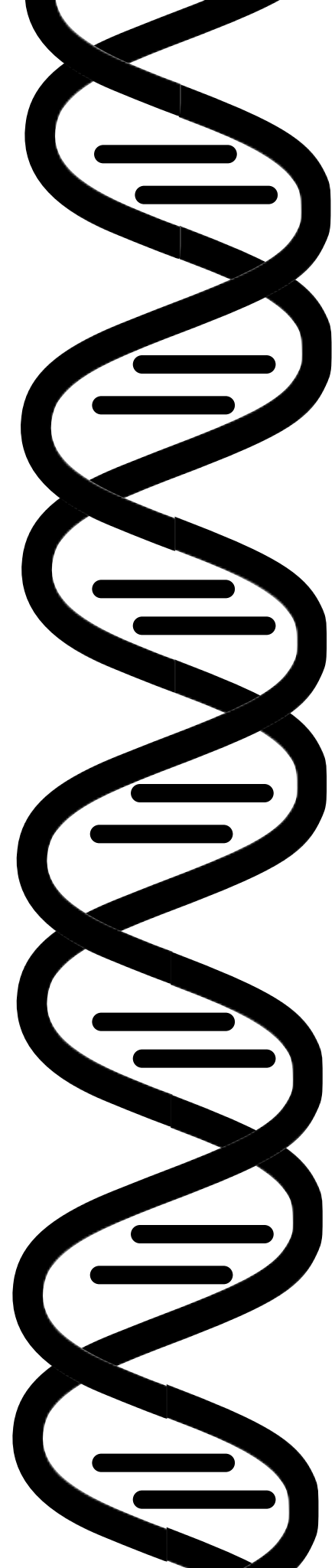


Matt Might, Ph.D.
Associate Professor
Computer Science
University of Utah
matt.might.net
[@mattmight](#)



Patient Experience; Patient Engagement

Matt Might, Ph.D.

Associate Professor

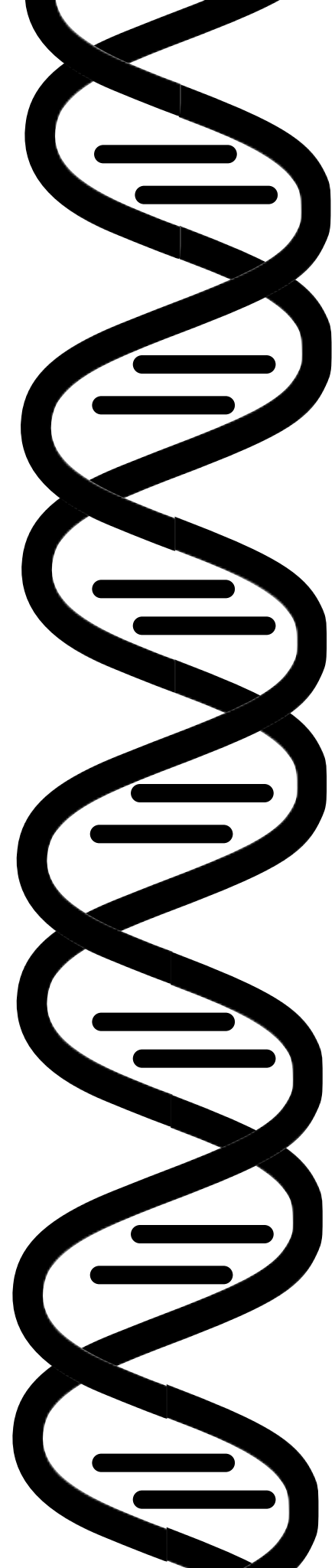
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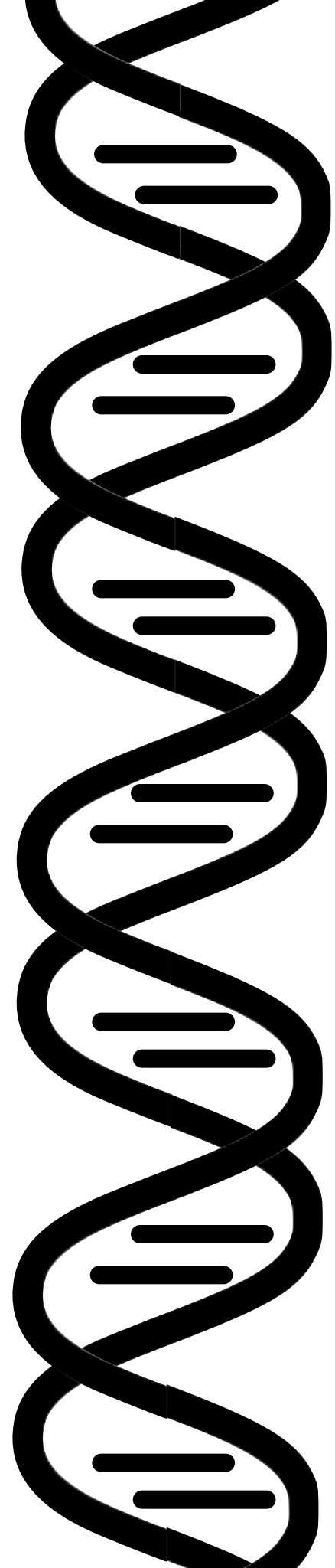
Associate Professor

Computer Science

University of Utah

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[@mattmight](https://twitter.com/mattmight)



Patient Experience; Patient Engagement

Matt Might , Dad







Undiagnosed

Patient Experience

Aftermath

“the island”

Patient Experience

Aftermath

“the island”

Logistics & Education

Aftermath

“the island”

Logistics & Education

Internet & Action

Undiagnosed Island





























4.5



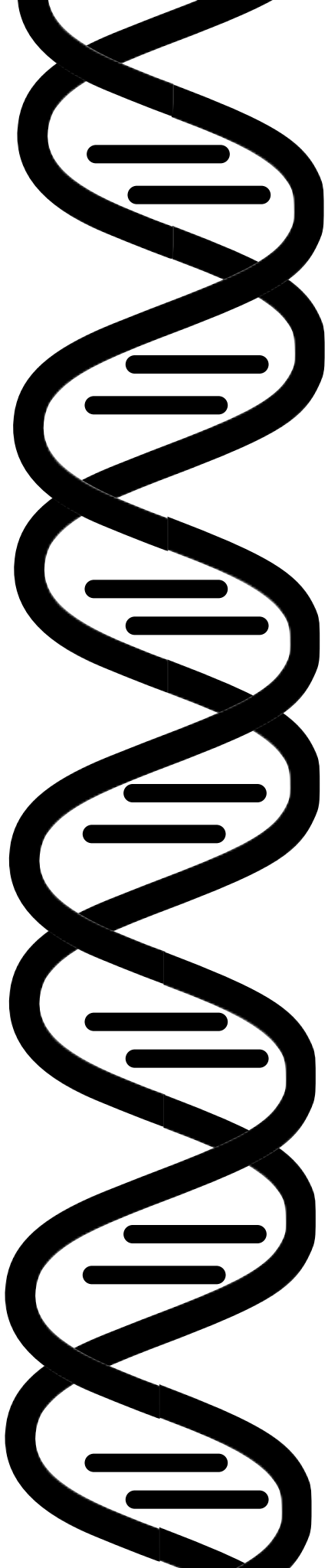
David Goldstein

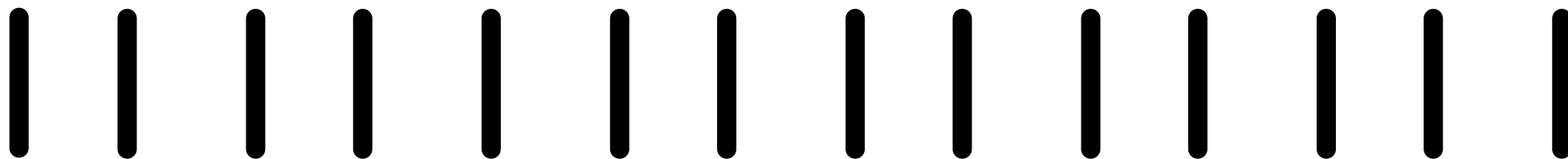


Kelly Schoch

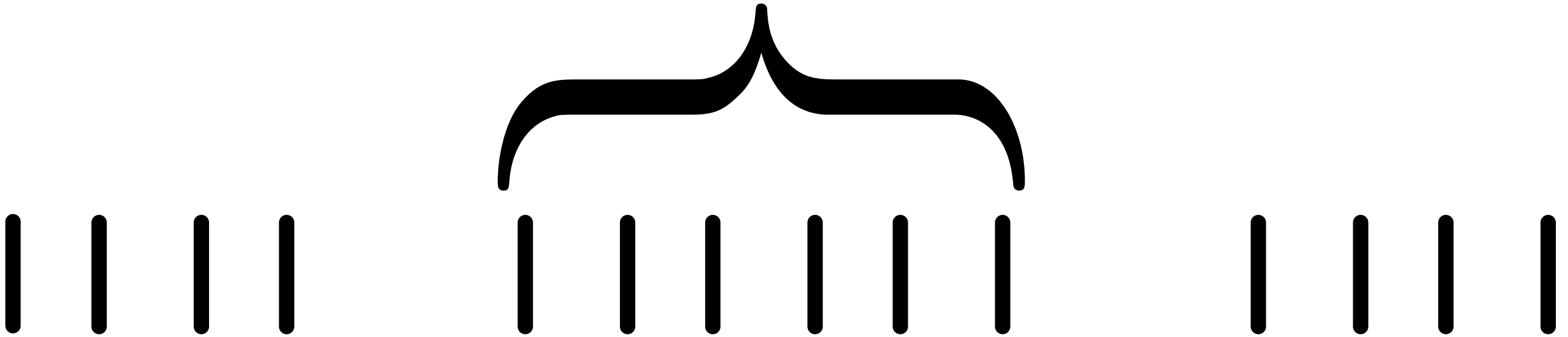


Vandana Shashi

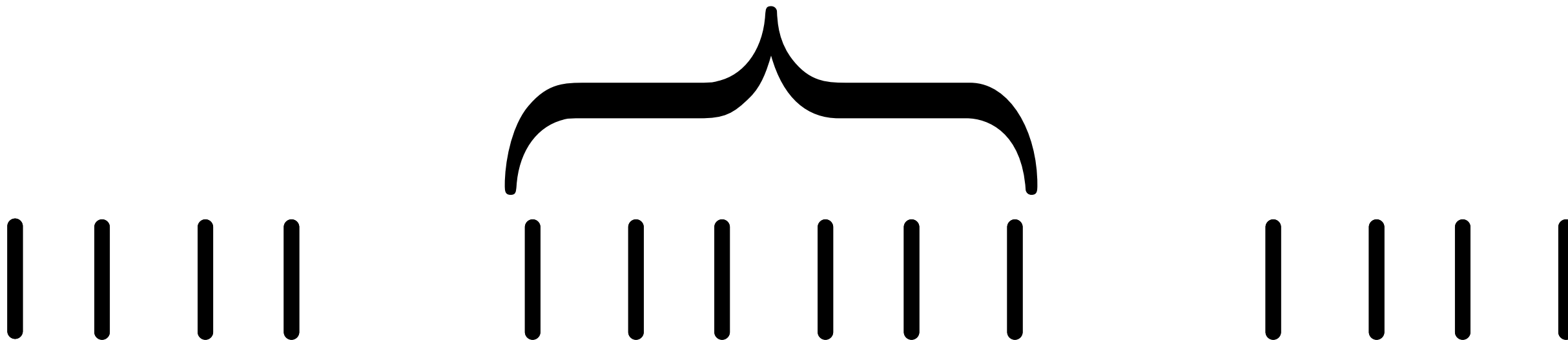




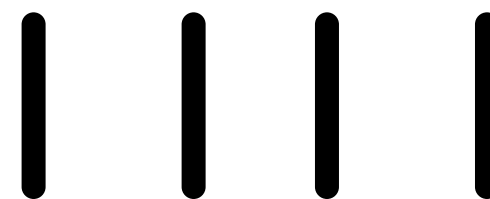
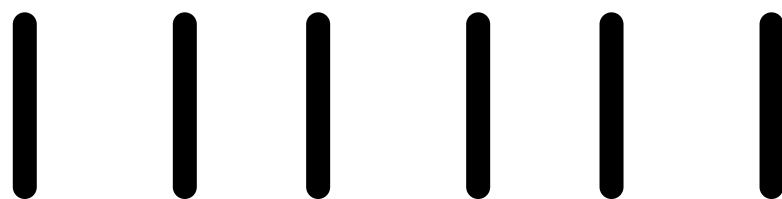
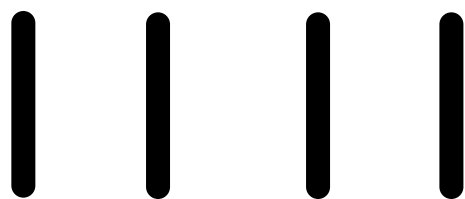
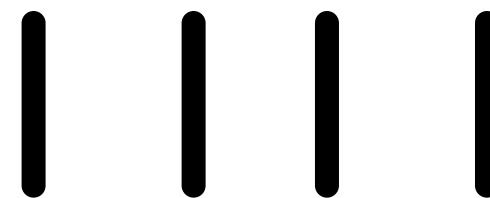
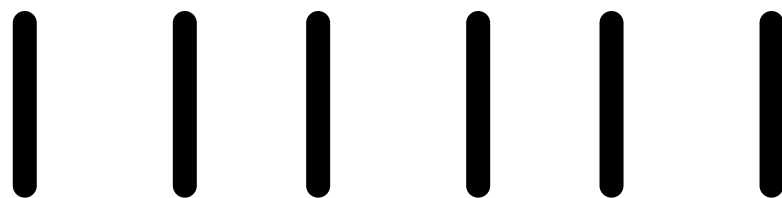
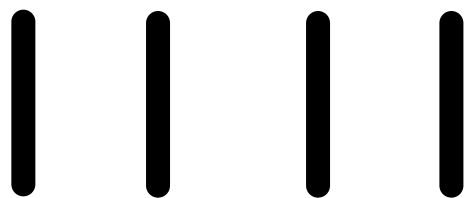
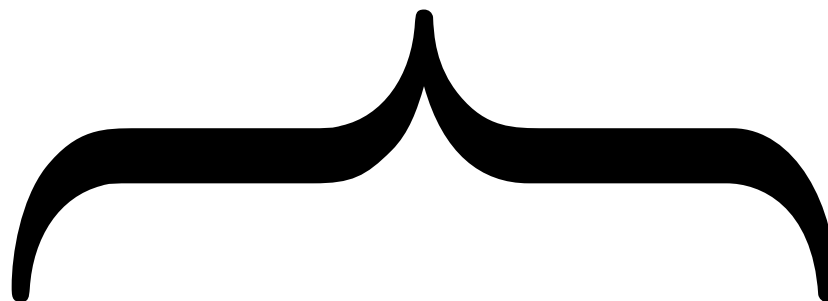
NGLY1



