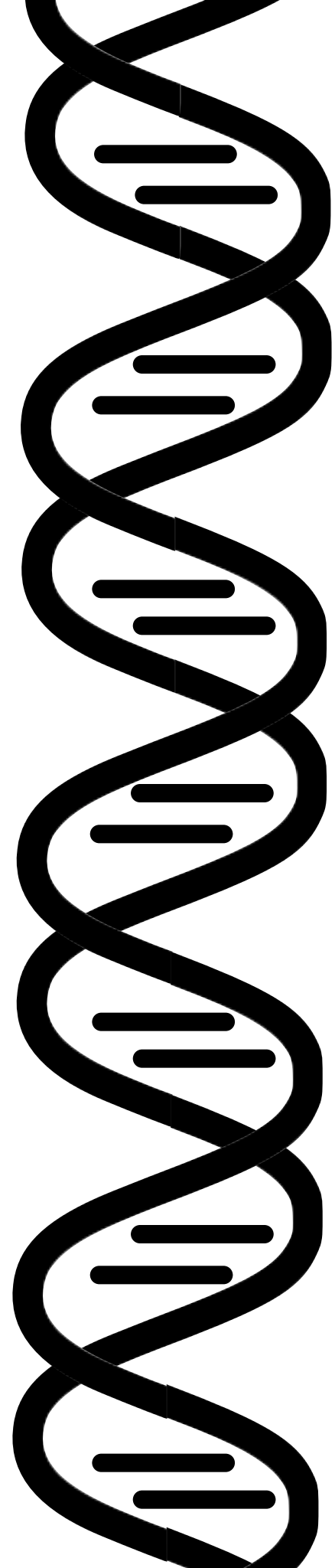


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Associate Professor
Presidential Scholar
University of Utah
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Partners in Research: Parents Accelerating Rare Disease Discoveries

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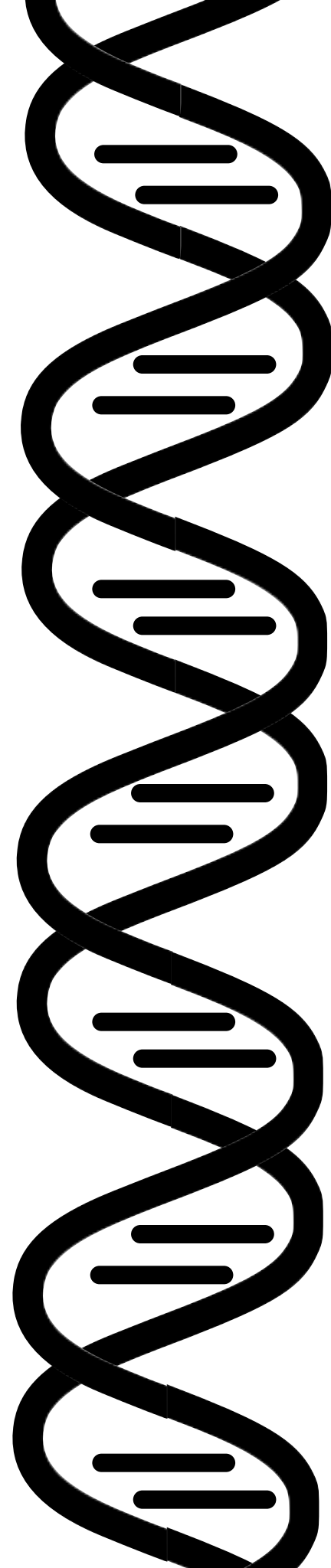
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Matt Might , Dad







Human genome

Rare disease

Curing cancer

Grand Challenge

Grand Challenge

Grand Challenge

Grand Challenge

Sanford|Burnham

Medical Research Institute

Human genome

20,000

1







Mario Capecchi



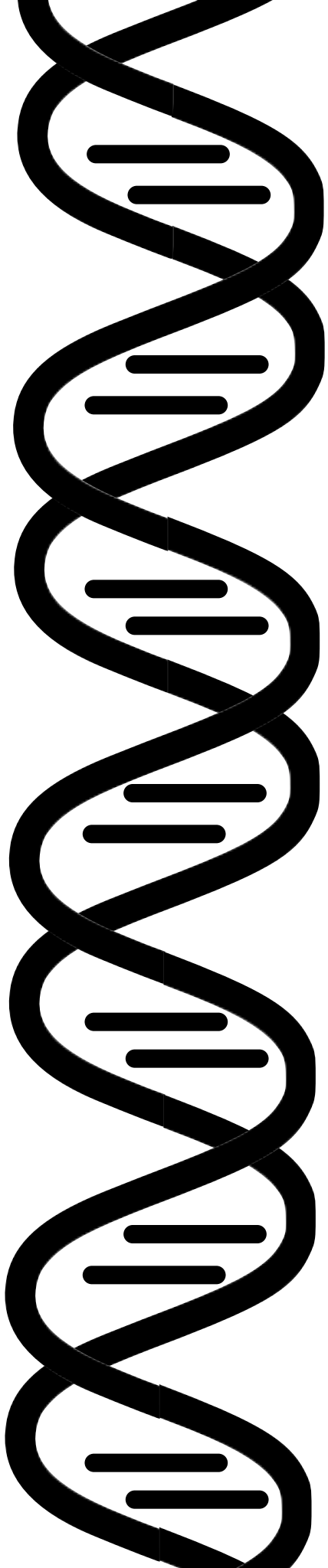
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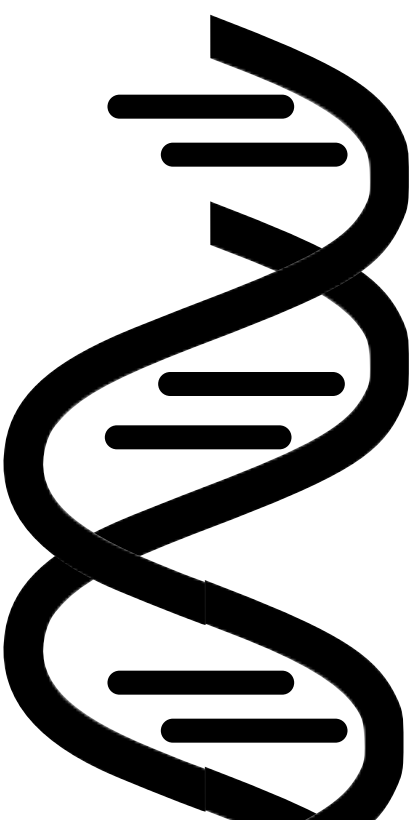
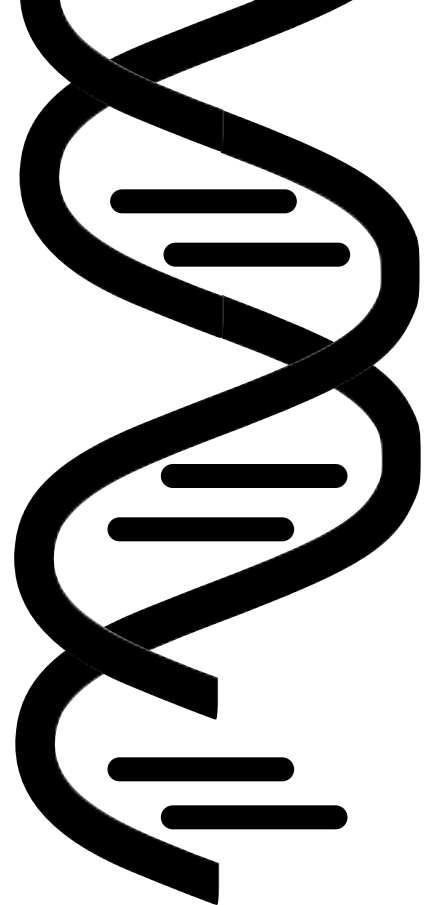


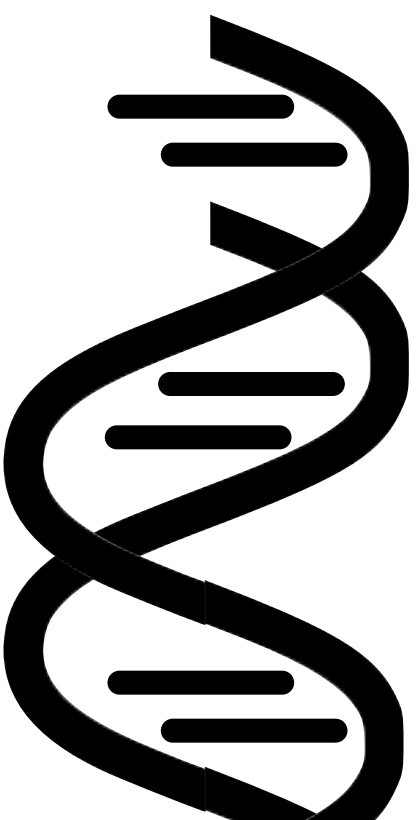
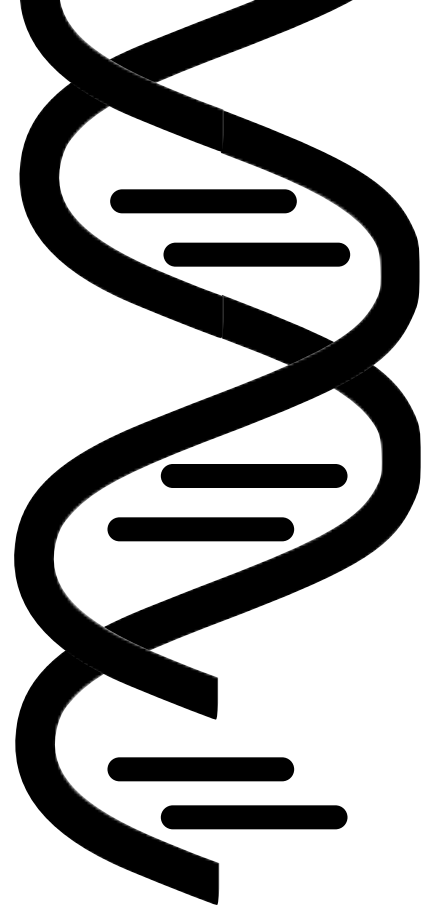
Martin Evans

