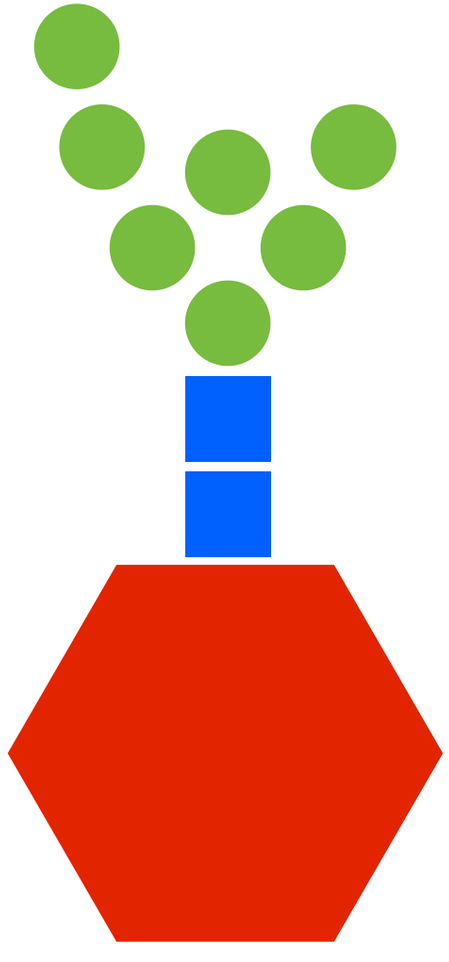
NGLY1

Bench to Bedside



Matt Might, Ph.D.

Associate Professor

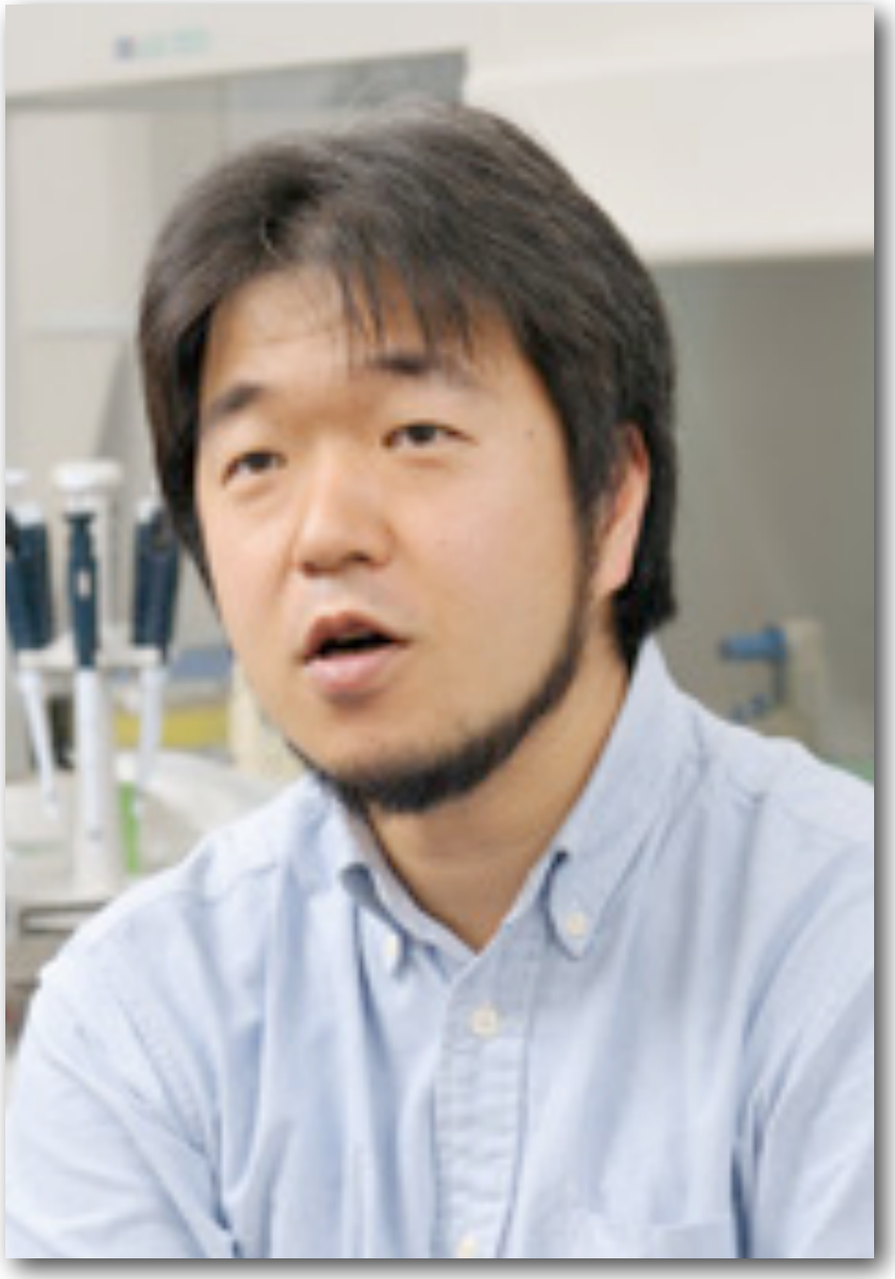
Computer Science

University of Utah

matt.might.net

@mattmight

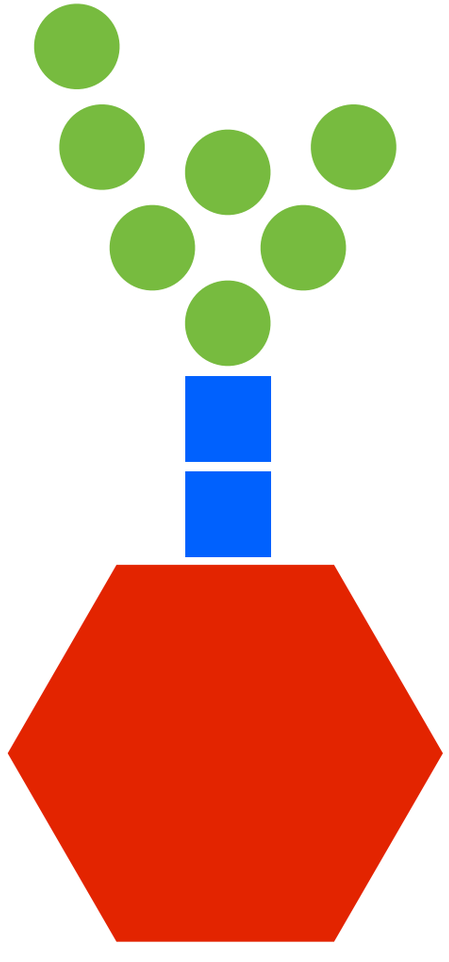
Real heroes don’t wear capes.



Real heroes do glycobiology.

NGLY1

Bench to Bedside



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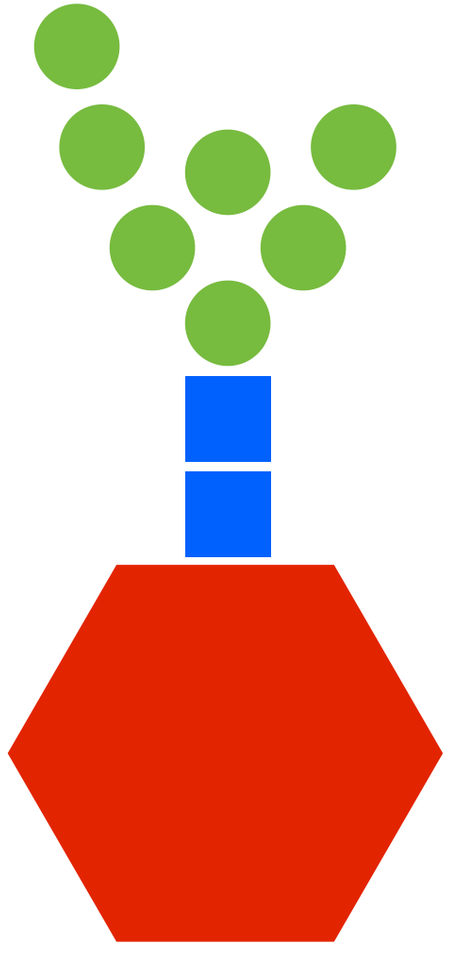
NGLY1