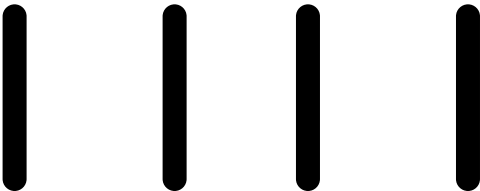
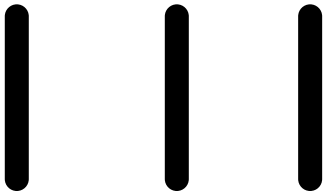
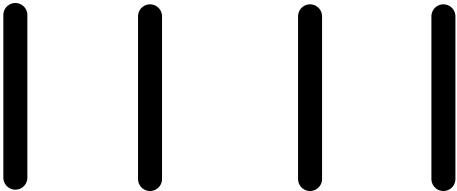
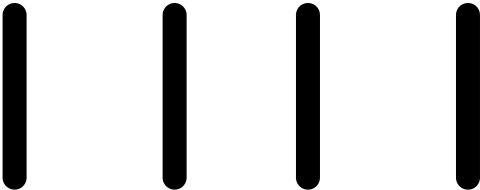
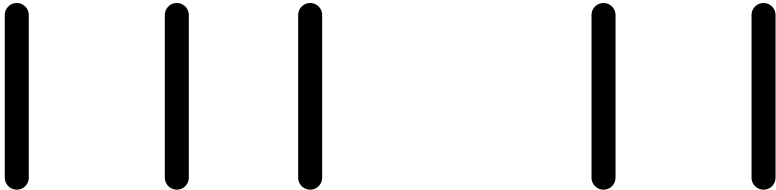
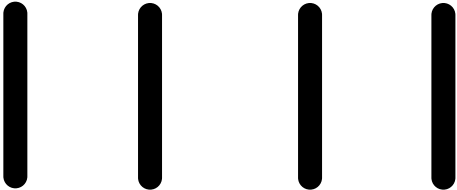
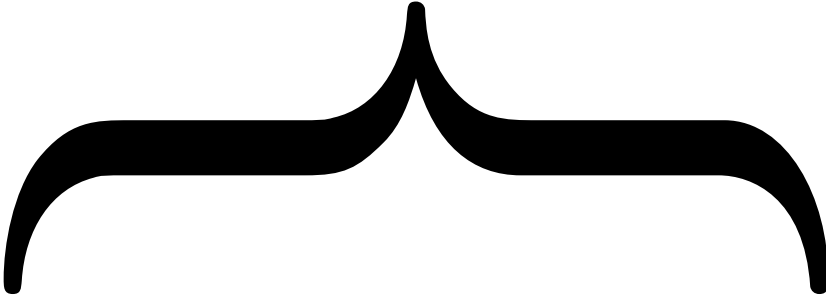
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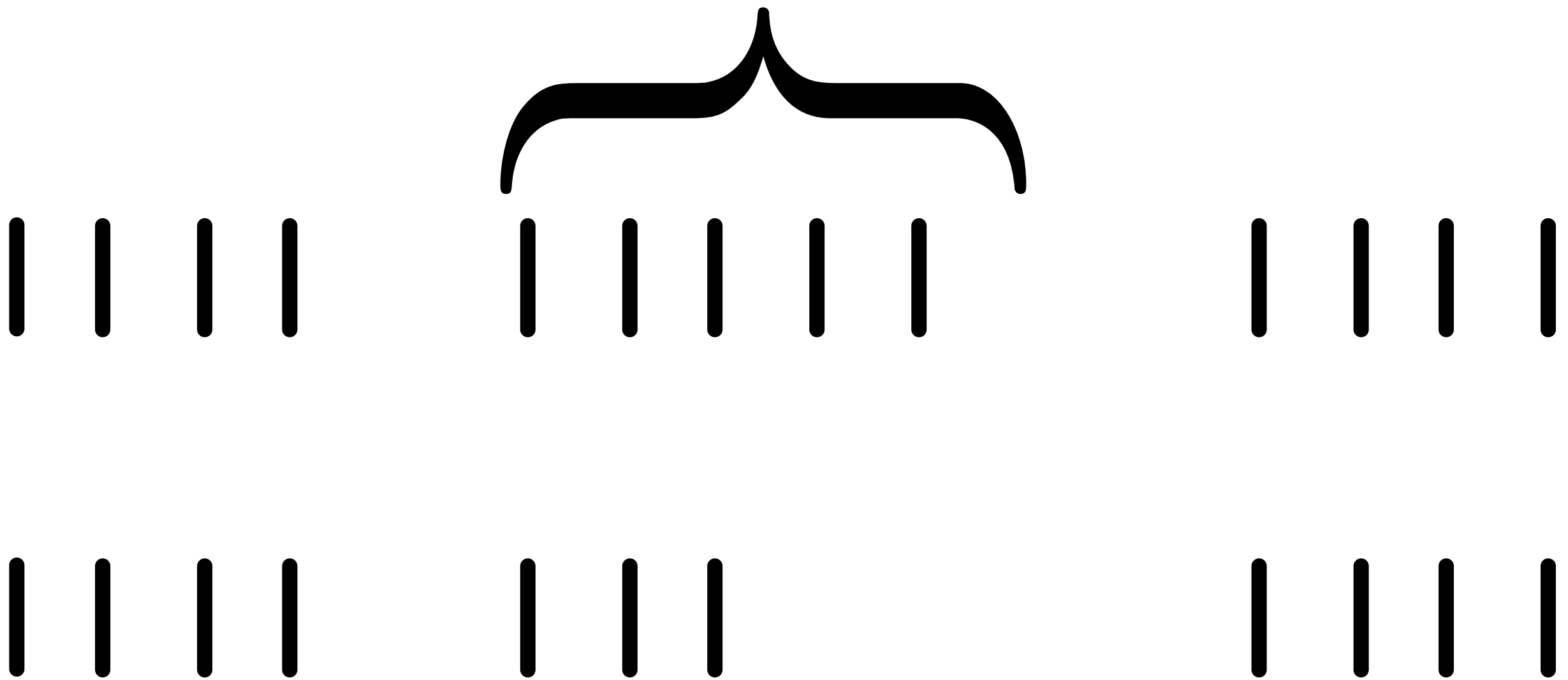
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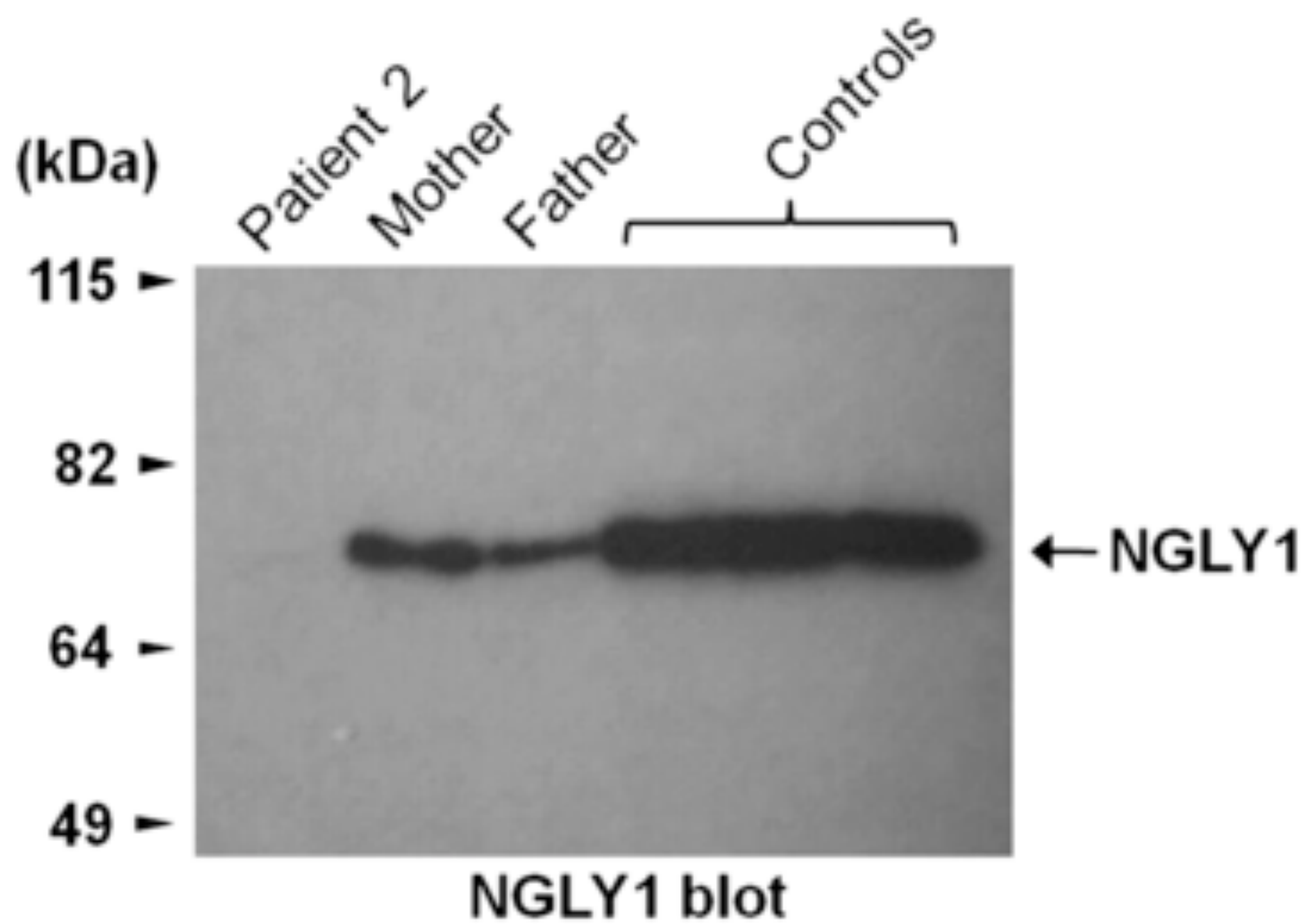
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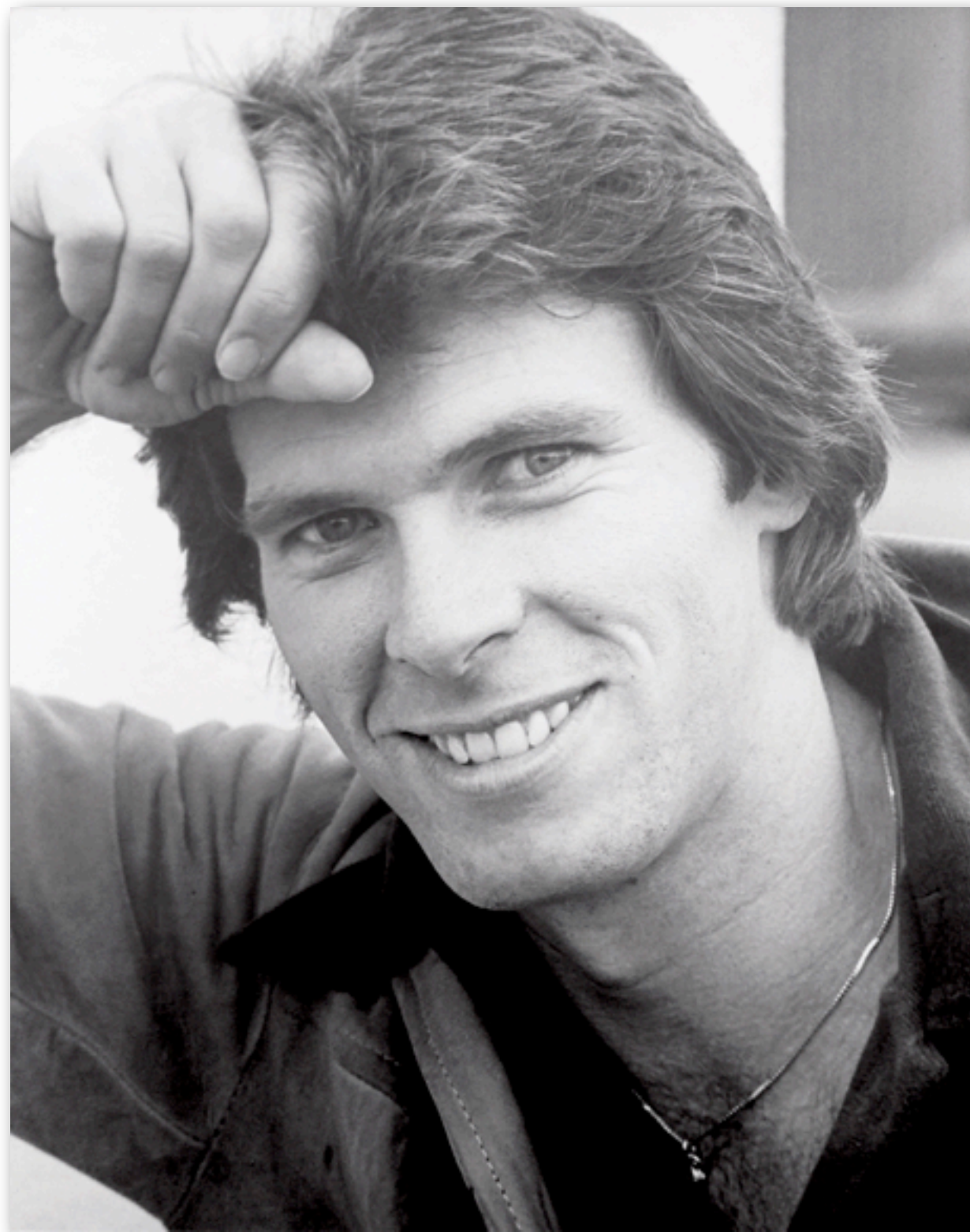


NGLY1

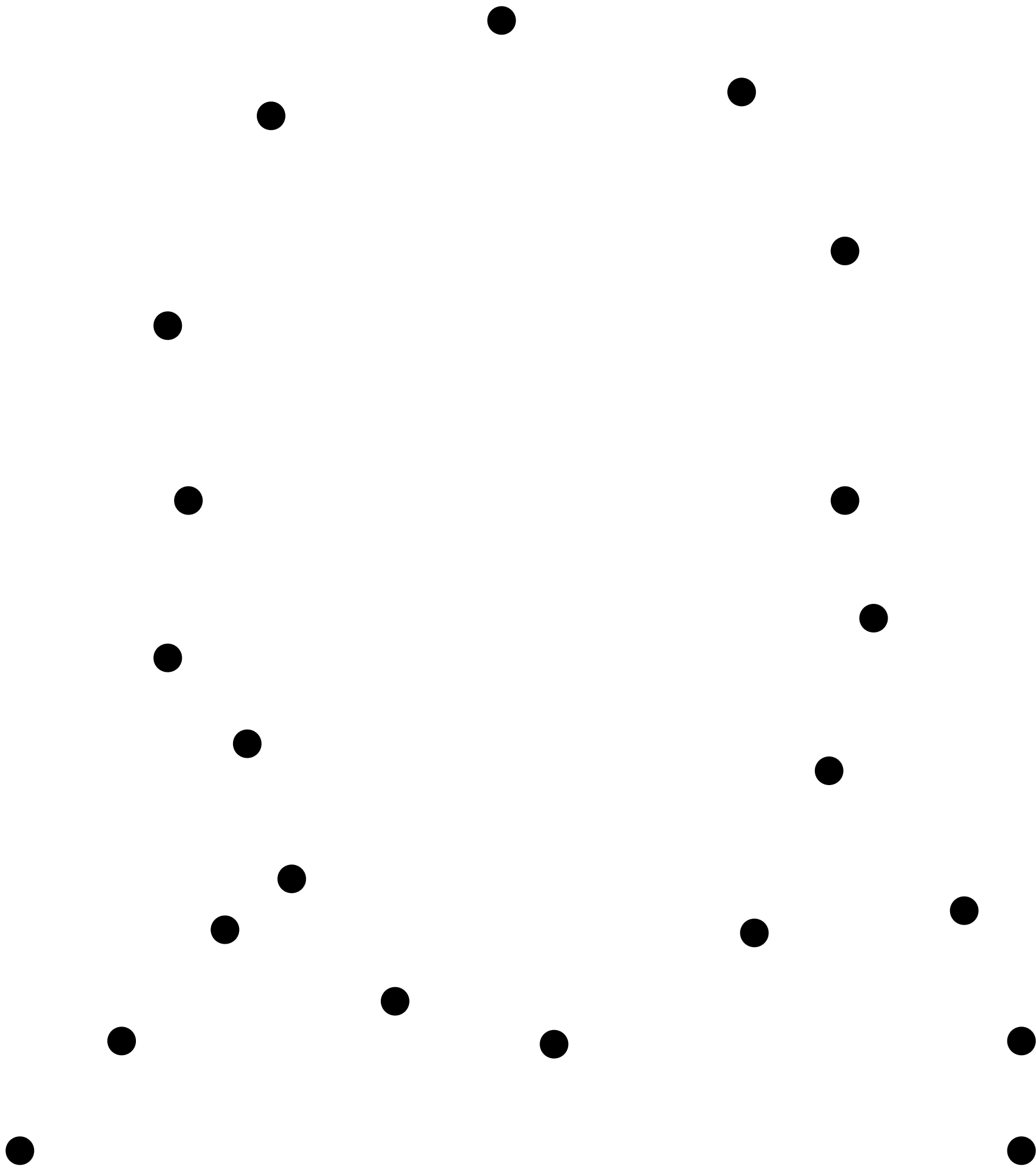


What if $n = 1$?





Hudson Freeze, Sanford-Burnham







Undiagnosed

Patient Experience

Aftermath

Patient Experience



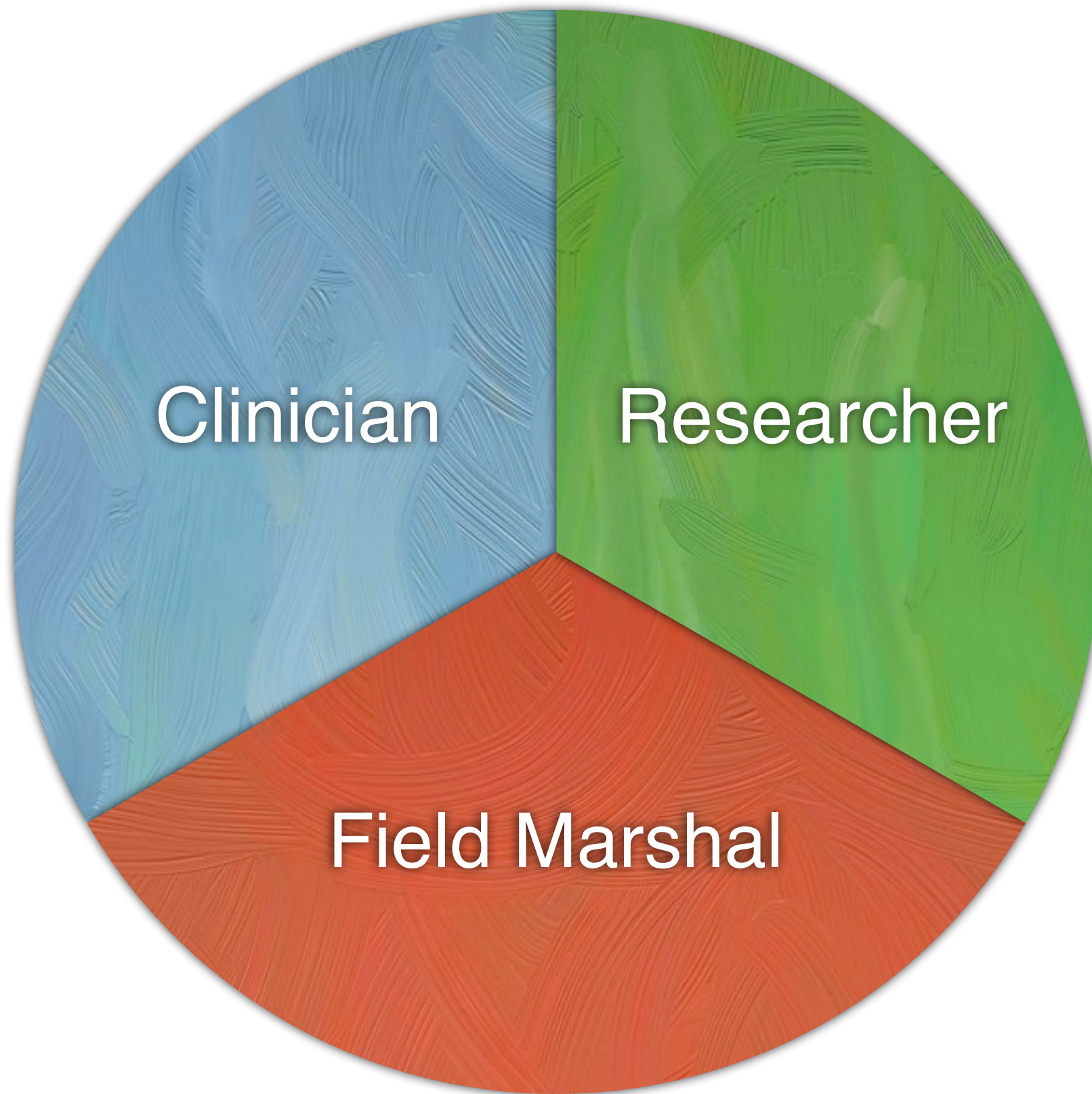




Logistics

Lynne Wolfe





What's it like?















Qty: 100
Reorder No. 8820000
Test Blue
Store in a cool, dry place.