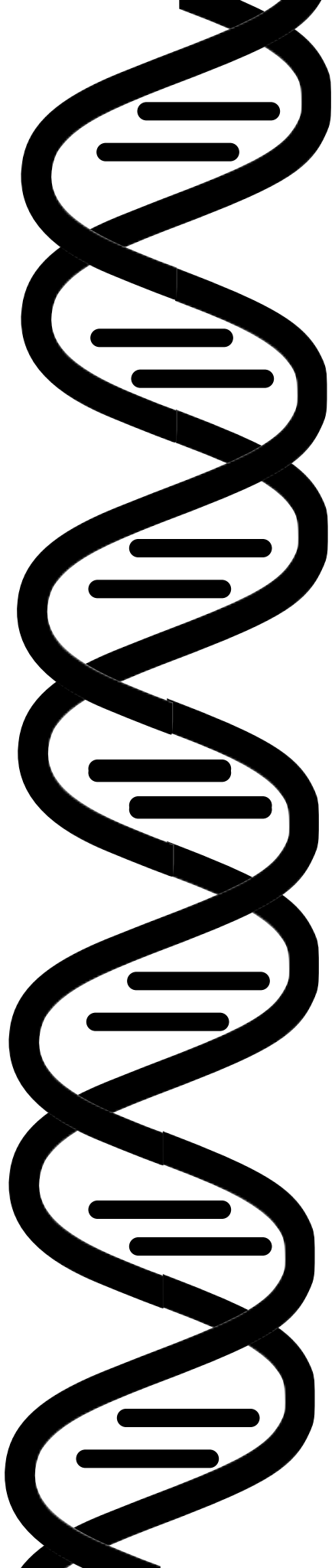
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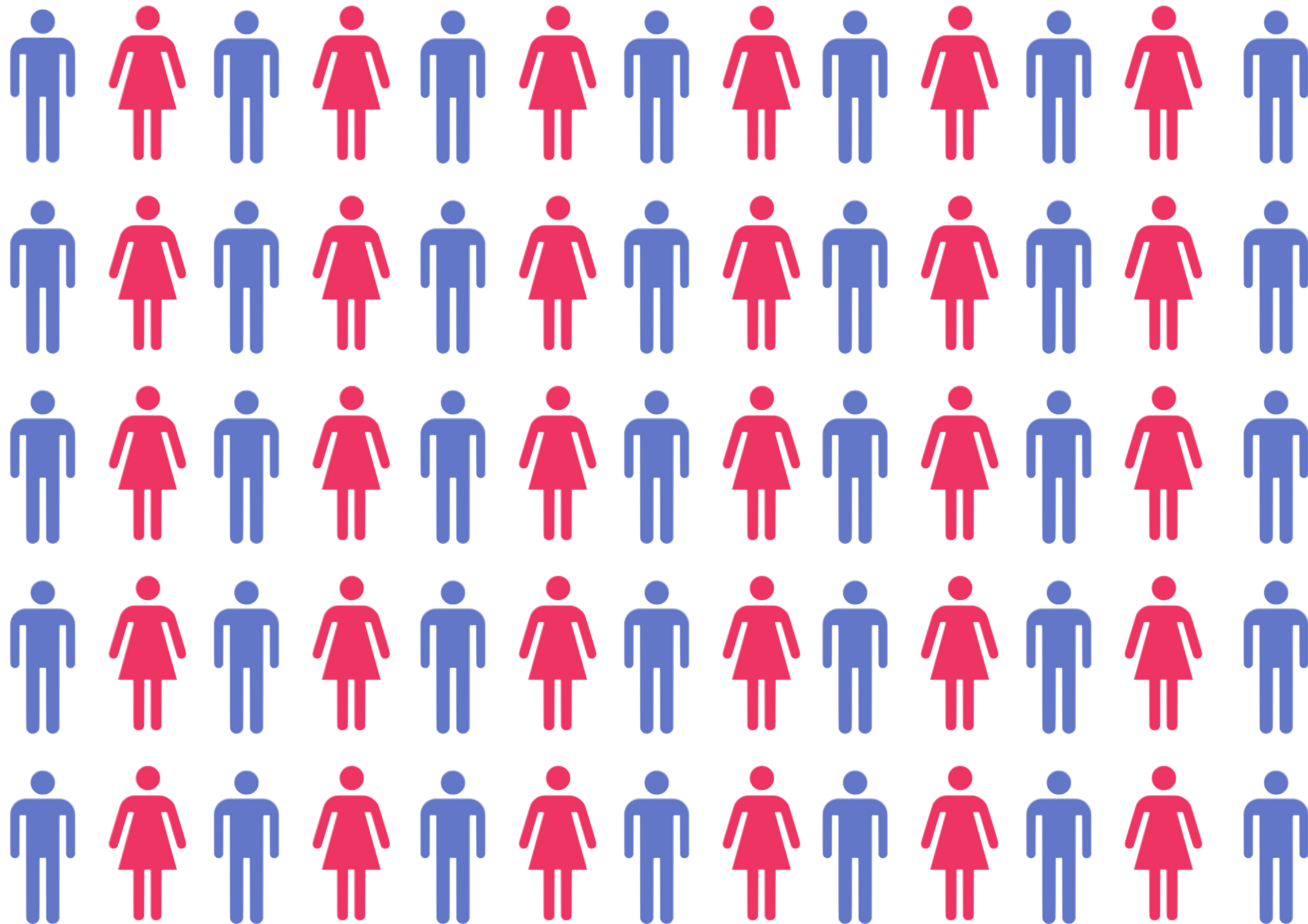


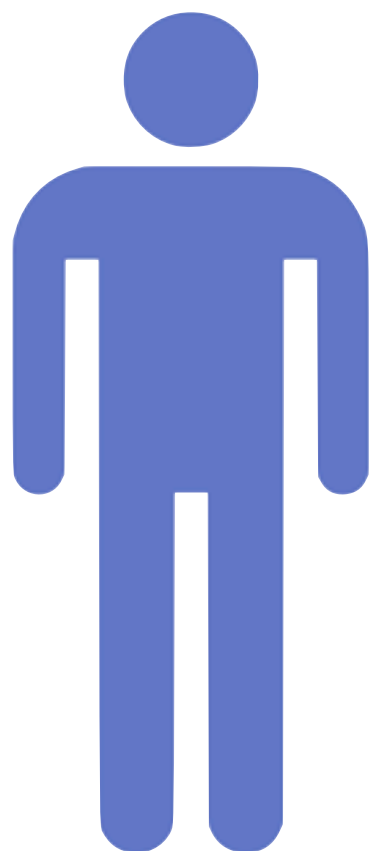


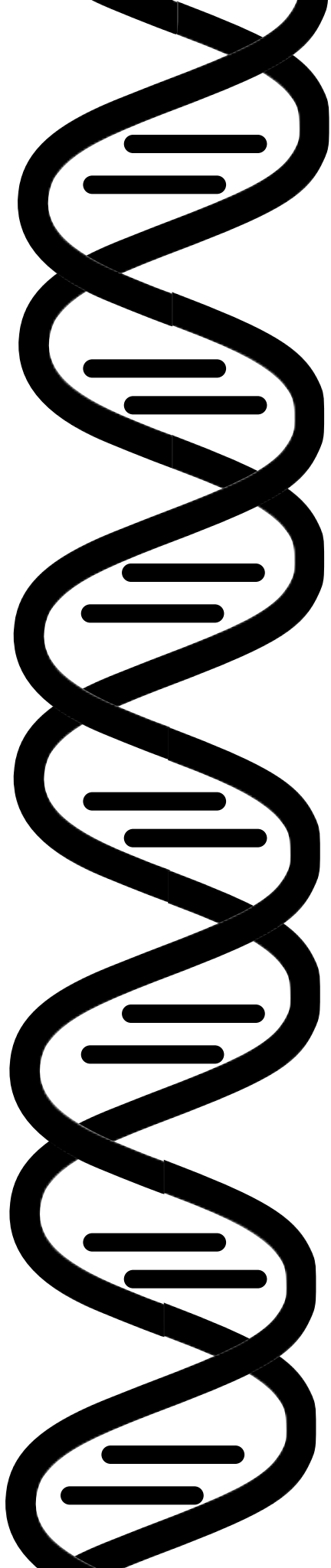
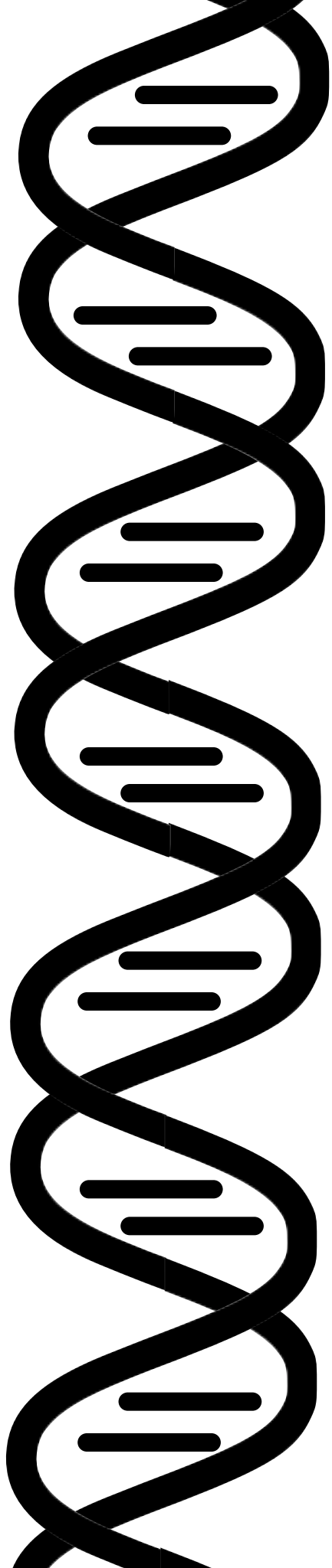
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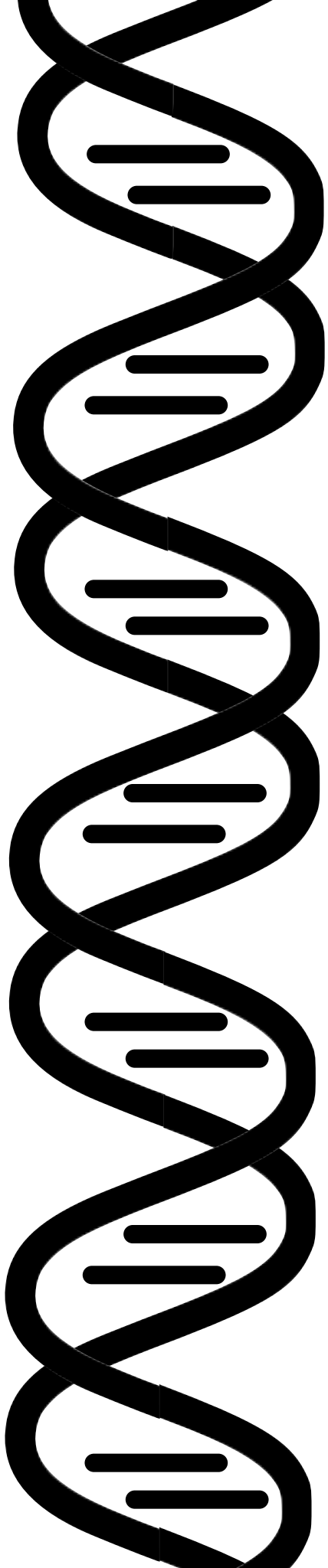




