

# Citizen Science: Empowering the Public to Help Solve Biomedical Challenges

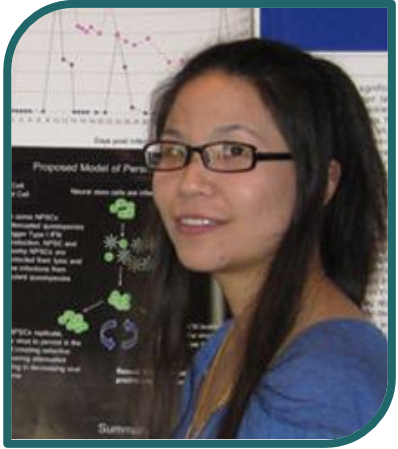
Andrew Su, Ph.D.

@andrewsu 

<http://sulab.org>

September 10, 2020

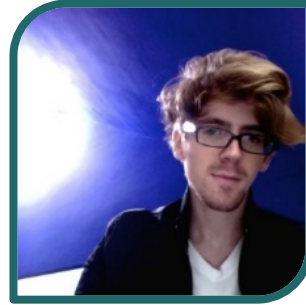
# Acknowledgements



Ginger Tsueng



Benjamin Good



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Kevin Xin  
Colleen Xu  
Jerry Zhou



National Institute of  
General Medical Sciences



National Center  
for Advancing  
Translational Sciences




National Institute  
on Aging



National Institute of  
Allergy and  
Infectious Diseases

# A quick poll

 Polls

—

□

×

Polling 1:

1. Is doing biomedical research part of your day job?

☐ Yes

☐ No







# GALAXY ZOO.org

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## Galaxy Analysis

Welcome to Galaxy Zoo's view of the Universe. If you're here you should already have seen the [Tutorial](#), but feel free to go and remind yourself. There's no need to agonise for too long over any one image, just make your best guess in each case.



☐ Show Grid Overlay on the next Image

Galaxy Ref:  
**587729387677679742**

Choose the Galaxy Profile  
by clicking the buttons  
below



# GALAXY ZOO.org

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## Galaxy Analysis

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"...within **24 hours** of launch we were stunned to be receiving almost **70,000 classifications an hour**. In the end, more than **50 million classifications** were received by the project during its first year, contributed by more than **150,000 people**."

☐ Show Grid Overlay on the next Image

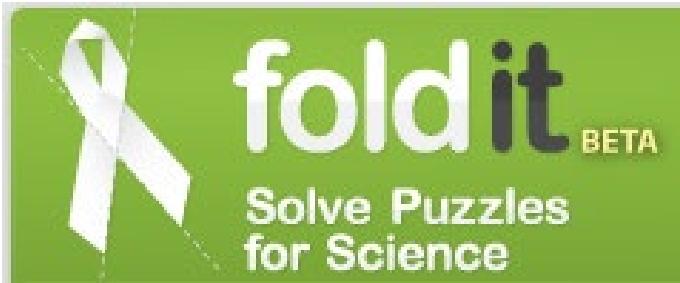
Galaxy Ref:

**587729387677679742**

Choose the Galaxy Profile by clicking the buttons below







Rank: 34

Score: 9455.364

Soloist

223: Core and Tail Design 2

► No conditions

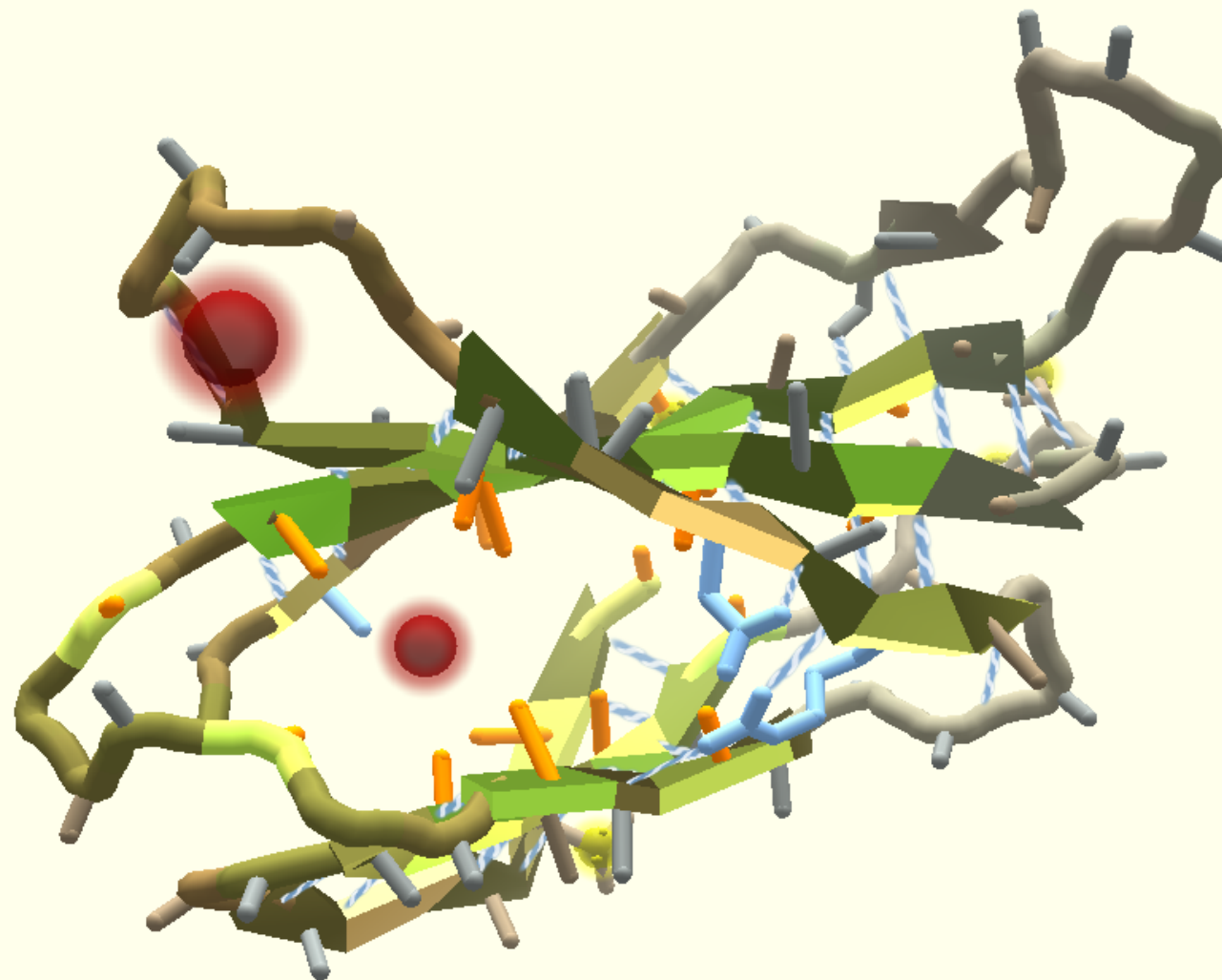
▼ Group Competition

#	Group Name	Score
1	Contenders	9628
2	Richard Dawkins Foundation	9627
3	GoFOLDers	9613
4	Natural Abilities	9611
5	Another Hour Another Point	9596
6	Czech National Team	9590
7	Void Crushers	9584

▼ Soloist Competition

#	Player Name	Current	Best
1	BootsMcGraw	-	9628
2	vertex	-	9627
3	themarquis	-	9625
4	Mark-	-	9624
5	infjamc	-	9613
6	Mr_Jolty	-	9611
7	kevpad5	-	9604

▼ Cookbook



- Chat - Group ⓘ ✕ auto show
- Chat - Puzzle ⓘ ✕ auto show

shpalmina: and so?

BletchleyPark: and left me with an unanswered question

Natanaell: what's up?

# Ribosome Challenge 1.5

AT MOST 30 CHANGES  
0/30 ✓

0 violations ✓



Total  
-111.5 kcal

Natural/Target Delta  
17.3 kcal

Explosion Factor

1 - +



Navigation and status bar containing icons for menu, lightning bolt, leaf, target, and a sequence of colored beads (A, U, G, C) with counts 24, 22, 58. It also includes zoom in (+), zoom out (-), and navigation arrows.