WEST PAS UN











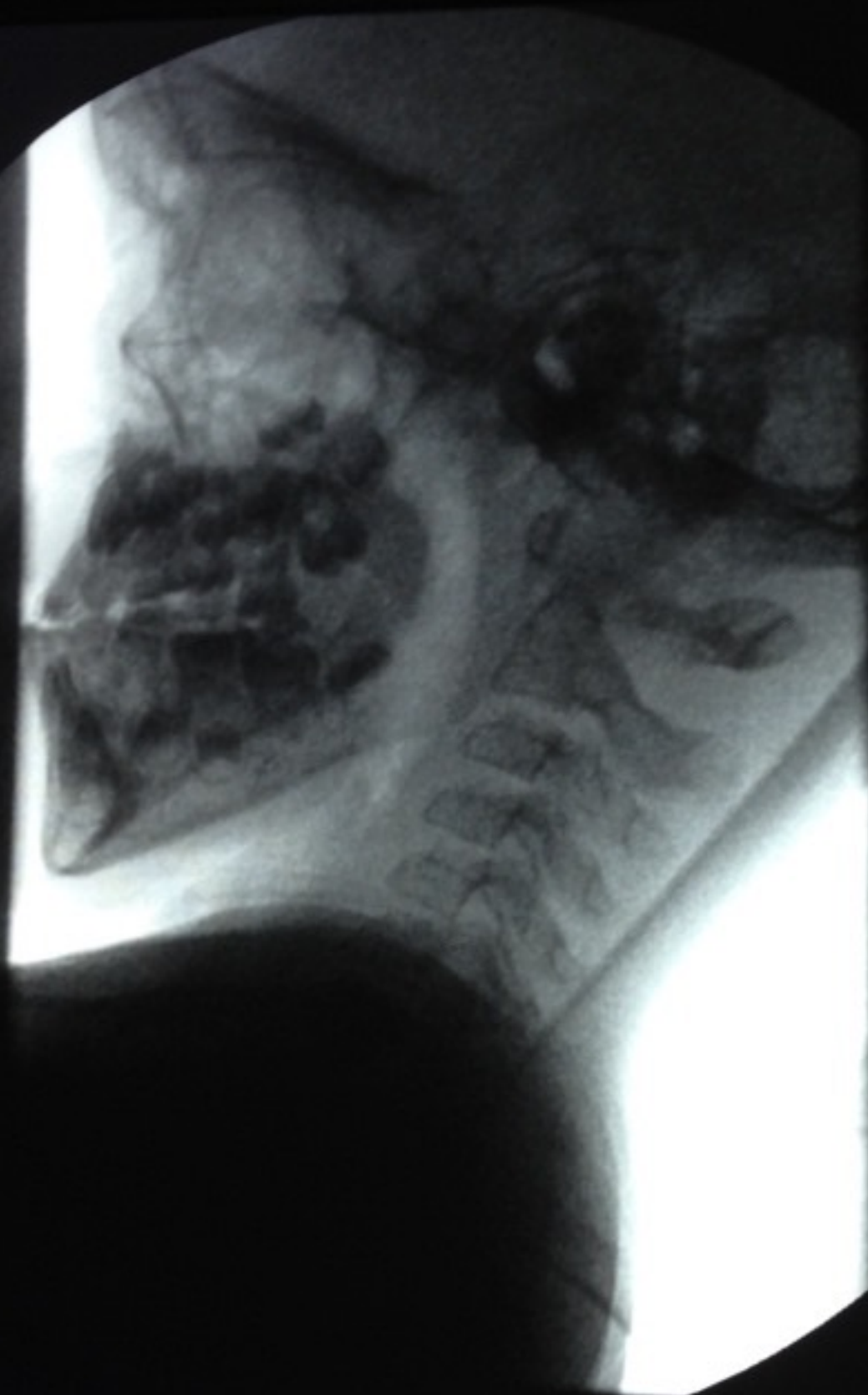












MIGHT
BERTRAND

Barium Single shot
Single shot
Continuous 25 f/s

0 0 1000



Patient

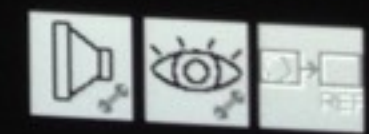
Examination

Postprocessing

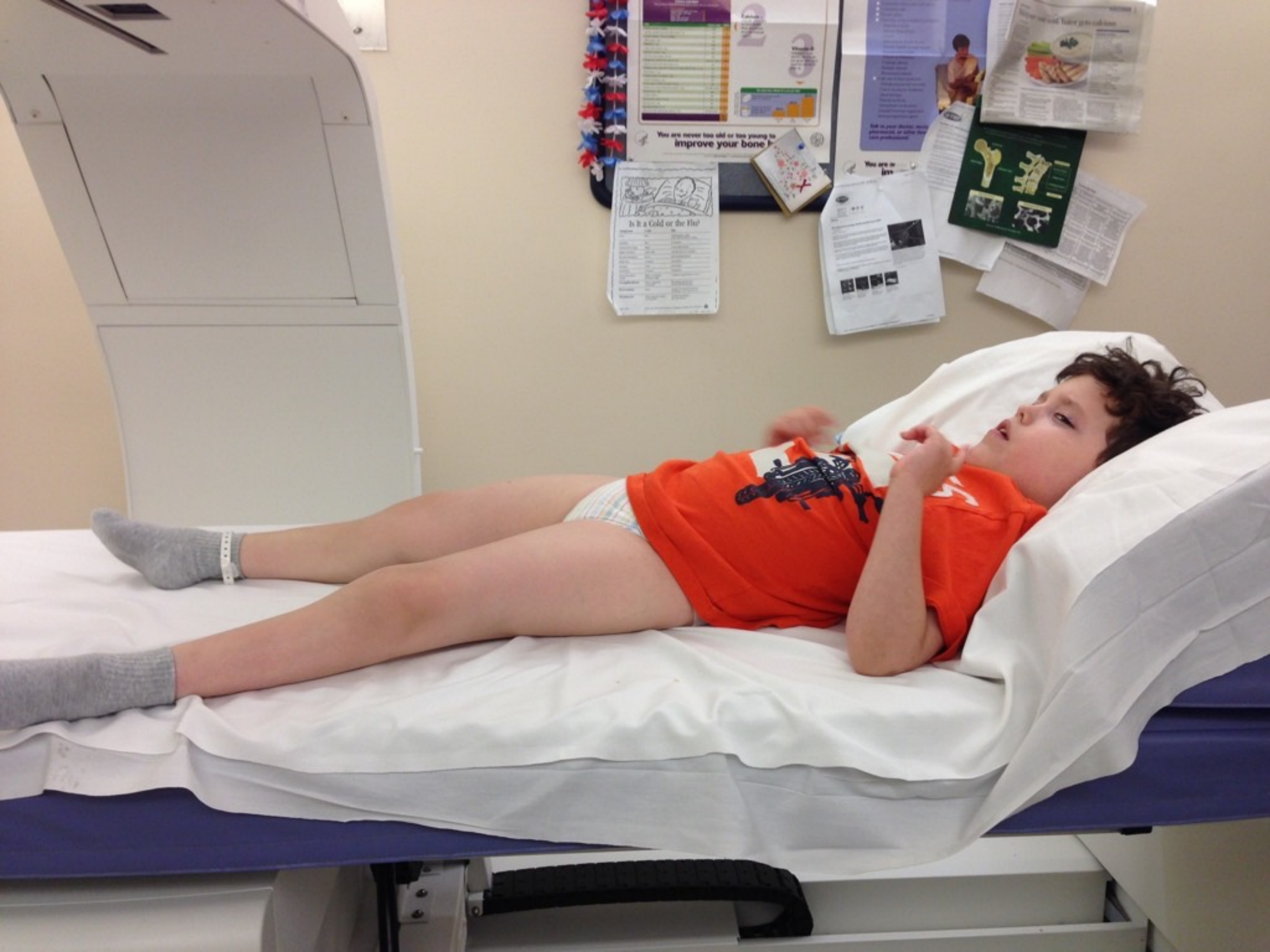
Documentation

LIH

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

































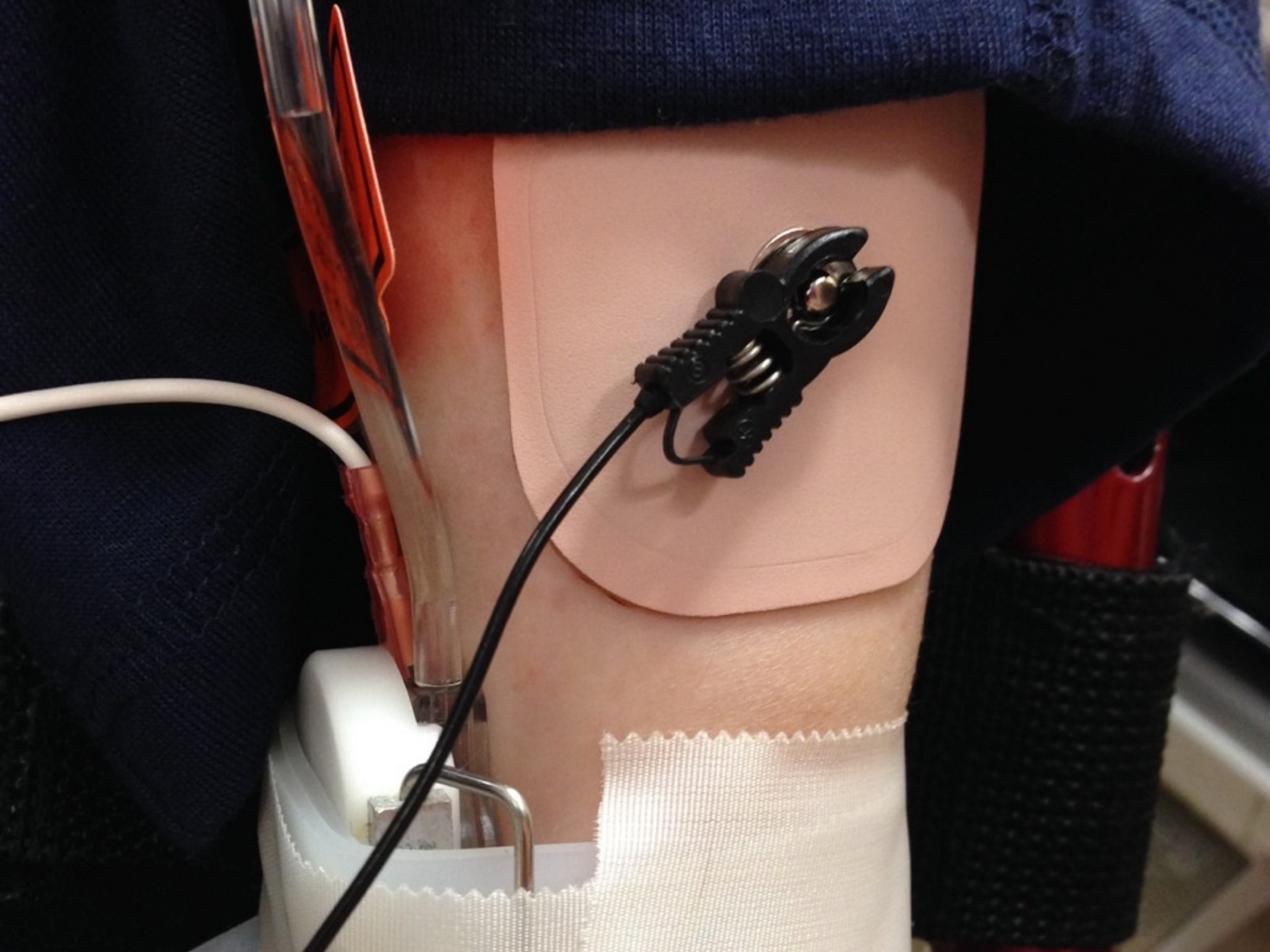
















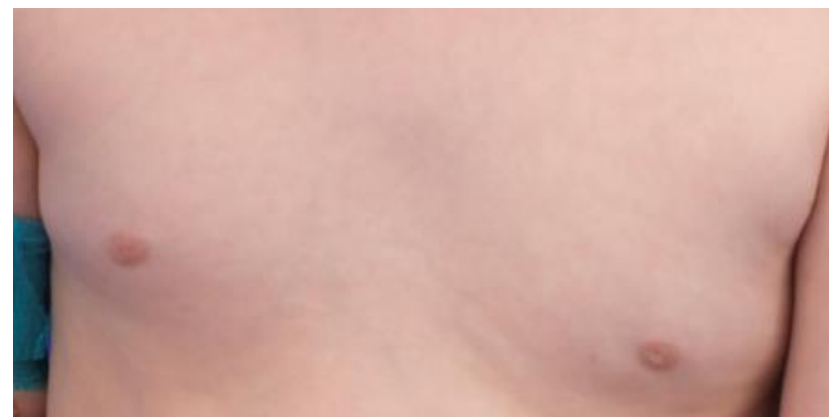


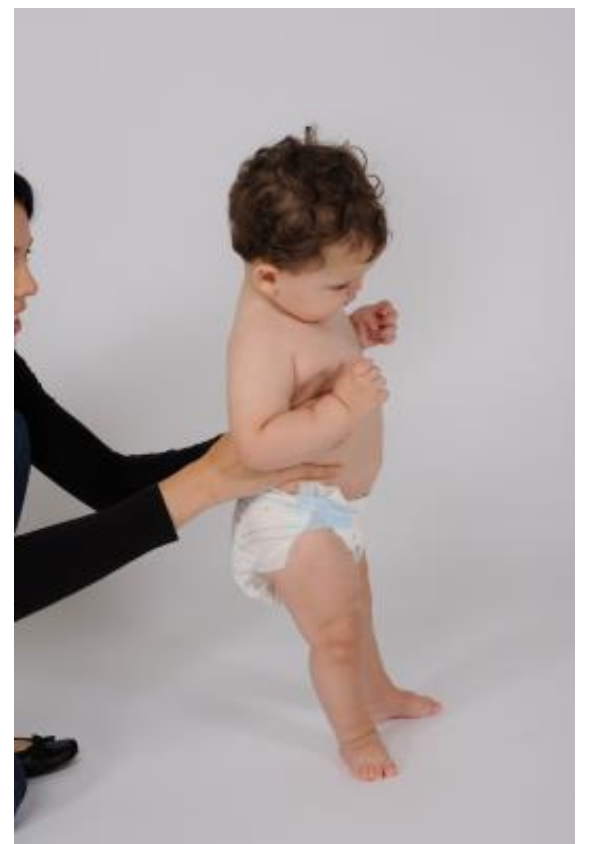
















**BRING
IT ON
72**





How to improve?

Exhaustion.

It takes two.

science lessons.

pseudo science lessons.

“Homework” for patients.

Undiagnosed

Patient Experience

Aftermath

Aftermath





NGLY1

“Actionable”







FASTEN SEAT BELT WHILE SEATED

USE BOTTOM OPTION FOR FLIGHT

SKYY

Patient-initiated science



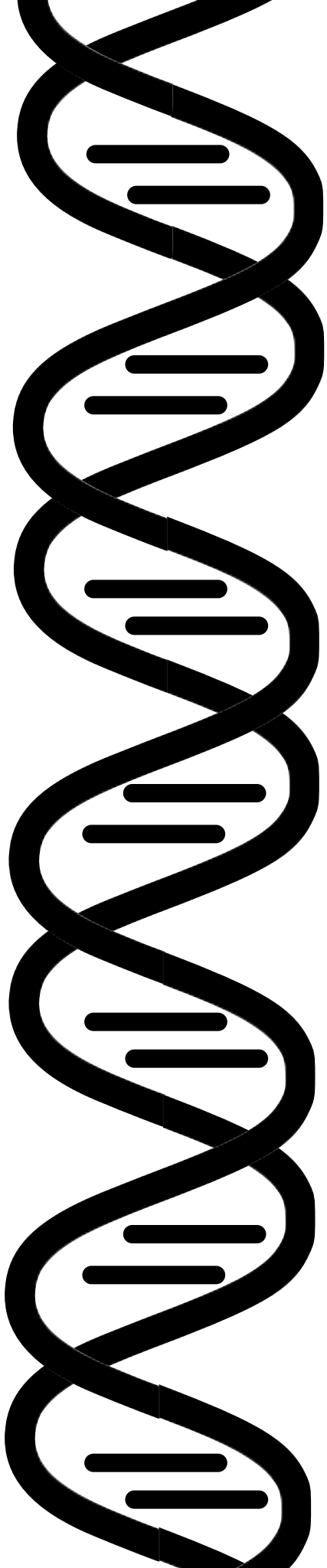
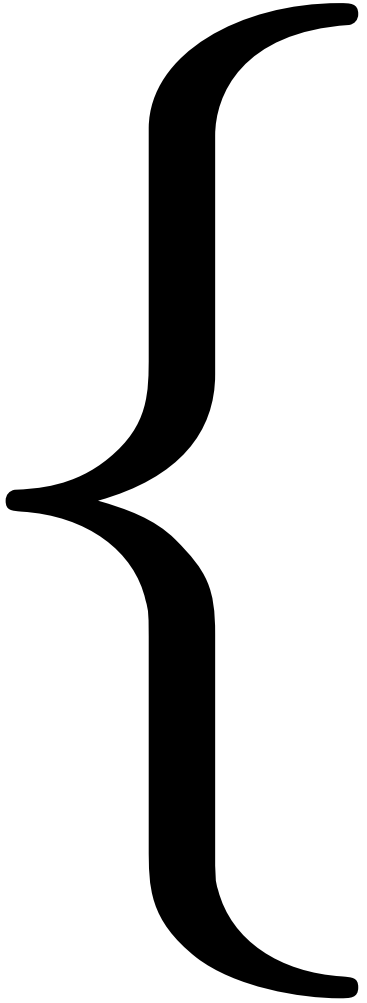