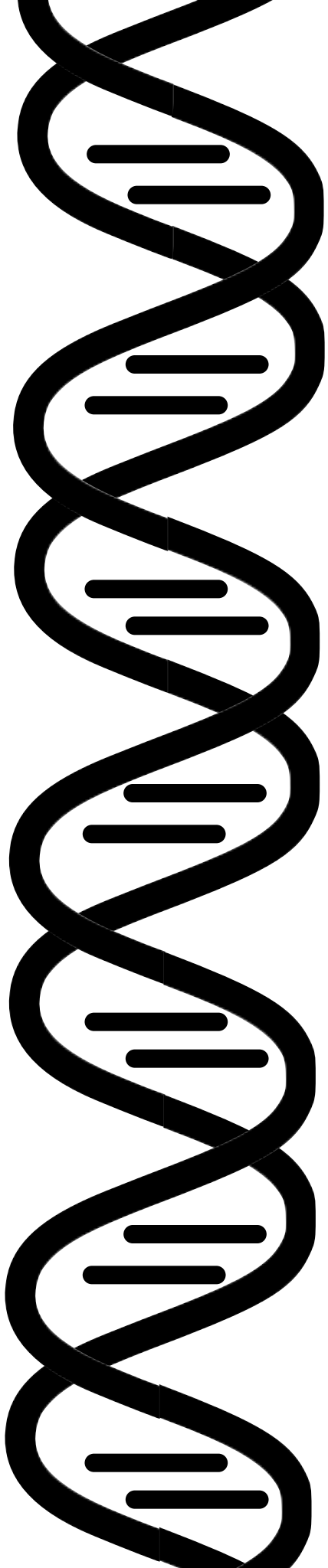


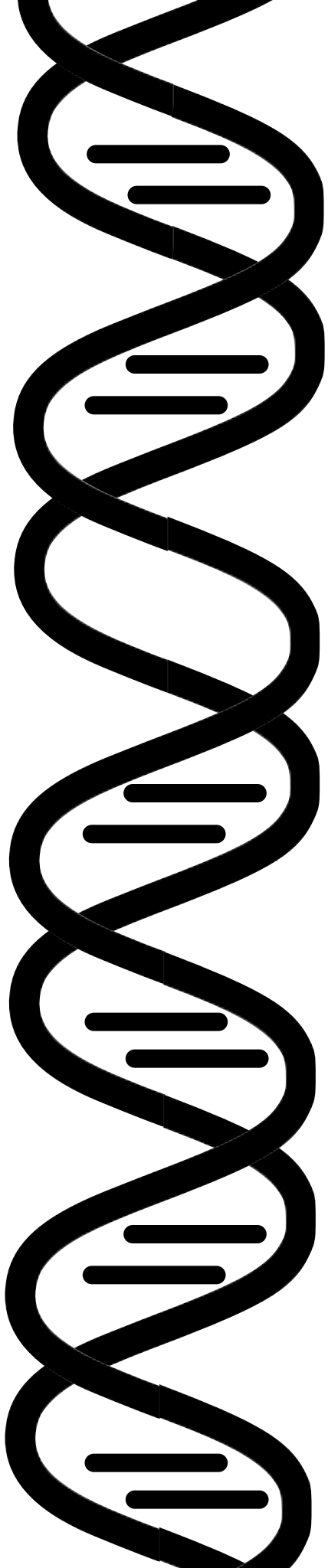


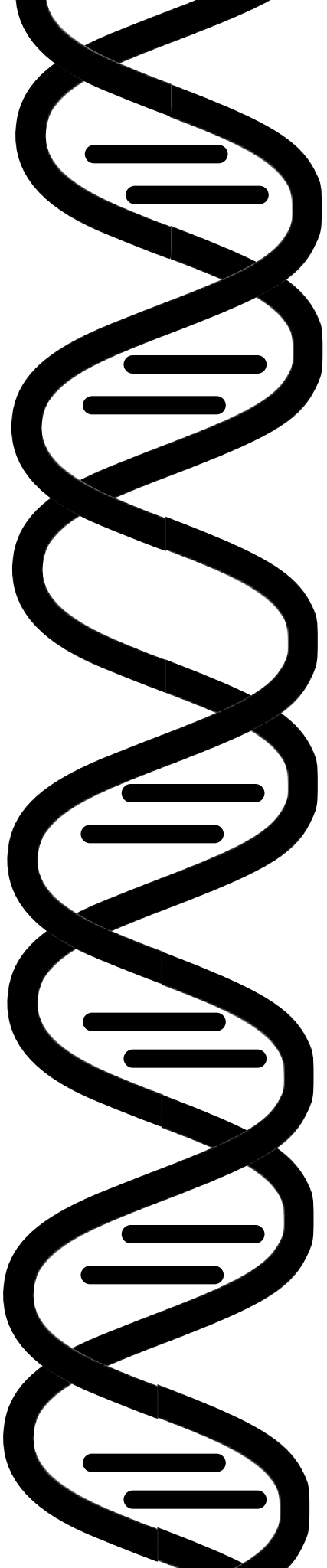
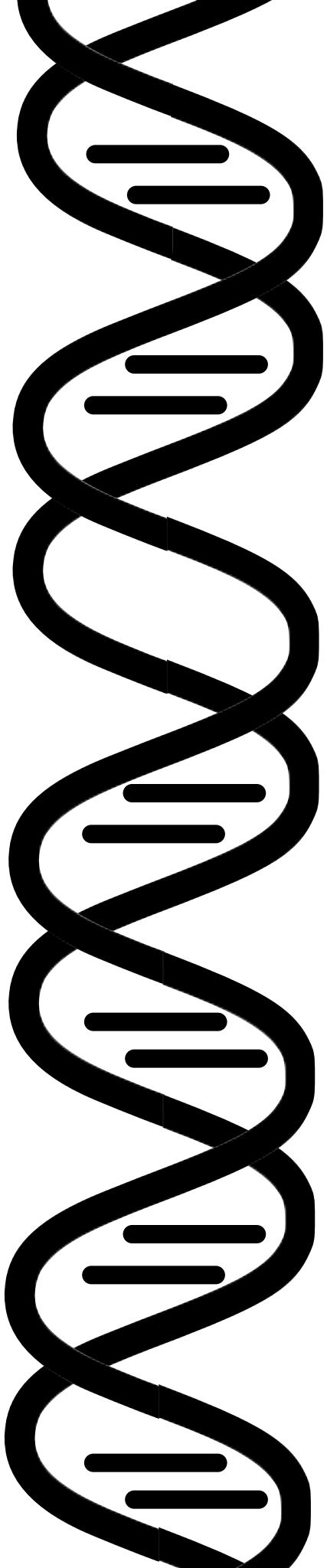
































Kids who don't cry: New genetic disorder discovered

By **Jacque Wilson**, CNN

updated 2:53 PM EDT, Thu March 20, 2014



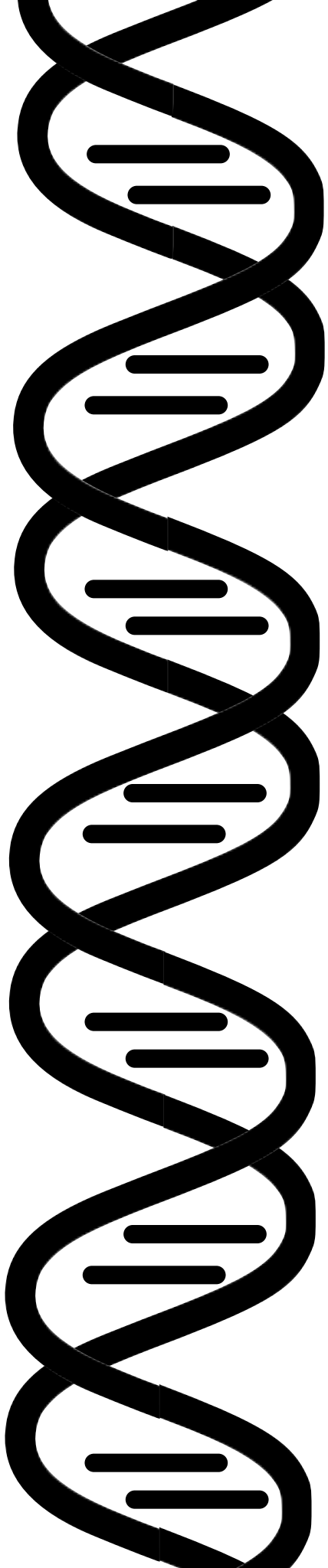


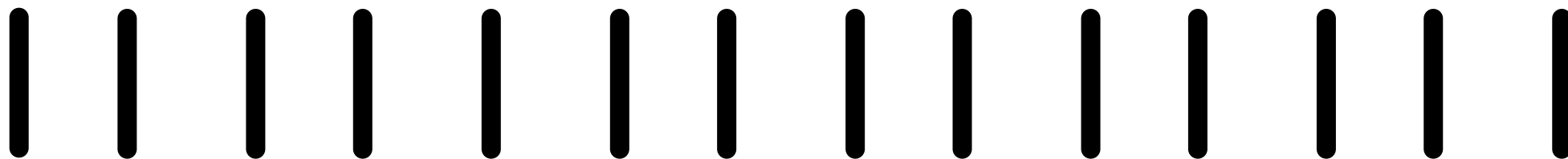




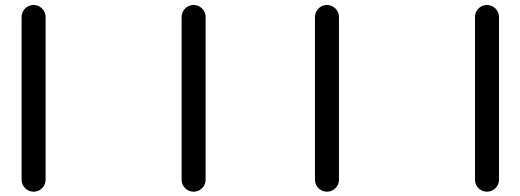
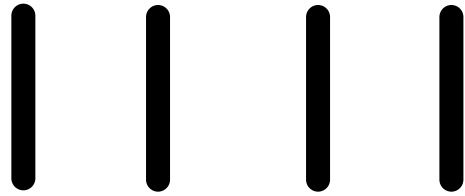
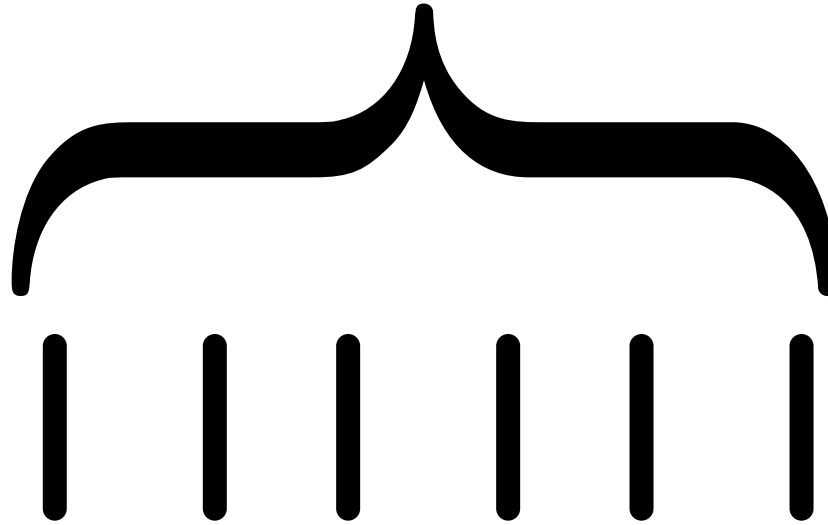
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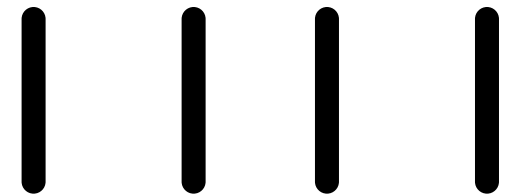
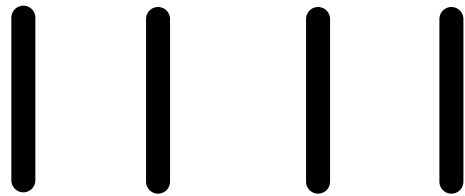
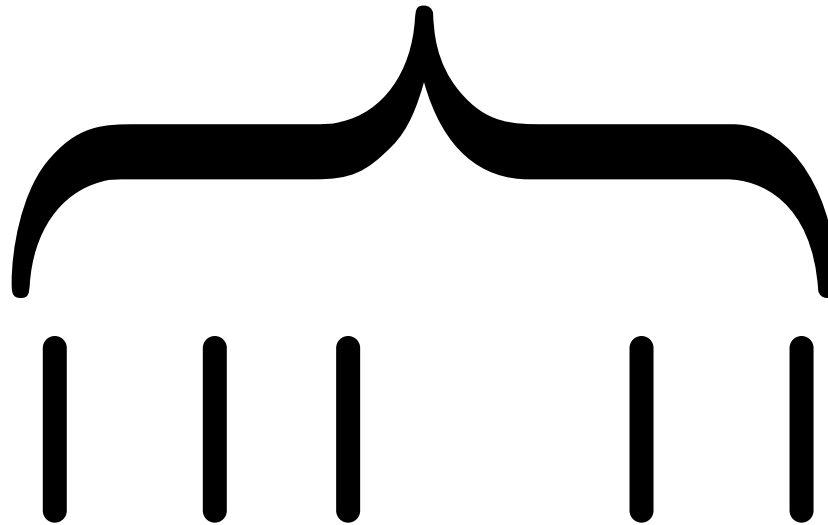