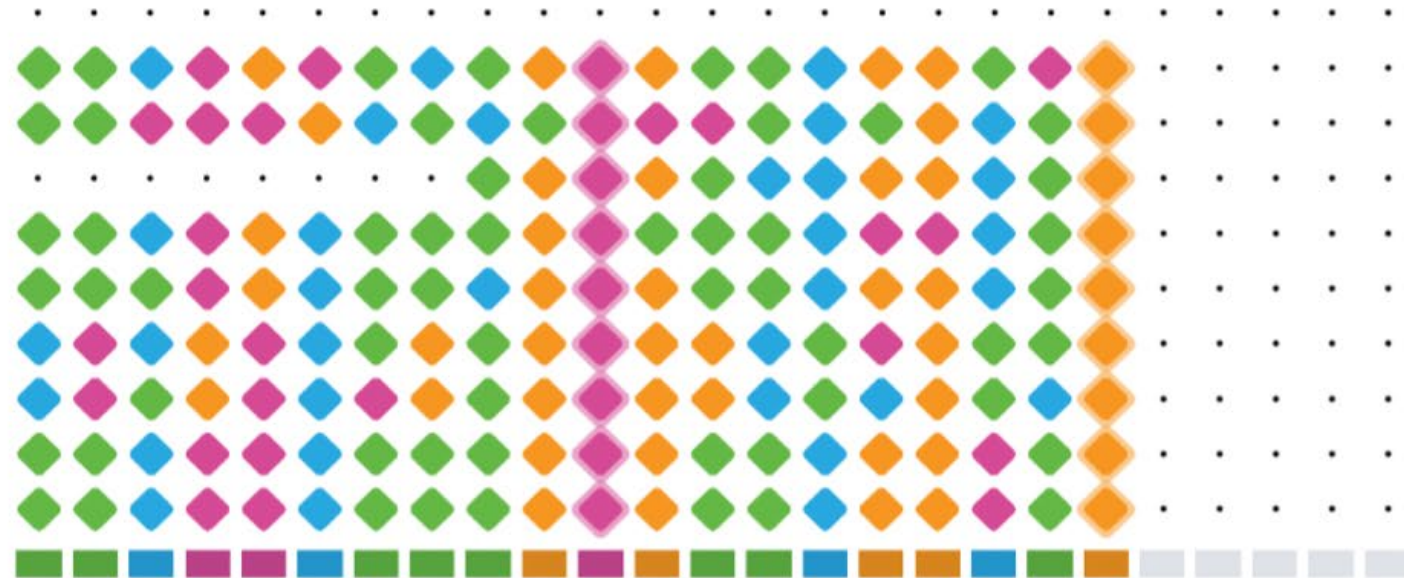
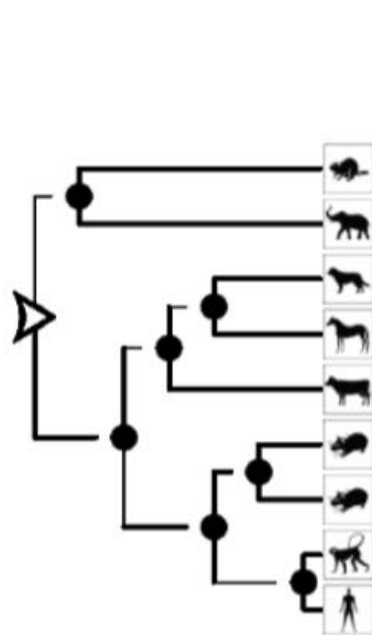
Credit: The Eterna Project



Goal 202

Score  
202

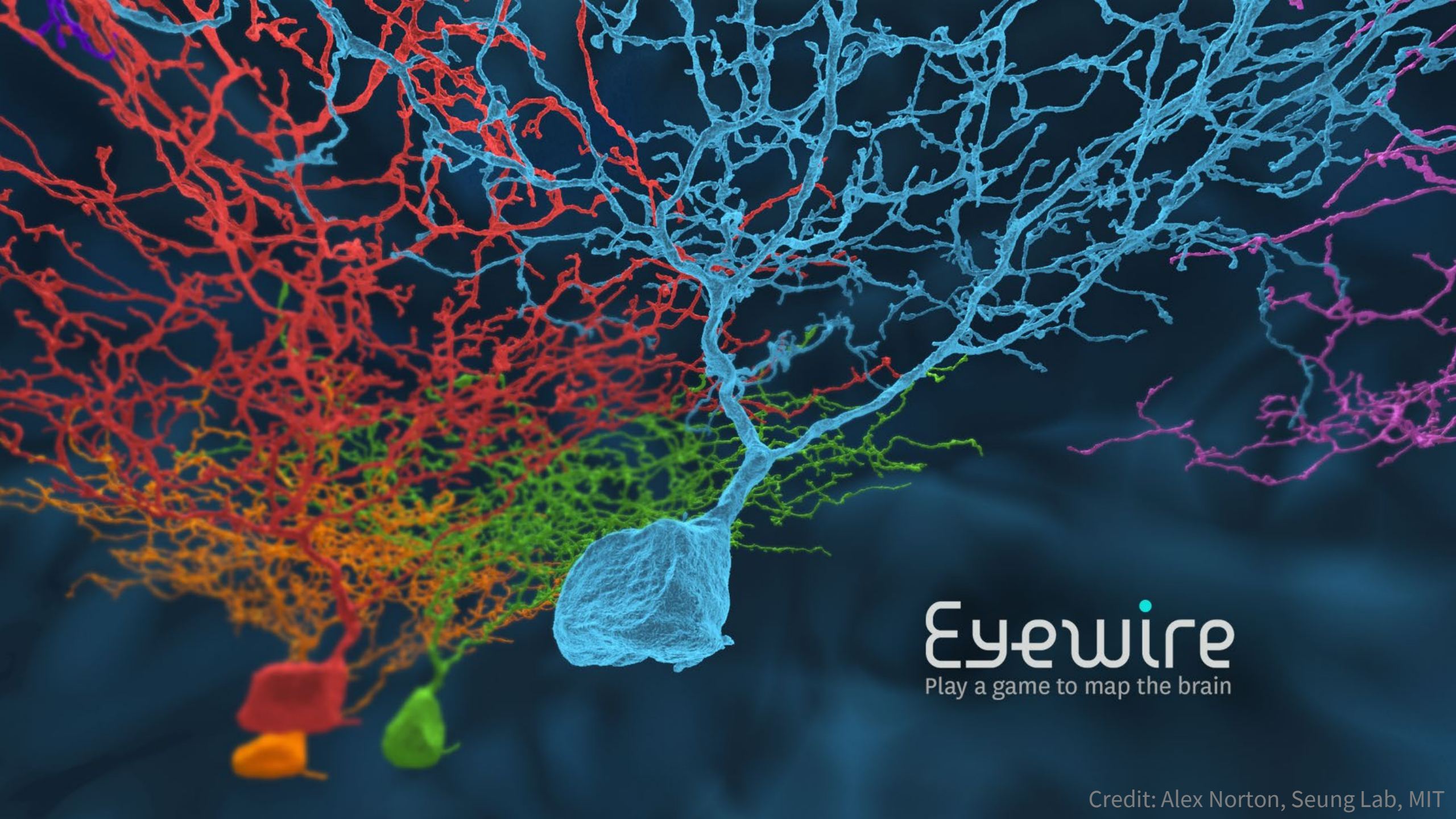
232 Top Score



NEXT STAGE







**Eyewire**  
Play a game to map the brain

Credit: Alex Norton, Seung Lab, MIT



# CITIZEN SCIENCE





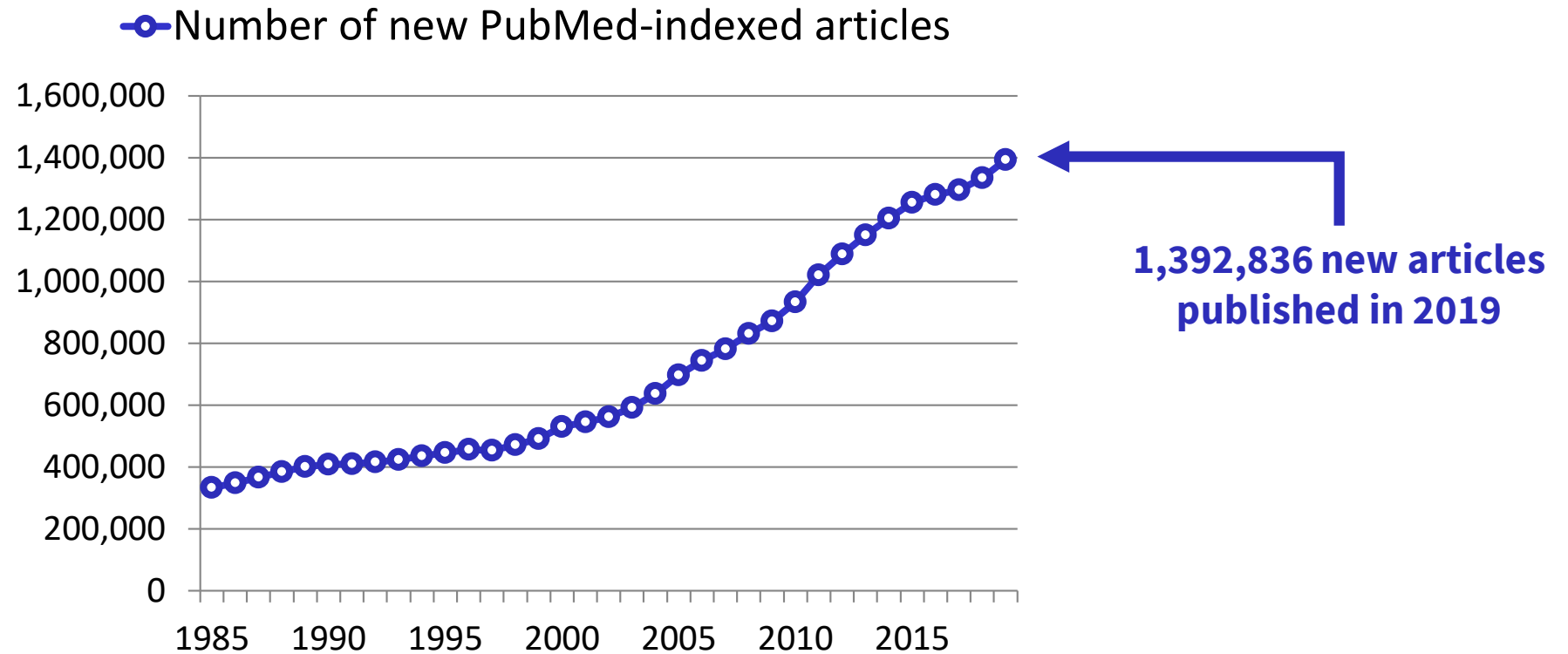








# The biomedical literature is massive



**Over 30 million articles total in PubMed**

# Citizen Science Demo

- Participate only if your day job does *NOT* include biomedical research
- We will read a bit of text together, and a zoom poll will ask you a series of questions
- Answer based only on what the text says, not what is “true”
- Don’t worry too much about being sure – make your best guess within the allotted time

