



**Health
Hack**



WHAT PUBMED SAID

Challenge presented to HealthHack Sydney 2014 by Dr Fabian Buske, Quek Xiu Cheng and Kenneth Sabir, of the Garvan Institute.

THE PROBLEM

“As a biomedical researcher it is difficult for me to digest the overwhelming amount of information there is on just one specific gene, let alone a group of them. I want a better way to see what makes a given large set of genes special.”

**Dr Fabian Buske,
The Garvan Institute**



THE PROBLEM

“As a PhD student, it’s hard to keep up with my experiments and what is happening in the literature. If only there was a way to summarize the all literature about my protein of interest.”

Quek Xiu Chen
PhD Student



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Show

Results: 1 to 20 of 75

<< First < Prev Page 1 of 4 Next > Last >>

- ☐ [Clonal Dominance With Retroviral Vector Insertions Near the ANGPT1 and ANGPT2 Genes in a Human Xenotransplant Mouse Model.](#)

Haemmerle R, Phaltane R, Rothe M, Schröder S, Schambach A, Moritz T, Modlich U.
Mol Ther Nucleic Acids. 2014 Oct 7;3:e200. doi: 10.1038/mtna.2014.51.

PMID: 25291142 [PubMed]

[Related citations](#)

- ☐ [Amniotic fluid cells have haematopoietic potential and engraft after autologous in utero transplantation.](#)

van der Griend MP, Pipino C, Shangaris P, Maghsoudlou P, Ramachandra DL, Georgiades F, van der Griend AJ, Porada CD, Almeida-Porada G, Cheng PJ, David AL, De Coppi P.
Stem Cells. 2014 Sep 3. doi: 10.1002/stem.1839. [Epub ahead of print]
PubMed - as supplied by publisher

- ☐ [Human hematopoietic stem cell maintenance is critical for acute myeloid leukemia.](#)

van der Griend MP, Bakker B, Rozenveld-Geugien M, Jaques J, de Haan G, Schuringa JJ, van der Griend H.
Stem Cells. 2014 Sep 3. doi: 10.1038/leu.2014.259. [Epub ahead of print]
PubMed - as supplied by publisher

- ☐ [Treatment of One Graft in a Double Cord Blood Transplant Provides Early Platelet Engraftment Contributing to Long-Term Engraftment in NSG Mice.](#)

van der Griend MP, van Hensbergen Y, Brand A, Slot MC, de Graaf-Dijkstra A, Mulder A, Watt SM, Zwaginga JJ.
Stem Cells Dev. 2014 Sep 29. [Epub ahead of print]

PMID: 25137252 [PubMed - as supplied by publisher]
[Related citations](#)

- ☐ [Ex vivo assays to study self-renewal, long-term expansion, and leukemic transformation of genetically modified human hematopoietic and patient-derived leukemic stem cells.](#)

Sontakke P, Carretta M, Capala M, Schepers H, Schuringa JJ.
Methods Mol Biol. 2014;1185:105-119. doi: 10.1007/978-1-4939-1188-9_10.

New feature

Try the new Display Settings option -
Sort by Relevance

Related searches[cd34 nsg mice](#)**Titles with your search terms**

Human **CD34+** CD133+ hematopoietic stem cells cultured with growth factors inc [PLoS One. 2011]

Human T cell development in the liver of humanized NOD/SCID/IL-2F [Clin Immunol. 2011]

Ex vivo rapamycin treatment of human cord blood **CD34+** cells en [Blood Cells Mol Dis. 2011]

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Sustained Engraftment of Cryopreserved Human Bone Marrow **CD34+** [Biores Open Access. 2014]

Analysis of the clonal growth and differentiation dynamics of primitive barcoded hurr [Blood. 2013]

Fucosyltransferase VII improves the function of selectin ligands on cord bloo [Glycobiology. 2013]

[See all \(35\)...](#)

Got a paper in
PubMed?



