

Novel Opportunities: Reaction

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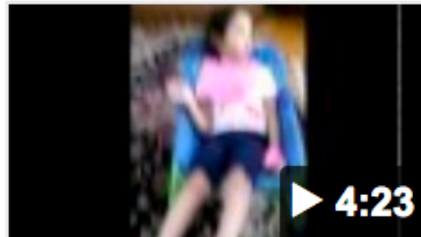
Patients

1. Novel sources of data
2. Deep learning in GM
3. Measuring outcomes

Novel sources of data

Self- or peer-to-peer phenotyping

Are these gelastic seizures ? - YouTube

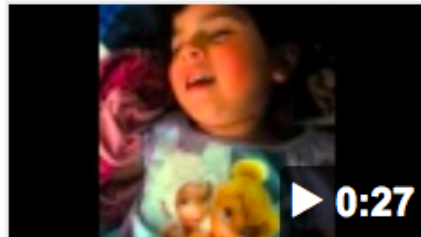


<https://www.youtube.com/watch?v=HezkuIVPLAY> ▼

Nov 16, 2014 - Uploaded by Mohanad el nokali

My sweet Malak has been having these weird attacks lately and i was wondering if anyone who is experienced ...

Evey possible gelastic seizure? - YouTube



<https://www.youtube.com/watch?v=EYZiz3KYyJU> ▼

Jul 17, 2013 - Uploaded by Star537

Nightline from ABC News S2012 • E65 Giggle **Seizures**: No Laughing Matter |

Nightline | ABC News - Duration ...

The Monarch Initiative

Overview



Diseases



Phenotypes



Models



Genes

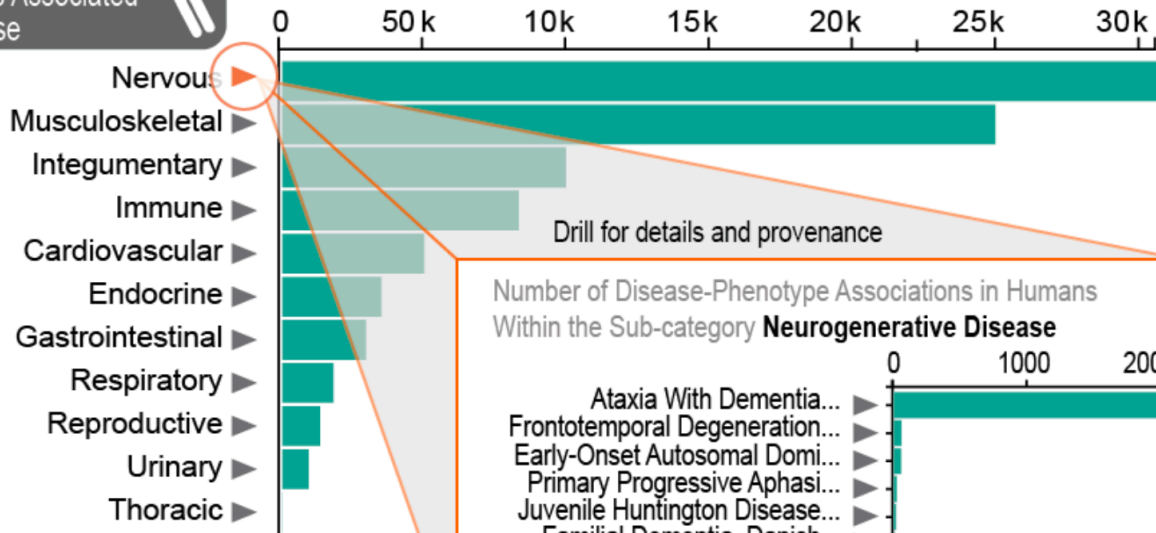


Investigate Phenotypes Associated With Disease



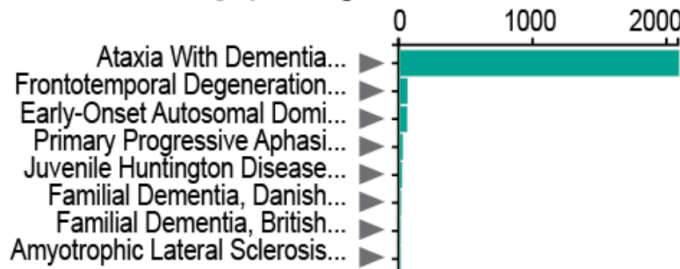
Number of Disease-Phenotype Associations in Humans

System Affected by Disease



Drill for details and provenance

Number of Disease-Phenotype Associations in Humans Within the Sub-category **Neurogenerative Disease**



Plain language for describing human diseases

We have developed the [Human Phenotype Ontology \(HPO\)](#), a vocabulary to describe human disease features (phenotypes). The HPO now includes synonyms that patients, doctors, and machines can all understand.