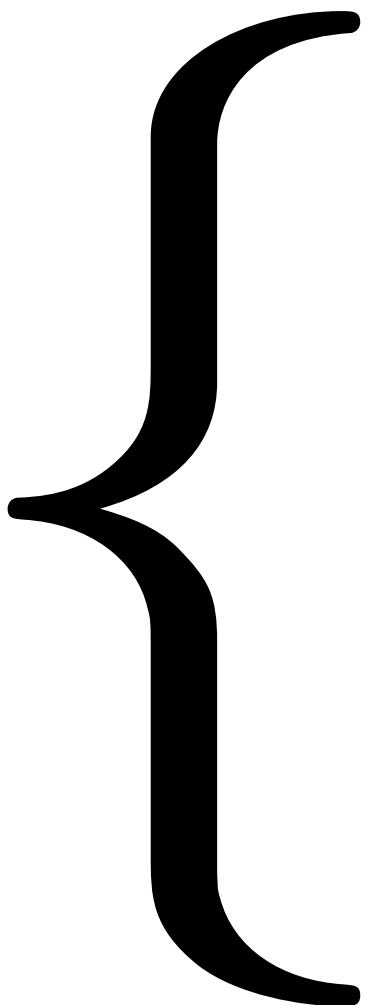
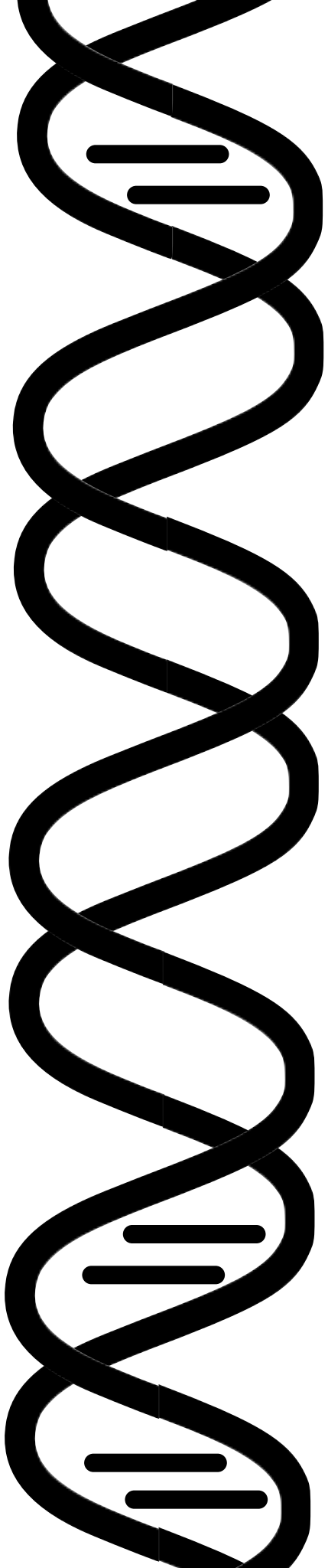
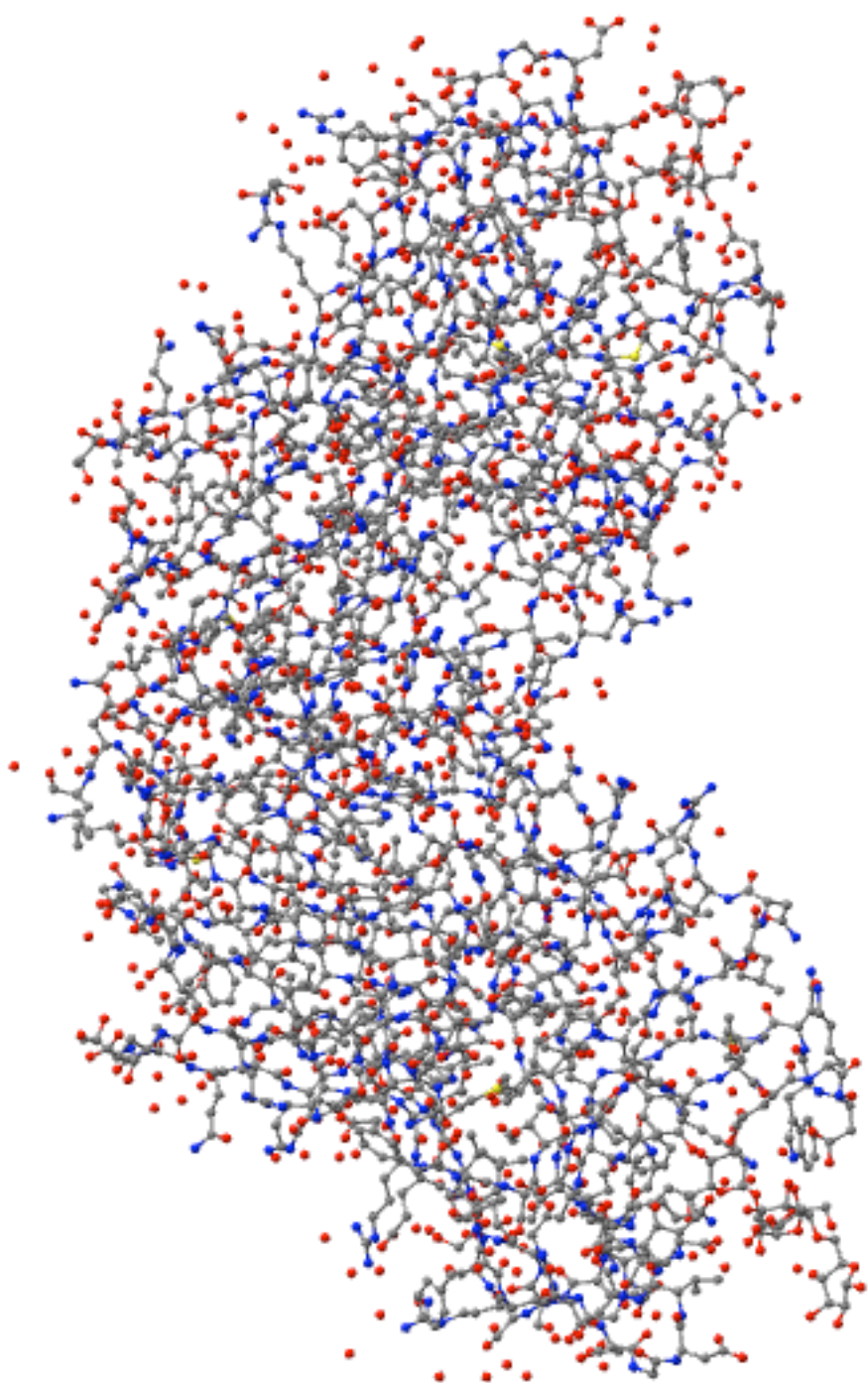




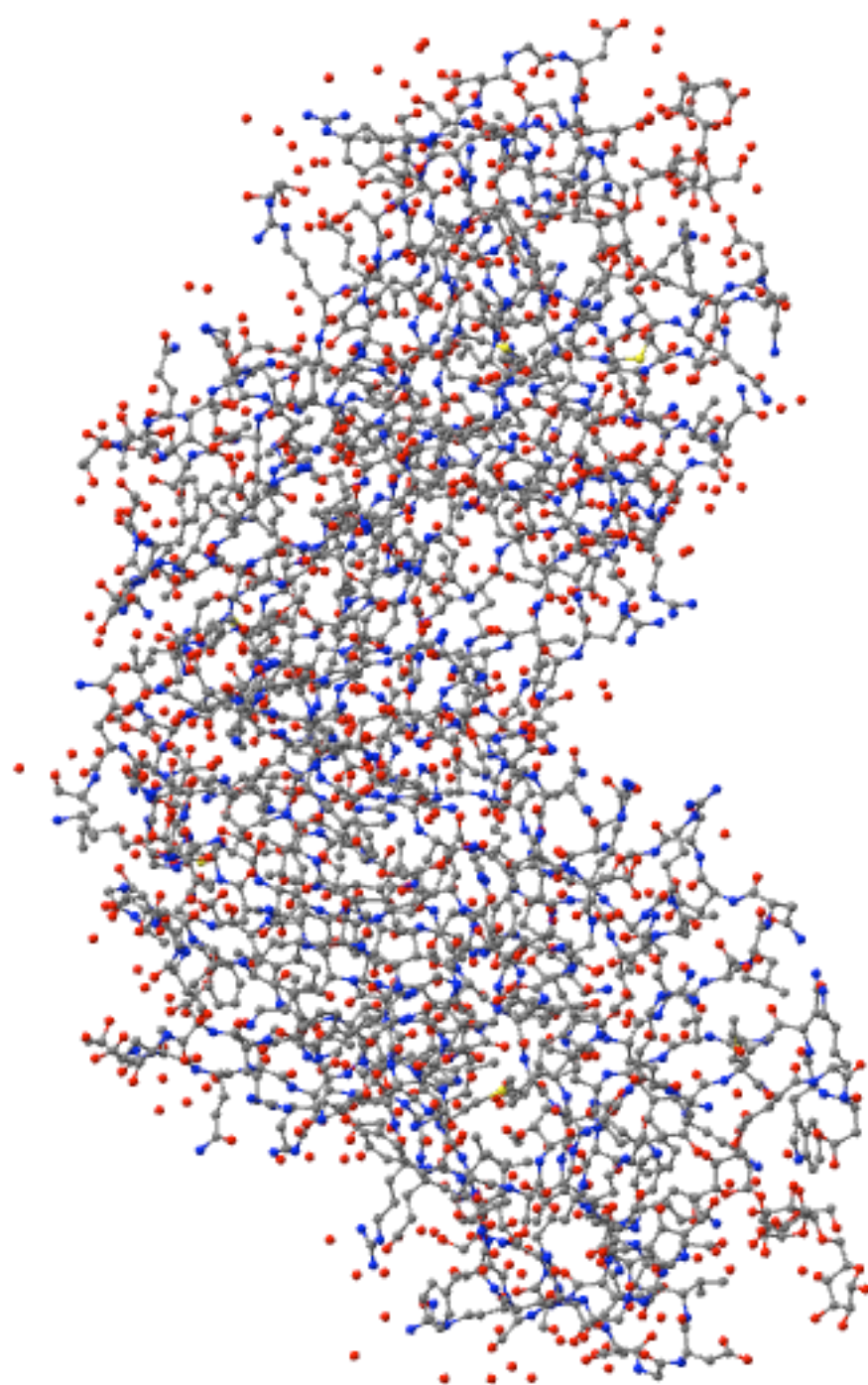


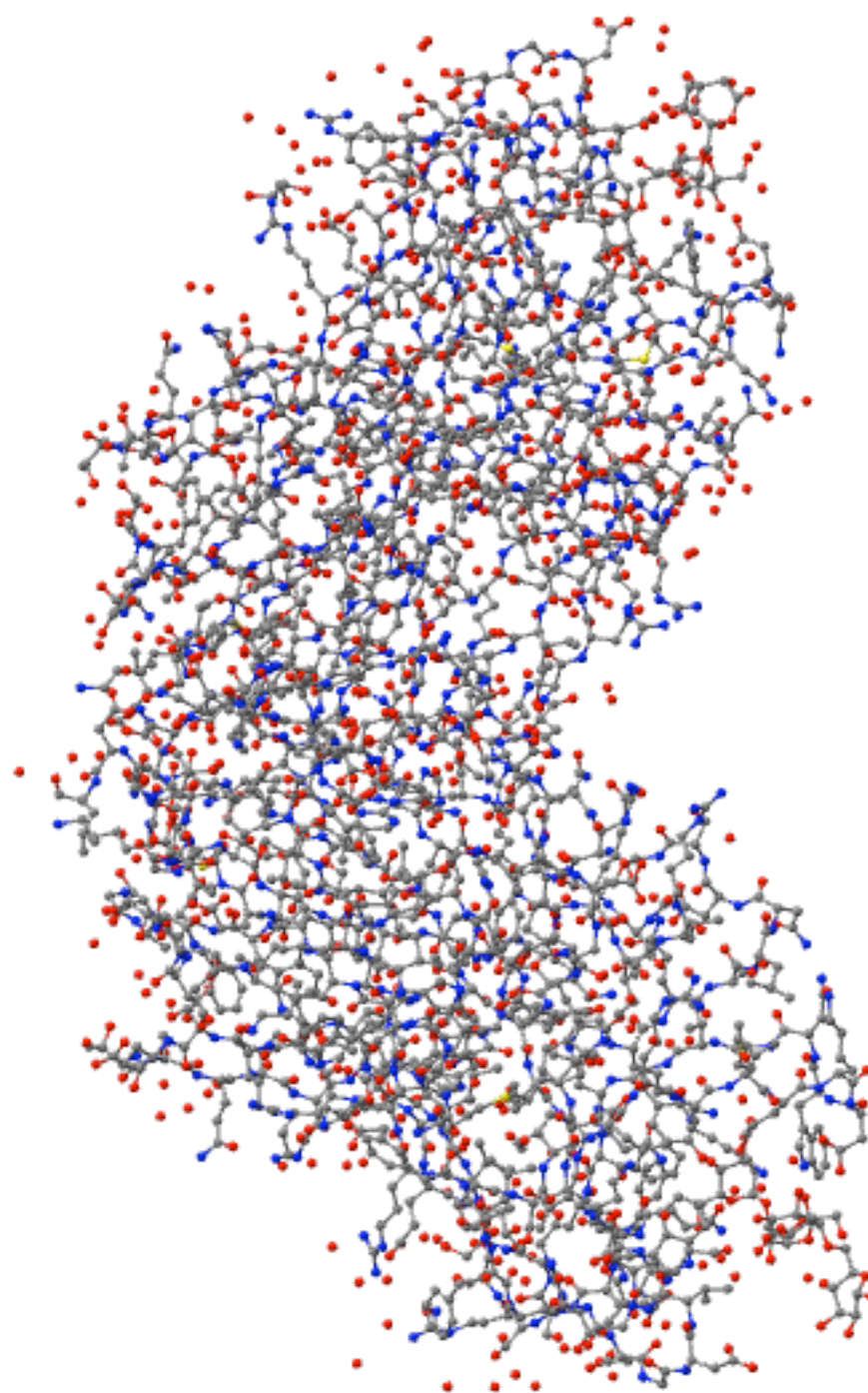
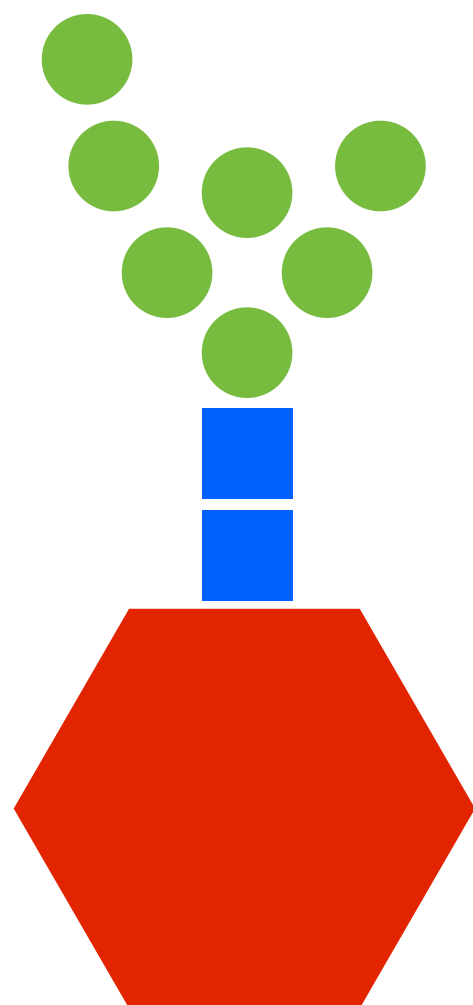
NGLY1

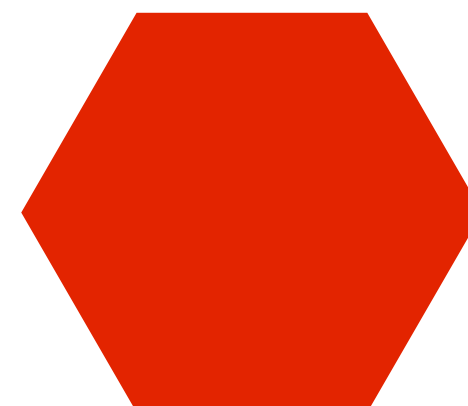
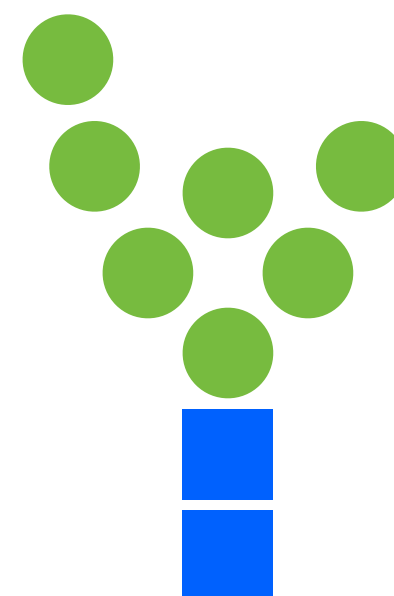
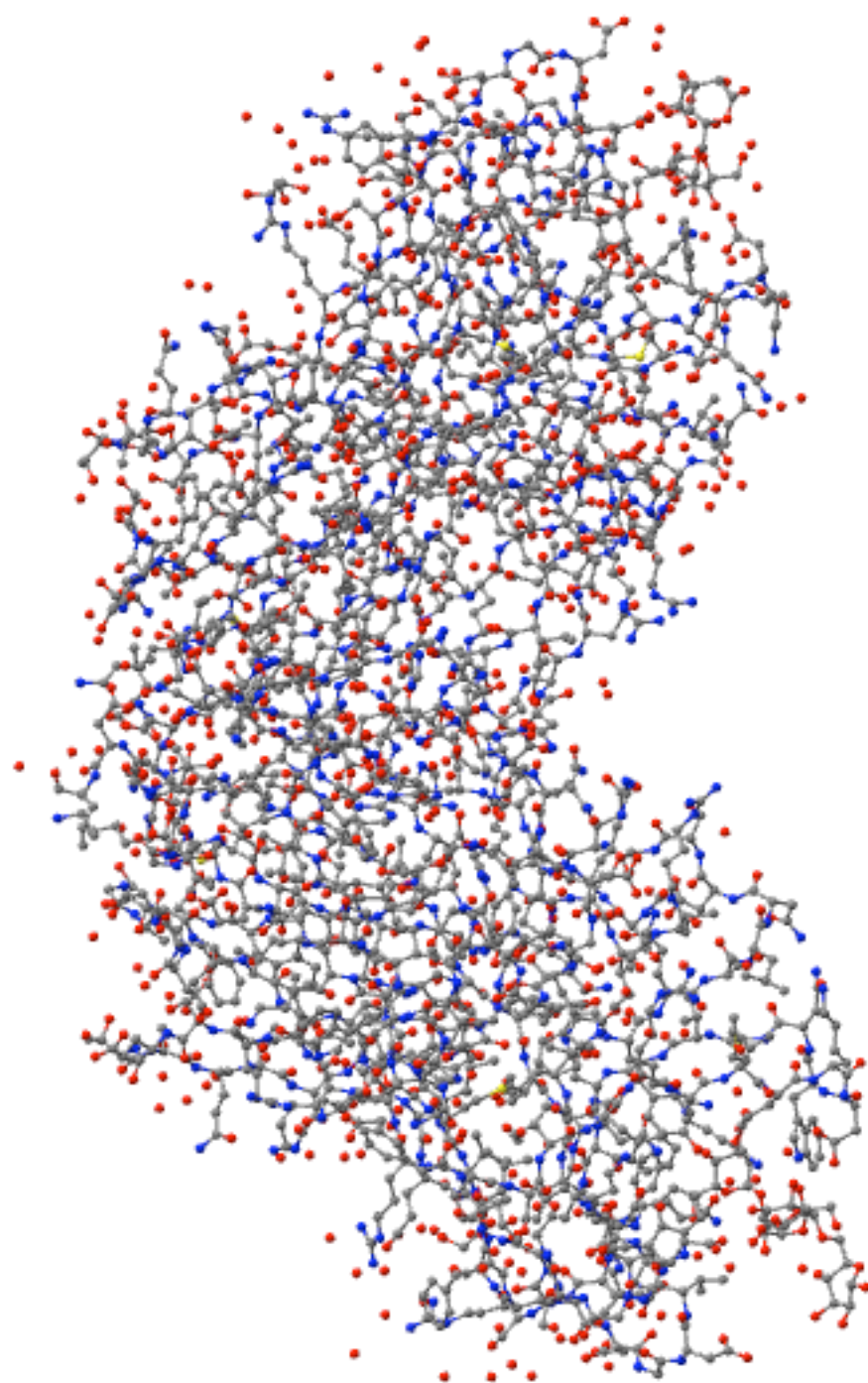