Bench to Bedside

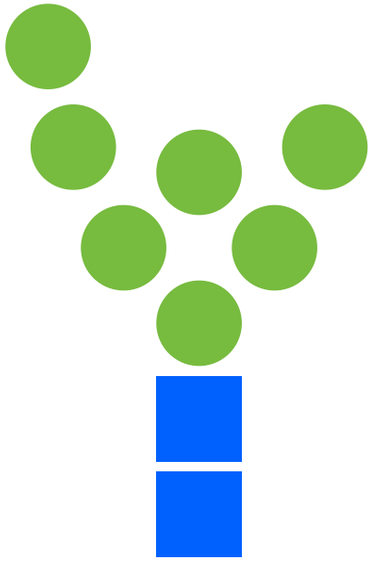


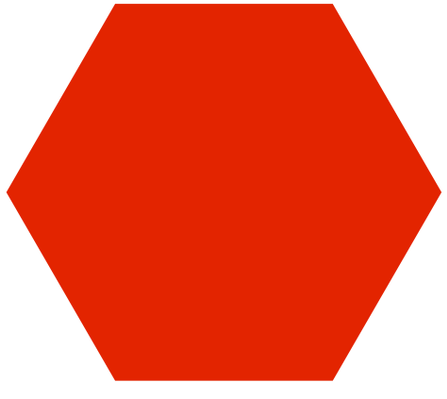
Matt Might, Dad

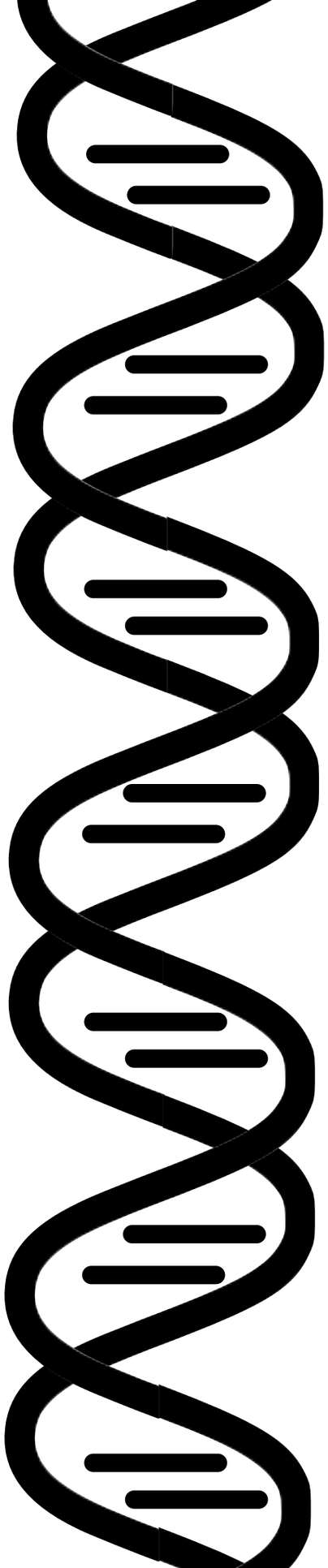
NGLY1.org

Bench to Bedside to Bench

Matt Might, Dad



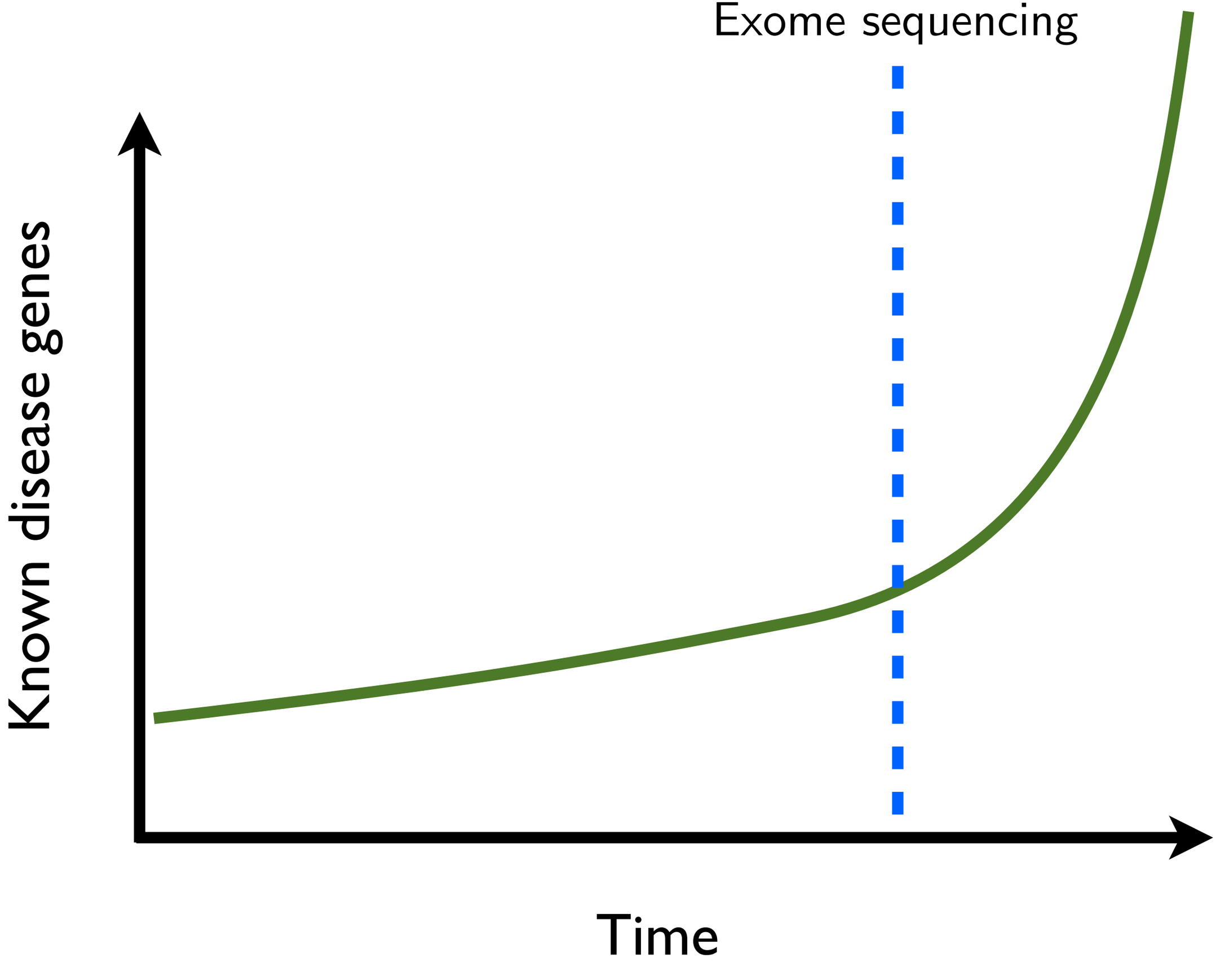




+

Exome sequencing

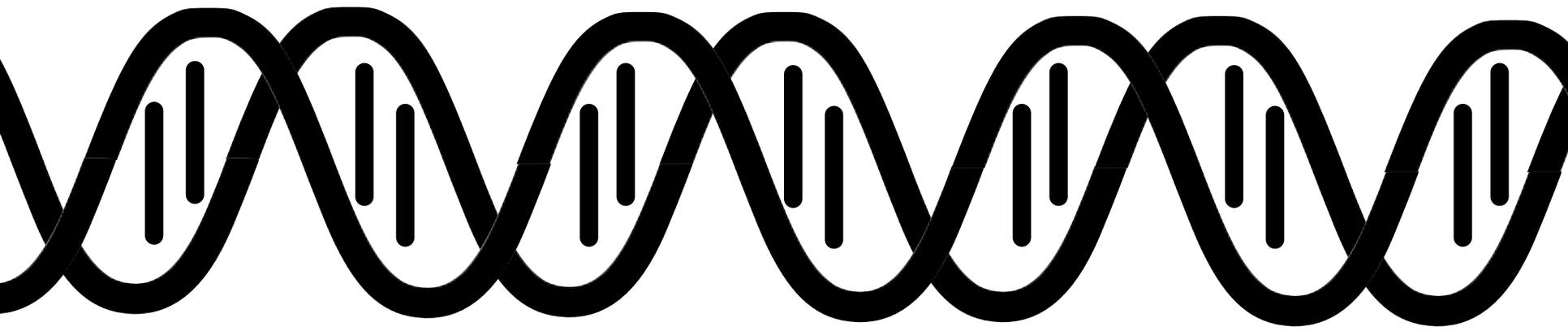
Exome sequencing

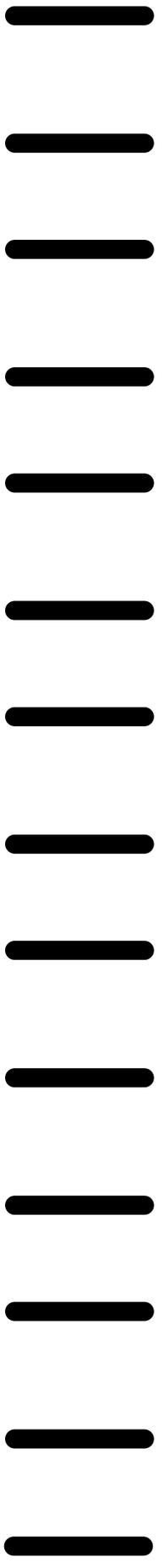

Exome sequencing
Known disease genes
Time

4.5

Duke

UNIVERSITY





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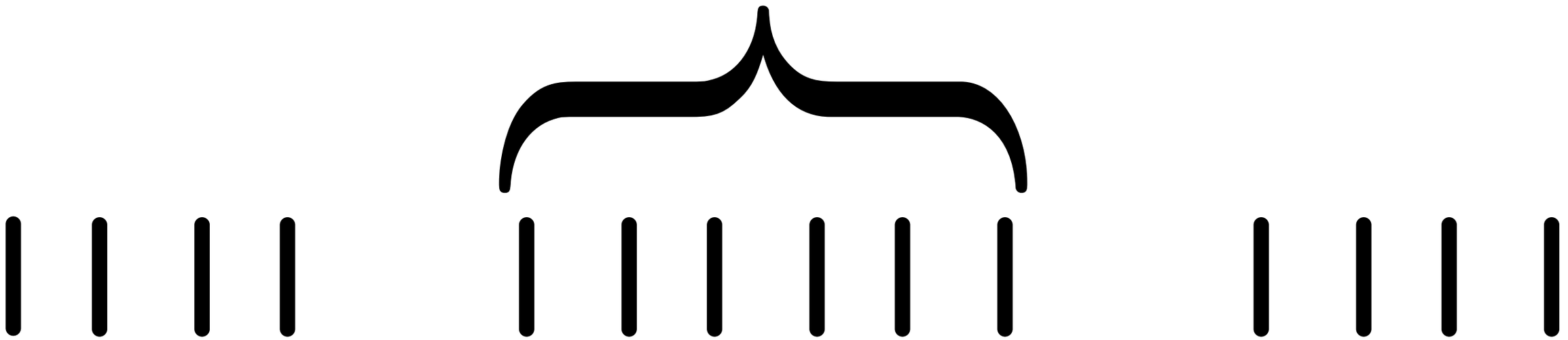