Apert's syndrome

	Plain language	Medical term
	Webbed toes	Syndactyly
	Deformity due to premature fusing of skull bones	Cranio-synostosis
	Wide-set eyes	Ocular hypertelorism

[View Announcement](#)

[Compare Phenotypes](#)

Internet/social-media-driven case-finding

Participant Web Pages



How do you get beyond n=1?

To help find patients with the same or similar condition, the Undiagnosed Diseases Network (UDN) is creating public web pages about participants in the study.

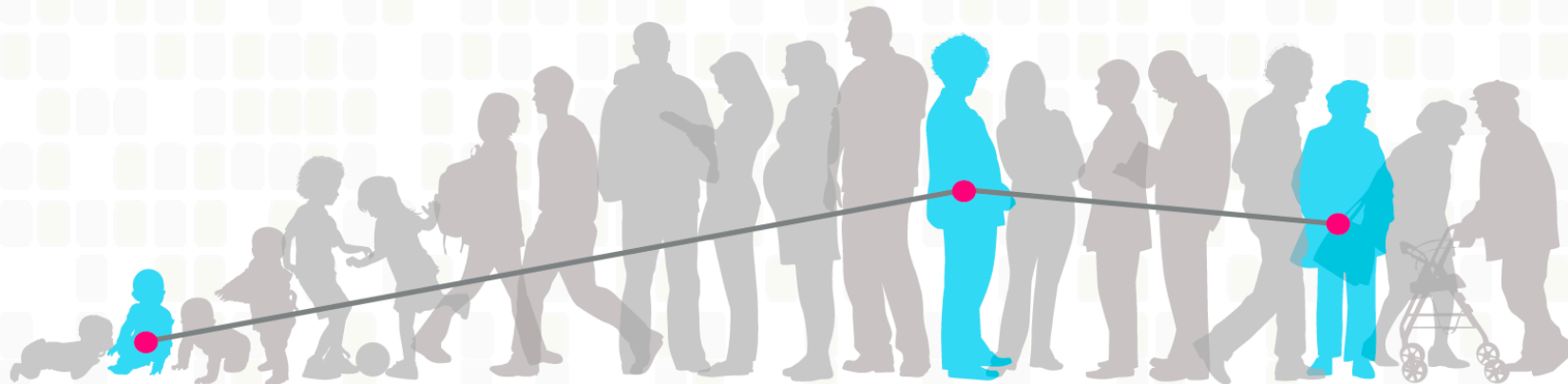
Ideally healthcare providers, researchers, and families who know similar patients will find these pages on the Internet. Connecting these individuals with UDN participants provides the hope of getting beyond n=1.

For more information, visit udnconnect.org!



Sharing information on our website is not a requirement of UDN study participation. Only descriptions of participants who give explicit consent will appear.

MyGene²



Search by gene (e.g., KDM1A), Family ID, or clinical finding



Data-driven genotype inference

Ex: Infer CYP variants from drug history?



Can we infer genotypes from from images?



high cholesterol

Google Search

I'm Feeling Lucky

Can search history tell you about genotype?

The Google logo is centered at the top of the page, rendered in its characteristic multi-colored font.