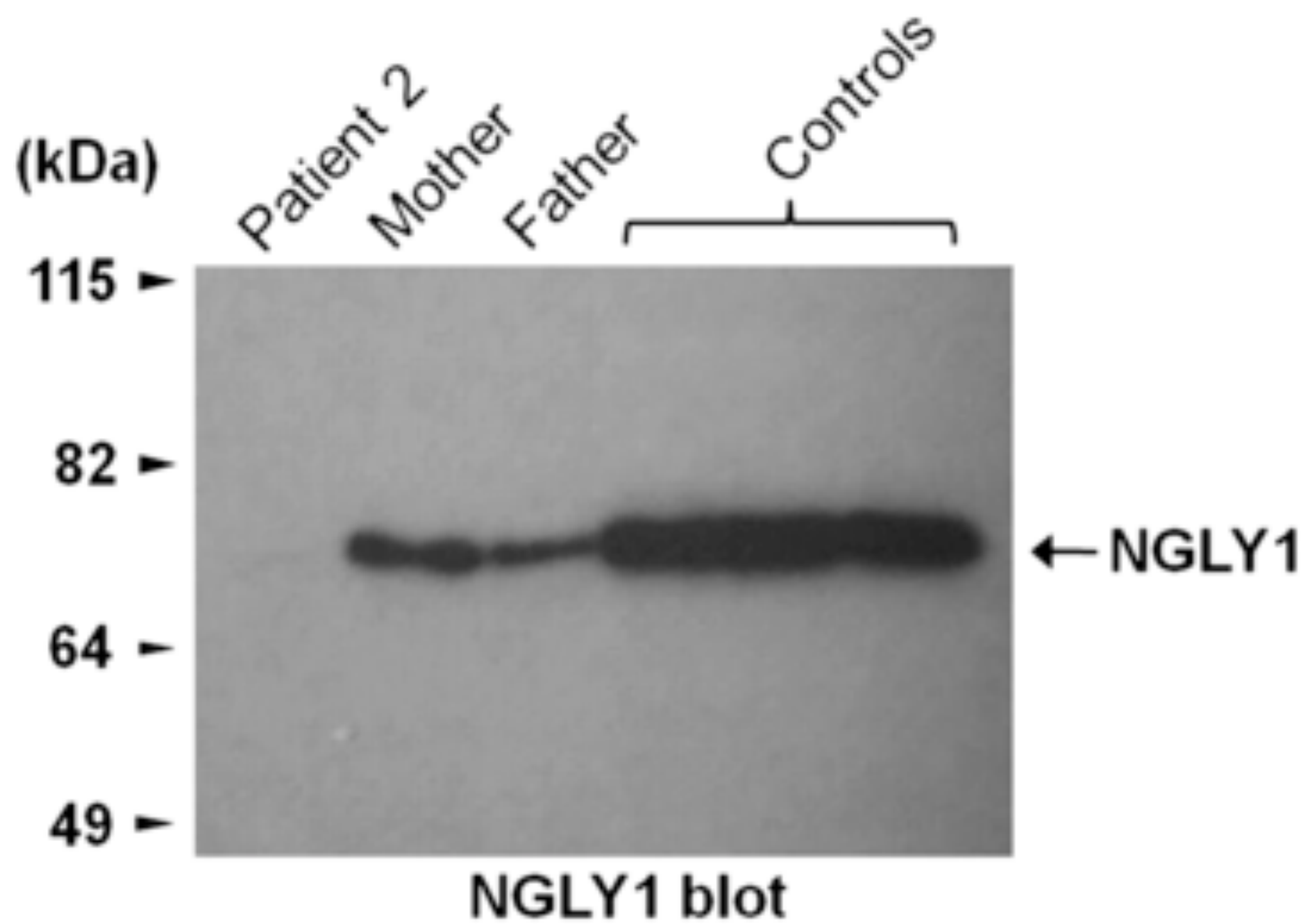


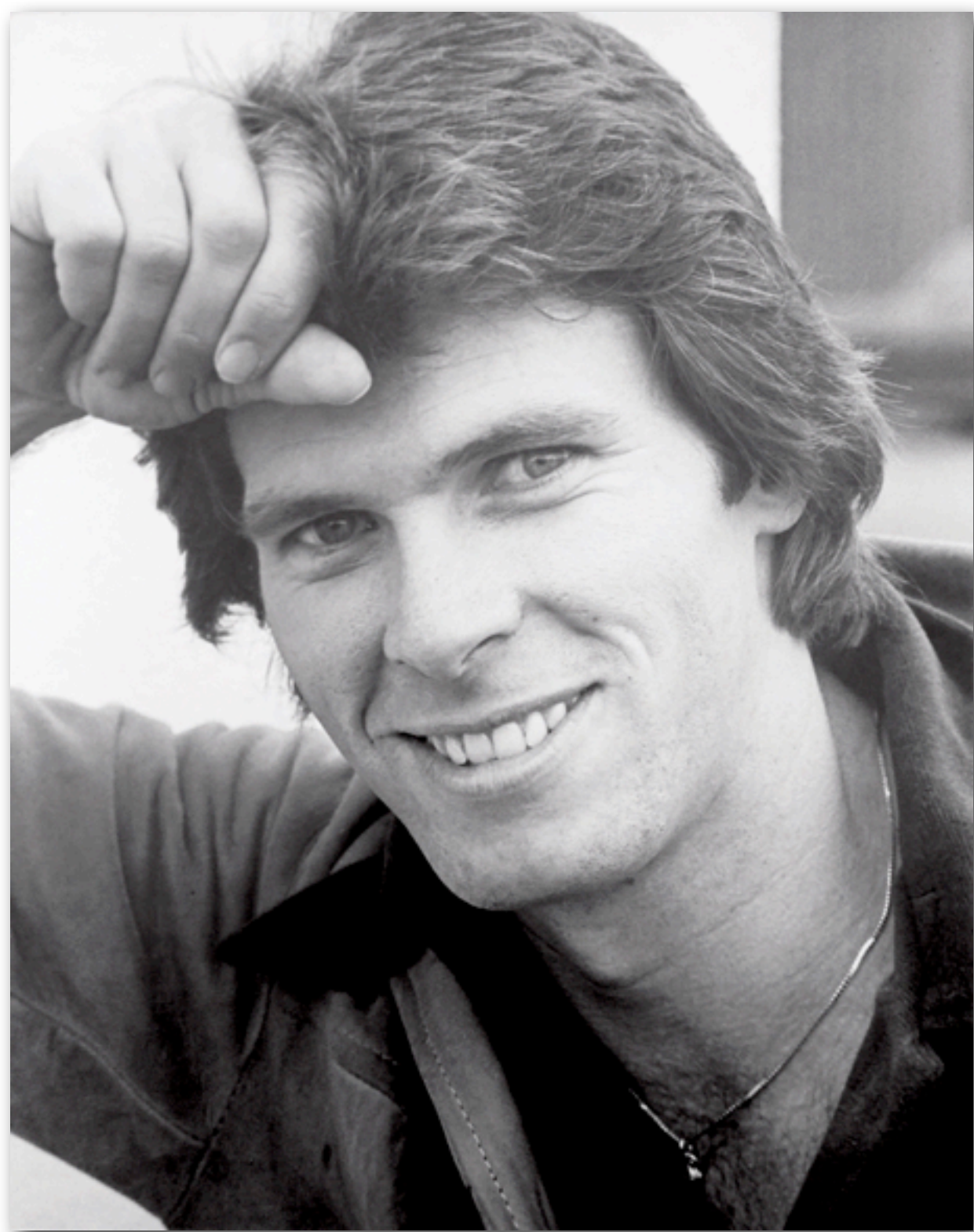
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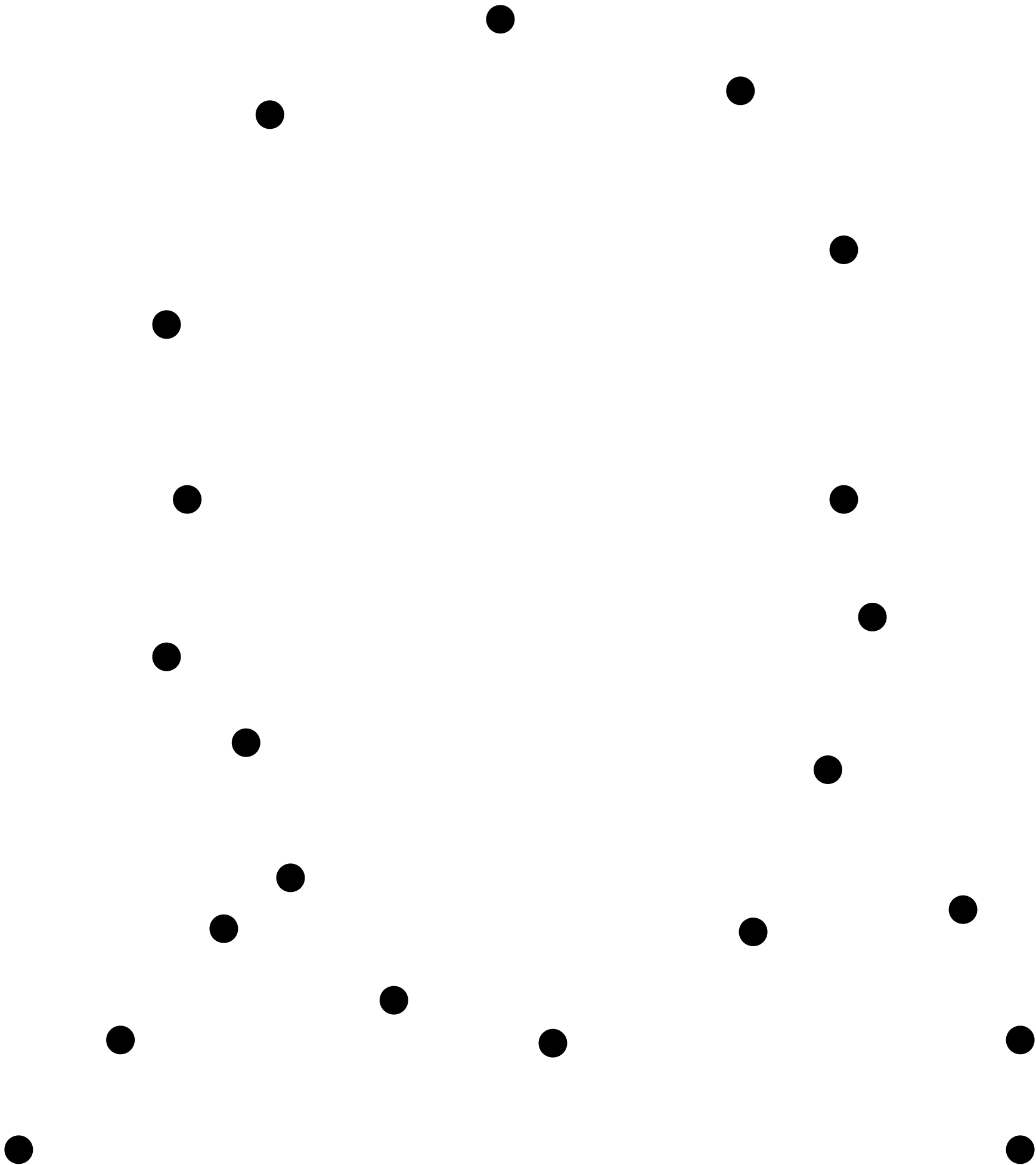


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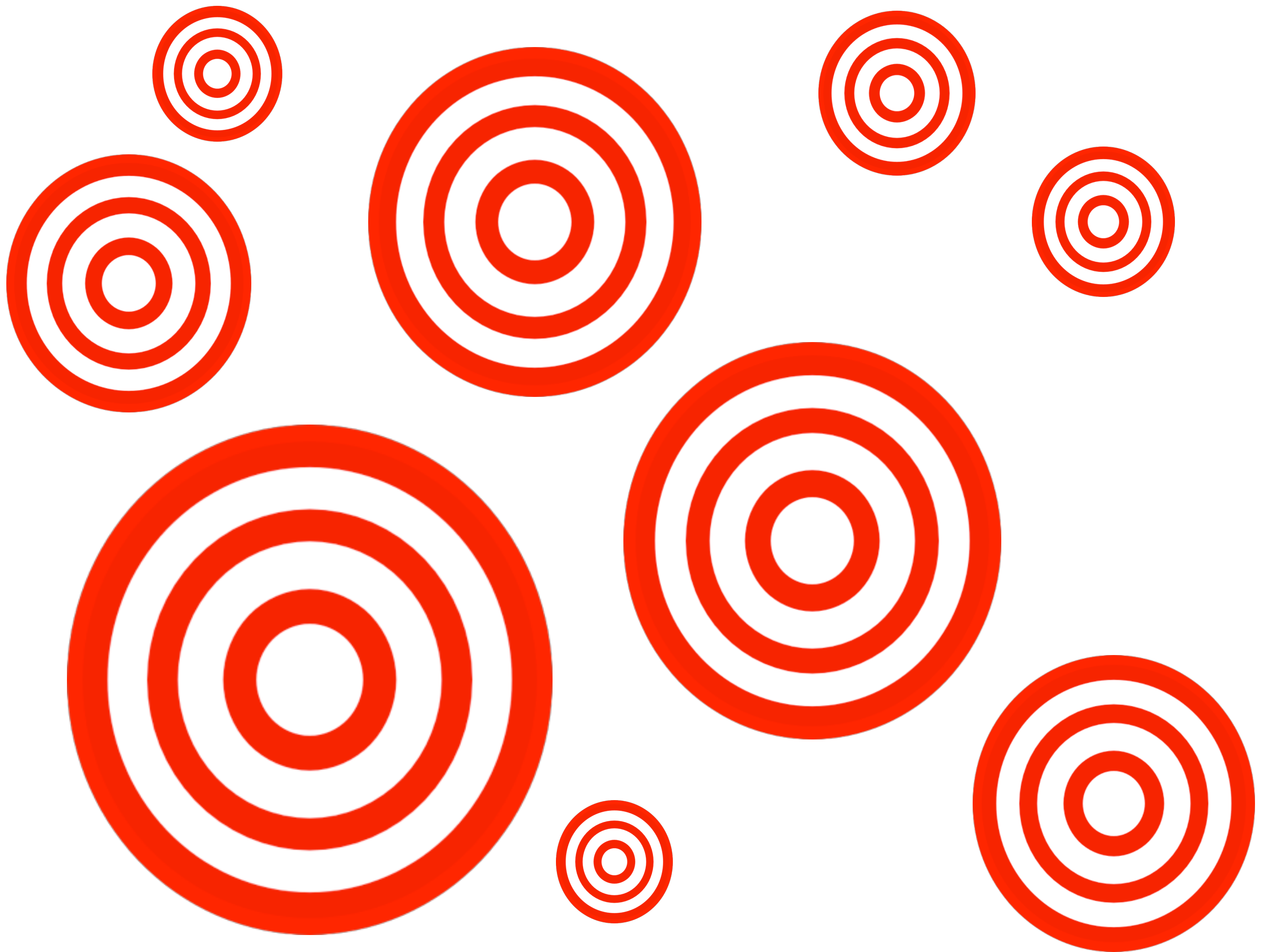