- The Garvan Institute has processed more than 2 million full-text scientific articles on biological and medical sciences from Pubmed Central
- Extracted meaningful keywords and associated these with categories such as “disease”. The result is a DB of 6 billion data points
- Each human protein-coding gene is now mapped to every PubMed article it appears in along with every term found in these papers

The challenge is to create a tool that allows us to query the DB categories via:

- Visual presentation layer to convey the returned information and illustrate the connectivity between the discoveries
- Ranked recommendations via collaborative filtering or consensus via aggregations
- Link back to the original publications to enable further in-depth studies.



THE TEAM

L-R: Quek Xiu Cheng (PhD Research Student), Andy Zeng (Data Scientist), Chunxiao Kin (Software Engineer), Paul Joseph (Software Developer), Svetlana Filimonova (Software Developer), Dr Fabian Buske (Garvan Institute), Amanda Wise (UX)

THE DATA

Data is in human unreadable objects.

The team needed to find the description for each term in the DB and associate this back to the data. We added statistical enrichment analysis of terms on some groups over others.

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{
  "ids": [
    "ENSP00000285930",
    "ENSP00000312606"
  ],
  "ensg_id": [
    "ENSG00000085662",
    "ENSG00000117448"
  ],
  "Disease": {
    "DOID:0014667": 11,
    "DOID:0050013": 11,
    "DOID:0060158": 11,
    "DOID:4": 60,
    "DOID:4194": 11,
    "DOID:9351": 9,
    "DOID:4195": 2,
    "DOID:0050155": 1,
    "DOID:1432": 1,
    "DOID:1492": 1,
    "DOID:7": 6,
```


CHOOSE A SET OF TERMS TO WORK WITH

✓ → DISEASE → ONTOLOGY

→ ENS

→ COG

→ KEGG

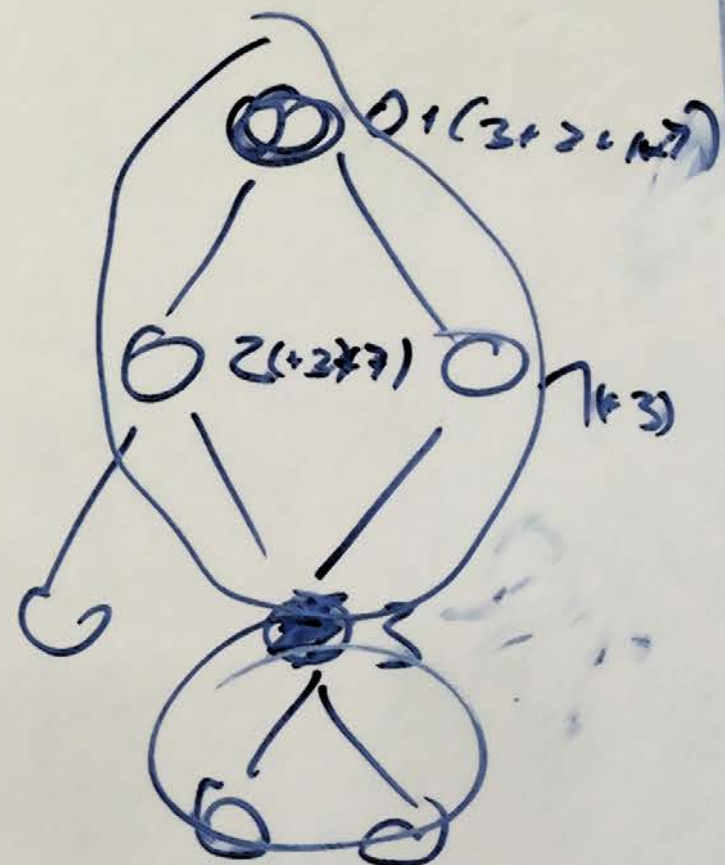
→ NOG

→ eu NOG

, → PubChem →

✓ → GO → ONTOLOGY

✓ → WIKI (NO MAPPING
FEED)



The challenge is to create a tool that allows us to query the DB categories via:

- Visual presentation layer to convey the returned information and illustrate the connectivity between the discoveries
- Ranked recommendations via collaborative filtering or consensus via aggregations
- Link back to the original publications to enable further in-depth studies.

SKETCHBOARDS

ref. VRh...

Ranking
list of