difficulty swallowing

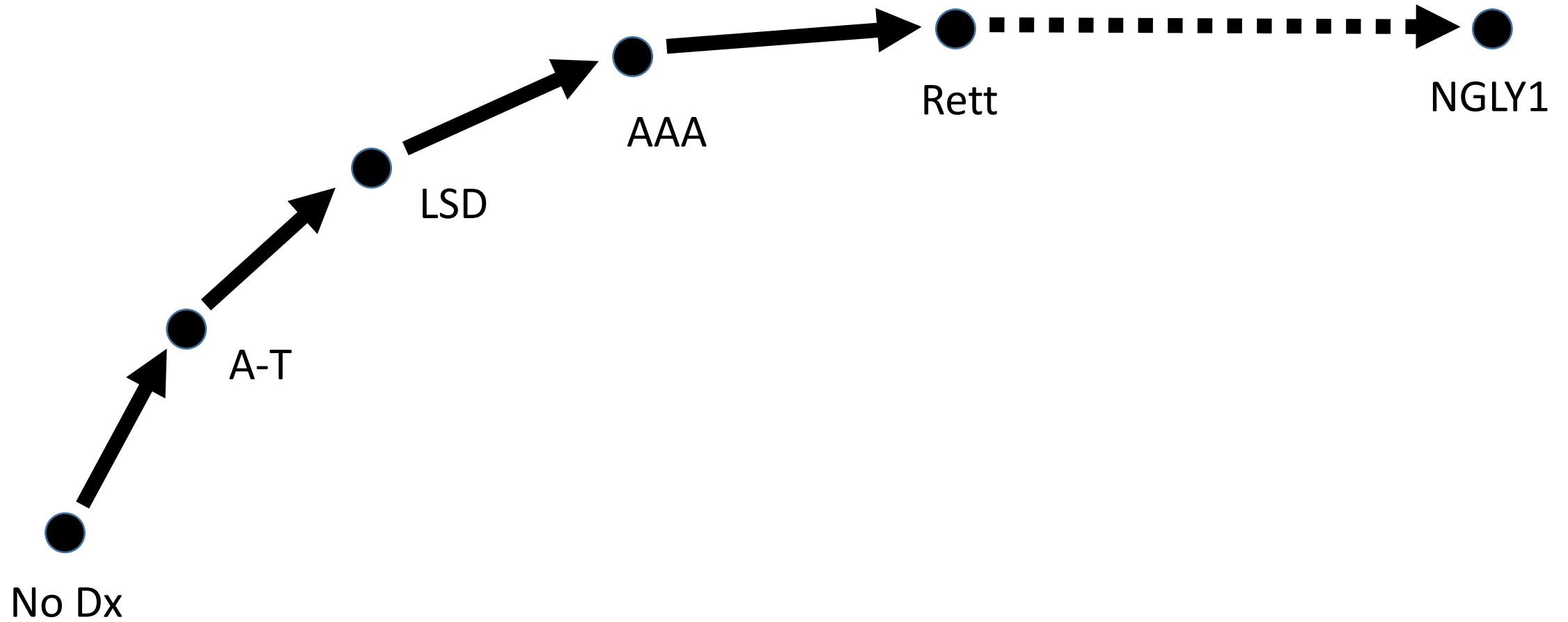
Google Search

I'm Feeling Lucky

Can search history tell you about genotype?