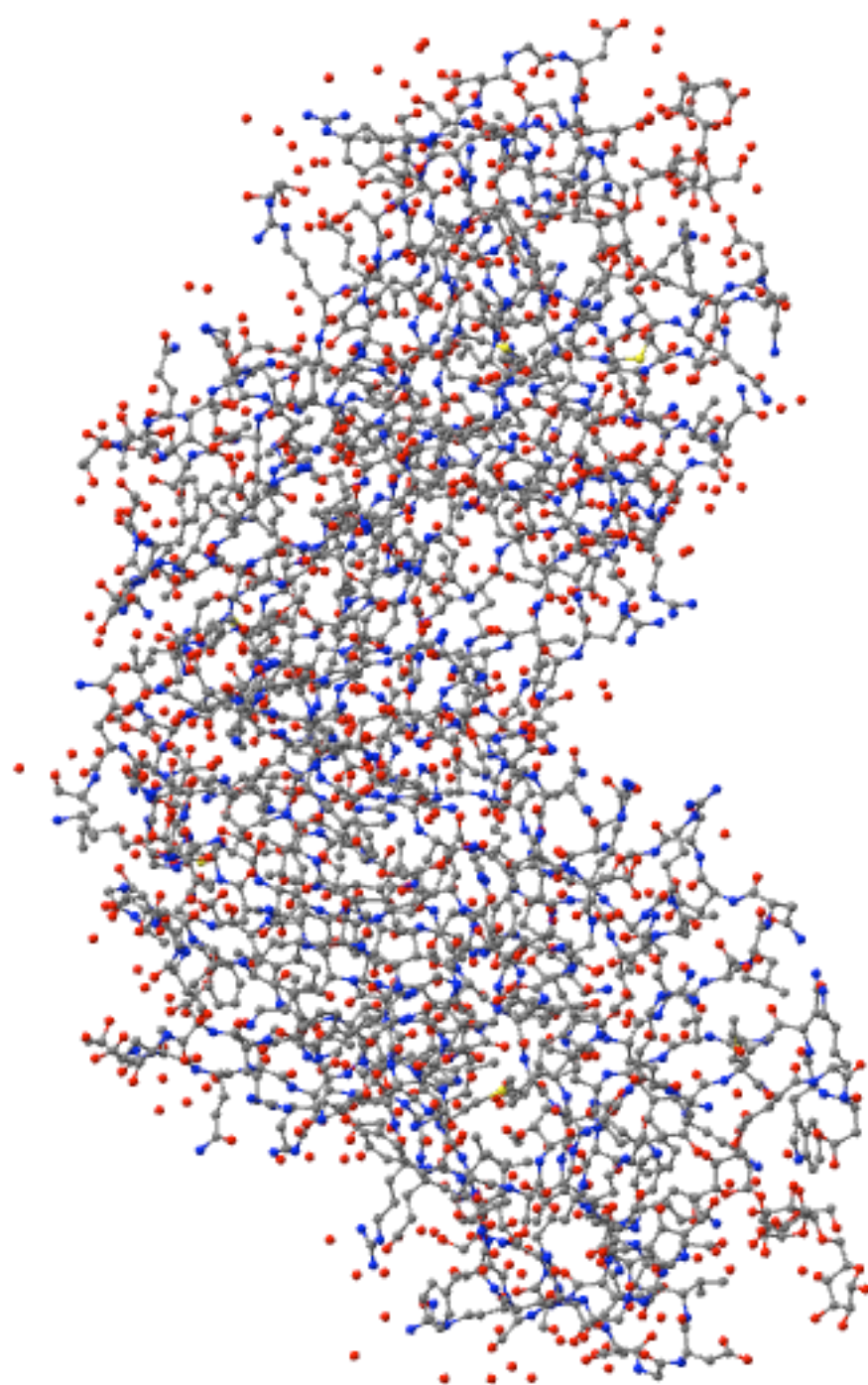


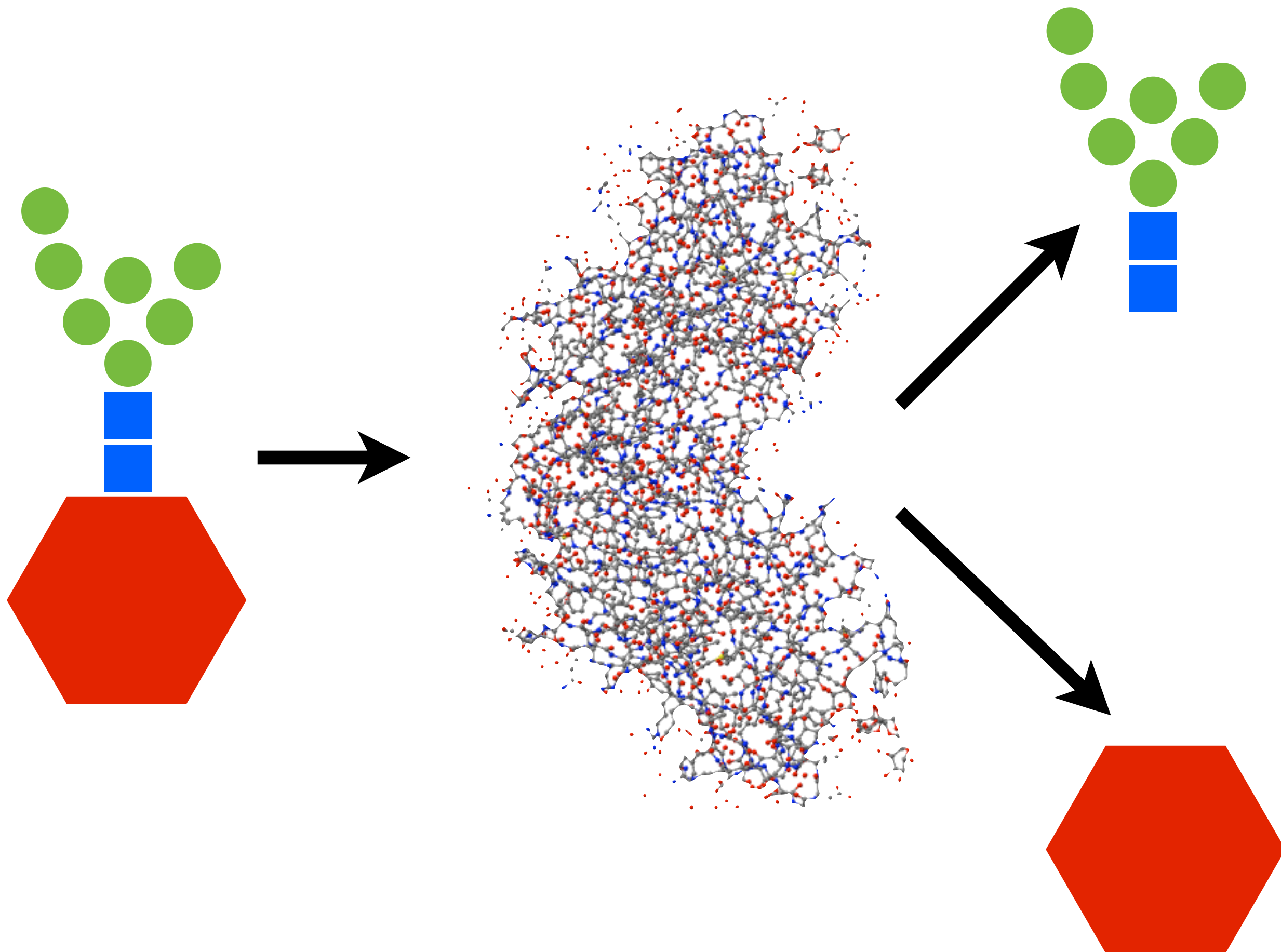
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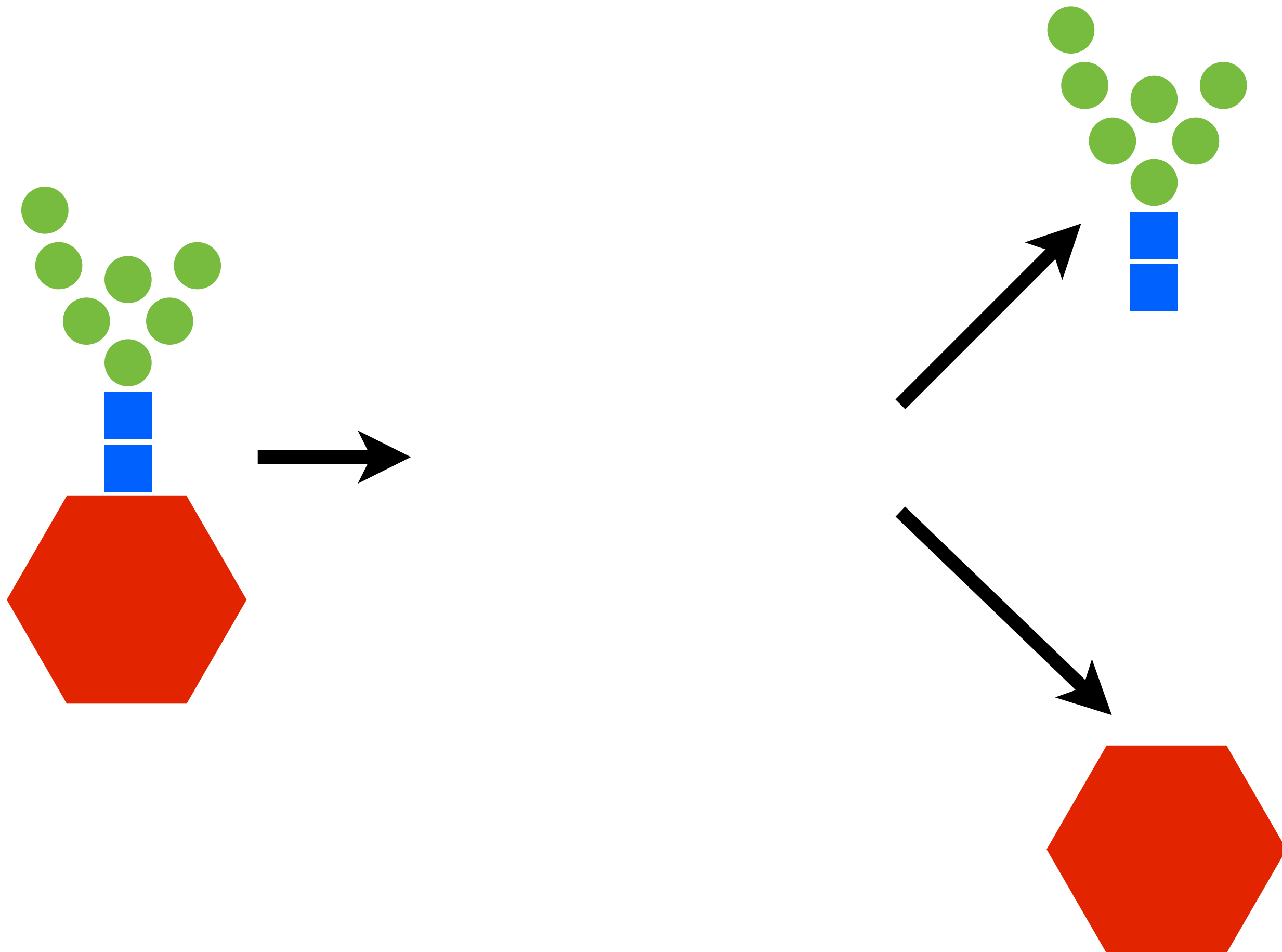


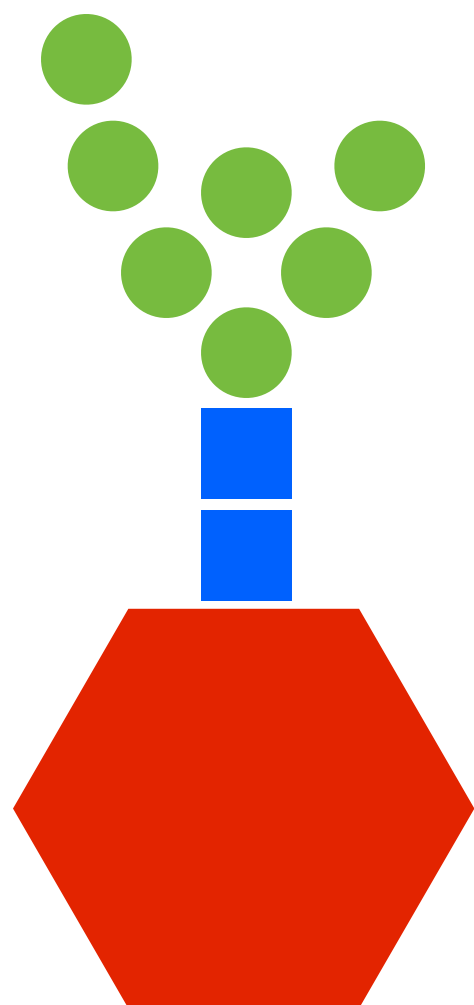


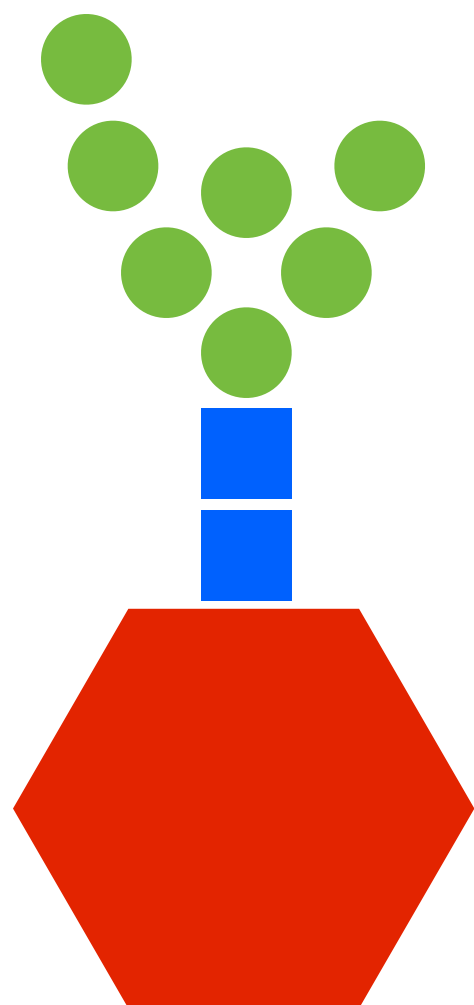


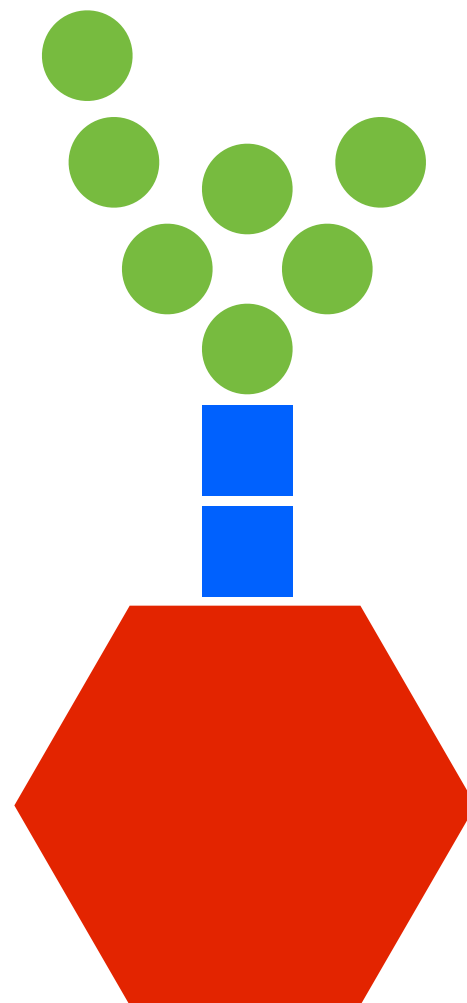
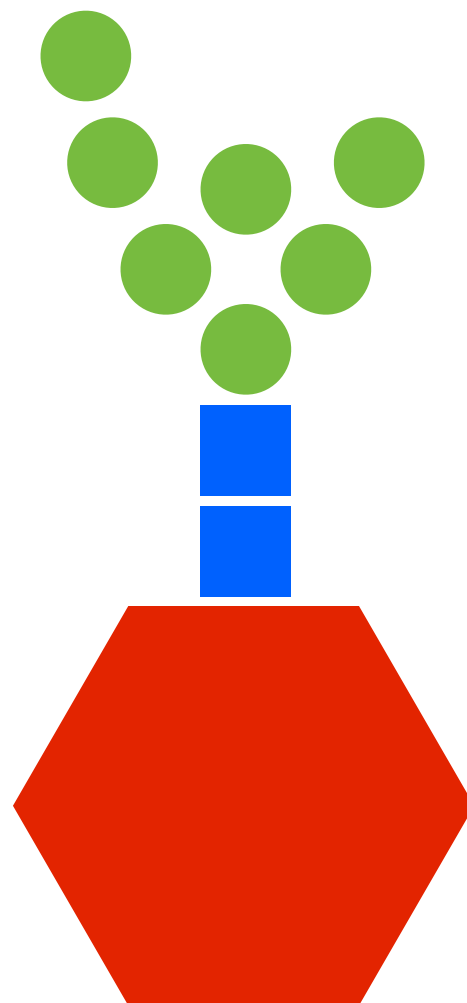
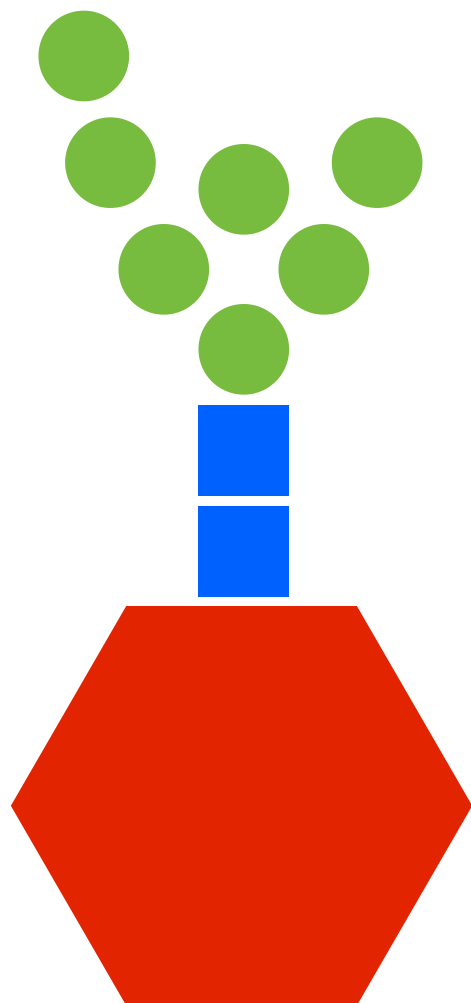
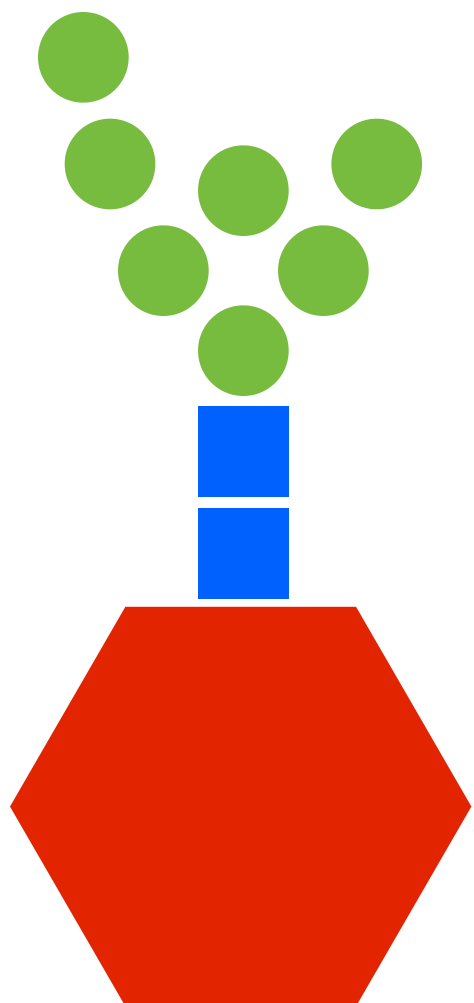












Hunting down my son's killer

[\[article index\]](#) [\[email me\]](#) [\[@mattmight\]](#) [\[+mattmight\]](#) [\[rss\]](#)

I found my son's killer.

