Novel Opportunities: Reaction

Matt Might, Ph.D. | @mattmight | might@uab.edu
Hugh Kaul Precision Medicine Institute
University of Alabama at Birmingham
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=UYZiz3KYyJU

Jul 17, 2013 - Uploaded by Star537

Nightline from ABC News S2012 • E65 Giggle Seizures: No Laughing Matter |
Nightline | ABC News - Duration ...
The Monarch Initiative

Investigate Phenotypes Associated With Disease

Number of Disease-Phenotype Associations in Humans

Overview
Diseases
Phenotypes
Models
Genes

System Affected by Disease

Nervous
Musculoskeletal
Integumentary
Immune
Cardiovascular
Endocrine
Gastrointestinal
Respiratory
Reproductive
Urinary
Thoracic

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
Webbed toes
Deformity due to premature fusing of skull bones
Wide-set eyes

Medical term
Syndactyly
Craniosynostosis
Ocular hypertelorism

Compare Phenotypes

View Announcement
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!

Collaborators: Kim Splinter, Cecilia Esteves
Data-driven genotype inference
Ex: Infer CYP variants from drug history?
Can we infer genotypes from images?

Source: FDNA
Can search history tell you about genotype?
Can search history tell you about genotype?
Role of deep learning
Diagnostic “trajectory” recognition
No Dx

A-T

LSD

AAA

Rett

NGLY1

Collaborators: Ken Mandl, Ben Raby
Variant interpretation
Structural inference
<table>
<thead>
<tr>
<th>Gene</th>
<th>Variant</th>
<th>Zygosity</th>
<th>Variant Classification</th>
</tr>
</thead>
<tbody>
<tr>
<td>RYR3</td>
<td>c.9443G&gt;T (p.Arg3148Leu)</td>
<td>heterozygous</td>
<td>Uncertain Significance</td>
</tr>
</tbody>
</table>
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

Adam D. I. Kramer, Jamie E. Guillory, and Jeffrey T. Hancock

*Core Data Science Team, Facebook, Inc., Menlo Park, CA 94025; and Departments of Communication and Information Science, Cornell University, Ithaca, NY 14853

Edited by Susan T. Fiske, Princeton University, Princeton, NJ, and approved March 25, 2014 (received for review October 23, 2013)

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 nonverbal cues are not strictly necessary for emotional contagion, and that the observation of others’ positive experiences constitutes a positive experience for people.

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!

Matt Might, Ph.D. | @mattmight | might@uab.edu
Hugh Kaul Precision Medicine Institute
University of Alabama at Birmingham