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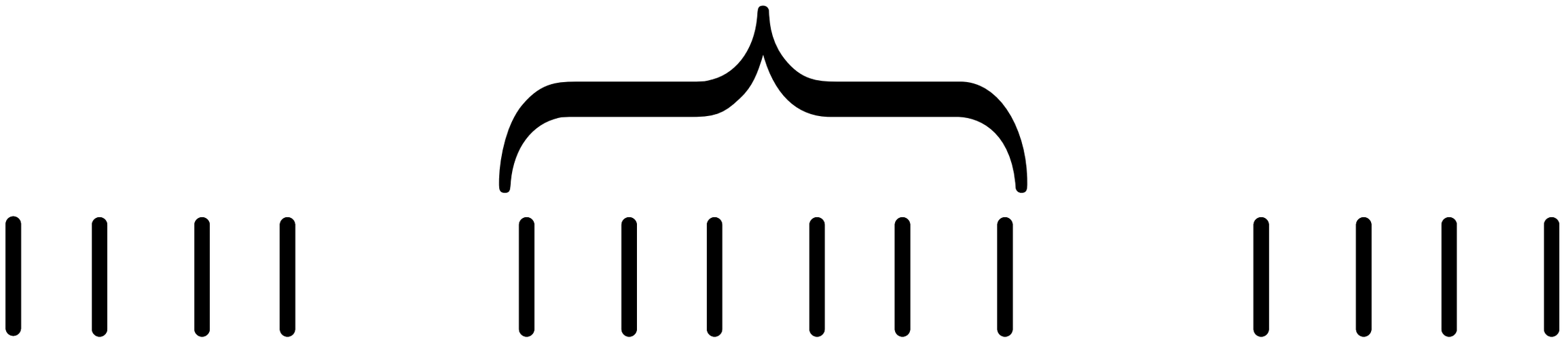

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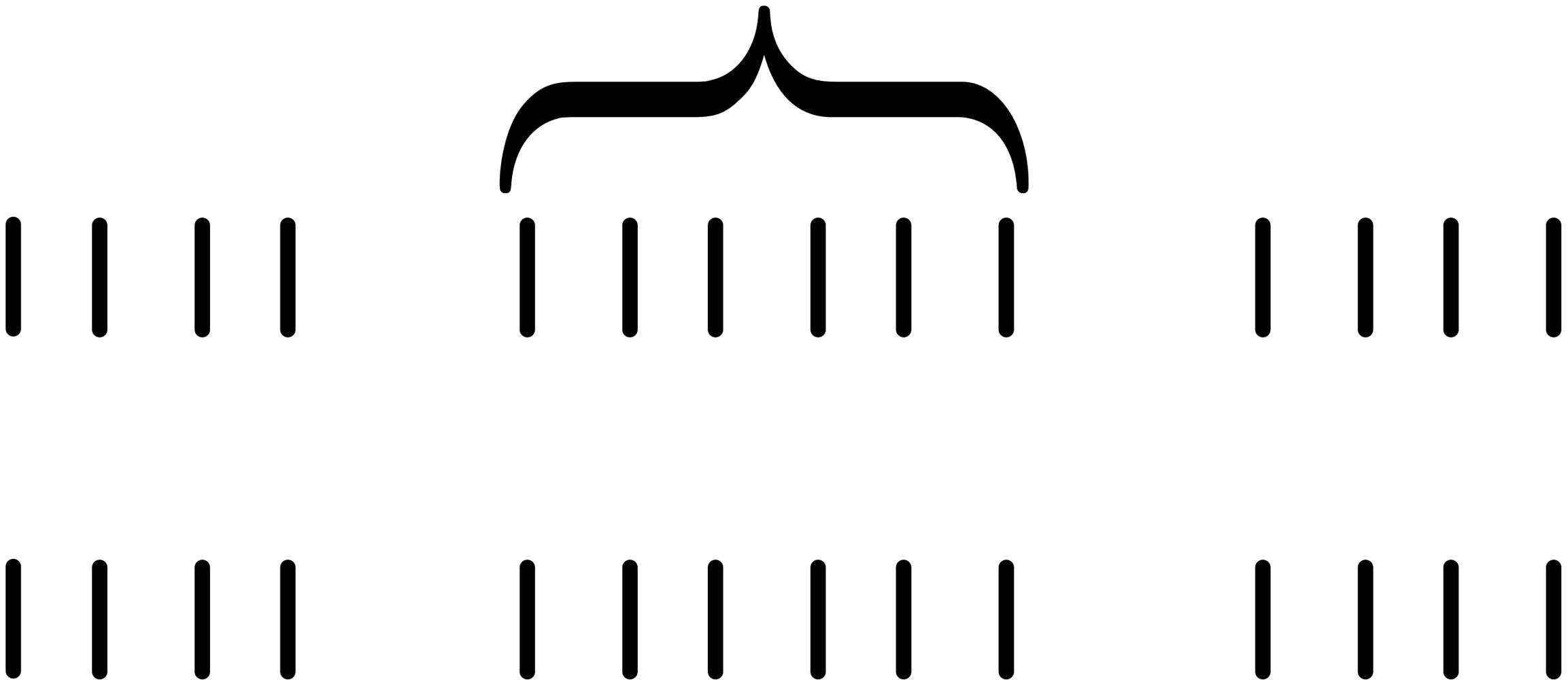