It took three years.

But we did it.



Not quite like this.

2,000,000+



NGLY1



Web

Maps

Images

Shopping

Videos

More ▾

Search tools

About 12,000 results (0.43 seconds)

[NGLY1 Gene - GeneCards | NGLY1 Protein | NGLY1 Antibody](#)

www.genecards.org/cgi-bin/carddisp.pl?gene=NGLY1 ▾

Complete information for **NGLY1** gene (protein-coding), N-glycanase 1, including: function, proteins, disorders, pathways, orthologs, and expression.

[NGLY1 - Wikipedia, the free encyclopedia](#)

en.wikipedia.org/wiki/NGLY1 ▾ Wikipedia ▾

Peptide-N(4)-(N-acetyl-beta-glucosaminy)asparagine amidase is an enzyme that in humans is encoded by the **NGLY1** gene.

[NGLY1 N-glycanase 1 \[Homo sapiens \(human\)\]](#)

www.ncbi.nlm.nih.gov/gene/55768 ▾ National Center for Biotec... ▾

5 days ago - This gene encodes an enzyme that catalyzes hydrolysis of an N(4)-(acetyl-beta-D-glucosaminy) asparagine residue to ...

[OMIM Entry - * 610661 - N-GLYCANASE 1; NGLY1](#)

www.omim.org/610661 ▾ OMIM : Online Mendelian... ▾

Jun 12, 2013 - (2000) identified several homologs of yeast Png1, including human **NGLY1**. In yeast, Png1 was expressed in both the cytoplasm and nucleus.

[Hunting down my son's killer - Matt Might](#)

matt.might.net/articles/my-sons-killer/ ▾

We discovered that my son inherited two different (thus-far-unique) mutations in the same gene--the **NGLY1** gene--which encodes the enzyme N-glycanase 1.



NGLY1



Web

Maps

Images

Shopping

Videos

More ▾

Search tools

About 12,000 results (0.43 seconds)

[NGLY1 Gene - GeneCards | NGLY1 Protein | NGLY1 Antibody](#)

www.genecards.org/cgi-bin/carddisp.pl?gene=NGLY1 ▾

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en.wikipedia.org/wiki/NGLY1 ▾ Wikipedia ▾

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[NGLY1 N-glycanase 1 \[Homo sapiens \(human\)\]](#)

www.ncbi.nlm.nih.gov/gene/55768 ▾ National Center for Biotec... ▾

5 days ago - This gene encodes an enzyme that catalyzes hydrolysis of an N(4)-(acetyl-beta-D-glucosaminy) asparagine residue to ...

[OMIM Entry - * 610661 - N-GLYCANASE 1; NGLY1](#)

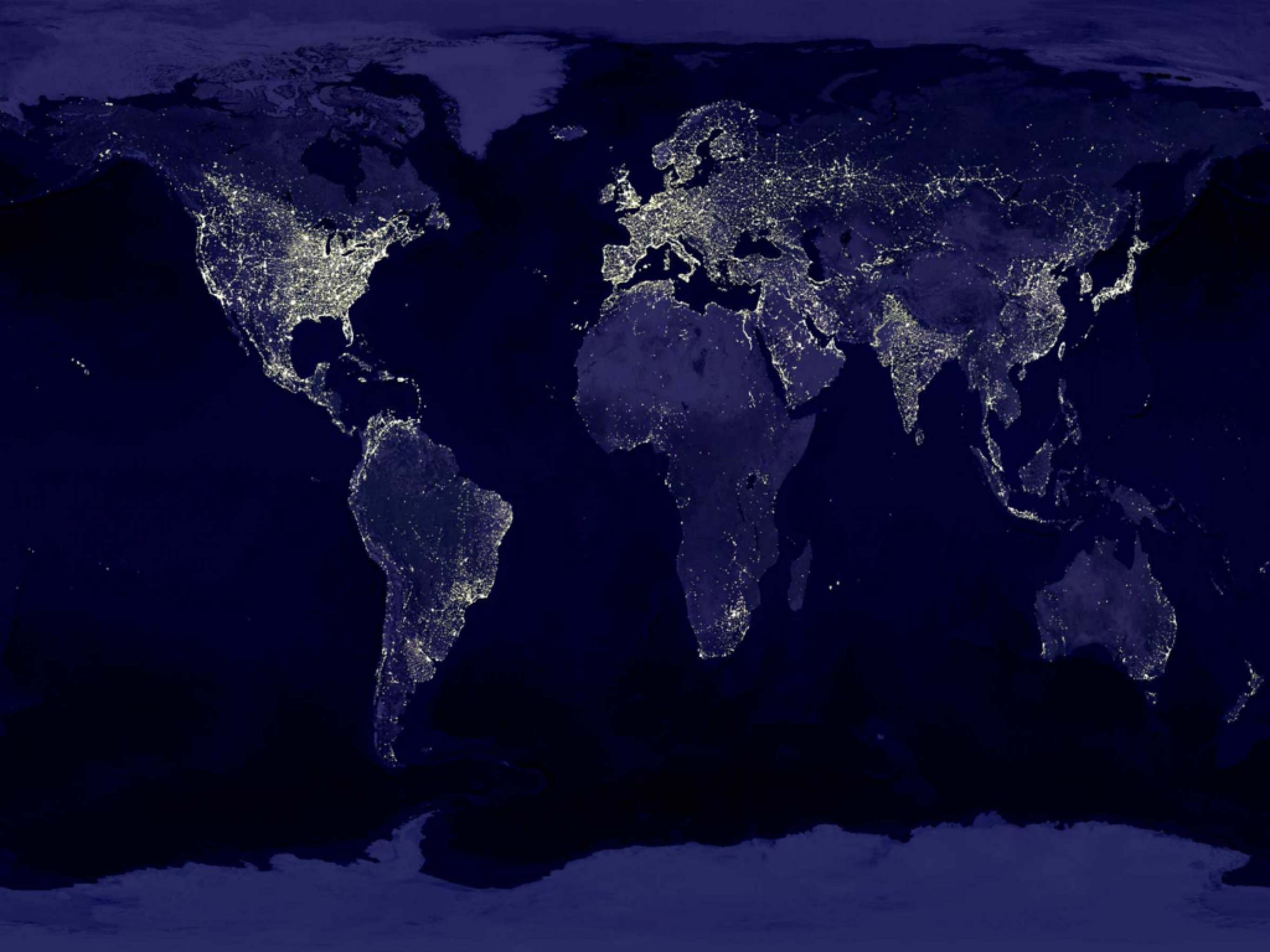
www.omim.org/610661 ▾ OMIM : Online Mendelian... ▾

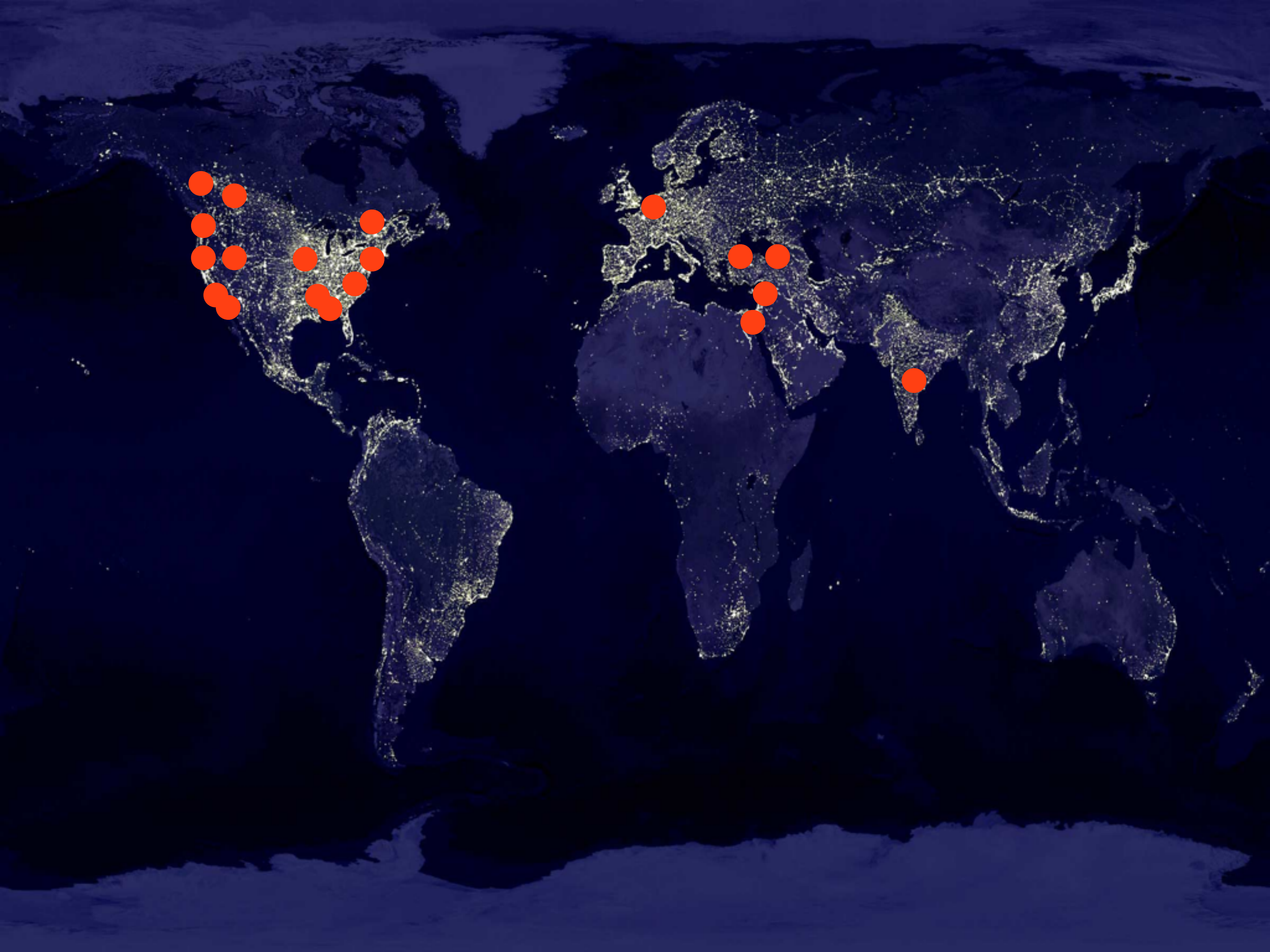
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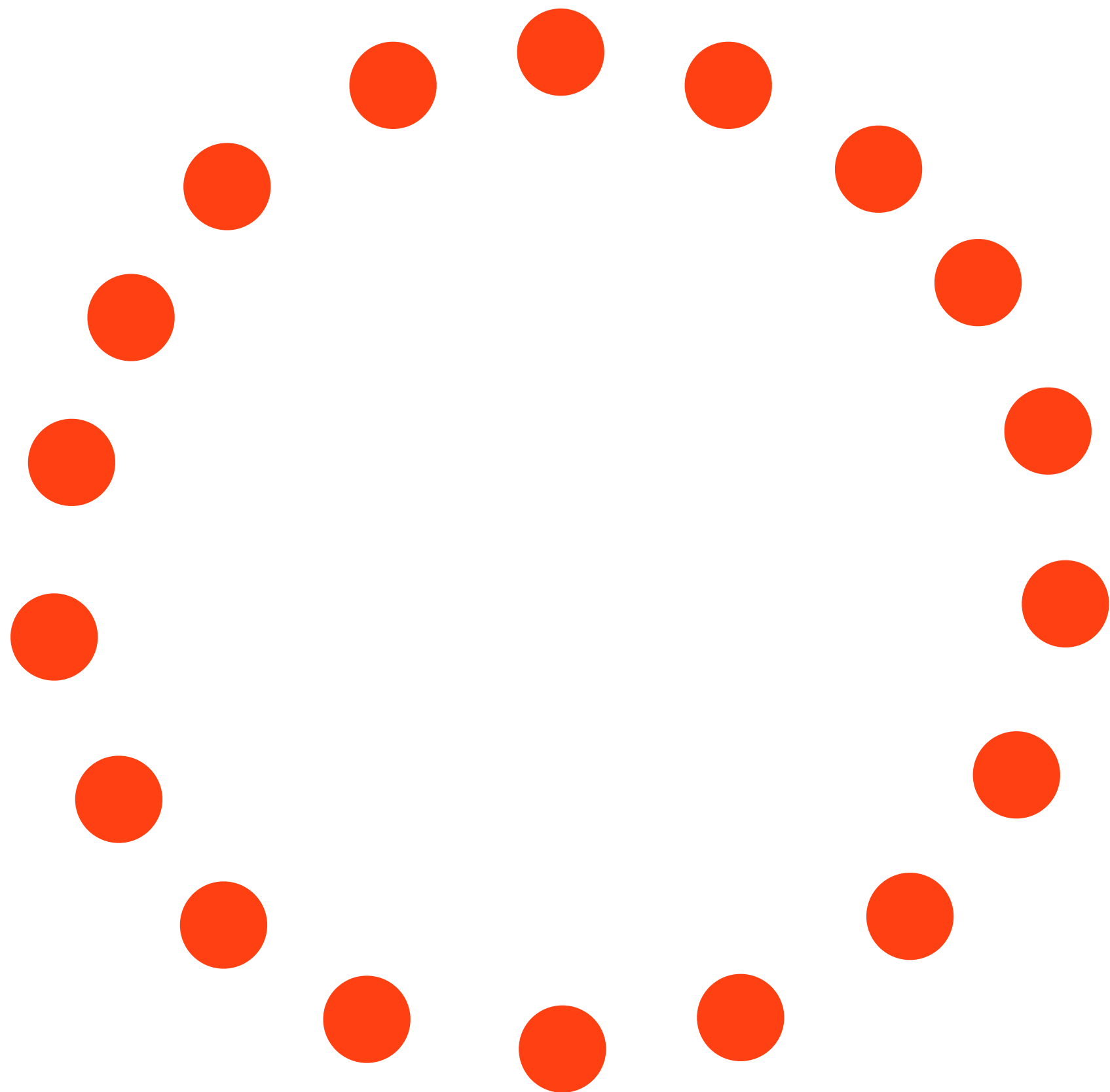
matt.might.net/articles/my-sons-killer/ ▾

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18

14



NGLY1

[HOME](#)[ABOUT](#)[RESEARCH](#)[PATIENTS](#)[TREATMENTS](#)[PRESS](#)[CONTACT](#)

MEDICAL DISPATCH

ONE OF A KIND

What do you do if your child has a condition that is new to science?

BY SETH MNOOKIN

Matt Might and Cristina Casanova met in the spring of 2002, as twenty-year-old undergraduates at the Georgia Institute of Technology. Cristina was an art history major, and Matt was an

worry. Matt and Cristina described Bertrand to friends as being "jiggly"; his body appeared always to be in motion, as if he were lying on a bed of Jell-O. He

City, and the first available appointment fell on the same day as a mandatory faculty retreat. That afternoon, when Matt was able to check his phone, he saw that Cristina had left several messages. "I didn't listen to them," he told me in an e-mail. "I didn't have to. The number of them told me this was really bad."

Bertrand had brain damage—or, at least, that was the diagnosis until an MRI revealed that his brain was perfectly normal. After a new round of lab work was done, Bertrand's doctors concluded that he likely had a rare, inherited movement disorder called ataxia-telangiectasia. A subsequent genetic screen

NGLY1 in The New Yorker!

Open science and social media were key in the discovery and growth of the NGLY1 rare disease community. Article by Seth Mnookin.

Dr. David Goldstein to direct Institute for Genomic Medicine at Columbia University

By [Cristina](#) on Saturday, October 25, 2014 [Columbia news](#) [No comments](#)



Do you know a NGLY1 patient?

If you know a patient with NGLY1-deficiency, or think you do, please contact us immediately.