# Familial systemic mastocytosis with germline KIT K509I mutation is sensitive to treatment with imatinib, dasatinib and PKC412



Paula de Melo Campos<sup>a</sup>, João A. Machado-Neto<sup>a</sup>, Renata Scopim-Ribeiro<sup>a</sup>, Valeria Visconte<sup>b</sup>, Ali Tabarroki<sup>b</sup>, Adriana S.S. Duarte<sup>a</sup>, Flávia F.C. Barra<sup>c</sup>, José Vassalo<sup>c</sup>, Heesun J. Rogers<sup>d</sup>, Irene Lorand-Metze<sup>a</sup>, Ramon V. Tiu<sup>b</sup>, Fernando F. Costa<sup>a</sup>, Sara T. Olalla Saad<sup>a</sup>, Fabiola Traina<sup>a,e,\*</sup>

<sup>a</sup> Hematology and Hemotherapy Center, University of Campinas/Hemocentro – Unicamp, Instituto Nacional de Ciência e Tecnologia do Sangue, Campinas, São Paulo, Brazil

<sup>b</sup> Department of Translational Hematology and Oncology Research, Taussig Cancer Institute, Cleveland Clinic, Cleveland, OH, USA

<sup>c</sup> Laboratory of Investigative and Molecular Pathology, CIPED, School of Medical Sciences, University of Campinas, Campinas, São Paulo, Brazil

<sup>d</sup> Department of Clinical Pathology, Cleveland Clinic, Cleveland, OH, USA

<sup>e</sup> Department of Internal Medicine, University of São Paulo at Ribeirão Preto Medical School, Ribeirão Preto, São Paulo, Brazil

## ARTICLE INFO

### Article history:

Received 7 February 2014

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### Keywords:

Familial mastocytosis

K509I KIT mutation

Tyrosine kinase inhibitors

Imatinib

Dasatinib

PKC412

## ABSTRACT

Mastocytosis are myeloproliferative neoplasms commonly related to gain-of-function mutations involving the tyrosine kinase domain of *KIT*. We herein report a case of familial systemic mastocytosis with the rare *KIT* K509I germ line mutation affecting two family members: mother and daughter. *In vitro* treatment with imatinib, dasatinib and PKC412 reduced cell viability of primary mast cells harboring *KIT* K509I mutation. However, imatinib was more effective in inducing apoptosis of neoplastic mast cells. Both patients with familial systemic mastocytosis had remarkable hematological and skin improvement after three months of imatinib treatment, suggesting that it may be an effective front line therapy for patients harboring *KIT* K509I mutation.

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<sup>b</sup> Department of Translational Hematology and Oncology Research, Taussig Cancer Institute, Cleveland Clinic, Cleveland, OH, USA

<sup>c</sup> Laboratory of Investigative and Molecular Pathology, CIPED, School of Medical Sciences, University of Campinas, Campinas, São Paulo, Brazil

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# Demo #1: Identifying the entities

... We report a case of familial systemic mastocytosis with the rare KIT K509I germ line mutation. In vitro treatment with imatinib, dasatinib and PKC412 reduced cell viability of primary mast cells harboring KIT K509I mutation. Both patients with familial systemic mastocytosis had remarkable hematological and skin improvement after three months of imatinib treatment.

# Demo #1: Identifying the entities

... We report a case of **familial systemic mastocytosis** with the rare KIT K509I germ line mutation. In vitro treatment with imatinib, dasatinib and PKC412 reduced cell viability of primary mast cells harboring KIT K509I mutation. Both patients with **familial systemic mastocytosis** had remarkable hematological and skin improvement after three months of imatinib treatment.

**familial systemic  
mastocytosis**



# Demo #1: Identifying the entities

... We report a case of **familial systemic mastocytosis** with the rare **KIT** K509I germ line mutation. In vitro treatment with imatinib, dasatinib and PKC412 reduced cell viability of primary mast cells harboring **KIT** K509I mutation. Both patients with **familial systemic mastocytosis** had remarkable hematological and skin improvement after three months of imatinib treatment.

**familial systemic  
mastocytosis**

**KIT**

# Demo #1: Identifying the entities

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**familial systemic  
mastocytosis**

**KIT**

**Imatinib**

# Demo #1: Instructions

For each of these  
entities ...



**familial systemic  
mastocytosis**

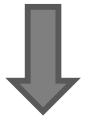
**KIT**

**Imatinib**



# Demo #1: Instructions

For each of these  
entities ...



**familial systemic  
mastocytosis**

**KIT**

**Imatinib**

... select the  
entity type ...



**GENE / PROTEIN**

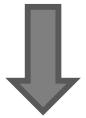
**DRUG / CHEMICAL**

**DISEASE**

**OTHER**

# Demo #1: Instructions

For each of these  
entities ...



**familial systemic  
mastocytosis**

**KIT**

**Imatinib**

... select the  
entity type ...



**GENE / PROTEIN**

**DRUG / CHEMICAL**

**DISEASE**

**OTHER**

... using this poll



Polls

Polling 1: Entities

1. "Familial systemic mastocytosis" is a

☐ Gene / Protein

☐ Drug / Chemical

☐ Disease

☐ Other

2. "KIT" is a

☐ Gene / Protein

☐ Drug / Chemical

☐ Disease

☐ Other

# Demo #1: Identifying the entities

... We report a case of **familial systemic mastocytosis** with the rare **KIT** K509I germ line mutation. In vitro treatment with **imatinib**, dasatinib and PKC412 reduced cell viability of primary mast cells harboring **KIT** K509I mutation. Both patients with **familial systemic mastocytosis** had remarkable hematological and skin improvement after three months of **imatinib** treatment.

**familial systemic  
mastocytosis**

**KIT**

**Imatinib**

**GENE / PROTEIN**

**DRUG / CHEMICAL**

**DISEASE**

**OTHER**



# Demo #1: --- 30 seconds, GO! ---

... We report a case of **familial systemic mastocytosis** with the rare **KIT** K509I germ line mutation. In vitro treatment with **imatinib**, dasatinib and PKC412 reduced cell viability of primary mast cells harboring **KIT** K509I mutation. Both patients with **familial systemic mastocytosis** had remarkable hematological and skin improvement after three months of **imatinib** treatment.