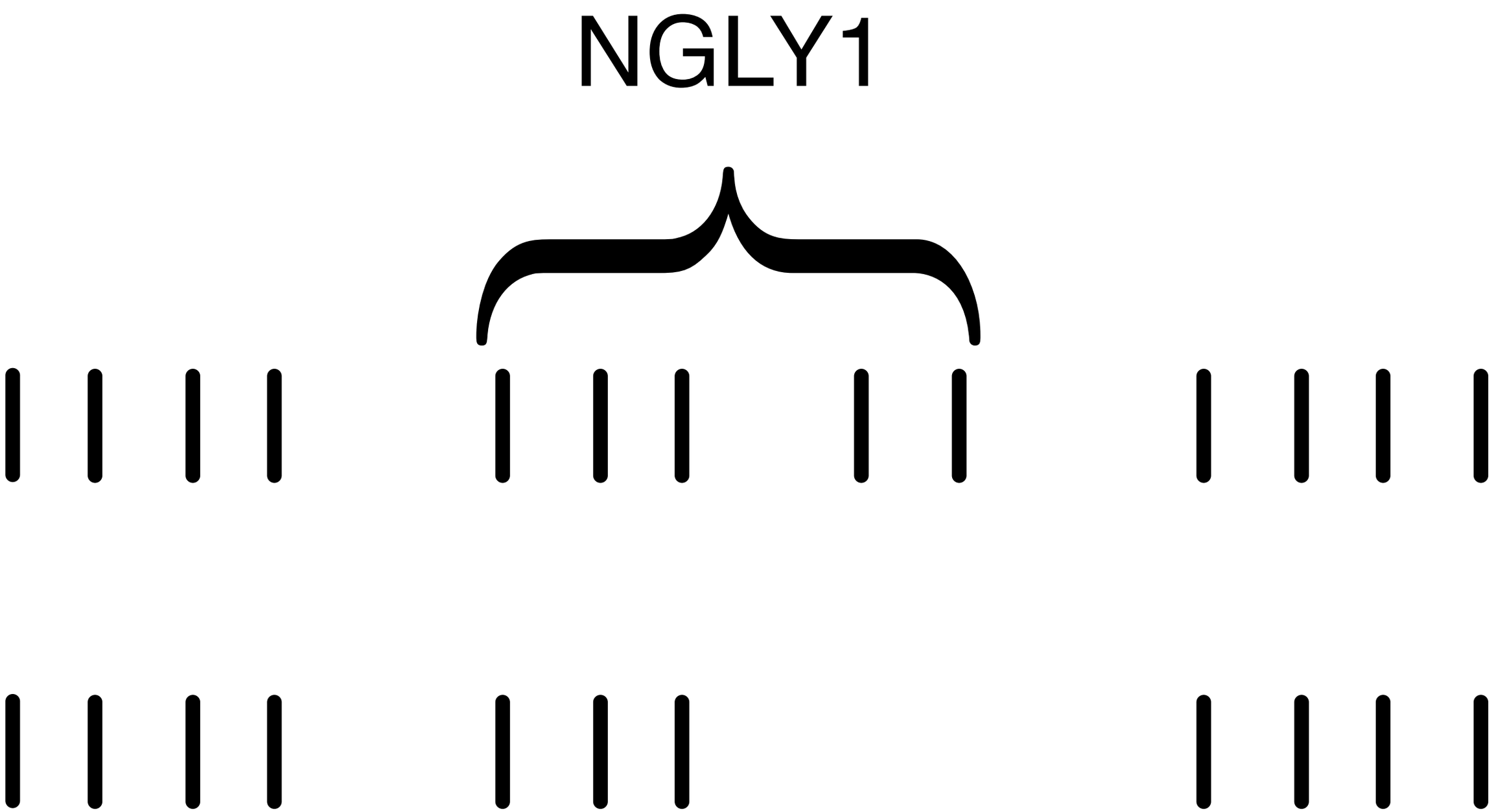
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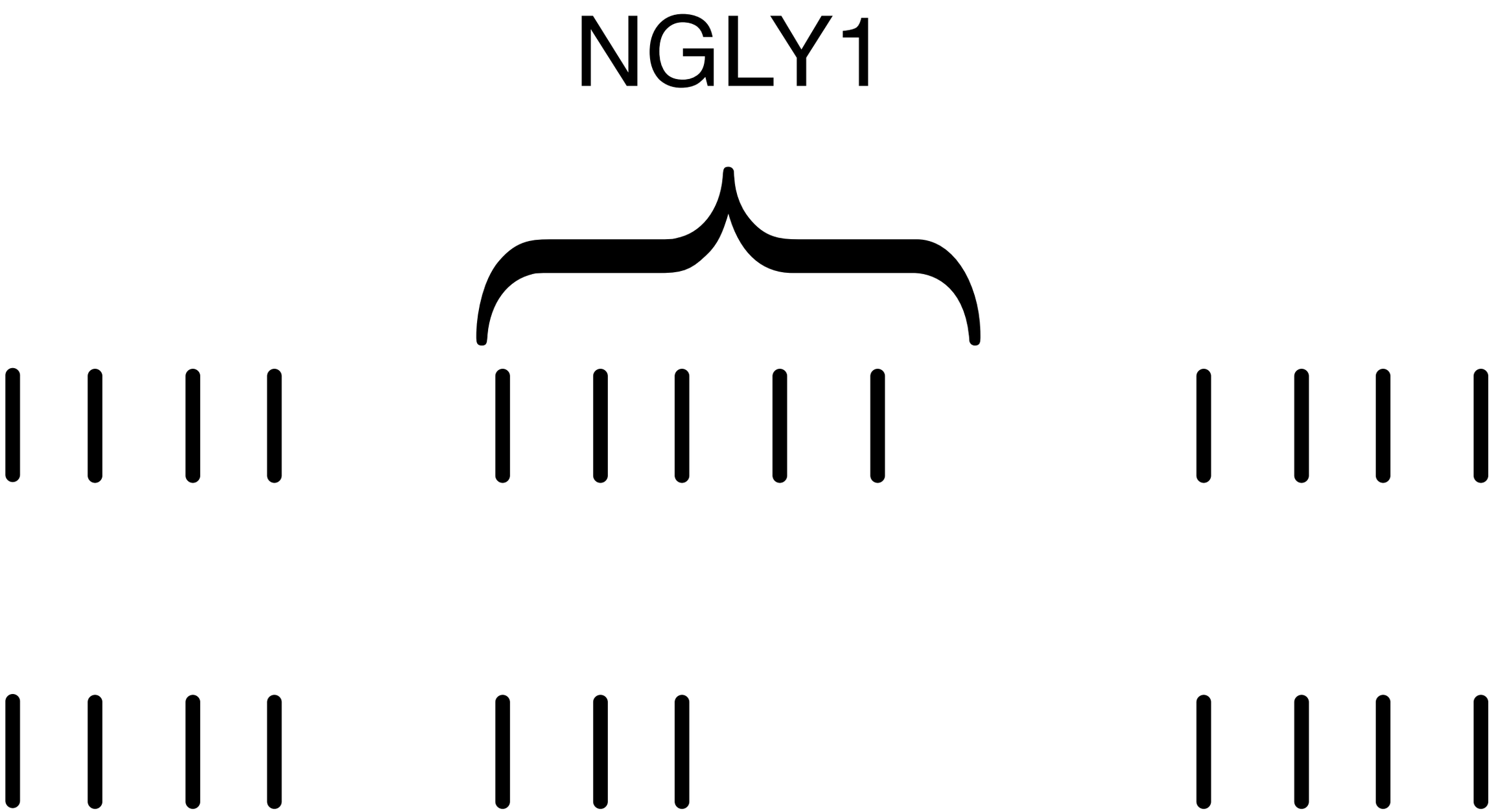
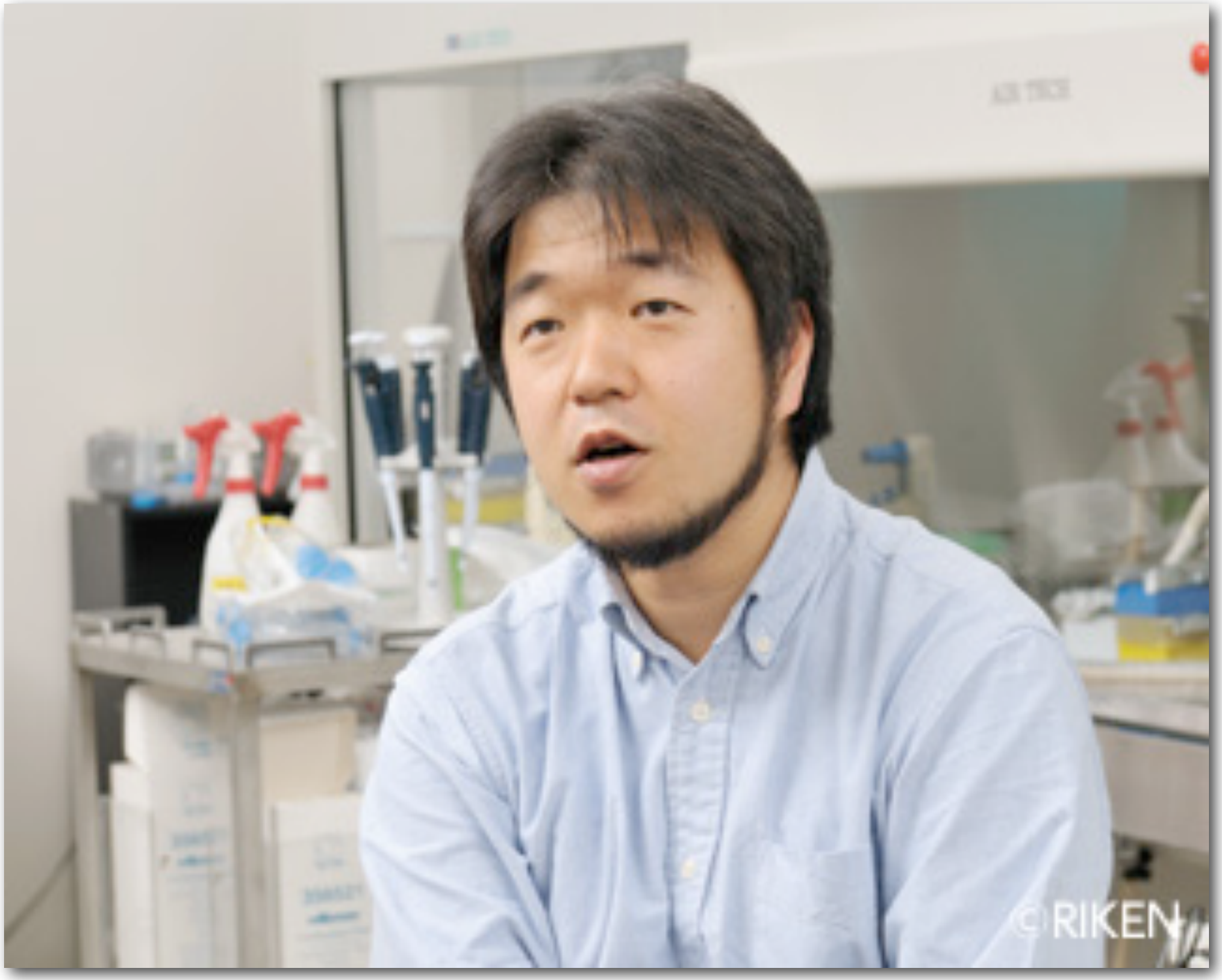
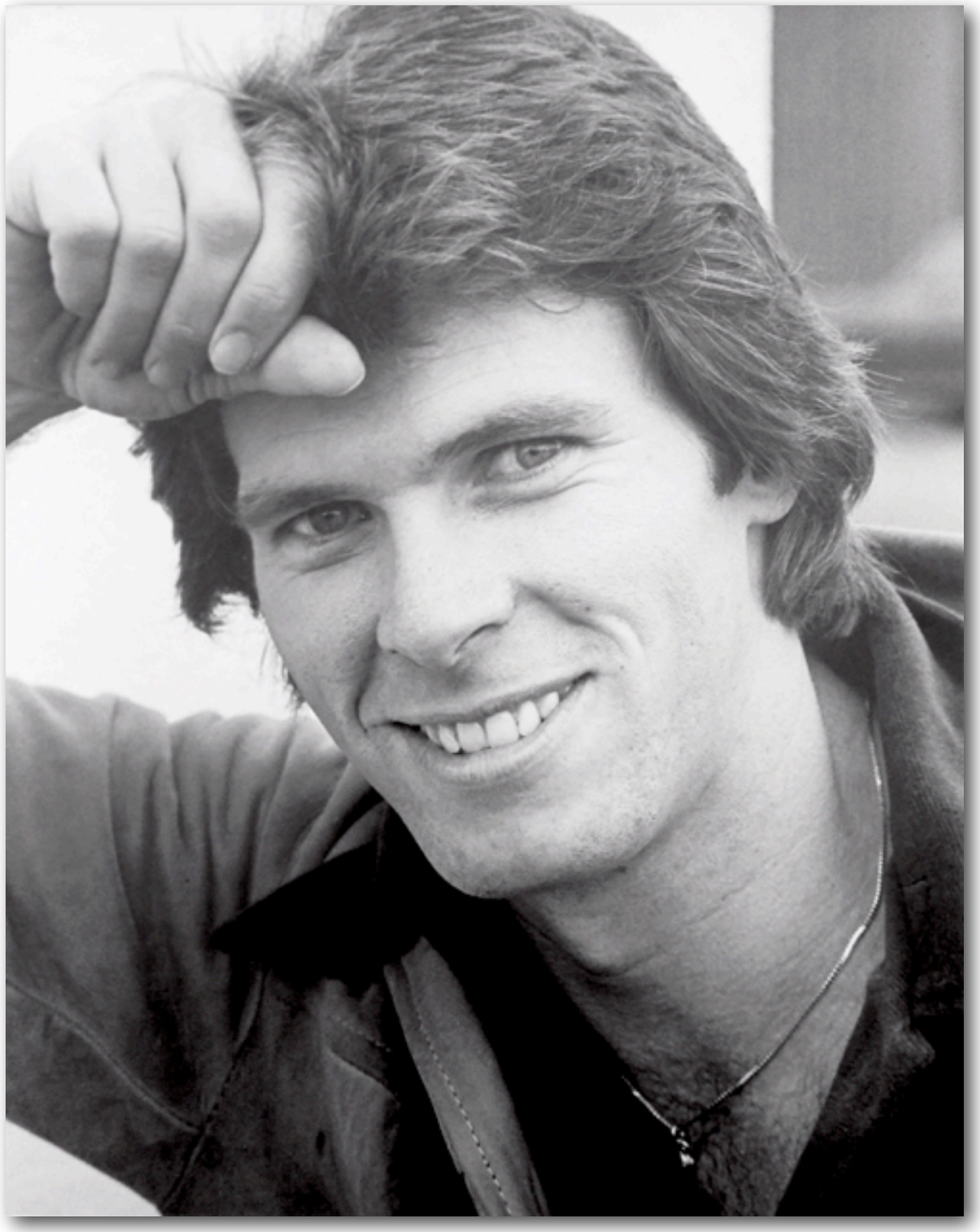