frequency

default search
manually
annotated
or full article
search

cd34 nsg
search terms

category analysis

Full

° P factor

Visualise

latest?

Score of
Confidence
"p"

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general/un-
wanted terms

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Article title

terms
definition

ID gene "term"

① what is
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or
under represents
② what is
common
amongst
search terms

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of
terms

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Elevator pitch

A visual search tool
for biomedical researcher
that enables search
of PubMed & rank
articles by specific
biomedical terms, fa

PUBMEDSAID

| ABOUT | EXAMPLE | CRED |
|---|--|--|
| A visual search tool that searches and ranks PubMed articles by specific biomedical terms. Etc. | See what you can do with PubMedSAID | Created by the Centre for Healthcare |

#354358

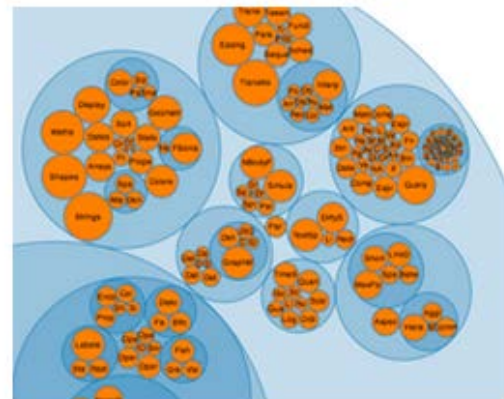
#298844

DISCUSSED WHICH SORT OF VISUALISATION WOULD COMMUNICATE BEST

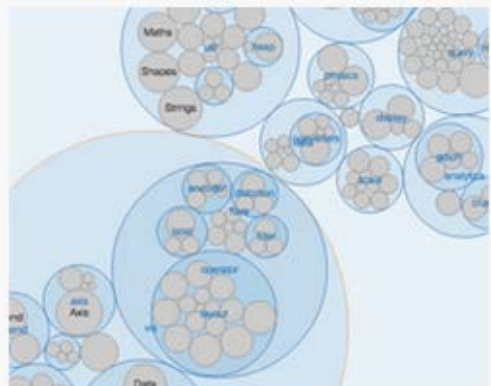
Bubble Chart



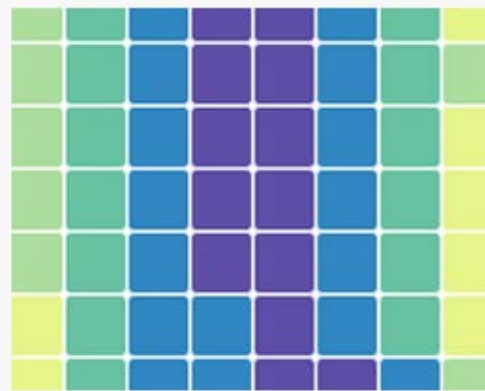
Circle Packing



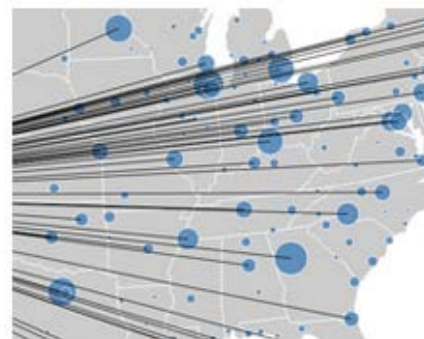
Layout



Trulia Trends



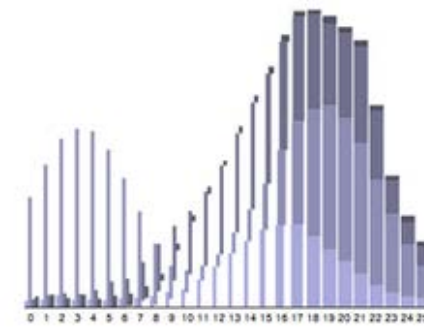
Symbol Map



Site or blog
concept browser



Stacked Bars



Gene Expression



Zoomable Treemap

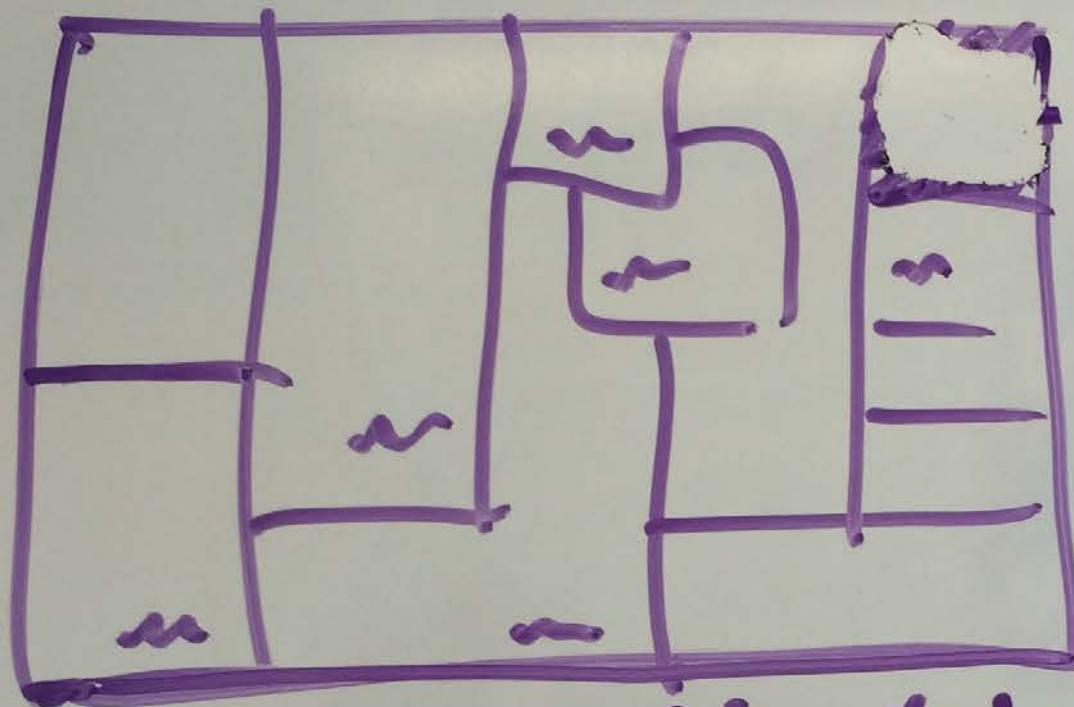


Zoomable Sunburst



USER INTERFACE

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- Terms proportionate to frequency in PubMed articles

Title....
Year

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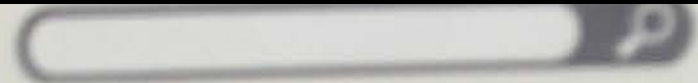
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Title
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WE DECIDED ON A ZOOMABLE TREEMAP



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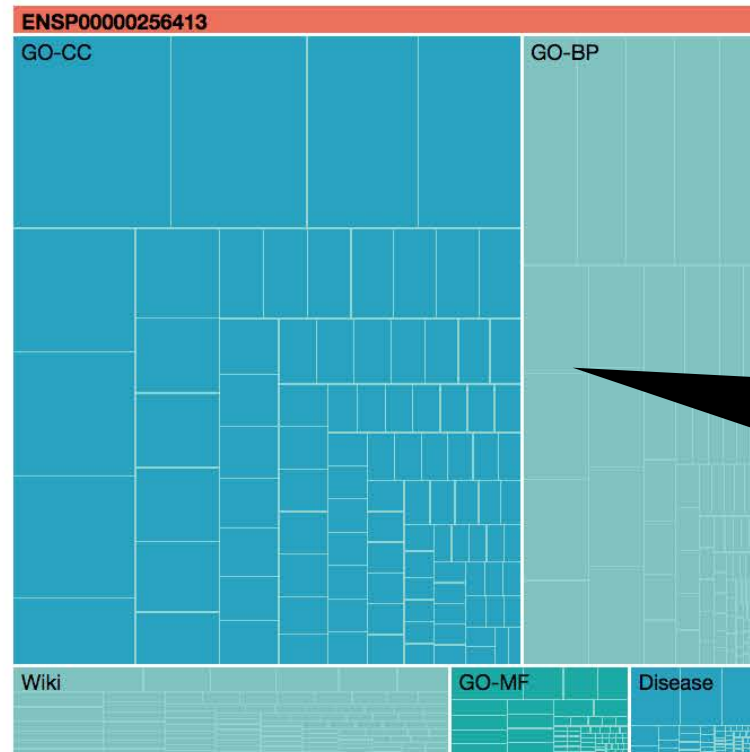
111 112 113 114 115

THE PROTOTYPE: <http://svetixbot.github.io/pubmedsaid>

- [ENSP00000256413](#)
- [ENSP00000308318](#)
- [ENSP00000362705](#)
- [ENSP00000371690](#)
- [ENSP00000458989](#)

PubMedSaid

Prototype presented by Dr Fabian Buske, Quek Xiu Cheng and Kenneth Sabir



Database
sources –
click to show
frequency of