**familial systemic  
mastocytosis**

**KIT**

**Imatinib**

**GENE / PROTEIN**

**DRUG / CHEMICAL**

**DISEASE**

**OTHER**

# Demo #1: Results

**familial systemic  
mastocytosis**

is a

**DISEASE**

**KIT**

is a

**GENE / PROTEIN**

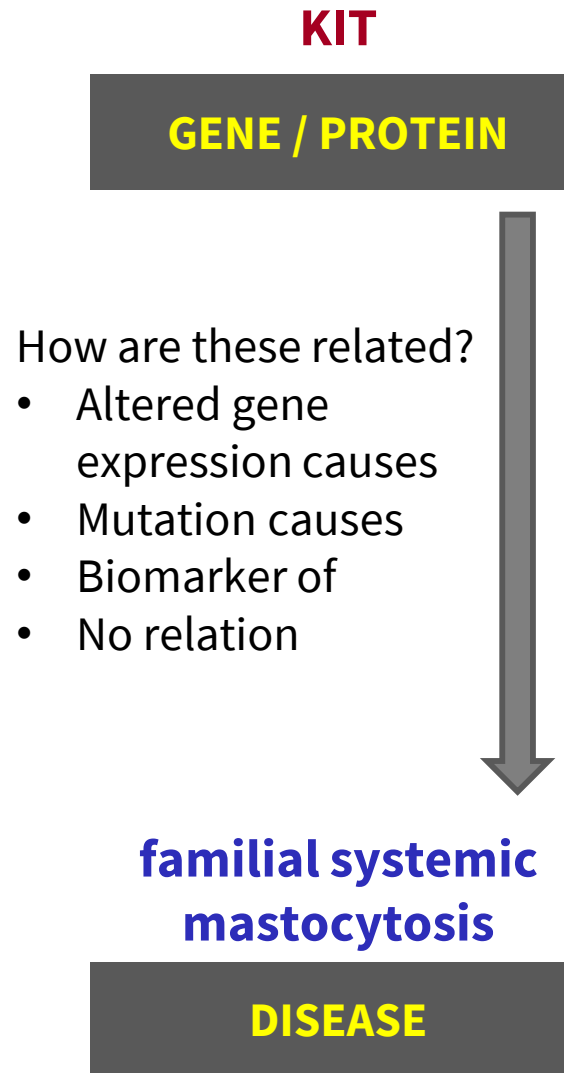
**Imatinib**

is a

**DRUG / CHEMICAL**

# Demo #2: Defining the relationships

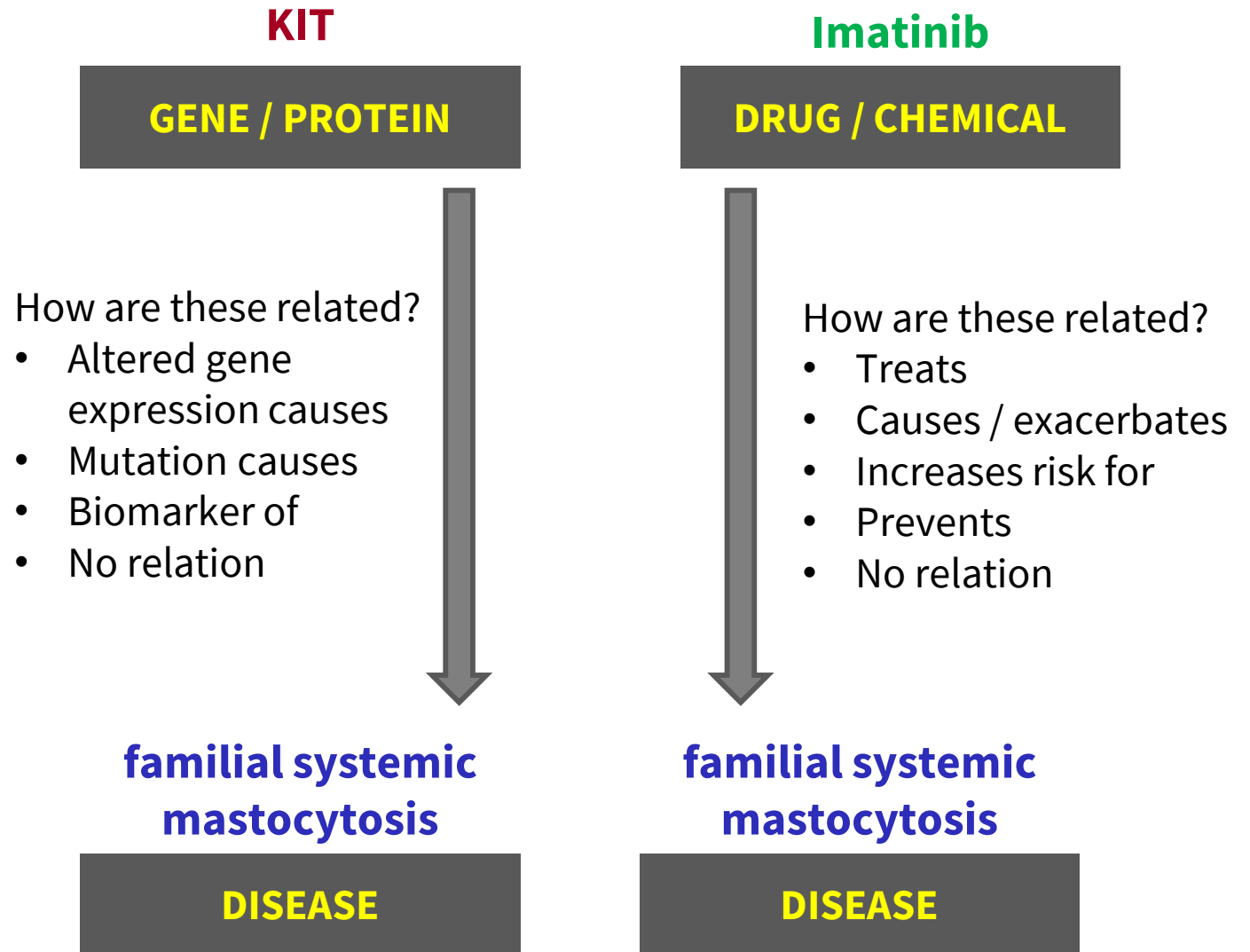
... We report a case of **familial systemic mastocytosis** with the rare **KIT** K509I germ line mutation. In vitro treatment with **imatinib**, dasatinib and PKC412 reduced cell viability of primary mast cells harboring **KIT** K509I mutation. Both patients with **familial systemic mastocytosis** had remarkable hematological and skin improvement after three months of **imatinib** treatment.





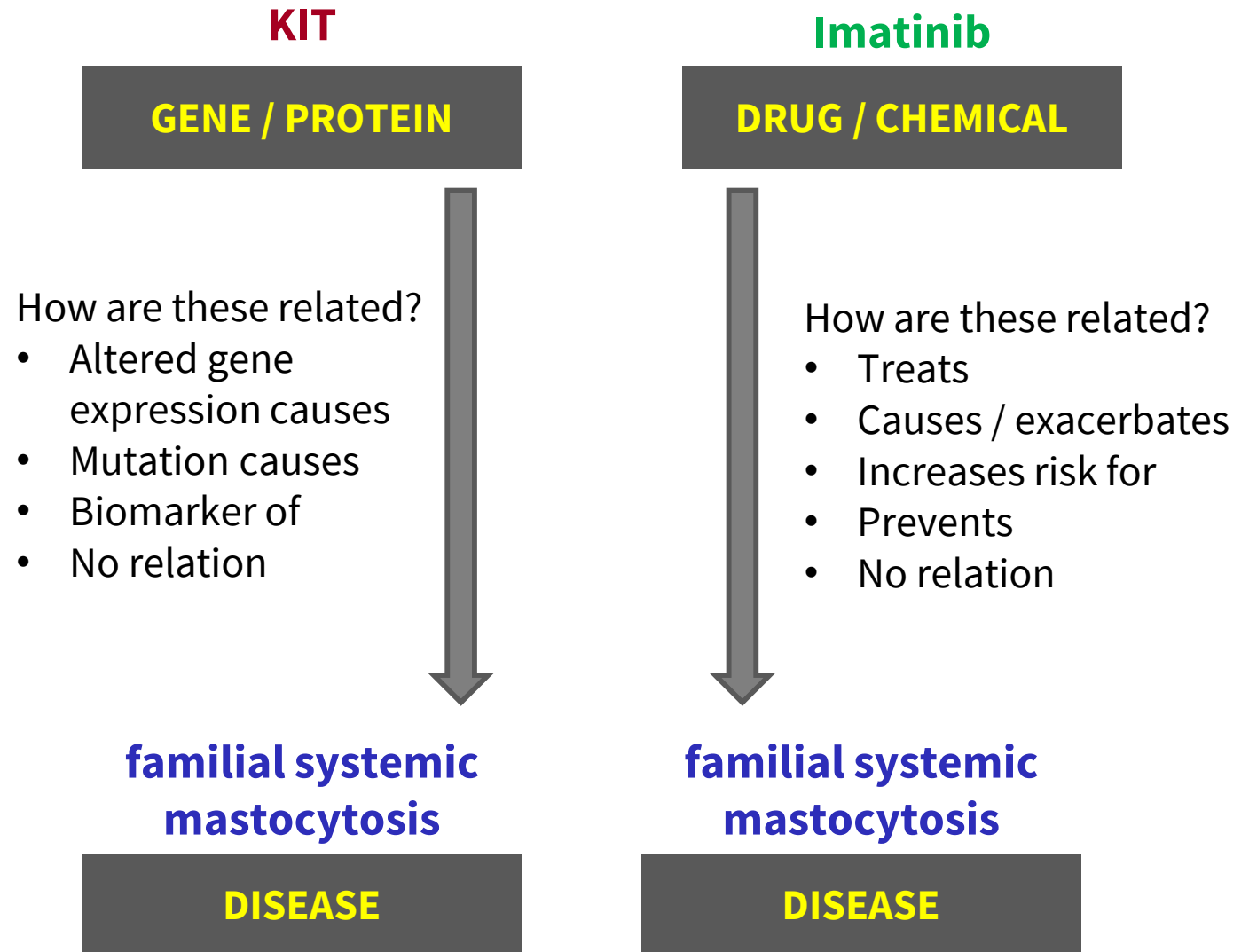
# Demo #2: Defining the relationships

... We report a case of **familial systemic mastocytosis** with the rare **KIT** K509I germ line mutation. In vitro treatment with **imatinib**, dasatinib and PKC412 reduced cell viability of primary mast cells harboring **KIT** K509I mutation. Both patients with **familial systemic mastocytosis** had remarkable hematological and skin improvement after three months of **imatinib** treatment.