NGLY1

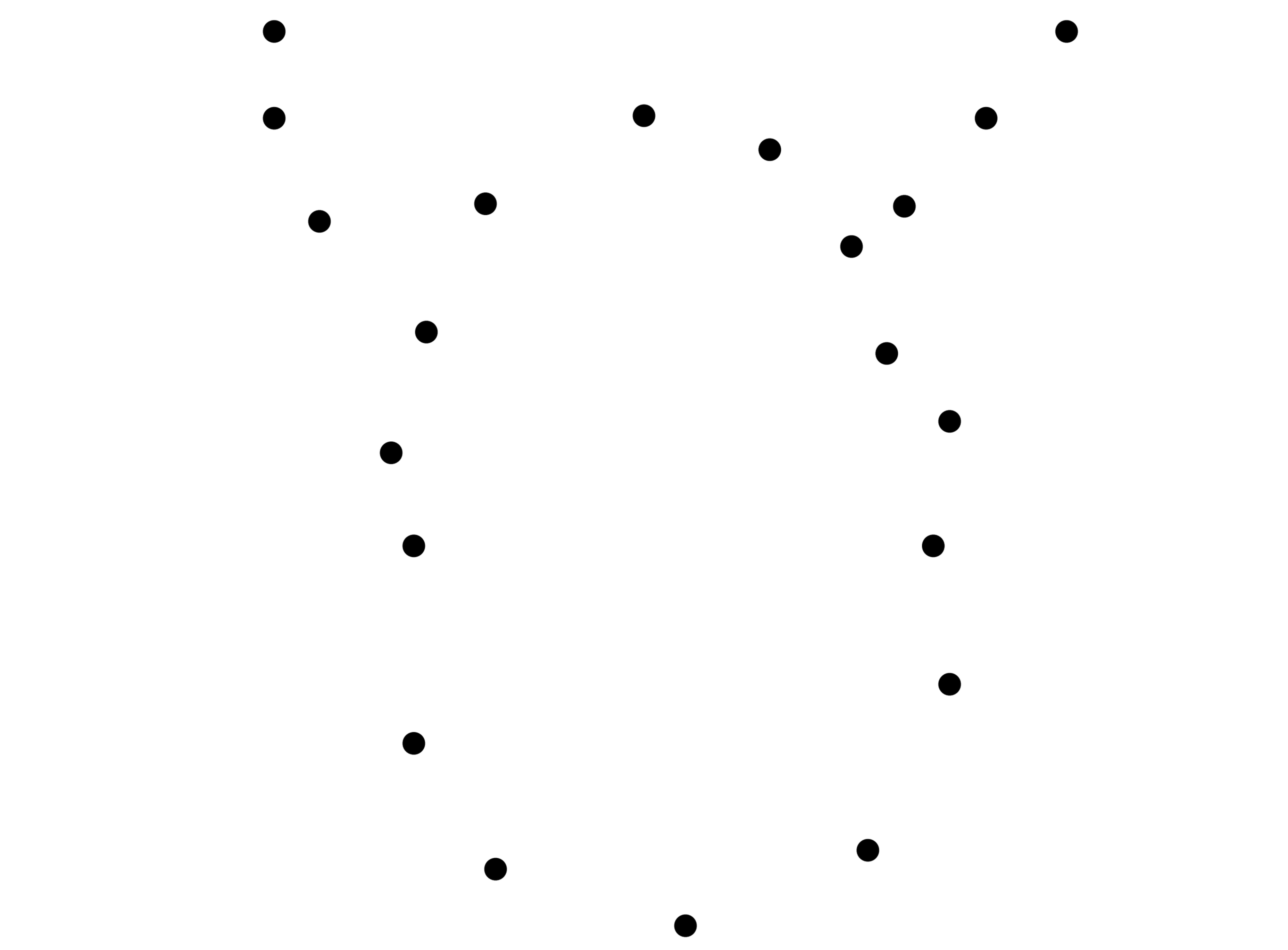
What if $n = 1$?



Tadashi Suzuki



Hudson Freeze



NGLY1

Why glycoscience matters

John "Rocket" Williams IV

Mt. Glyco

We can’t do this alone.