NGLY I



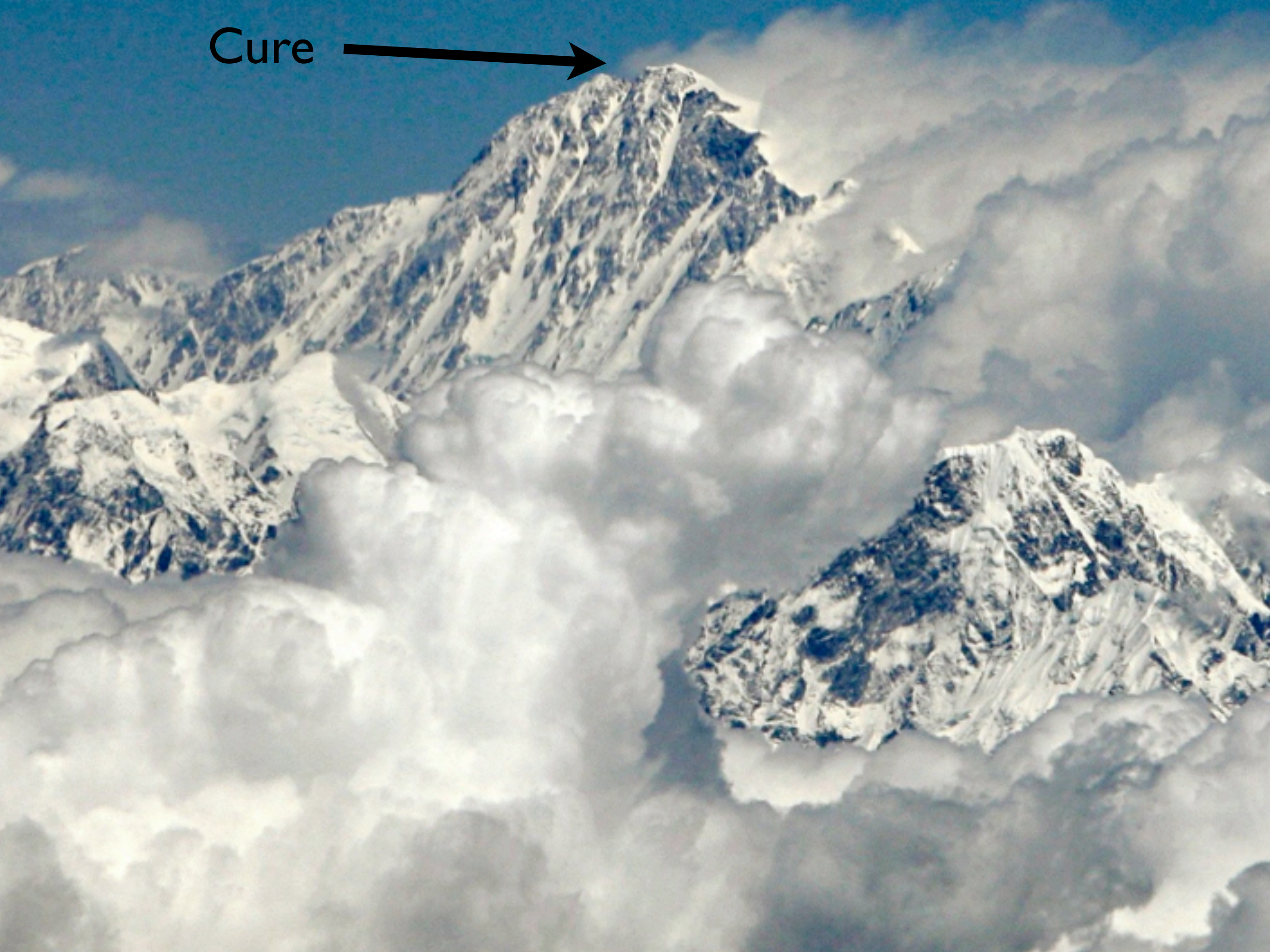
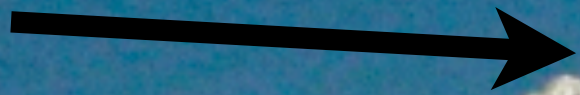




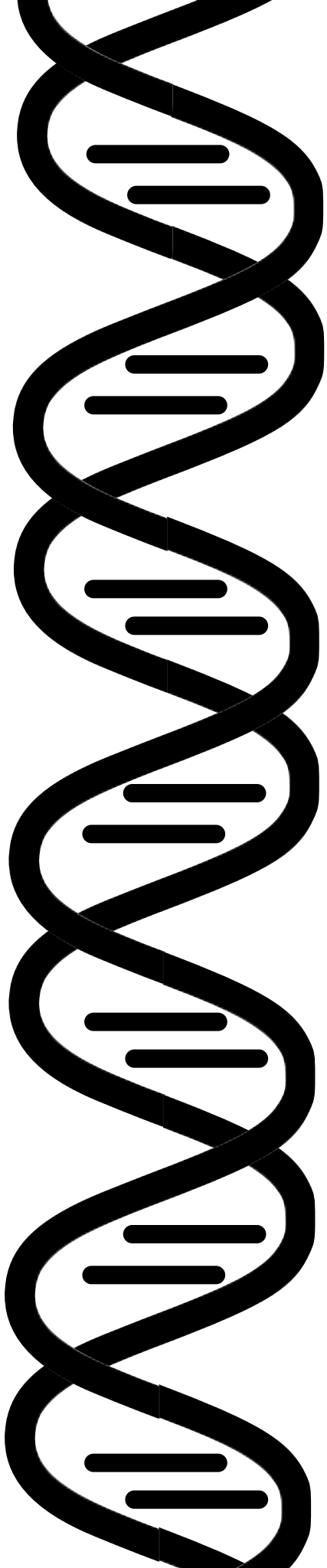
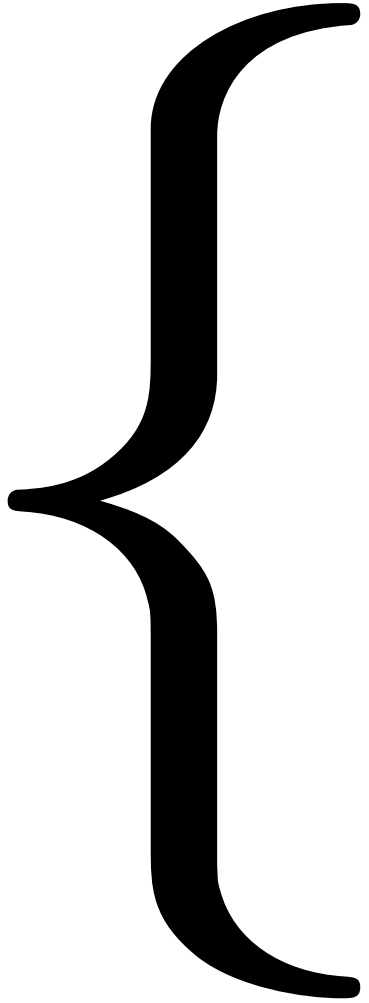




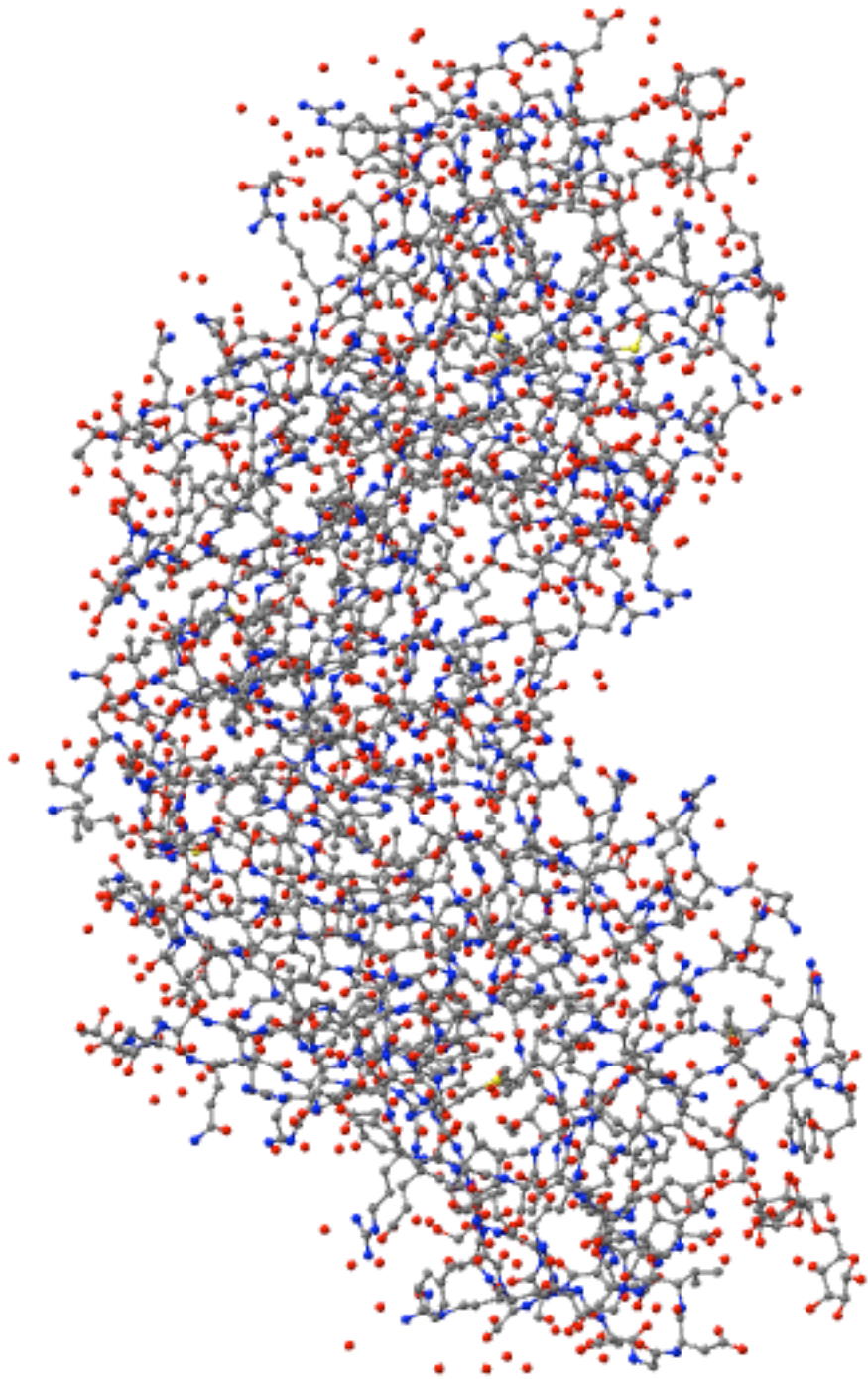
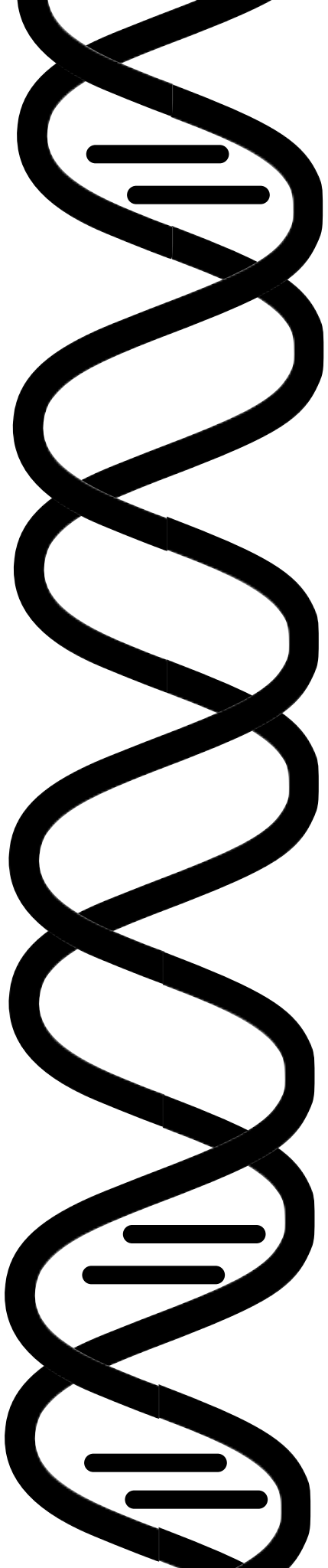
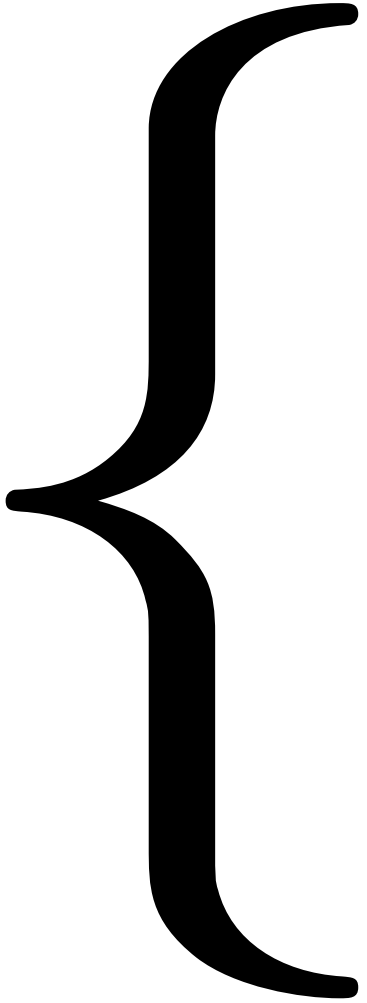
Cure