You can reach us directly at info@ngly1.org



NGLY1 Symptoms

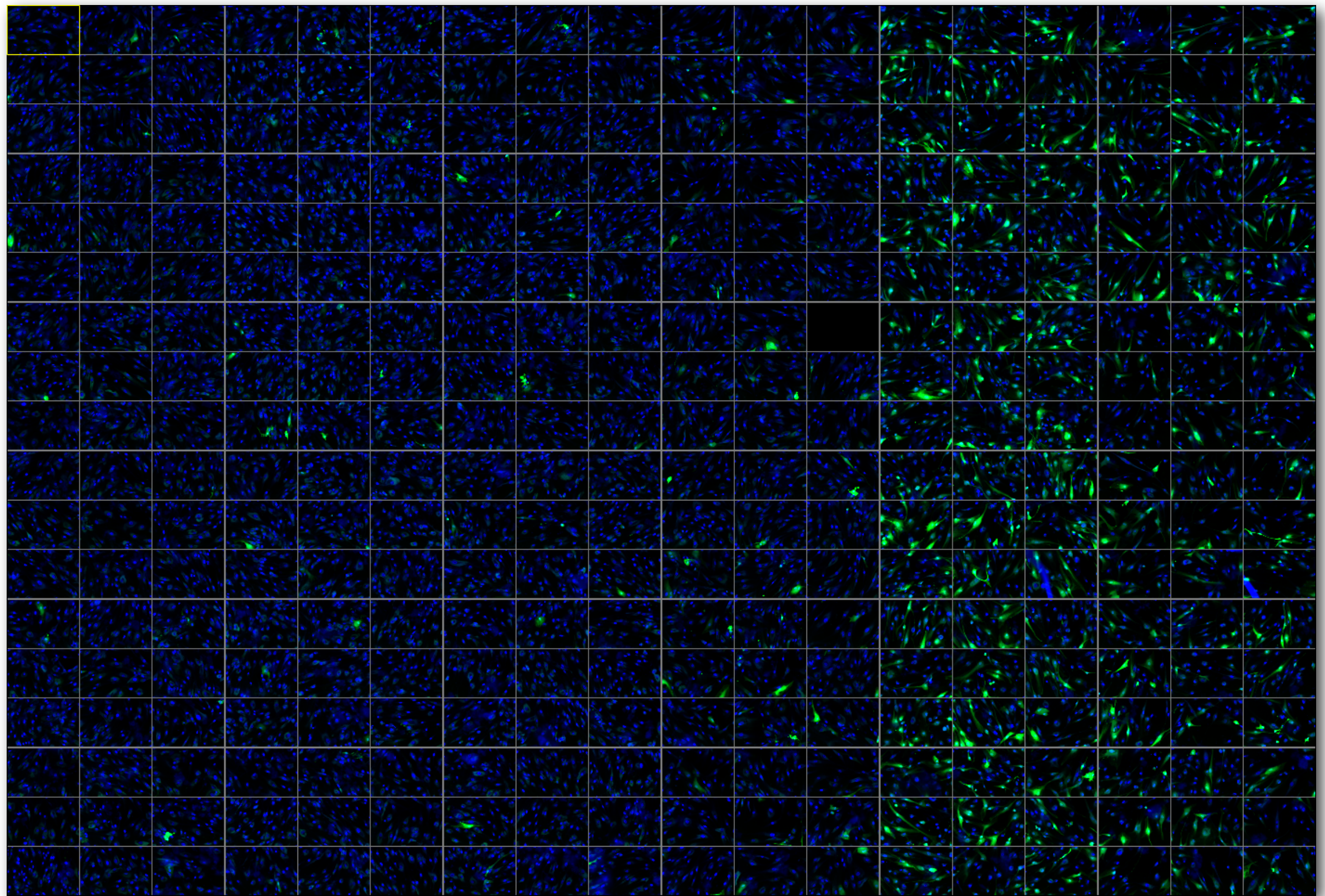
Global Developmental Delays

Movement Disorder

Hypotonia

For patients, functional studies beat case-finding!





Source: Hudson Freeze



Animal models









NUTRITIONAL



YEAST

Flakes



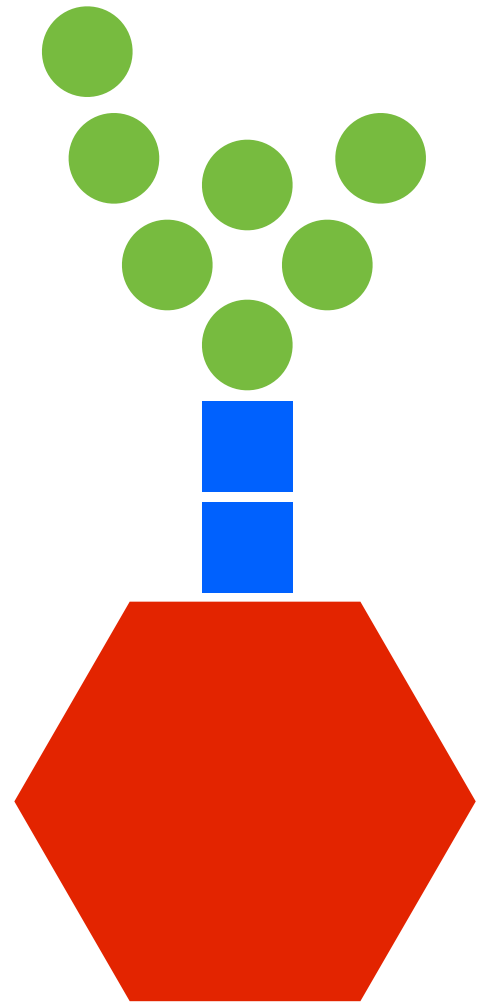
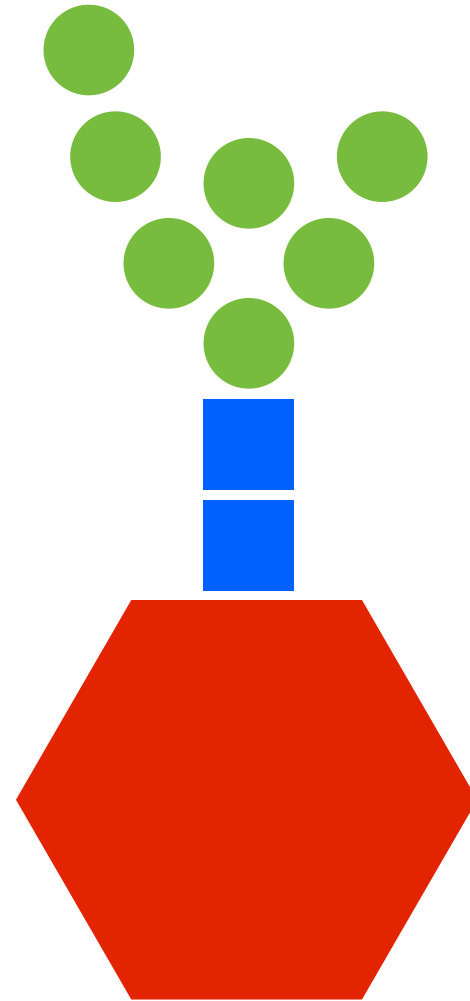
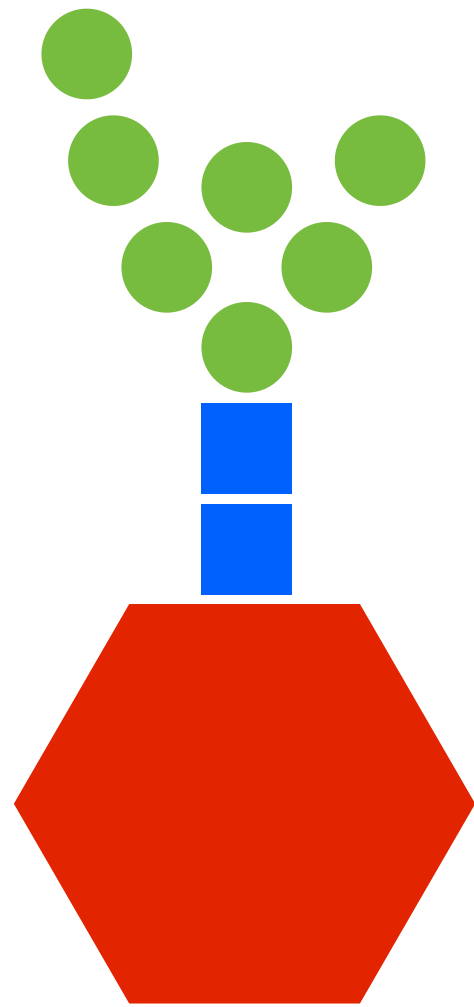
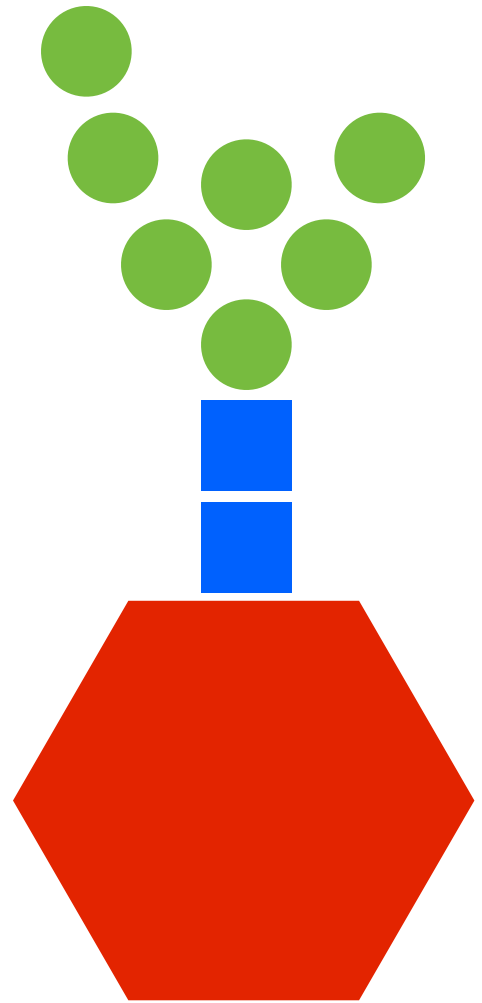
PREMIUM • UNSWEETENED • FORTIFIED

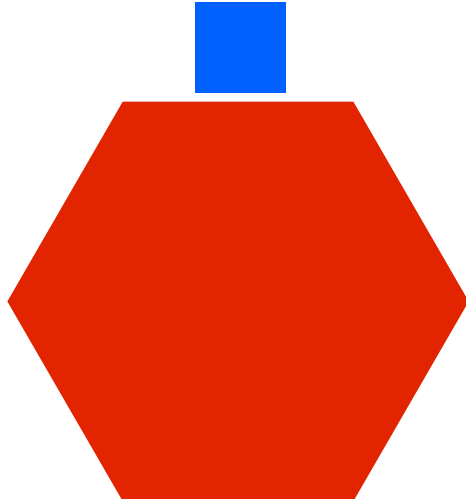
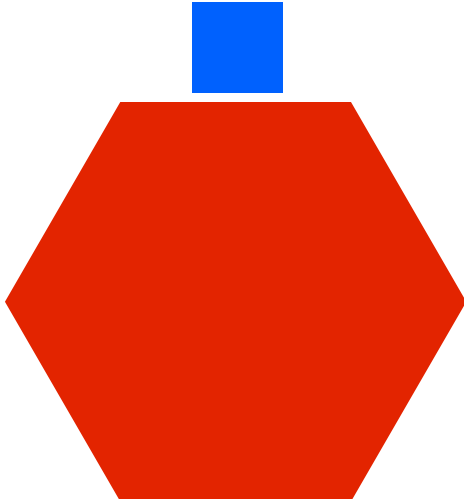
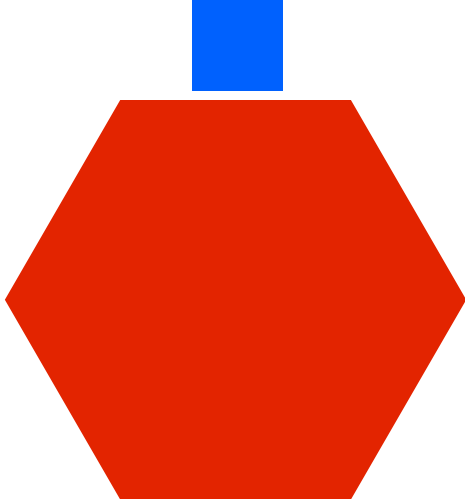
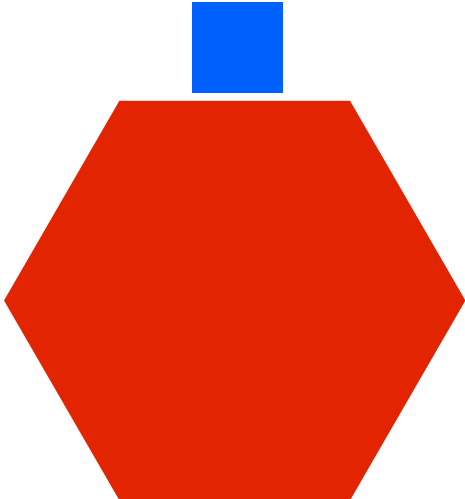
Gluten Free

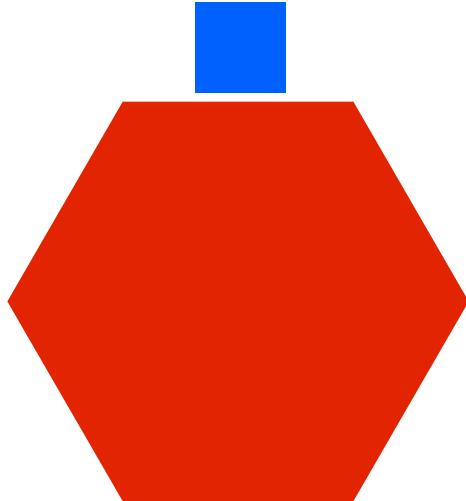
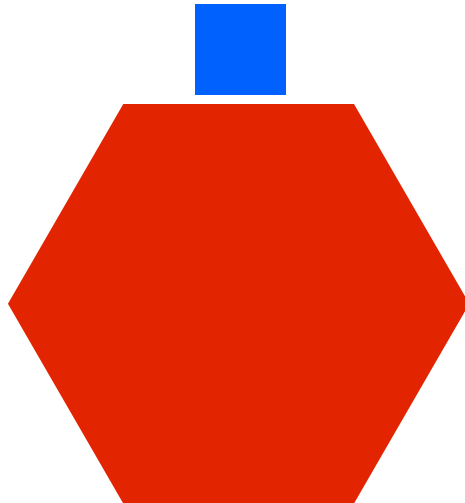
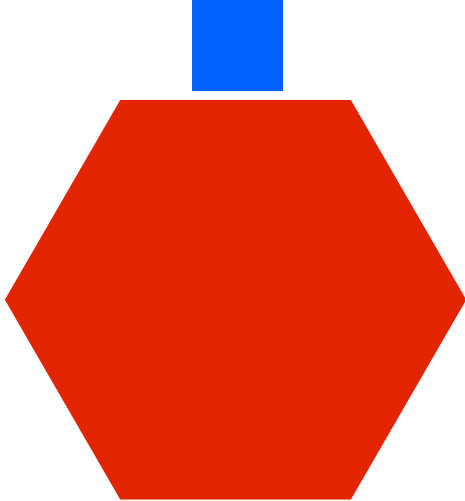
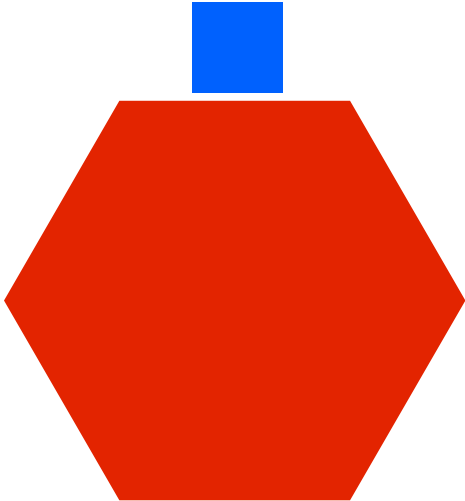
Wonderful Nutty Flavor
Non-GMO

NET WT. 22 oz. (624 g)