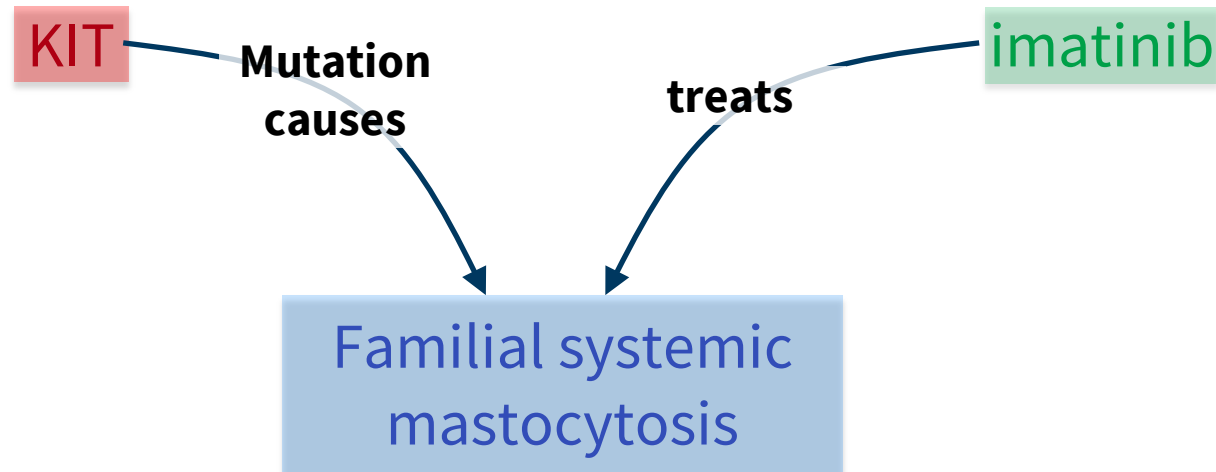
# Demo #2: Defining the relationships – 30 s --- GO! ---

... We report a case of **familial systemic mastocytosis** with the rare **KIT** K509I germ line mutation. In vitro treatment with **imatinib**, dasatinib and PKC412 reduced cell viability of primary mast cells harboring **KIT** K509I mutation. Both patients with **familial systemic mastocytosis** had remarkable hematological and skin improvement after three months of **imatinib** treatment.



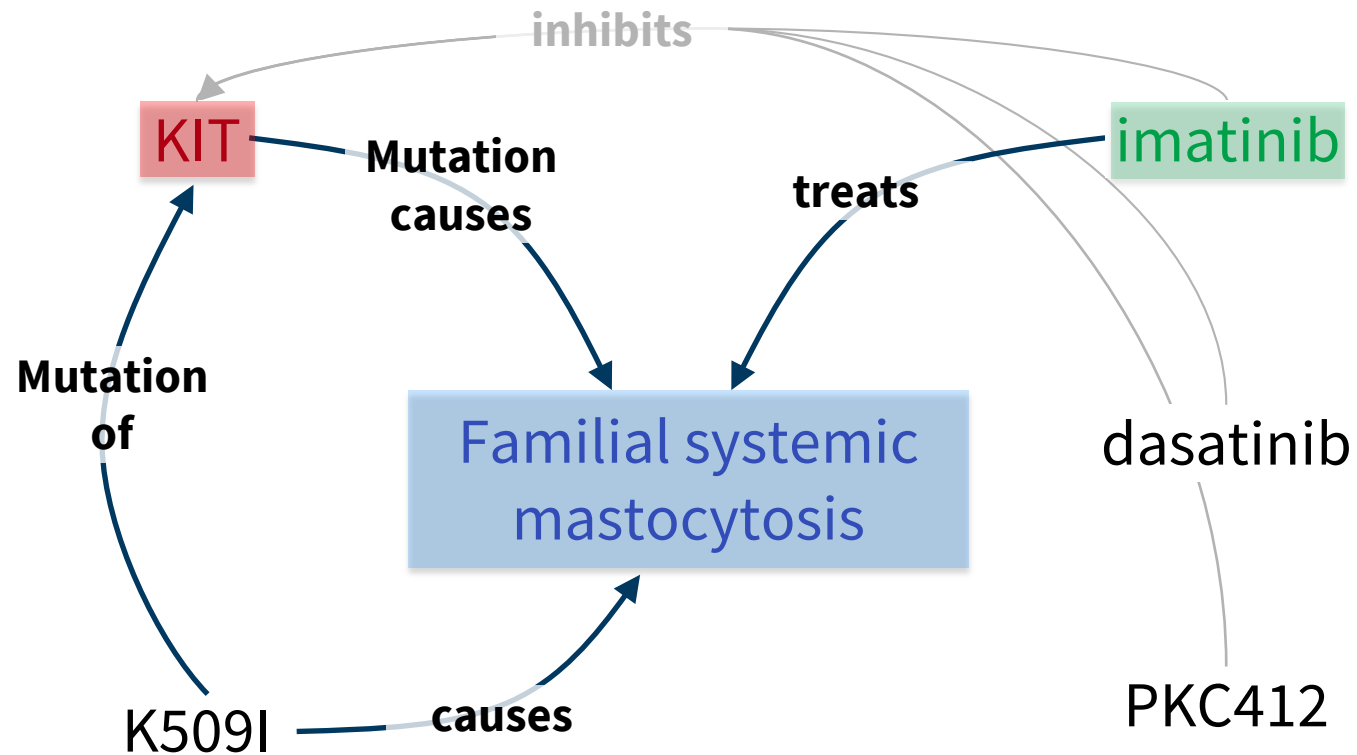
# Demo #2: Results

32



# Demo #2: Results

33





# Bertrand was the first case of NGLY1, but he is not alone.

NGLY1 Researchers are racing to find clues in biomedical literature and need your help to uncover hidden links. If you can read, you can help.

[About NGLY1](#)[Get Started](#)[▶ Watch Video](#)

787,684 annotations have been submitted so far, but we're not done! Your help is still needed... [Learn More >](#)

## Current MISSIONS.



Stress Response and  
glnac  
Total Docs: 73



Stress Response and  
glnac  
Total Docs: 75



Galactosemia and  
Oxidative Stress  
Total Docs: 36



<http://mark2cure.org>

Total Docs: 75



Total Docs: 48



GlcNAc and lipocalin  
Total Docs: 68



Oxidative Stress  
Total Docs: 444



3,200 documents  
1,300 contributors  
787,400 annotations

# “Why do you Mark2Cure?”

In memory of my daughter who  
had Cystic Fibrosis

Studied biology in college  
and I really miss it!

My 4 year old daughter  
Phoebe is living with and  
battling rare disease.

I have Ehlers Danlos Syndrome. I hope to help people  
learn about this painful and debilitating disorder, so that  
others like me can receive more effective medical care.

I am retired, have a doctorate in  
medical humanities, and have two  
children with Gaucher disease. I am  
just looking for some way to put my  
education to use.

## Give back

I Mark2Cure in memory of my son  
Mike who had type 1 diabetes.





Technical Curiosity  
... and a chance  
to help others

To use my  
brain to help  
others

To be a citizen  
scientist and help  
save lives

TO HELP  
SCIENCE  
(& STRETCH MY  
BRAIN)