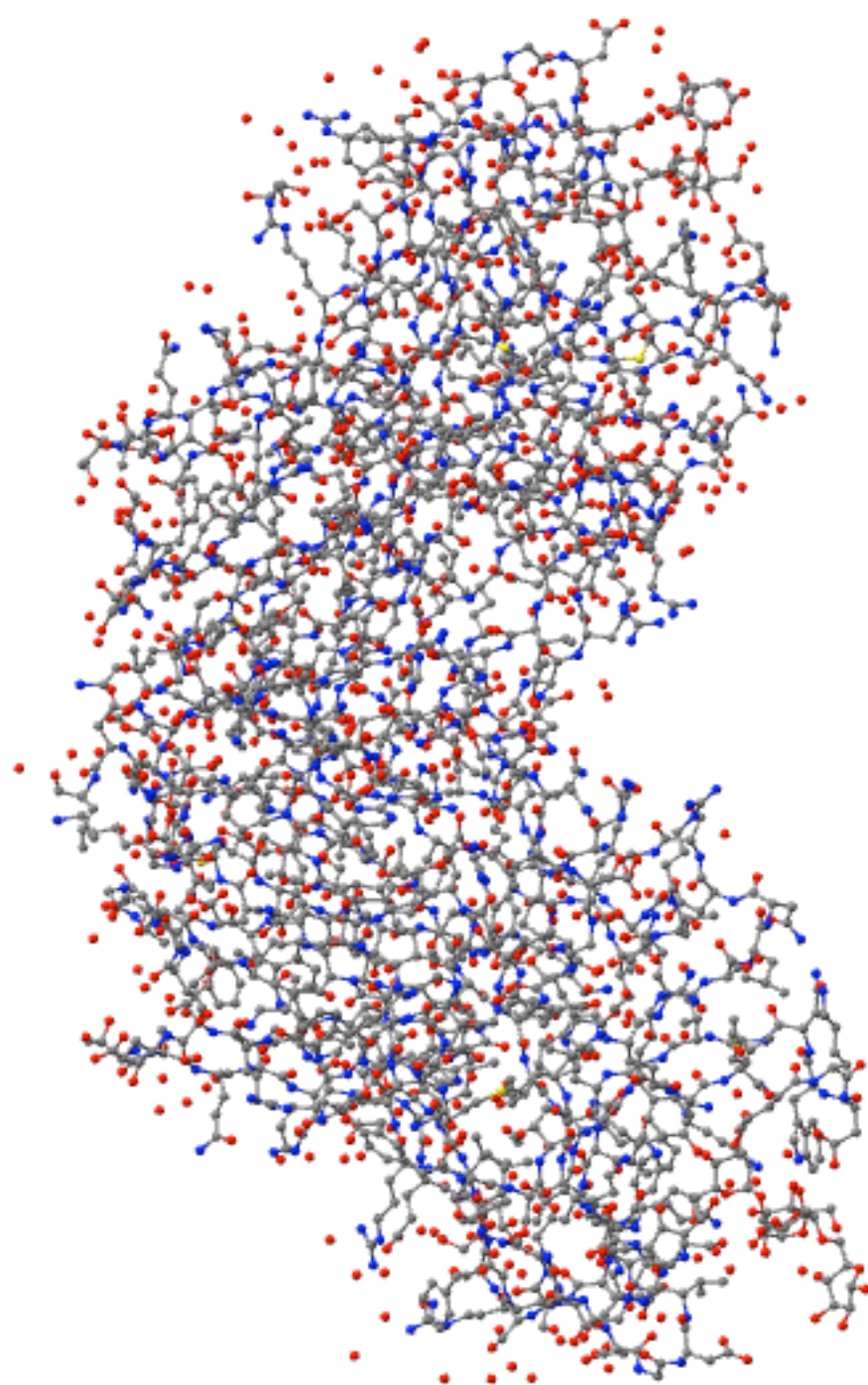


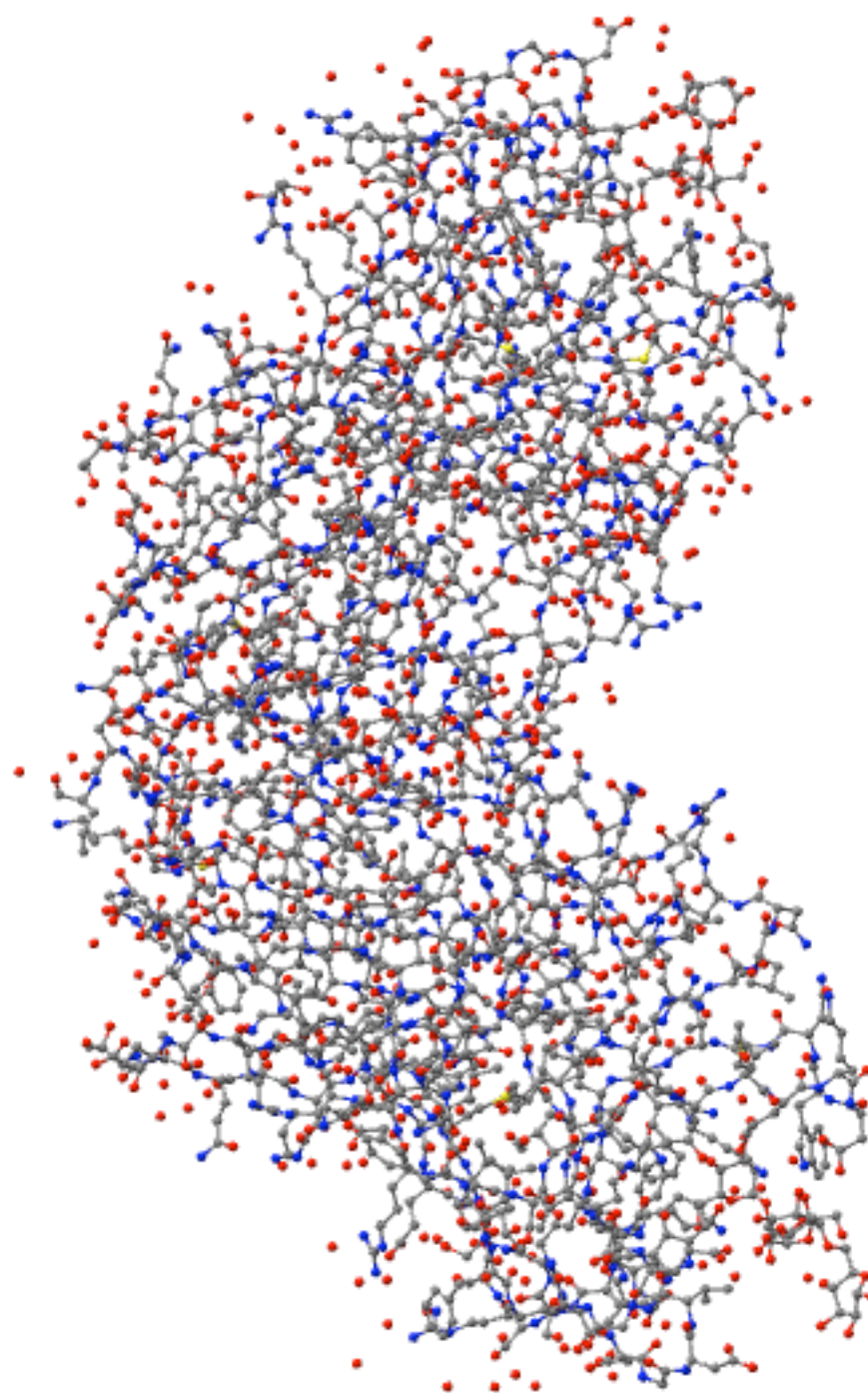
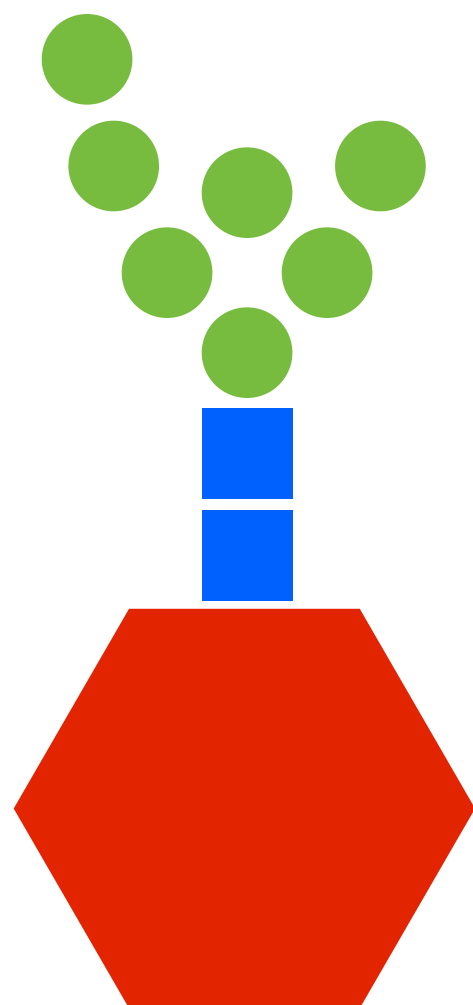
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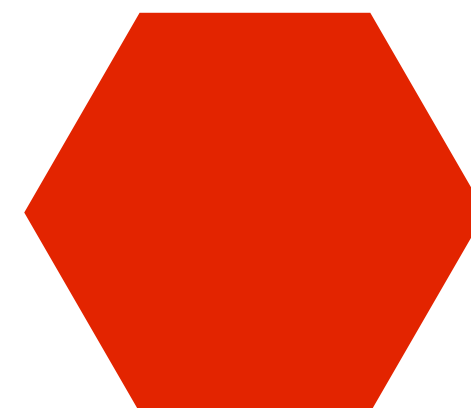
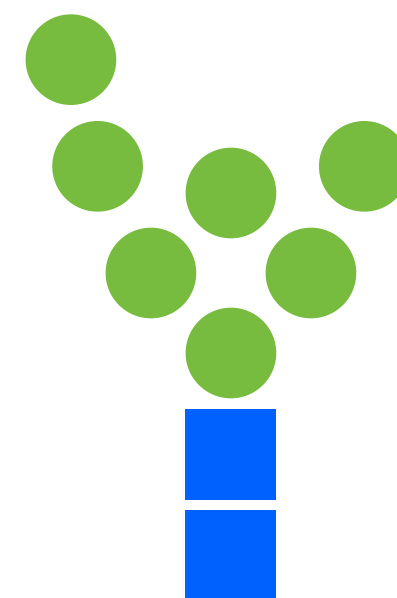
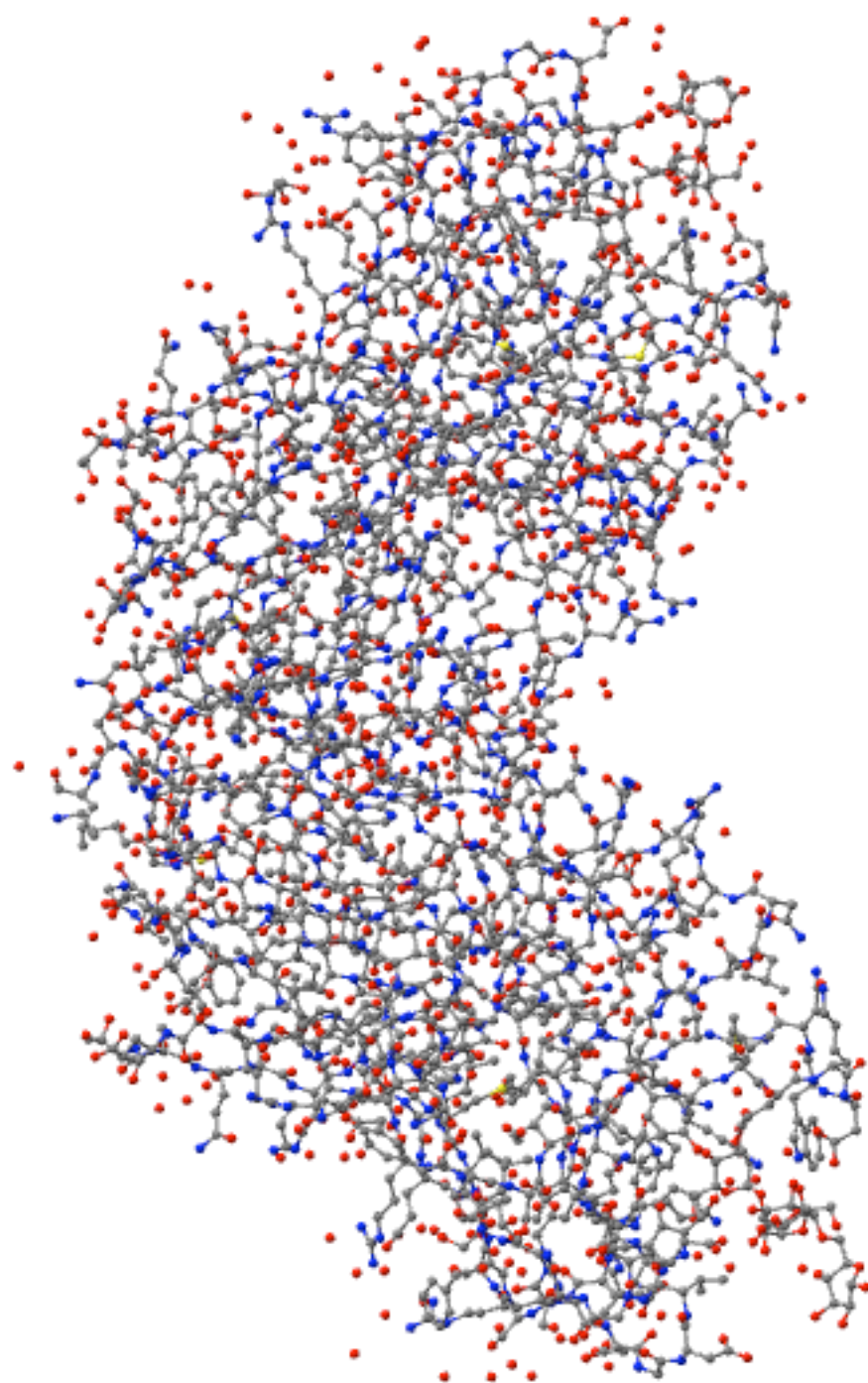