terms within
that DB

THE PROTOTYPE: <http://svetixbot.github.io/pubmedsaid>

- ENSP00000256413
- ENSP00000308318
- ENSP00000362705
- ENSP00000371690
- ENSP00000458989

Data sets

PubMedSaid

Prototype presented by Dr Fabian Buske, Quek Xiu Cheng and Kenneth Sabir

Term and definition

Ranked frequency of terms

Click block to zoom into more references

Articles ranked by date

cellular macromolecule biosynthetic process

cellular process

GO:0009987 "Any process that is carried out at the cellular level, but not necessarily restricted to a single cell. For example, cell communication occurs among more than one cell, but occurs at the cellular level." [GOC:go_curators, GOC:isa_complete]

Found 357 articles

- **FGFR1 mutations cause Hartsfield syndrome: the unique association of holoprosencephaly and ectrodactyly.**

Author: Simonis N,Migeotte I,Lambert N,Perazzolo C,de Silva DC,Dimitrov B,Heinrichs C,Janssens S,Kerr B,Mortier G,Van Vliet G,Lepage P,Casimir G,Abramowicz M,Smits G,Vilain C

Journal: Journal of medical genetics

Published: 2013 08

- **Tespa1 is a novel inositol 1,4,5-trisphosphate receptor binding protein in T and B lymphocytes.**

Author: Matsuzaki H,Fujimoto T,Ota T,Ogawa M,Tsunoda T,Doi K,Hamabashiri M,Tanaka M,Shirasawa S

Journal: FEBS open bio

Published: 2013 05

- **Mutations in FGF17, IL17RD, DUSP6, SPRY4, and FLRT3 are identified in individuals with congenital hypogonadotropic hypogonadism.**

Author: Miraoui H,Dwyer AA,Sykitis GP,Plummer L,Chung W,Feng B,Beenken A,Clarke J,Pers TH,Dworzynski P,Keefe K,Niedziela M,Raivio T,Crowley WF,Seminara SB,Quinton R,Hughes VA,Kumanov P,Young J,Yialamas MA,Hall JE,Van Vliet G,Chanoine JP,Rubenstein J,Mohammadi M,Tsai PS,Sidis Y,Lage K,Pitteloud N

Journal: American journal of human genetics

Published: 2013 05

- **Defining regulatory and phosphoinositide-**

Article abstract

Congenital hypogonadotropic hypogonadism (CHH) and its anosmia-associated form (Kallmann syndrome [KS]) are genetically heterogeneous. Among the >15 genes implicated in these conditions, mutations in FGF8 and FGFR1 account for ~12% of cases; notably, KAL1 and HS6ST1 are also involved in FGFR1 signaling and can be involved in CHH. We therefore hypothesized that mutations in genes encoding a broader set of the