Basic science















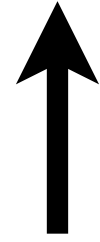


**National Institutes
of Health**

CDG Protocol



Oligos



GPI

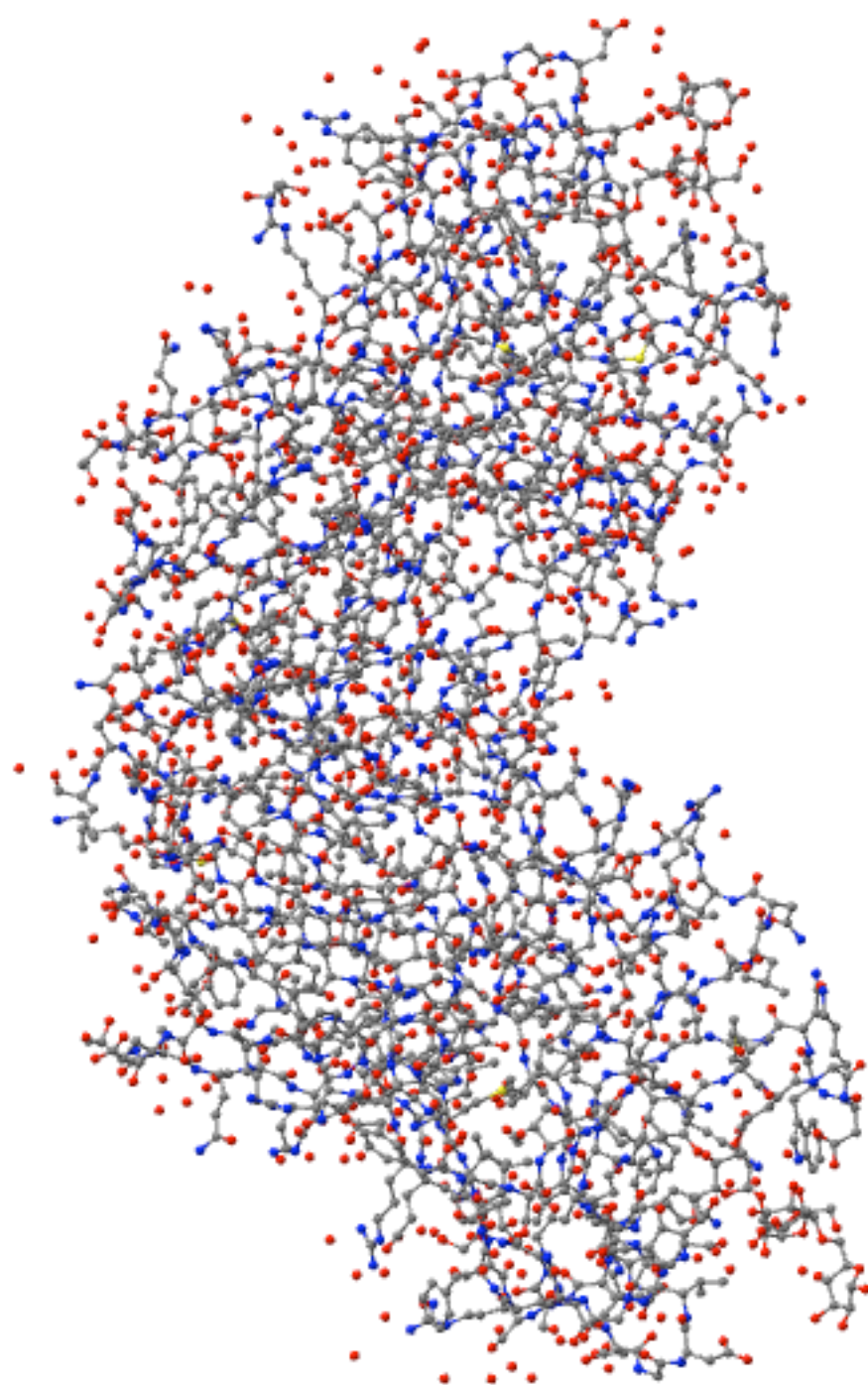


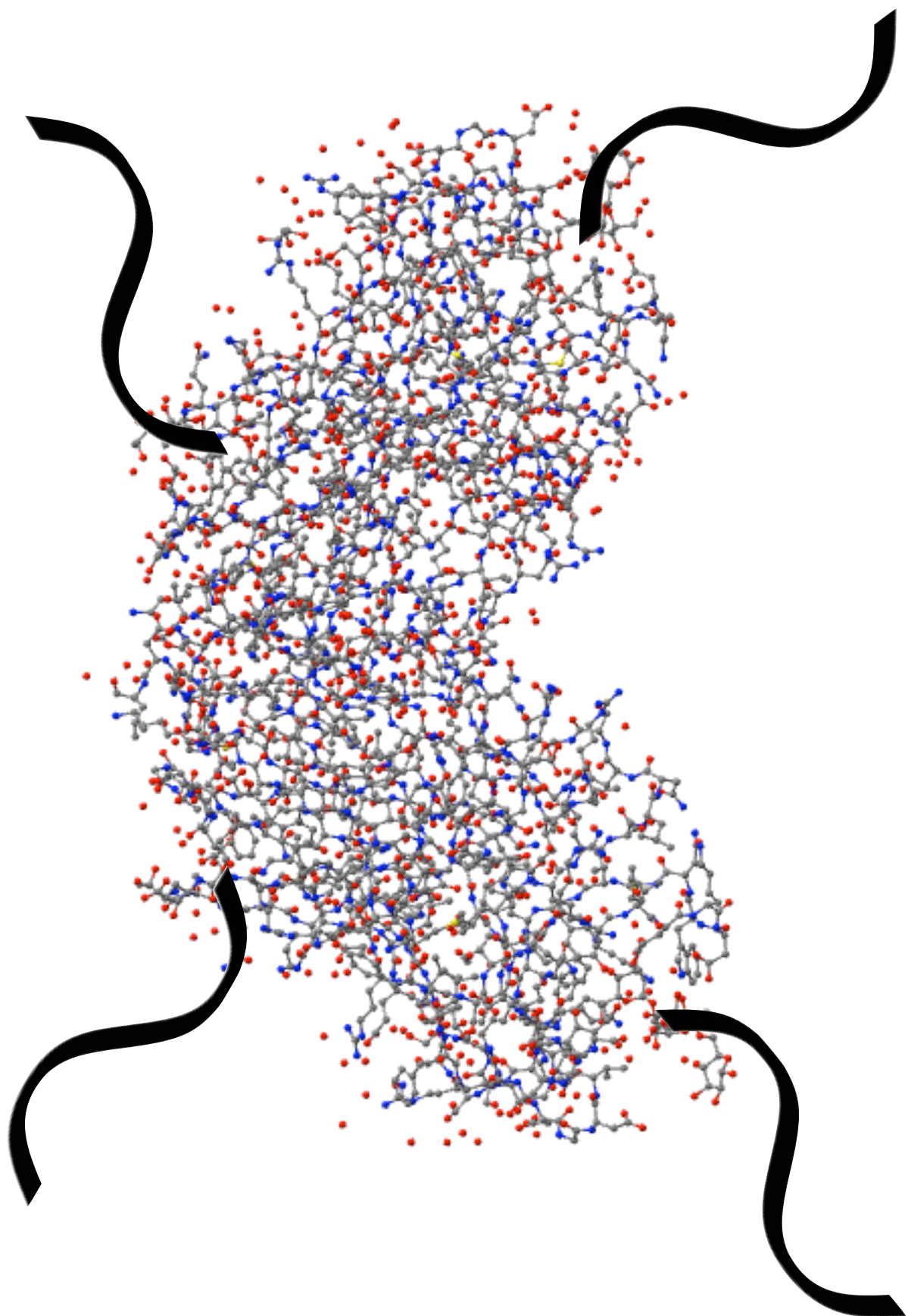
Mito



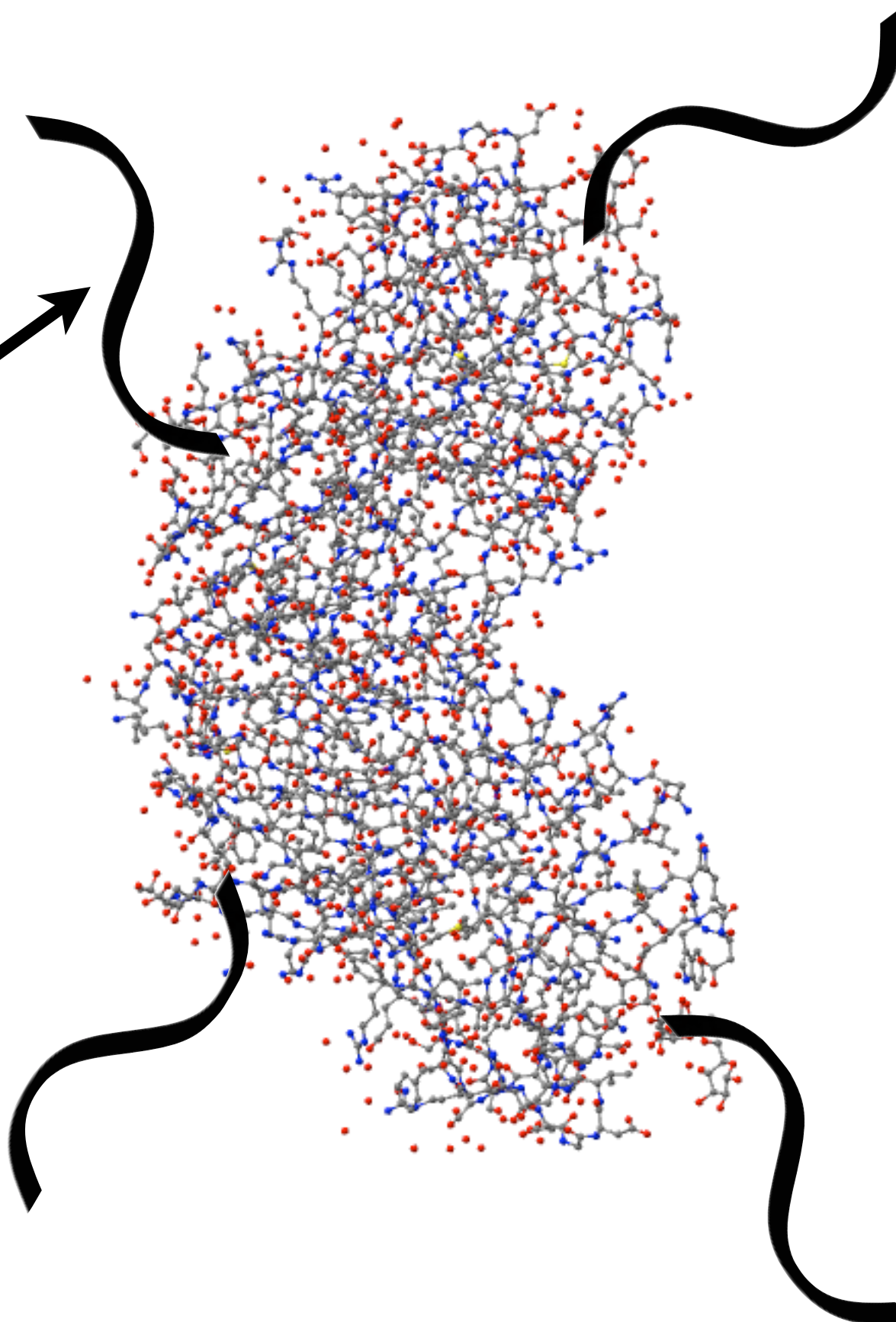
CSF

Enzyme replacement



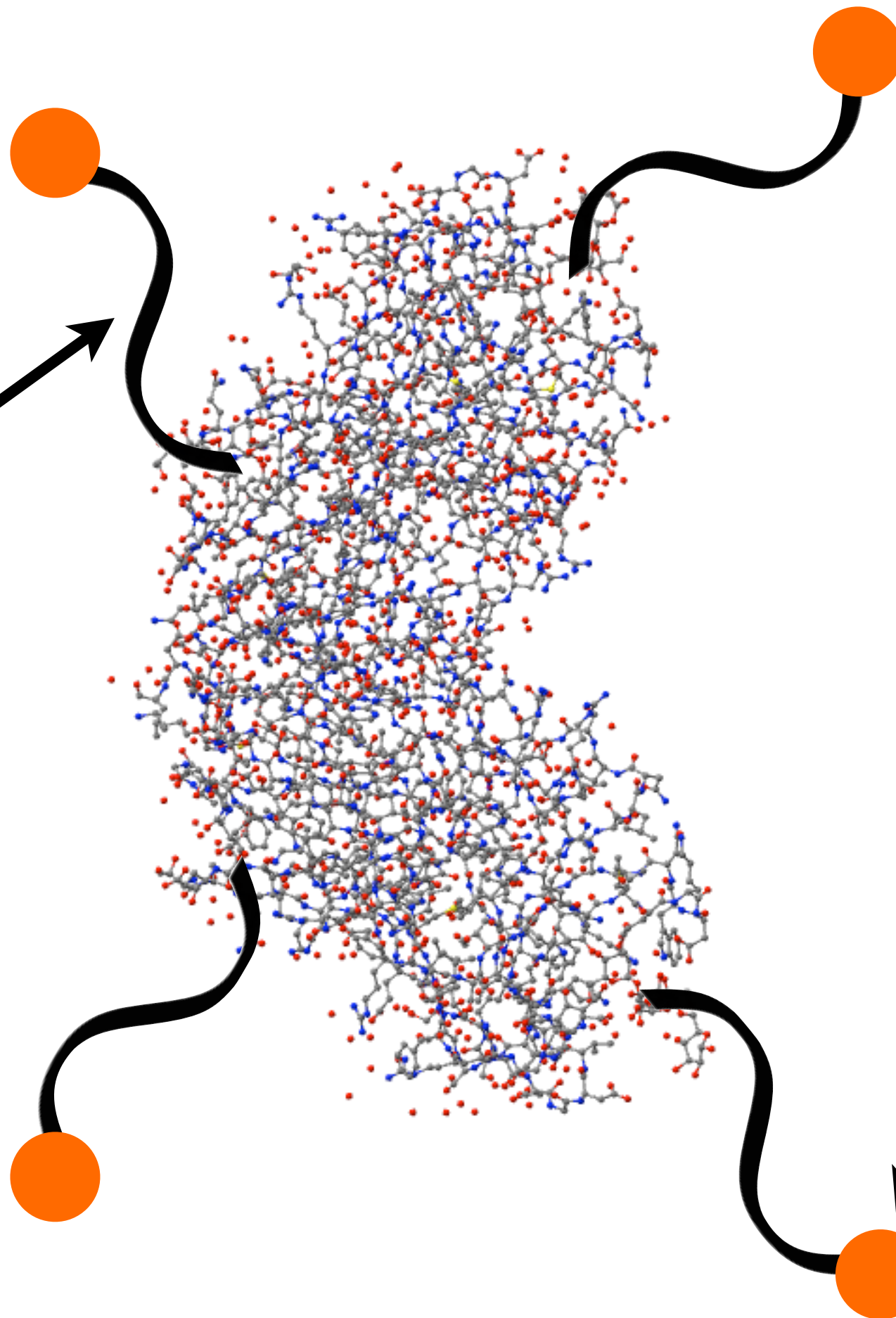


PEG polymer



PEG polymer

TAT peptide



Replicable?

Yes!!



....*looking for companions*

Reaching out

This page is for parents, doctors, or researchers who may know of other children like our son, Milo. If you know of a similar case, please get in touch with us. The more cases we have, the more opportunities we will have to improve our understanding of his condition and facilitate research that can help him and others.

Find out more

- [What this site is for](#)
- [Case study:](#)



Finding others like our Milo

Currently, at age 3, Milo's primary challenges are global developmental delay and significant hypotonia. He has had surgical repairs for a minor cleft in his soft palate, for ptosis, for C1 stenosis, for a tethered

De novo gain of function in Gene X

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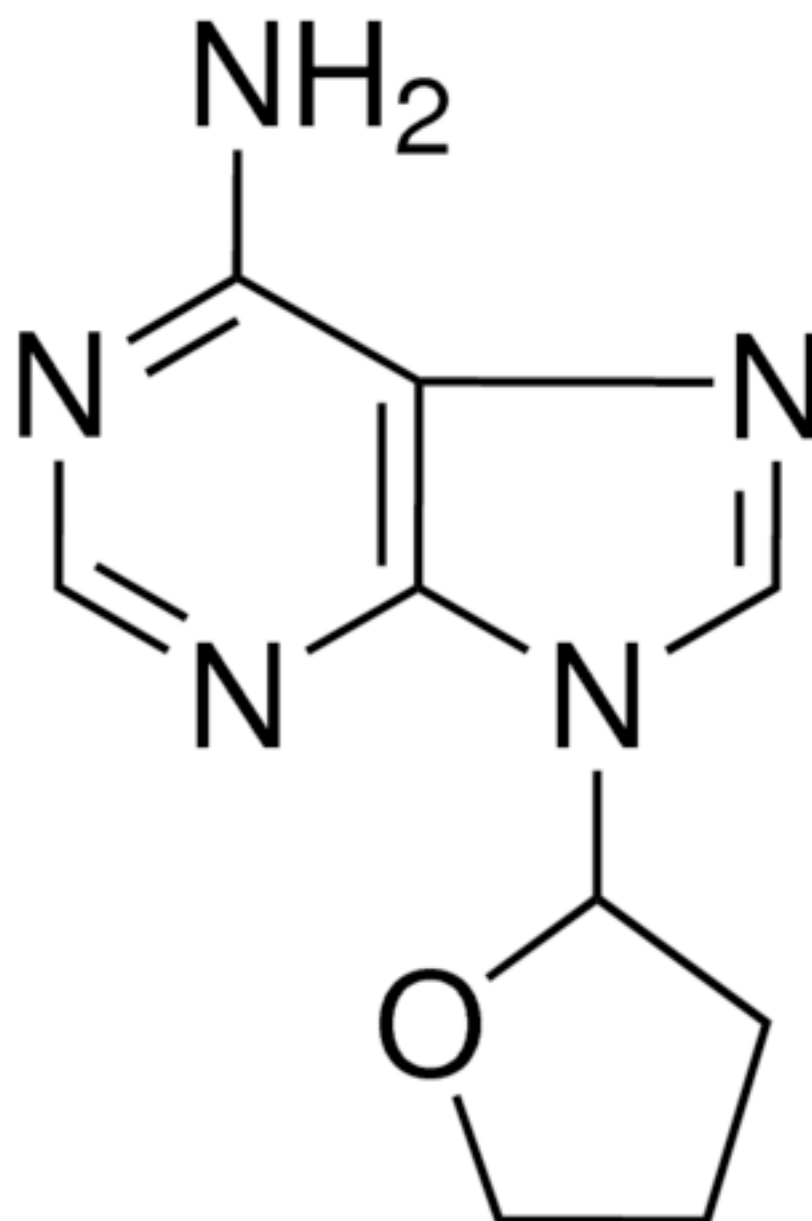
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Gene X inhibitor

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Gene X inhibitor

What do patients need?

“next step”

NGLY1

From Wikipedia, the free encyclopedia

Peptide-N(4)-(N-acetyl-beta-glucosaminyl)asparagine amidase is an **enzyme** that in humans is encoded by the ***NGLY1*** gene.^[1]

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Further reading

Function [edit]

This gene encodes an enzyme that catalyzes hydrolysis of an N(4)-(acetyl-beta-D-glucosaminyl) asparagine residue to N-acetyl-beta-D-glucosaminyamine and a peptide containing an **aspartate** residue. The encoded enzyme may play a role in the **proteasome**-mediated degradation of misfolded **glycoproteins**.^[1]

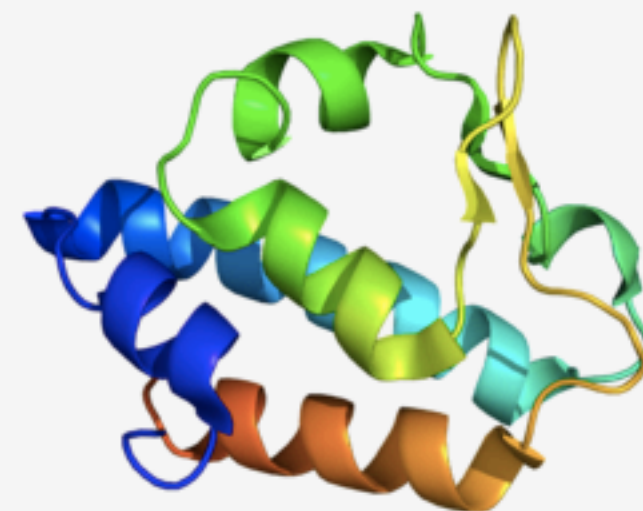
Clinical significance [edit]

In 2012, by means of **exome sequencing** it was determined that a genetic mutation of the *NGLY1* gene, resulting in inability to synthesise this enzyme, is the cause of an extremely rare **congenital disorder of glycosylation** variant.^[2]

In 2014, a study of eight recently discovered patients with mutations in the *NGLY1* gene established a phenotype for NGLY1 deficiency.^[3] NGLY1 deficiency is characterized by global developmental delay (often severe), neurological impairment, movement disorder and hypotonia. Almost all patients have difficulty producing tears and present abnormally on EEGs.

The site **ngly1.org** serves as a hub for N-Glycanase deficient patients. The Grace Wilsey Foundation (**gracewilsey.org**) has been established to raise awareness and support research.

N-glycanase 1



PDB rendering based on 2ccq.

Available structures

PDB Ortholog search: [PDBe](#) , [RCSB](#)

List of PDB id codes [[show](#)]

Identifiers

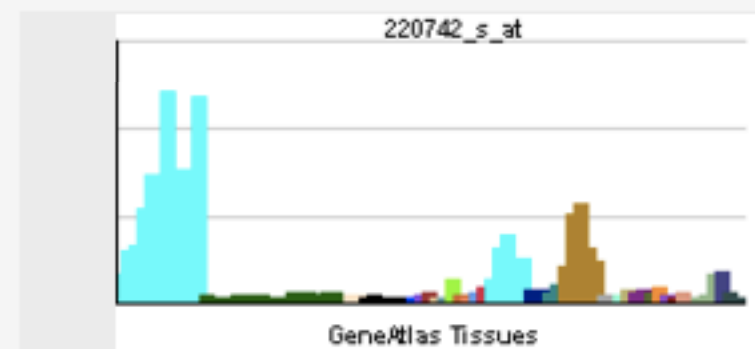
Symbols [NGLY1](#) ; [CDG1V](#); [PNG1](#); [PNGase](#)

External IDs [OMIM: 610661](#) [MGI: 1913276](#)
[HomoloGene: 10117](#) [GeneCards: NGLY1 Gene](#)

EC number [3.5.1.52](#)

Gene ontology [[show](#)]

RNA expression pattern



How to create a web site

What information to share

All variants

HPO

PPO

Parent Phenotyping Ontology

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why does my child have no tears?

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