Role of deep learning

Diagnostic “trajectory” recognition

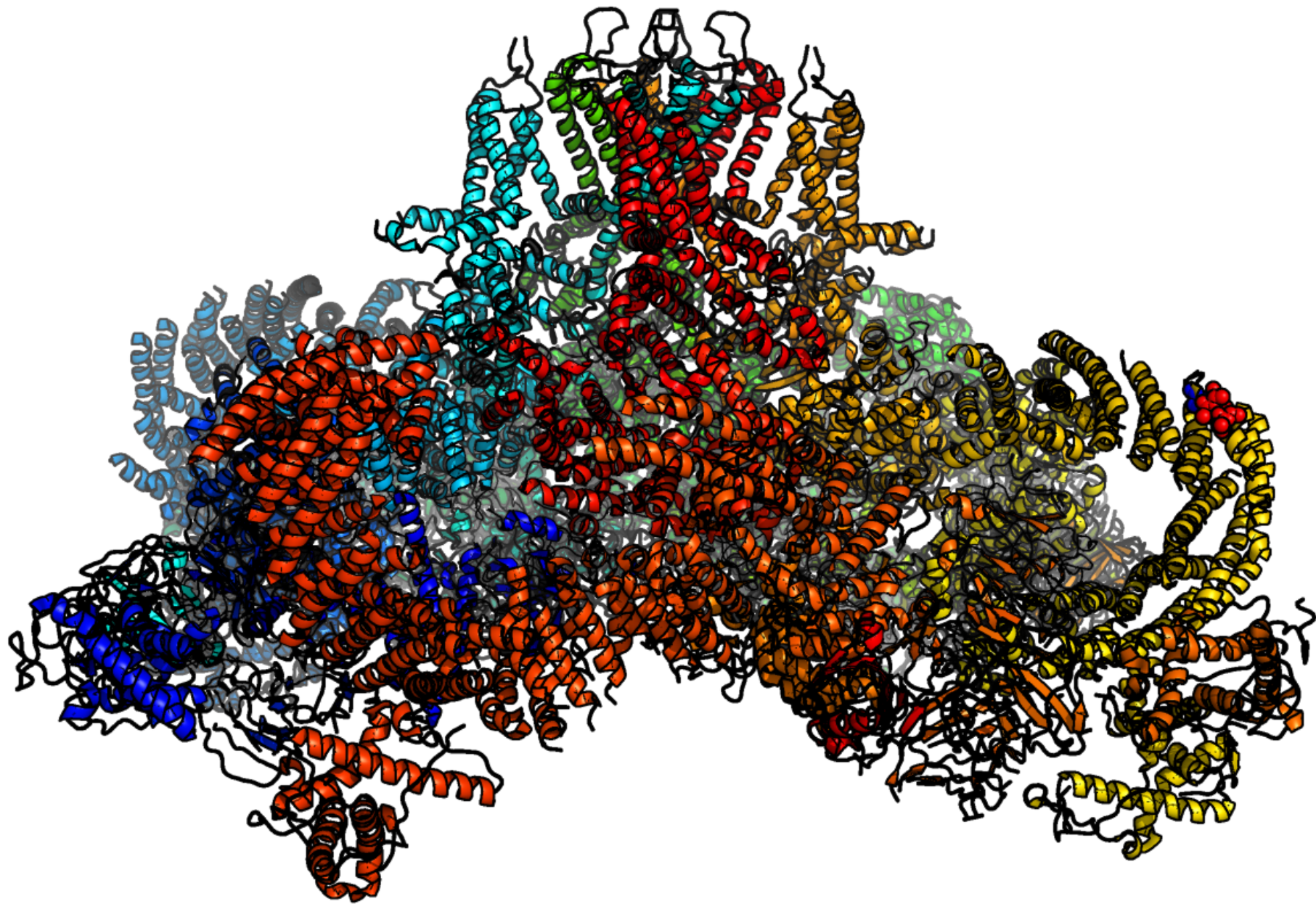


Collaborators: Ken Mandl, Ben Raby

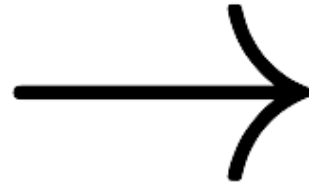
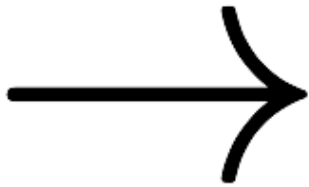
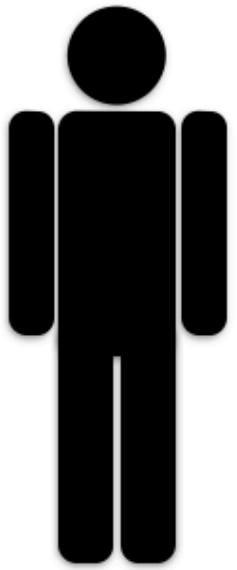
Variant interpretation