Hunting down my son's killer

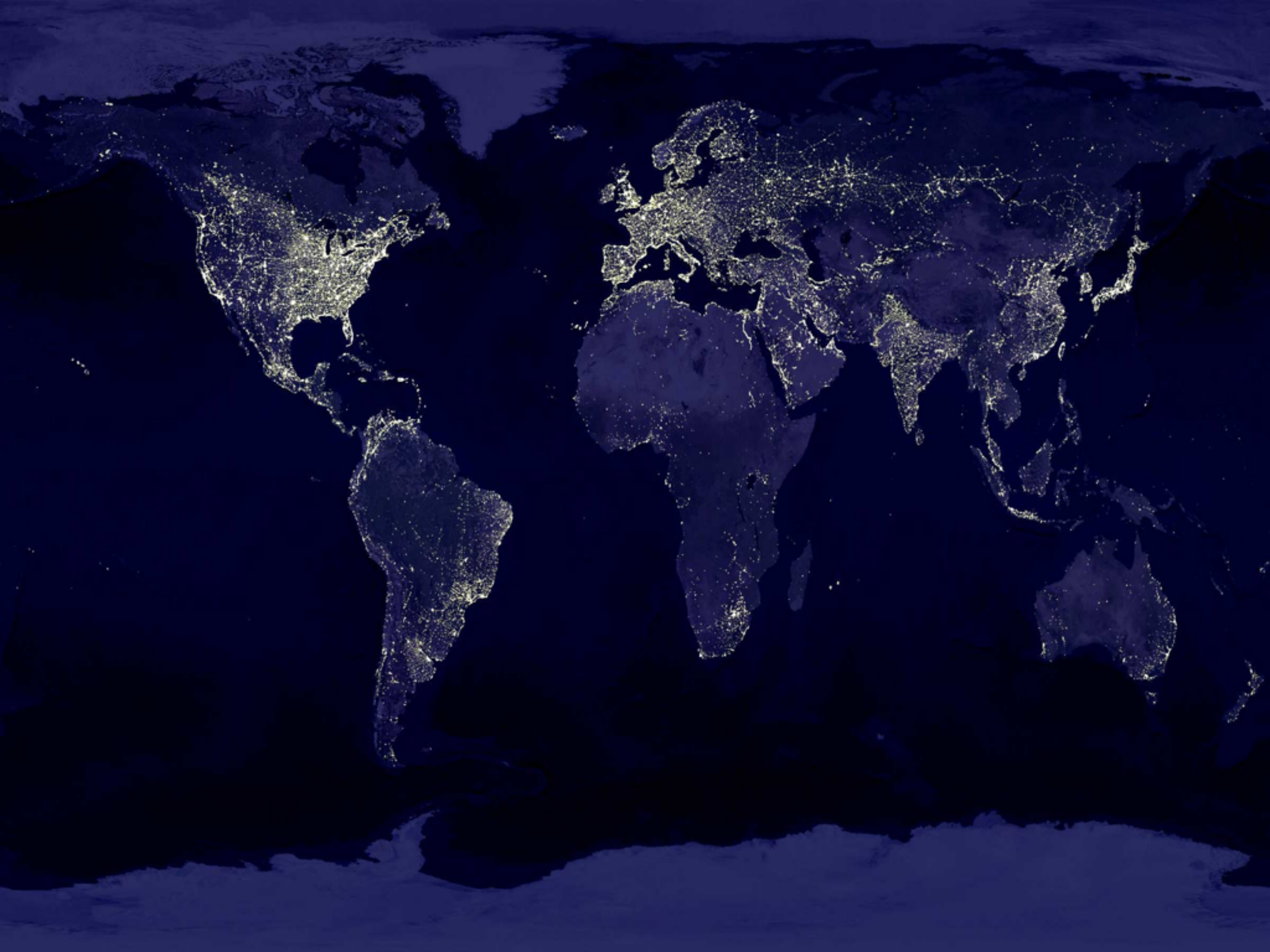
I found my son's killer.

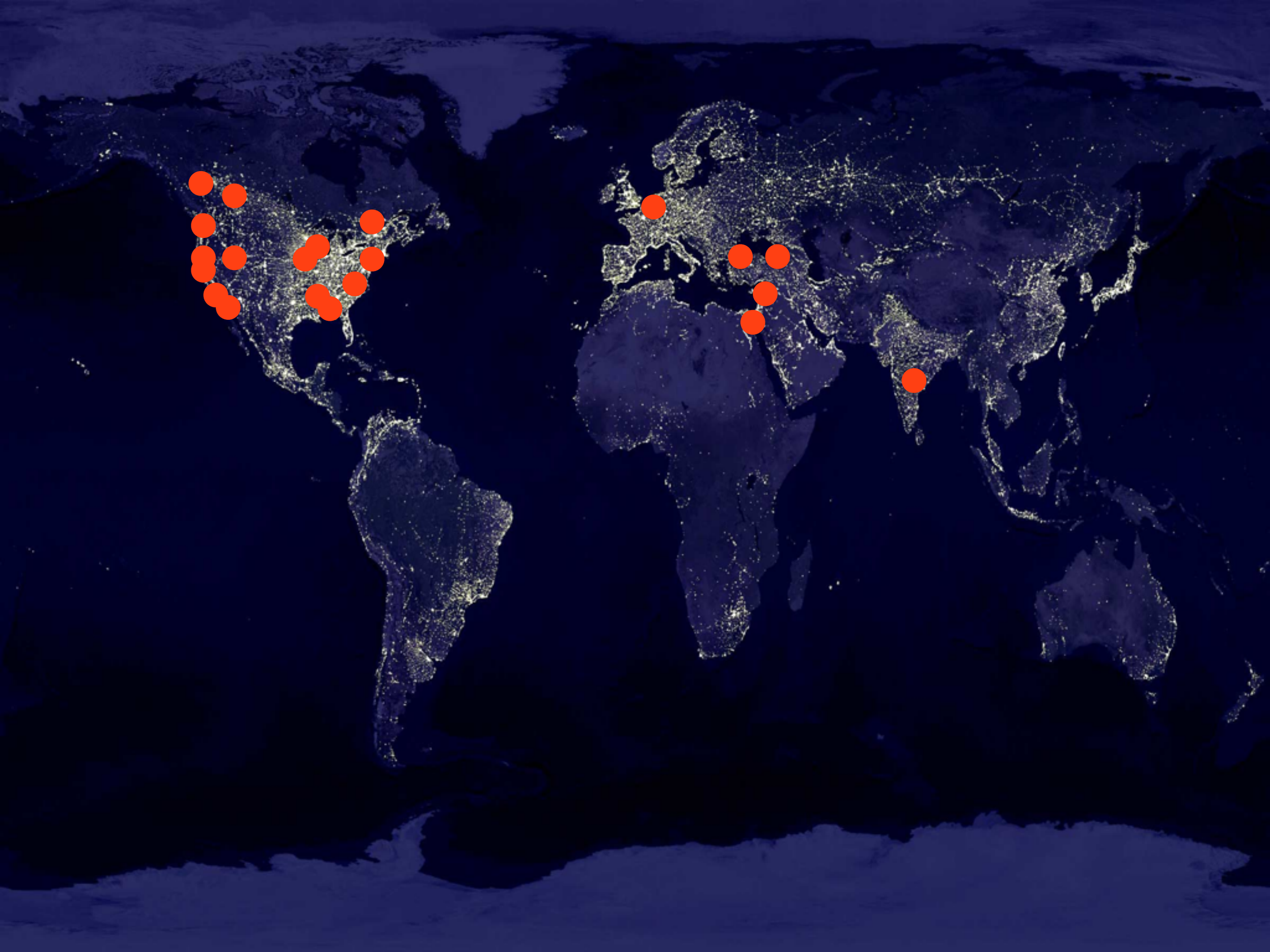
It took three years.

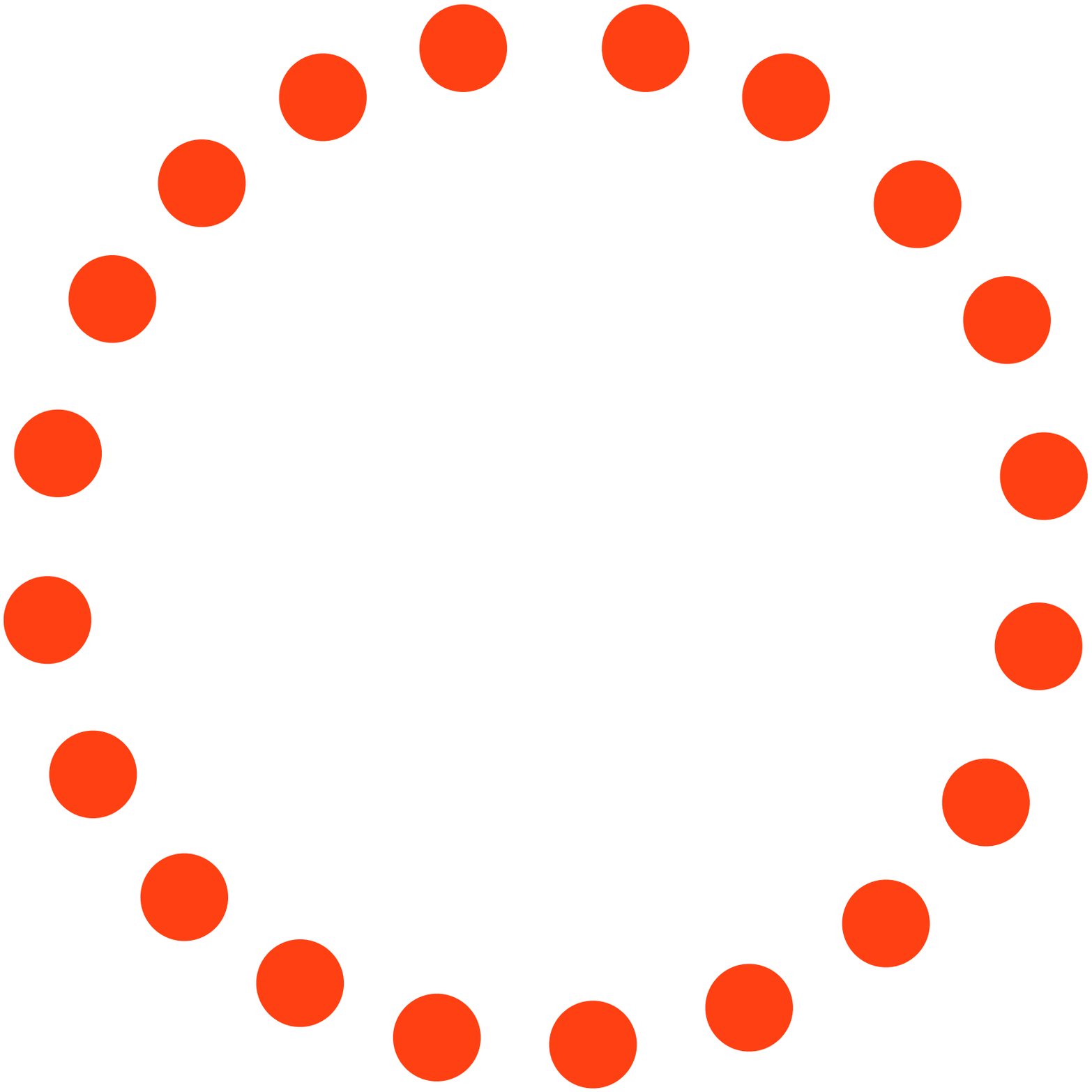
But we did it.