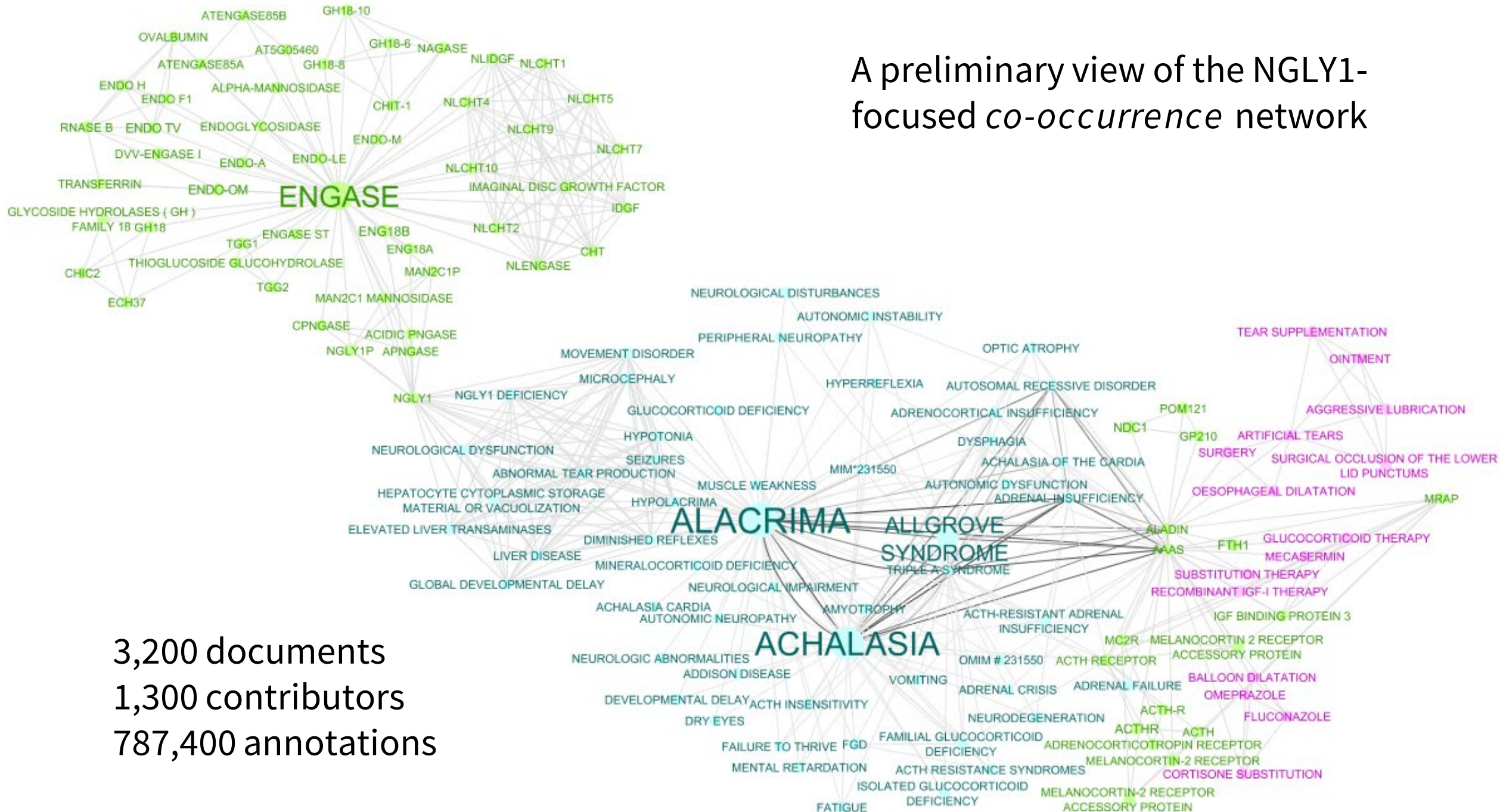
I MISSED SCIENCE  
I WANTED TO HAVE  
FUN WHILE HELPING

To give back  
to the  
community

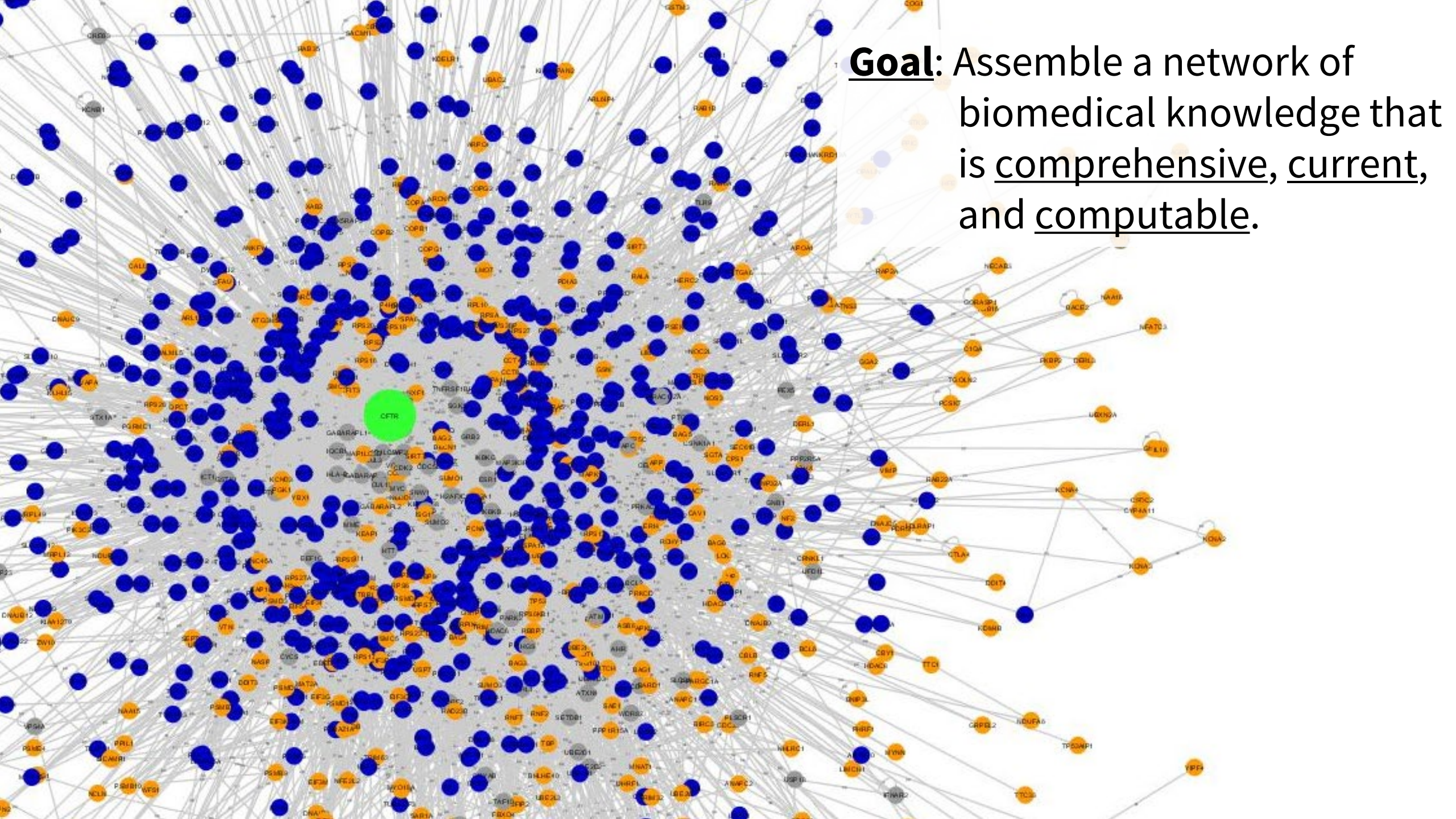
I was/am  
curious



A preliminary view of the NGLY1-  
focused *co-occurrence* network







**Goal:** Assemble a network of biomedical knowledge that is comprehensive, current, and computable.



# Drug repurposing



... identifying  
and developing  
new uses for  
existing drugs

# Drug repurposing using knowledge graphs



**DRUGS**



# Drug repurposing using knowledge graphs

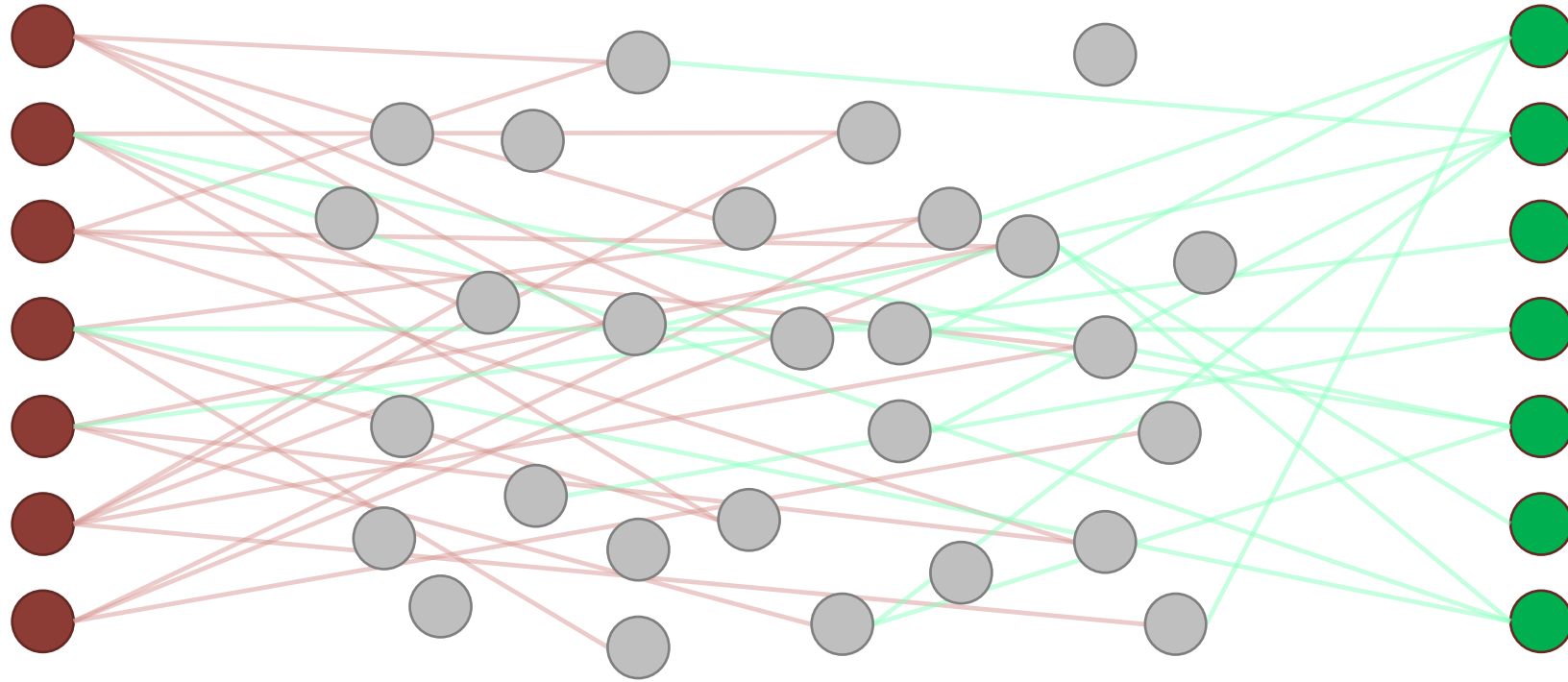


**DRUGS**



**DISEASES**

# Drug repurposing using knowledge graphs

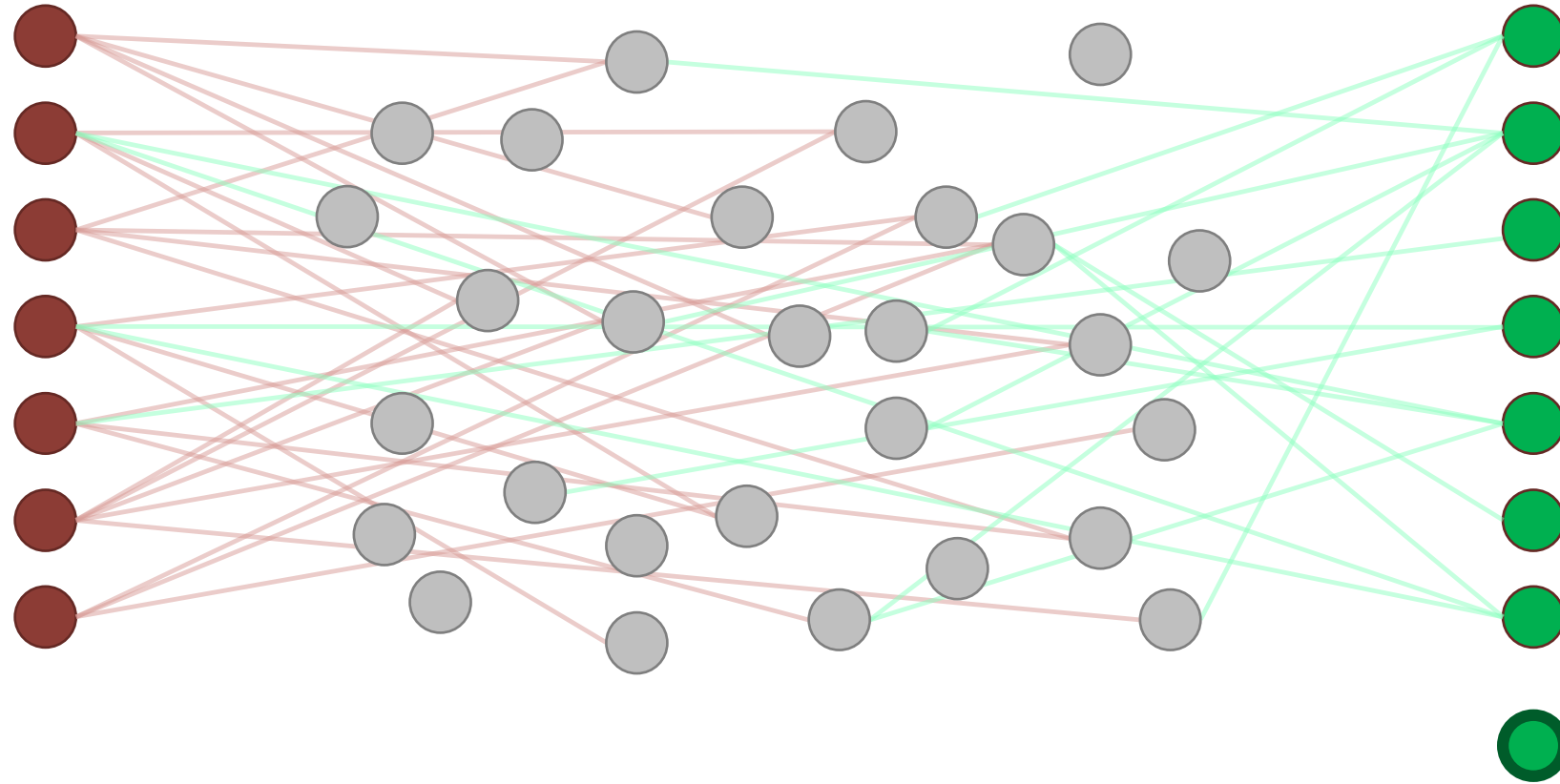


**DRUGS**

**Genes, proteins, pathways,  
genetic variants, metabolites, ...**

**DISEASES**

# Drug repurposing using knowledge graphs

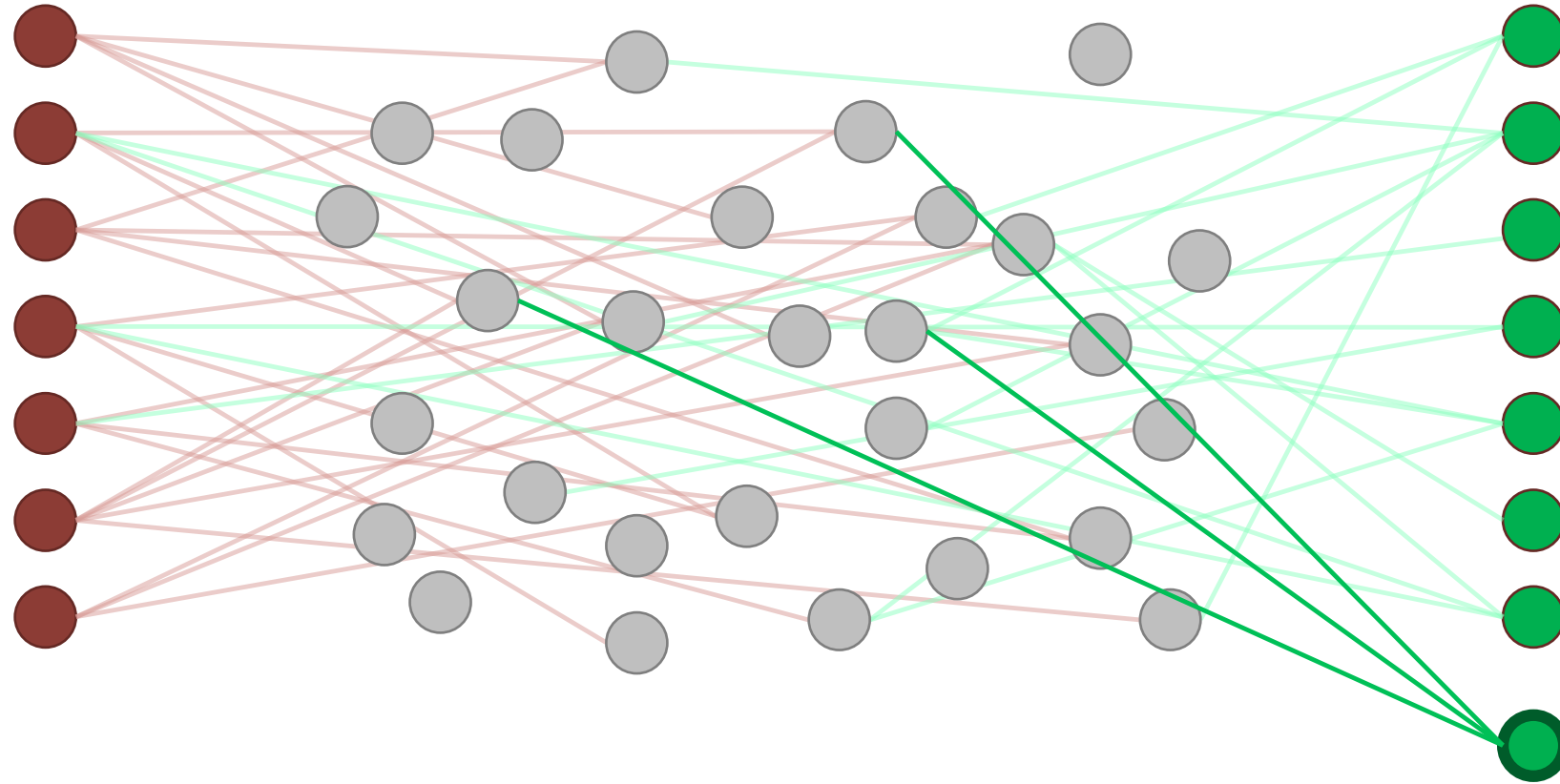


**DRUGS**

**Genes, proteins, pathways,  
genetic variants, metabolites, ...**

**DISEASES**

# Drug repurposing using knowledge graphs



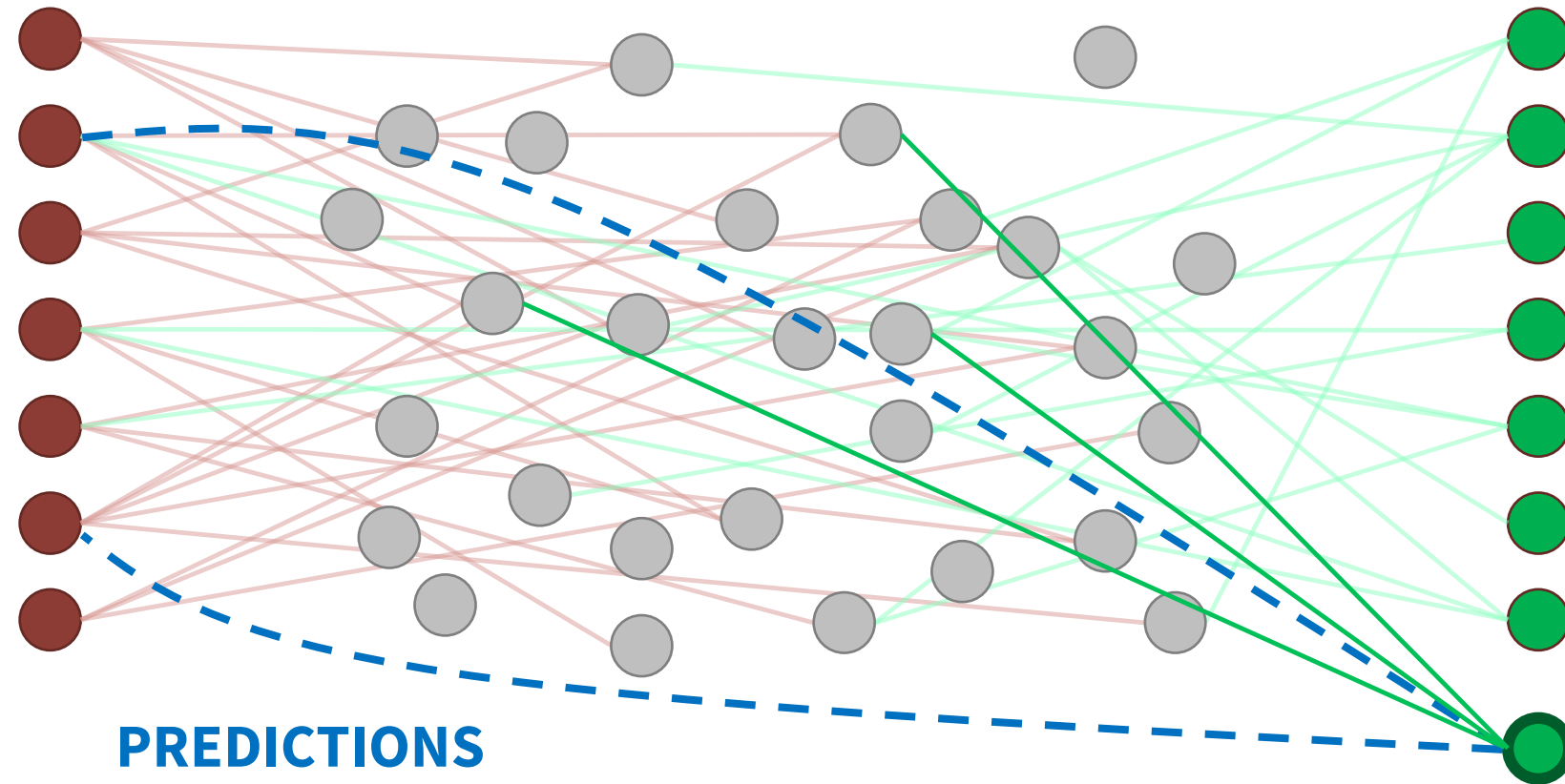
**DRUGS**

**Genes, proteins, pathways,  
genetic variants, metabolites, ...**