FGFR1 pathway might contribute to the genetics of CHH as well as mutations. Thus, we aimed to (1) investigate whether CHH individuals have mutations in members of the so-called "FGF8 synexpression" group and (2) develop a bioinformatics algorithm on the basis of protein-protein interaction data from the interactome-based affiliation scoring (IBAS) to identify high-quality candidates on the basis of sequence homology, expression, and structural and functional data. Genes were selected and sequenced in 386 unrelated CHH individuals and 155 controls. Except for FGF18 and SPRY2, all other genes were found to be mutated in CHH individuals: FGF17 (n = 3 individuals), IL17RD (n = 8), DUSP6 (n = 5), SPRY4 (n = 1), and FLRT3 (n = 3). Independently, IBAS predicted FGF17 and IL17RD as the two top candidates in the entire proteome on the basis of a statistical test of their protein-protein interaction patterns to proteins known to be altered in CHH. Most of the FGF17 and IL17RD mutations altered protein function in vitro. IL17RD mutations were found only in KS individuals and were strongly linked to hearing loss (6/8 individuals). Mutations in genes encoding components of the FGF pathway are associated with complex modes of CHH inheritance and act primarily as contributors to an oligogenic genetic architecture underlying CHH.

NEXT STEPS: UI ENHANCEMENTS

- ENSP00000256413
- ENSP00000308318
- ENSP00000362705
- ENSP00000371690
- ENSP00000458989

SEARCH FIELD

PubMedSaid

presented by Dr Fabian Buske, Quek Xiu Cheng and Kenneth Sabir

Show term frequency in colour as well as size



Congenital hypogonadotropic hypogonadism (CHH) and Kallmann syndrome (KS) are genetically heterogeneous conditions. In these conditions, mutations in FGF8 and FGF17 are implicated; notably, KAL1 and HS6ST1 are also involved in CHH. We therefore hypothesized that mutations in a range of modulators of the FGFR1 pathway might contribute to causal or modifier mutations. Thus, we aimed to (1) investigate harbor mutations in members of the so-called "FGF8 syndrome" and (2) validate the ability of a bioinformatics algorithm on the basis of interactome data (interactome-based affiliation scoring [IBAS]) to predict candidate genes. On the basis of sequence homology, expression data, and