regulome
- How will I be able to figure out when a patient has a pathological regulome mutation?
- Actionable?
- High-throughput assays for regulomopathies?

How many types of cells?

- 400 different types of cells in the body

How many types of cells?

- It depends….
- activation vs. differentiation
- Diverse modes of activation, receptors, signals
- Cell-cycle, metabolism, senescence
- Integration of new datasets with "Encode/Roadmap" data?

How do I get my data into "Encode". Quality control vs. crowd sourcing

- Long tailed data - diverse and heterogeneous small data sets produced by individual scientists
- Represent the vast majority of scientific data

How do I get my data into "Encode"? (Should I be able?)

- Level of coordination?
- Assurance that all appropriate data are included: source/preparation of cells, purity, cell stimulation, etc
- Better "history" of acquired cells (dz history, allergies, infectious disease, geography, smoking, etc)
- Quality control of archived regulomes?
- Lost opportunities vs Garbage in/Garbage out
- Ease of analysis/comparisons
- Development of tools/visualization for meta-analysis

What technological breakthroughs would really change how biological questions can be addressed?

- Epigenomics of single cells
- Dynamic views of the epigenome
  - signal transduction meets chromatin biology
  - What changes quickly, what doesn't
  - Rapid analysis/measurement in realtime
  - Longitudinal studies? Autoantibodies precede dz by many years
  - Aging/twin (again – example of lupus – 25% concordance)
- Better use of natural variation of *Mus musculus* Primates?
  - ENU mutagenesis
- Humanized mice to understand segments human regulomes
  - E.g. polymorphic regions associated with disease (e.g. *IL10*)
  - Moving from association to causality
- Other model organisms, optimal use of evolutionary lessons