Not quite like this.

2,000,000+







20

National Institutes of Health

CDG Natural History

PATIENT PORTAL

Check-In

DUDES

DUDES
CAPE
HAARTJE

Pocket
Mask

EXIT

Patient | Examination | Postprocessing | Documentation

MIGHT BERTRAND

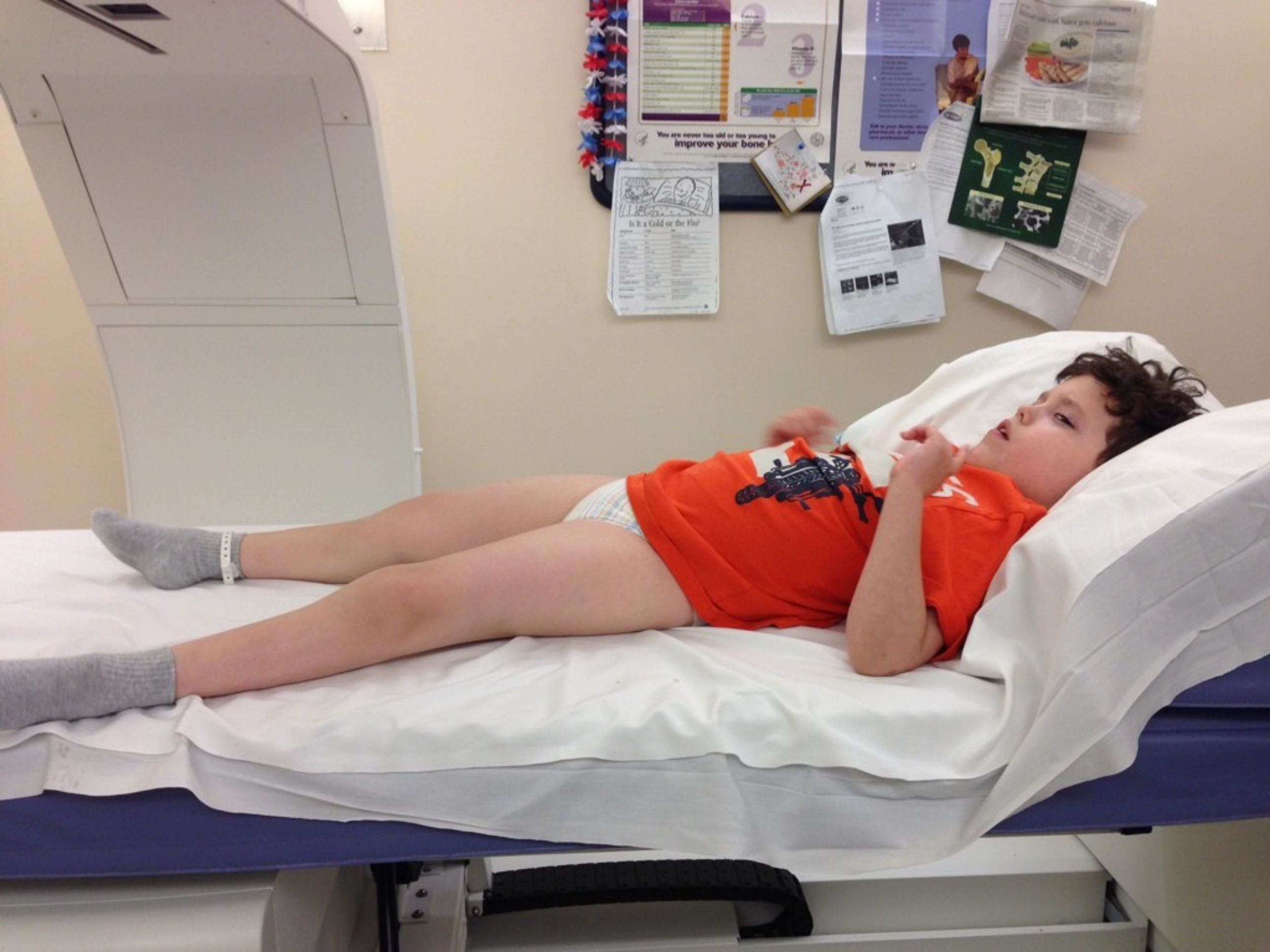
Barium
Single shot
Single shot
Continuous
25 f/s
1000
0
0

Control

Fluoro: Cu 0.0mm
Acquis: Cu 0.0mm
25.4 cGycm2

LIH

CYCLE
ES


Is It a Cold or the Flu?