functional data, seven genes were selected and sequenced in 155 CHH individuals and 155 controls. Except for FGF18 and SPRY4, no mutations were found to be mutated in CHH individuals: FGF17 (n = 3 individuals), IL17RD (n = 8), DUSP6 (n = 5), SPRY4 (n = 14), and FLRT3 (n = 3). Independently, IBAS predicted FGF17 and IL17RD as the two top candidates in the entire proteome on the basis of a statistical test of their protein-protein interaction patterns to proteins known to be altered in CHH. Most of the FGF17 and IL17RD mutations altered protein function in vitro. IL17RD mutations were found only in KS individuals and were strongly linked to hearing loss (6/8 individuals). Mutations in genes encoding components of the FGF pathway are associated with complex modes of CHH inheritance and act primarily as contributors to an oligogenic genetic architecture underlying CHH.

Term and definition on roll-over of each block

cellular process

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Author: Simonis N, Migeotte I, Lambert N, Perazzolo C, de Silva DC, Dimitrov B, Heinrichs C, Janssens S, Kerr B, Mortier G, Van Vliet G, Lepage P, Castaigne G, Abramowicz M, Smits G, Vilain C

Journal: Journal of medical genetics

Published: 2013 08

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cells.

Author: H, Fujimoto T, Ota T, Ogawa M, Tsunoda T, Doi K, Hamabashiri S, Sawada S

Journal: J Biol Chem

2013

in FGF17, IL17RD, DUSP6, SPRY4, and FLRT3 are identified in individuals with hypogonadotropic hypogonadism.

Author: Miraoui H, Dwyer AA, Sykiotis GP, Plummer L, Chung W, Feng B, Beenken A, Clarke J, Pers TH, Dworzynski P, Keefe K, Niedziela M, Raivio T, Crowley WF, Seminara SB, Quinton R, Hughes VA, Kumanov P, Young J, Yialamas MA, Hall JE, Van Vliet G, Chanoine JP, Rubenstein J, Mohammadi M, Tsai PS, Sidis Y, Lage K, Pitteloud N

Journal: American journal of human genetics

Published: 2013 05

- **Defining regulatory and phosphoinositide-**

Layout of Article summary

“Not being able to go after the answer at the opportune moment an idea strikes can stop new discoveries in its tracks.

Thus, a framework that can reveal the connections between distinct scientific fields or discoveries on the spot has the potential to facilitate future Eureka moments.”