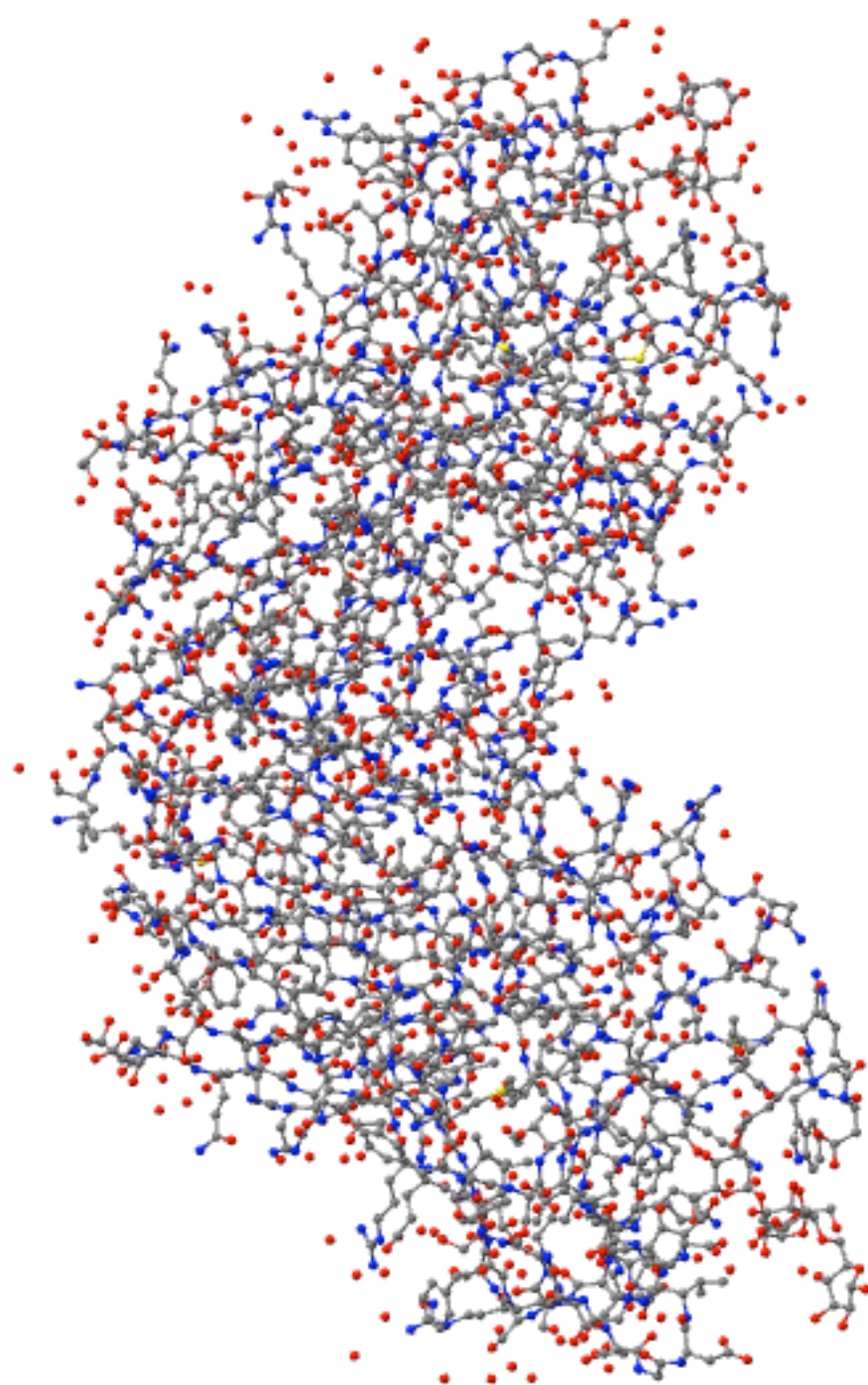
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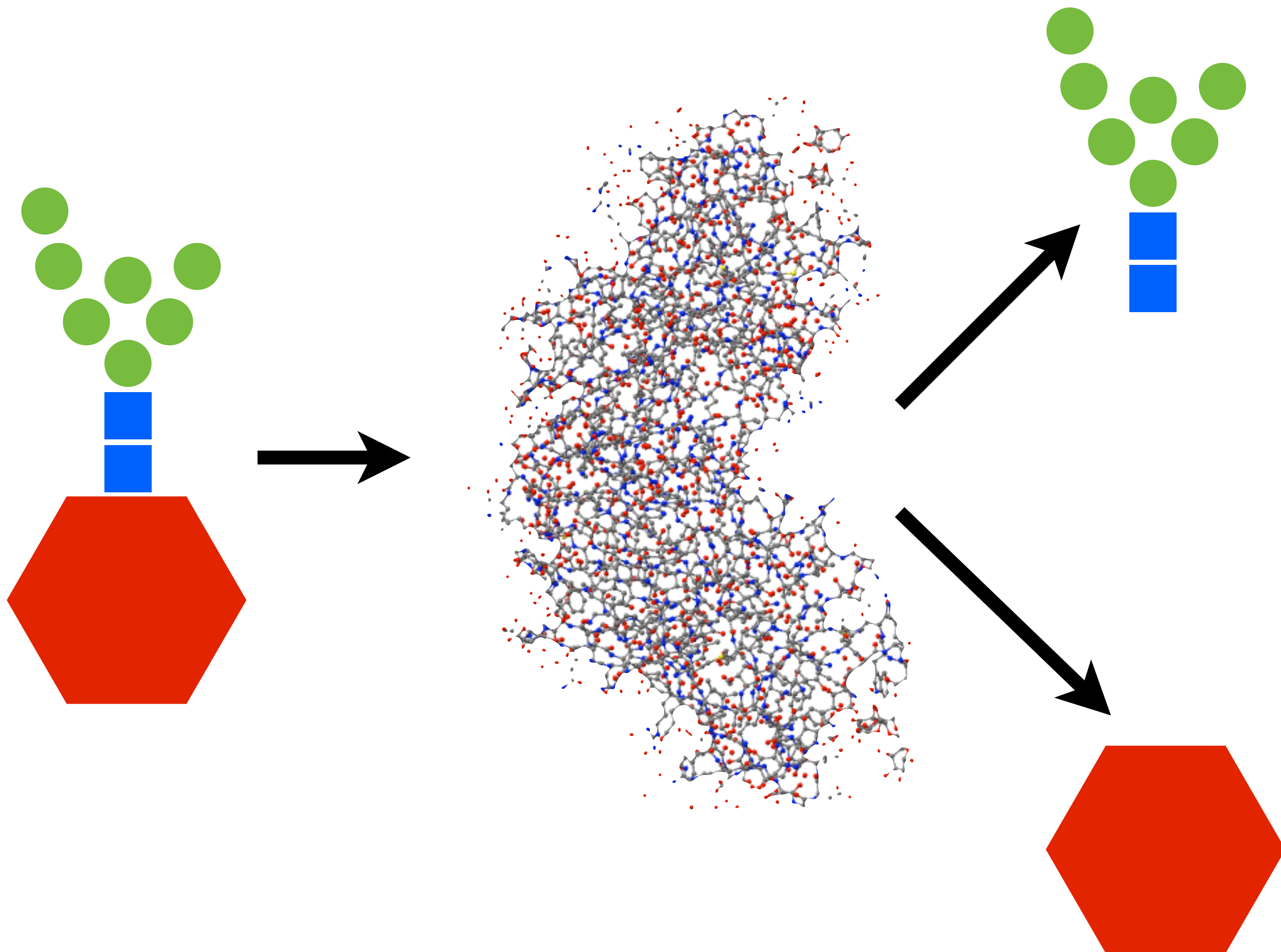