CYCLES

DELIVER TO

CYCLES

N.I.H. MEDICAL RECORD: U.S. GOVER
TO BE OPENED BY ADDRE
RUN

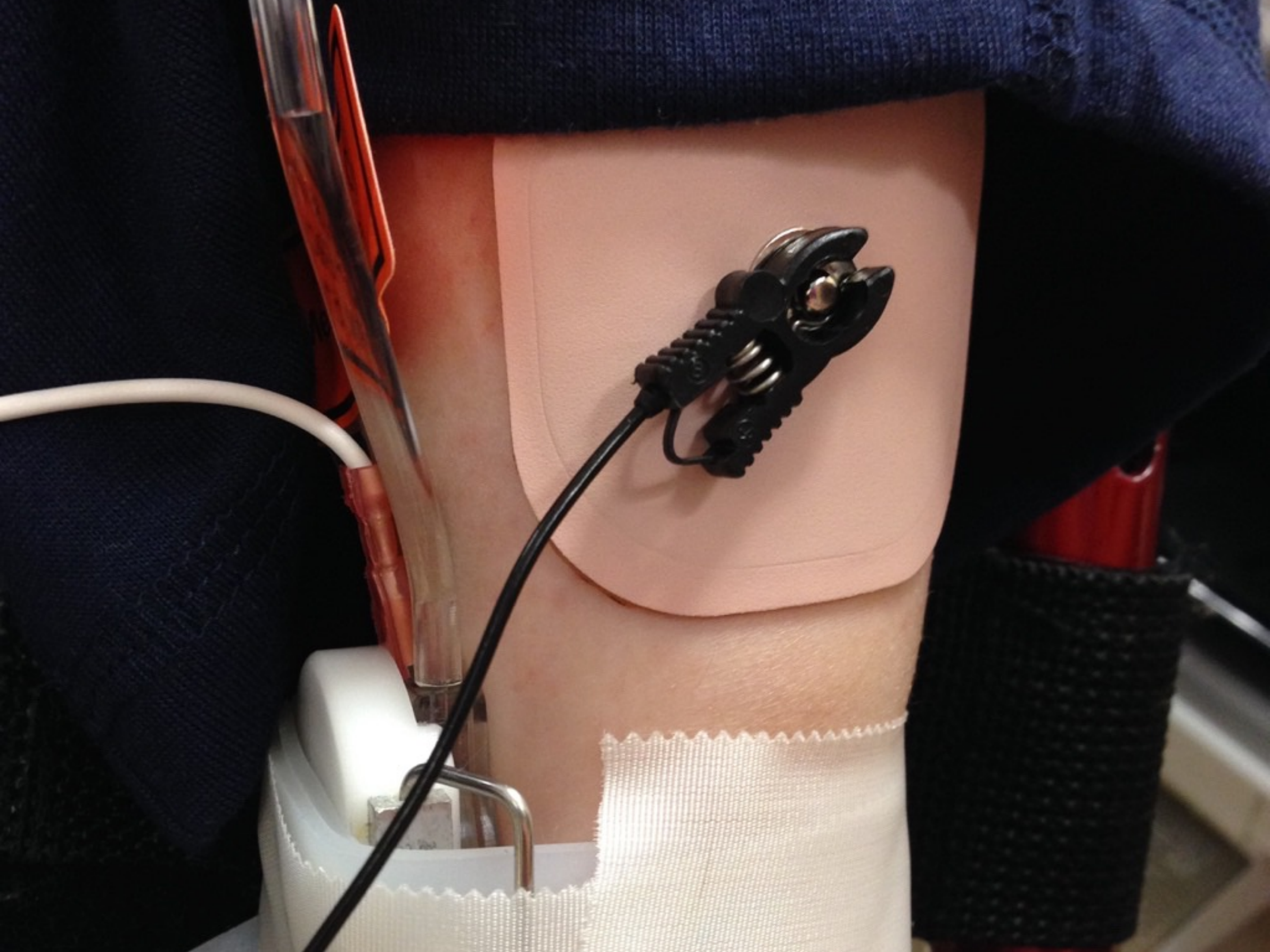
BRING

ALARIS
65

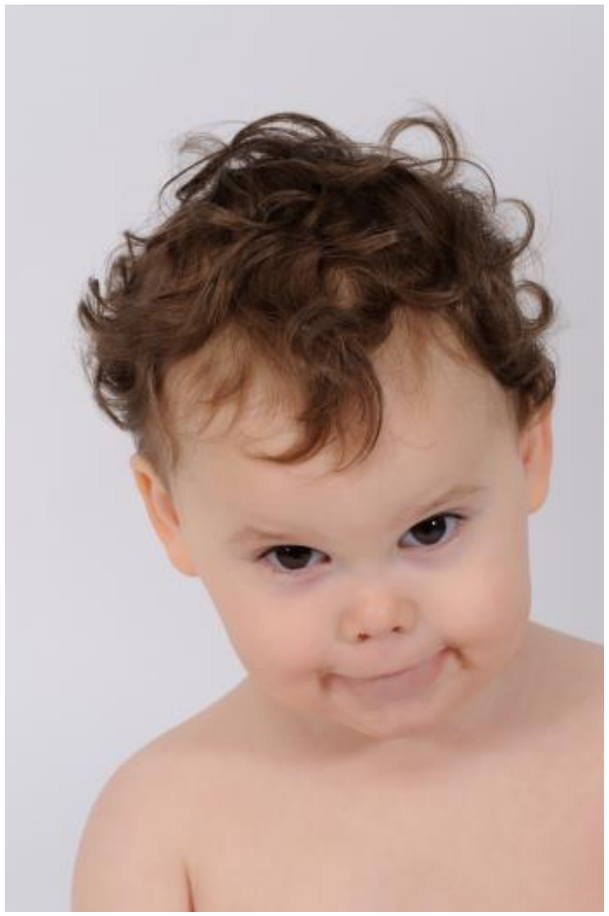
DELIVER TO
GOVERNMENT PROPERTY
ESSEE ONLY

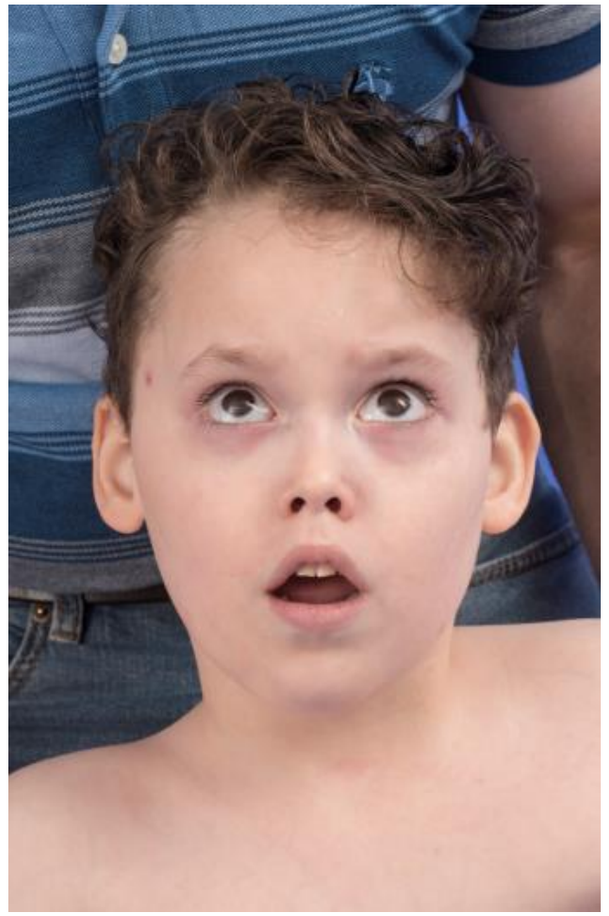
MAGNETOM Skyra
SIEMENS

SECU
SCREE
BAGGA
ONLY



BRING





BRING
IT ON
72

BRING
IT ON
72

BRING
IT ON
72

BRING

Biomarkers!

NGLY1

Oligos

GPI

NGLY1

Mito

CSF protein

Community science

Nature

Patients

max(mom,dad)

max(mom,dad)

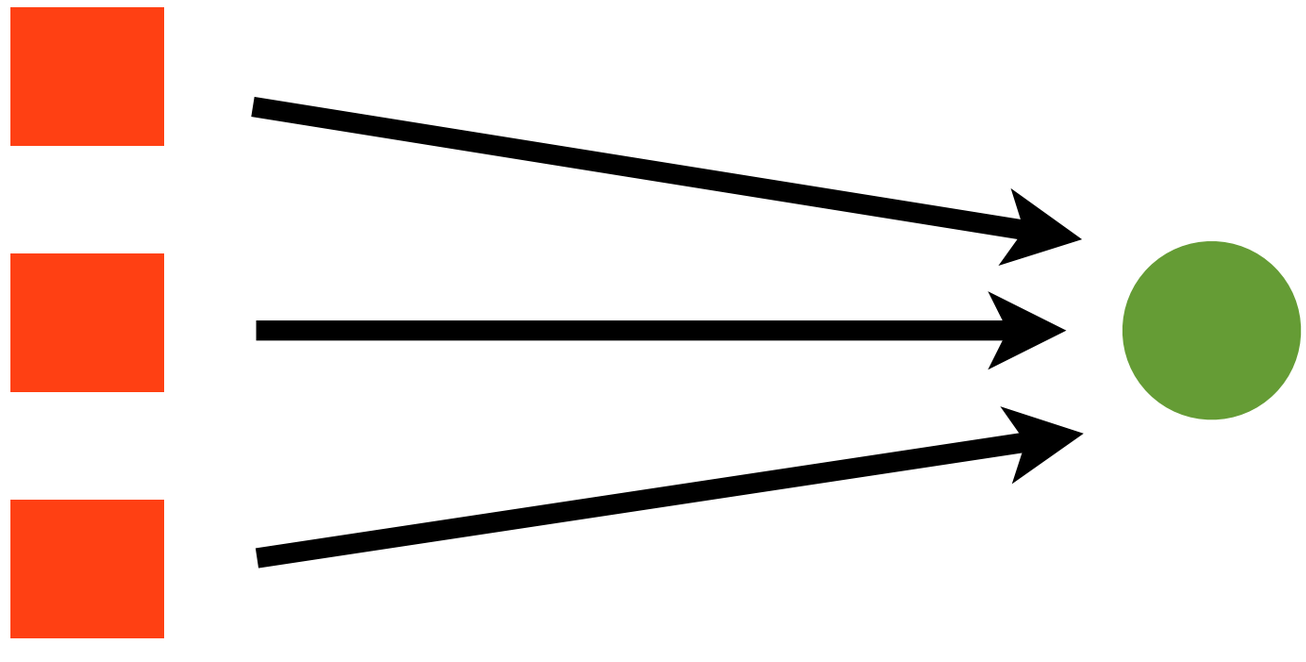
max(mom,dad)

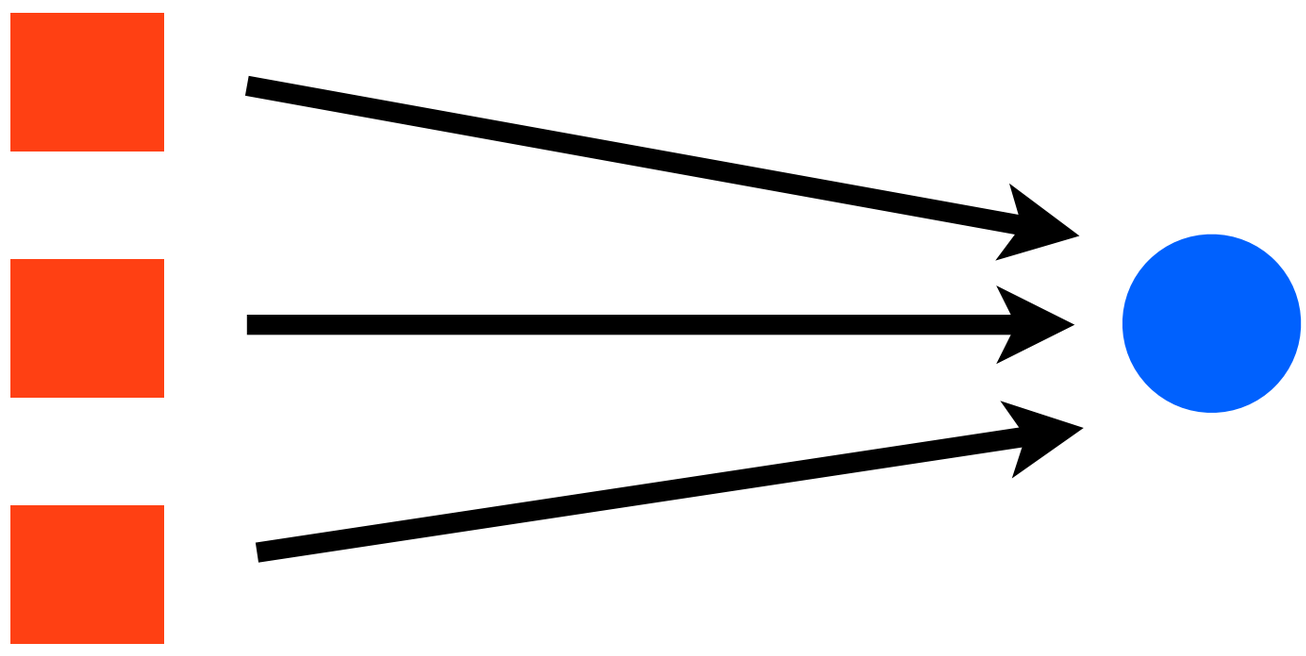
active site

Severity

More

Less





KEEP

CALM

AND

GLYCO

ON