Structural inference

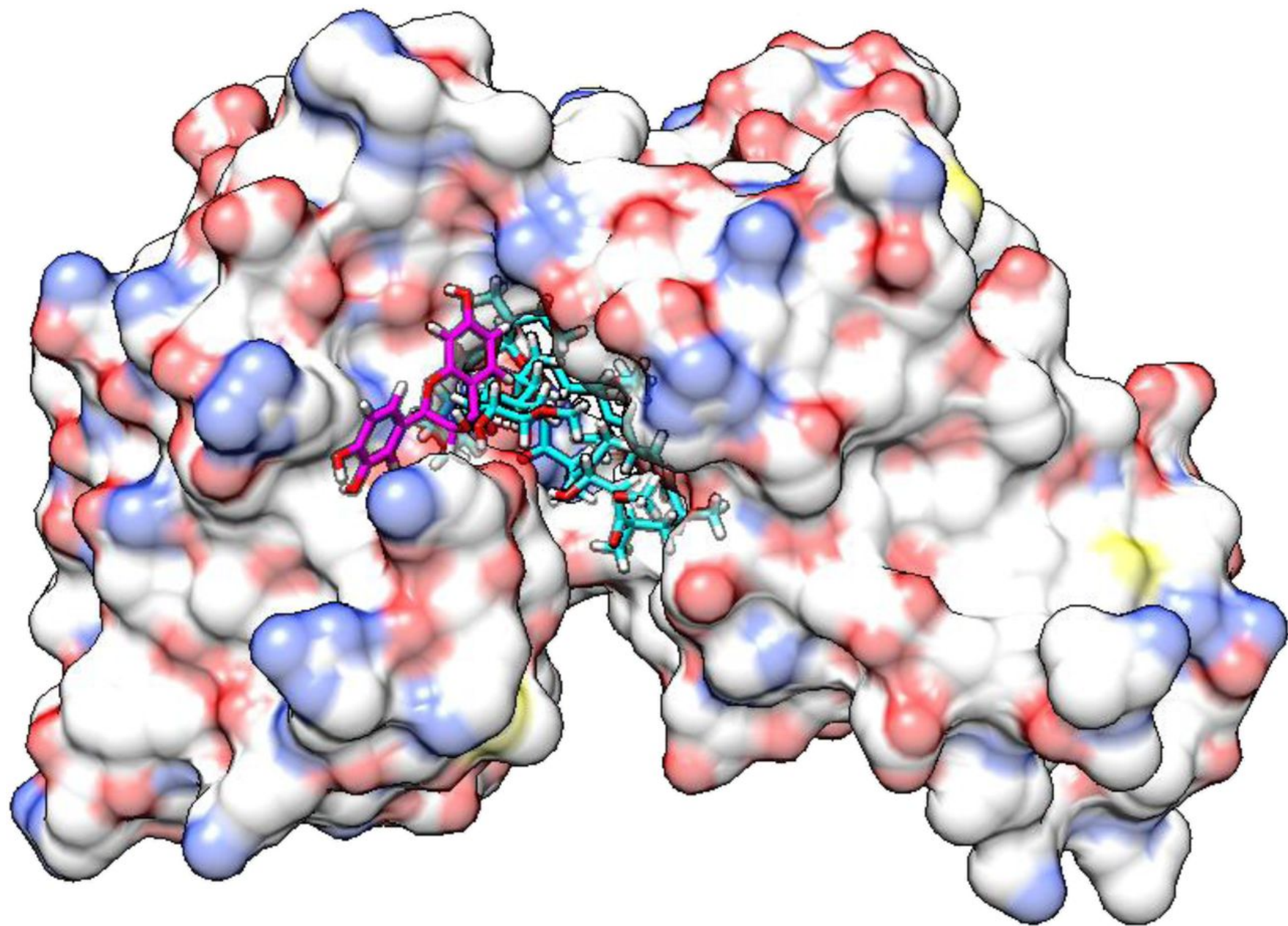
Gene	Variant	Zygoty	Variant Classification
RYR3	c.9443G>T (p.Arg3148Leu)	heterozygous	Uncertain Significance



Toward therapies



Docking simulations



Tox predictions

Structural pharmacogenomics

Novel methods to study impact

Post-intervention Rx change

Improved survival

Improved quality of life



Sentiment and mood inference



Experimental evidence of massive-scale emotional contagion through social networks

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Emotional states can be transferred to others via emotional contagion, leading people to experience the same emotions without their awareness. Emotional contagion is well established in laboratory experiments, with people transferring positive and negative emotions to others. Data from a large real-world social network, collected over a 20-y period suggests that longer-lasting moods (e.g., depression, happiness) can be transferred through networks [Fowler JH, Christakis NA (2008) *BMJ* 337:a2338], although the results are controversial. In an experiment with people who use Facebook, we test whether emotional contagion occurs outside of in-person interaction between individuals by reducing the amount of emotional content in the News Feed. When positive expressions were reduced, people produced fewer positive posts and more negative posts; when negative expressions were reduced, the opposite pattern occurred. These results indicate that emotions expressed by others on Facebook influence our own emotions, constituting experimental evidence for massive-scale contagion via social networks. This work also suggests that, in contrast to prevailing assumptions, in-person interaction and non-verbal cues are not strictly necessary for emotional contagion, and that the observation of others' positive experiences constitutes a positive experience for people.

demonstrated that (i) emotional contagion occurs via text-based computer-mediated communication (7); (ii) contagion of psychological and physiological qualities has been suggested based on correlational data for social networks generally (7, 8); and (iii) people's emotional expressions on Facebook predict friends' emotional expressions, even days later (7) (although some shared experiences may in fact last several days). To date, however, there is no experimental evidence that emotions or moods are contagious in the absence of direct interaction between experiencer and target.

On Facebook, people frequently express emotions, which are later seen by their friends via Facebook's "News Feed" product (8). Because people's friends frequently produce much more content than one person can view, the News Feed filters posts, stories, and activities undertaken by friends. News Feed is the primary manner by which people see content that friends share. Which content is shown or omitted in the News Feed is determined via a ranking algorithm that Facebook continually develops and tests in the interest of showing viewers the content they will find most relevant and engaging. One such test is reported in this study: A test of whether posts with emotional content are more engaging.

The experiment manipulated the extent to which people ($N =$

Financial impact on patients

Patient engagement with science

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](#)

Development of communities

Development of new therapies

Thank you!

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