**DISEASES**



# Drug repurposing using knowledge graphs



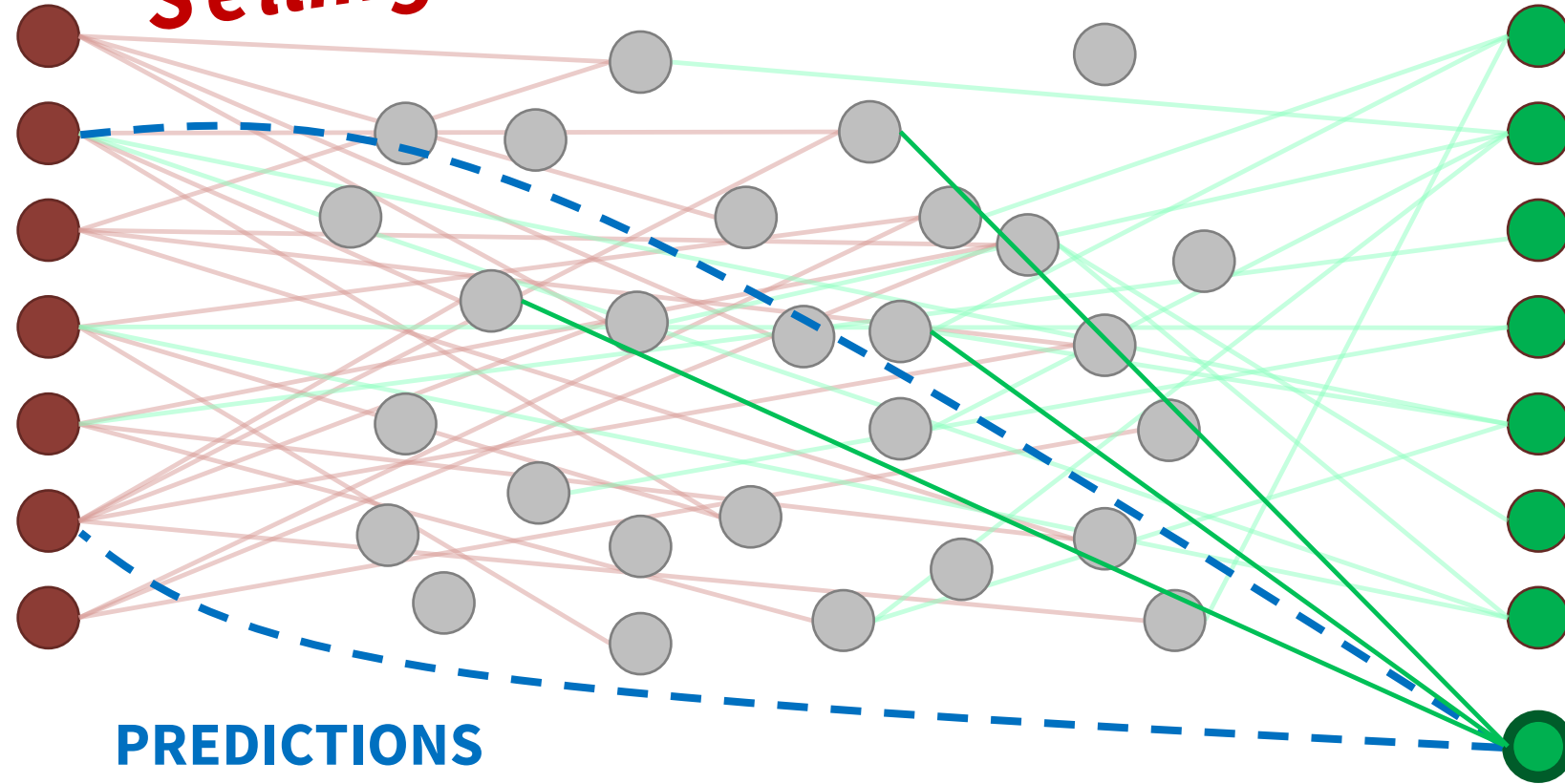
**DRUGS**

Genes, proteins, pathways,  
genetic variants, metabolites, ...

**DISEASES**

# ~~Drug repurposing~~ using knowledge graphs

*Selling stuff!*



**DRUGS**

Genes, proteins, pathways,  
genetic variants, metabolites, ...

**DISEASES**



# Knowledge graphs in high tech

## Model Ensemble for Click Prediction in Bing Search Ads

### ABSTRACT

Accurate estimation of the click-through rate (CTR) in sponsored ads significantly impacts the user search experience and businesses' revenue, even 0.1% of accuracy improvement would yield greater earnings in the hundreds of millions of dollars. CTR prediction is generally formulated as a supervised classification problem. In this paper, we share our experience and learning on model ensemble design and our innovation. Specifically, we present 8 ensemble methods and evaluate them on our production data. Boosting neural networks with gradient boosting decision trees turns out to be the best. With larger training data, there is a nearly 0.9% AUC improvement in offline testing and significant click yield gains in online traffic. In addition, we share our experience and learning on improving the quality of training.

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ACM 978-1-4503-4914-7/17/04.  
<http://dx.doi.org/10.1145/3041021.3054192>

<https://www.microsoft.com/en-us/research/publication/model-ensemble-click-prediction-bing-search-ads/>







# How you can engage in research

## **Citizen Science Portals**

- [www.citizenscience.gov](http://www.citizenscience.gov)
- [scistarter.org](http://scistarter.org)
- [zooniverse.org](http://zooniverse.org)

## **Biomedical Citizen Science**

- [crowd.cochrane.org](http://crowd.cochrane.org) – organize healthcare evidence
- [curate.outbreak.info](http://curate.outbreak.info) – COVID-19 resources
- [eternagame.org](http://eternagame.org) – RNA folding
- [eyewire.org](http://eyewire.org) – 3D neuronal structure
- [foldit.it](http://foldit.it) – protein folding
- [phylogame.org](http://phylogame.org) – sequence alignment
- [stallcatchers.com](http://stallcatchers.com) – Alzheimer's disease

# UPCOMING LECTURE



## Harnessing Chemical Biology for Cancer Drug Discovery

**Speaker:**

**Michael Erb, PhD**

Assistant Professor  
Department of Chemistry

WEDNESDAY, OCTOBER 14, 2020