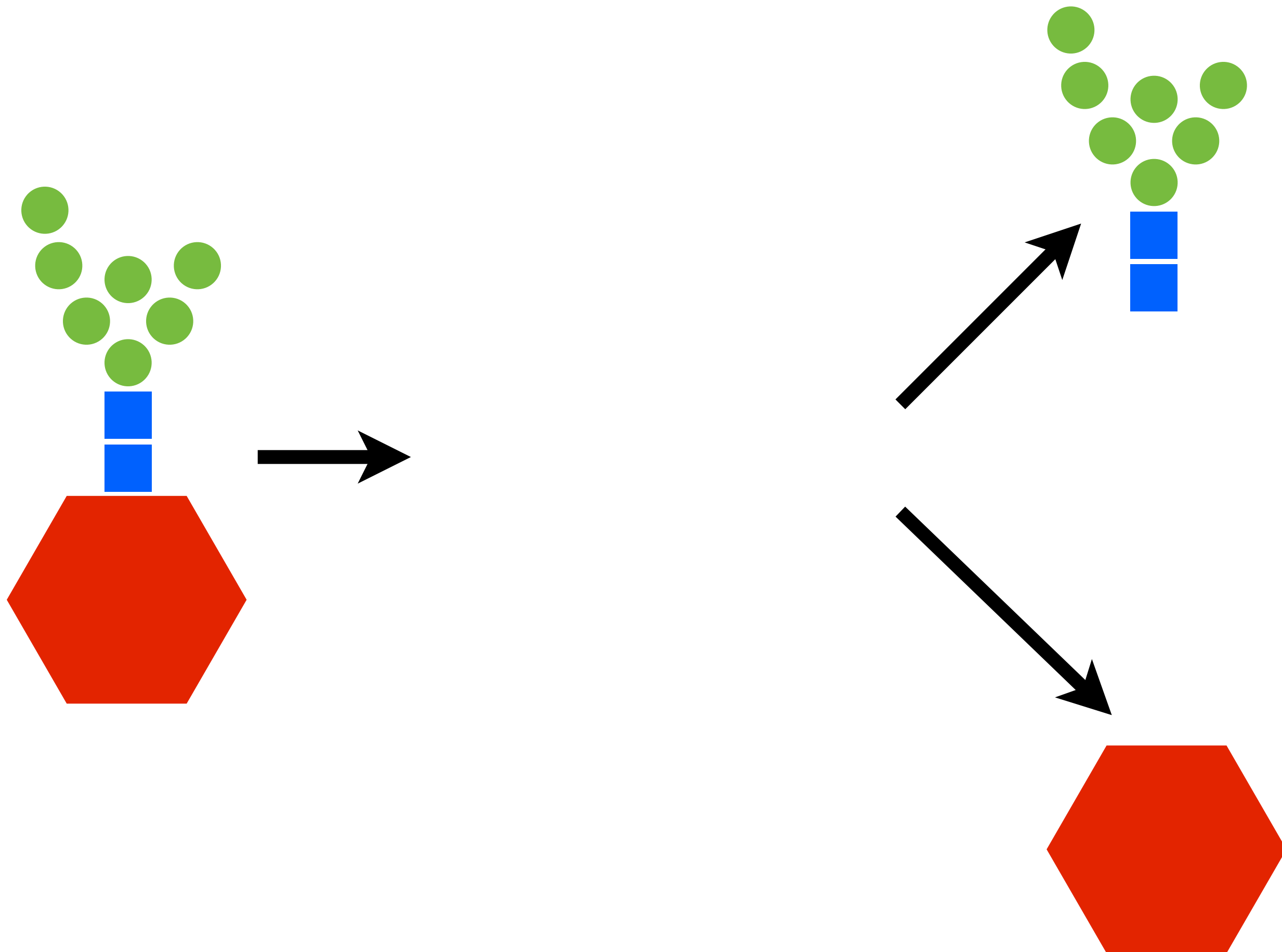


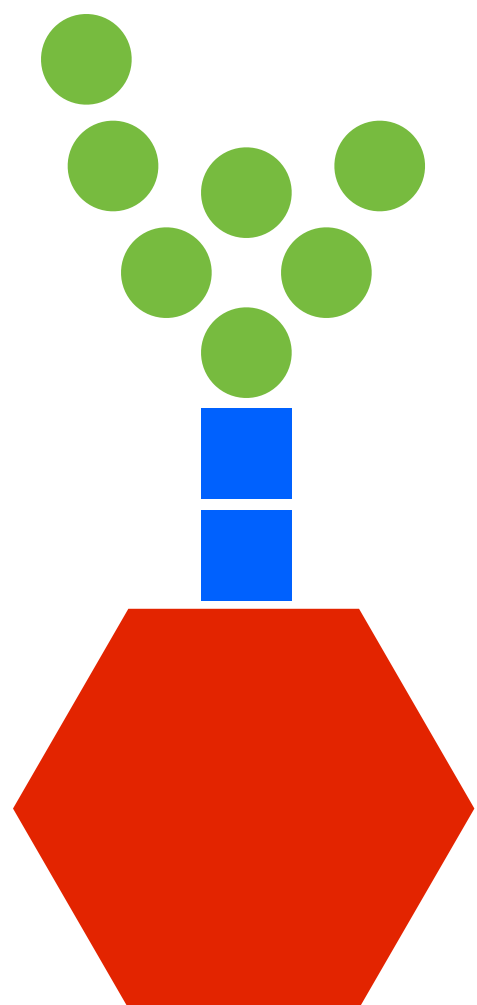


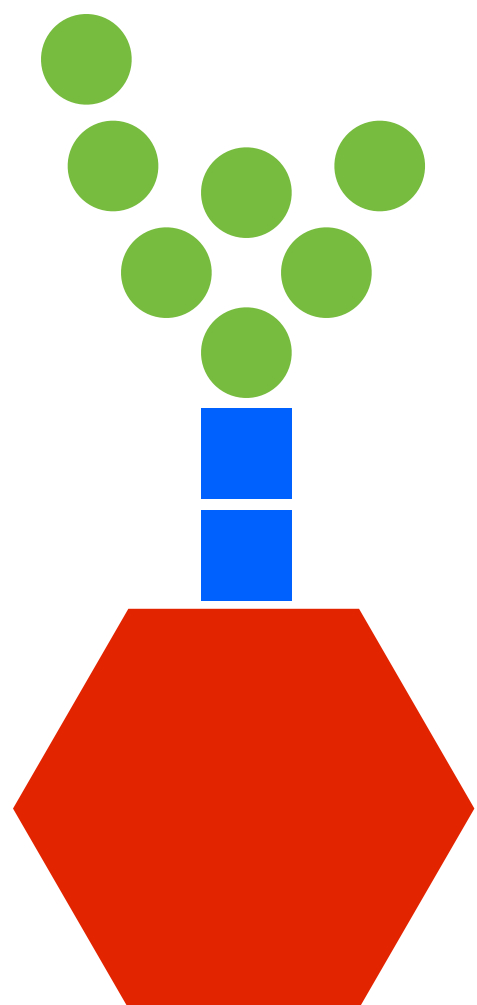


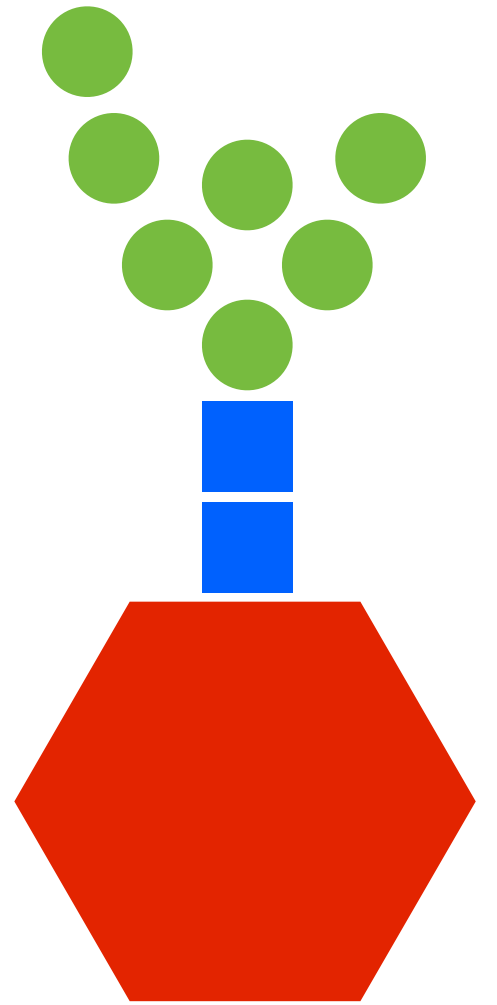
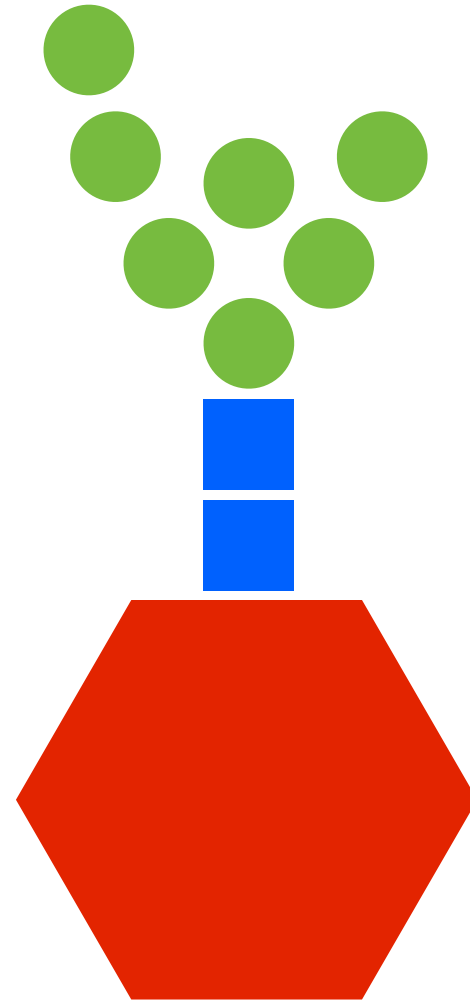
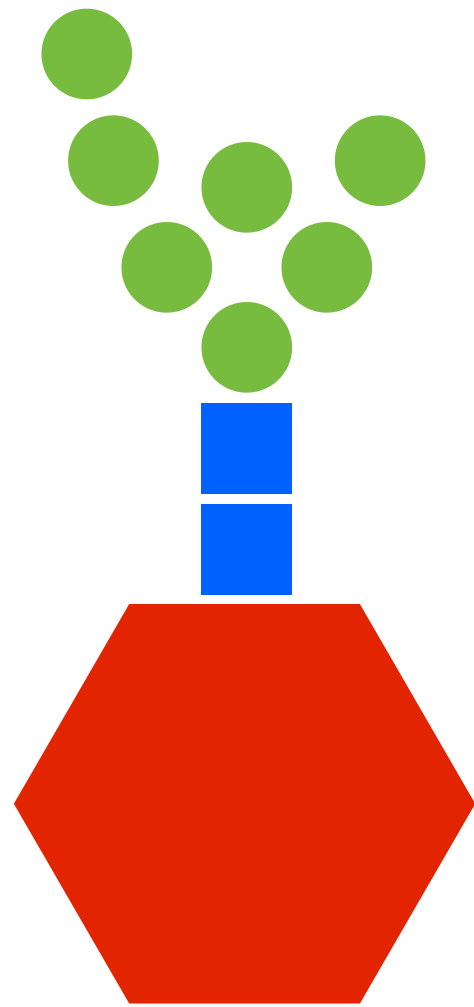
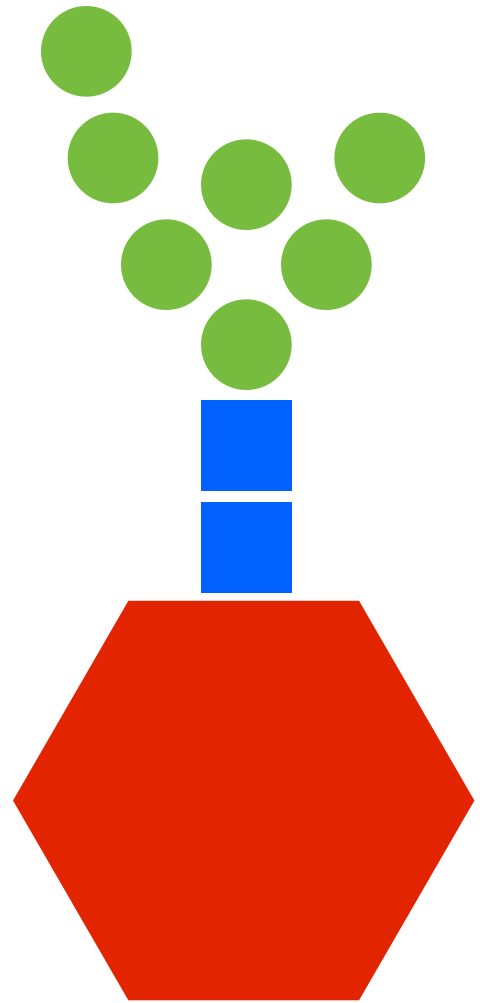












Hunting down my son's killer

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I found my son's killer.

It took three years.

But we did it.



Not quite like this.

2,000,000+



NGLY1



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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.

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

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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.



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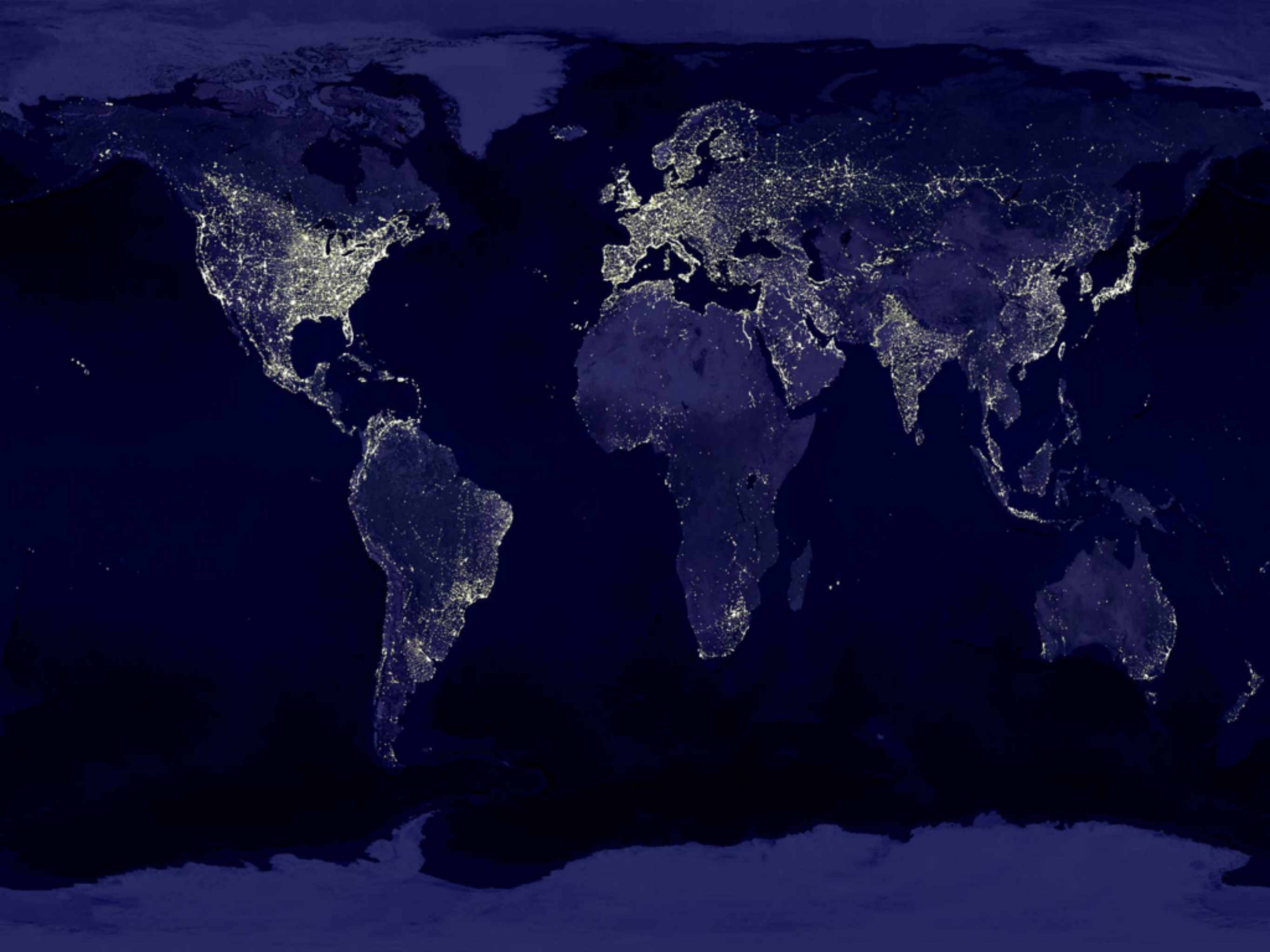
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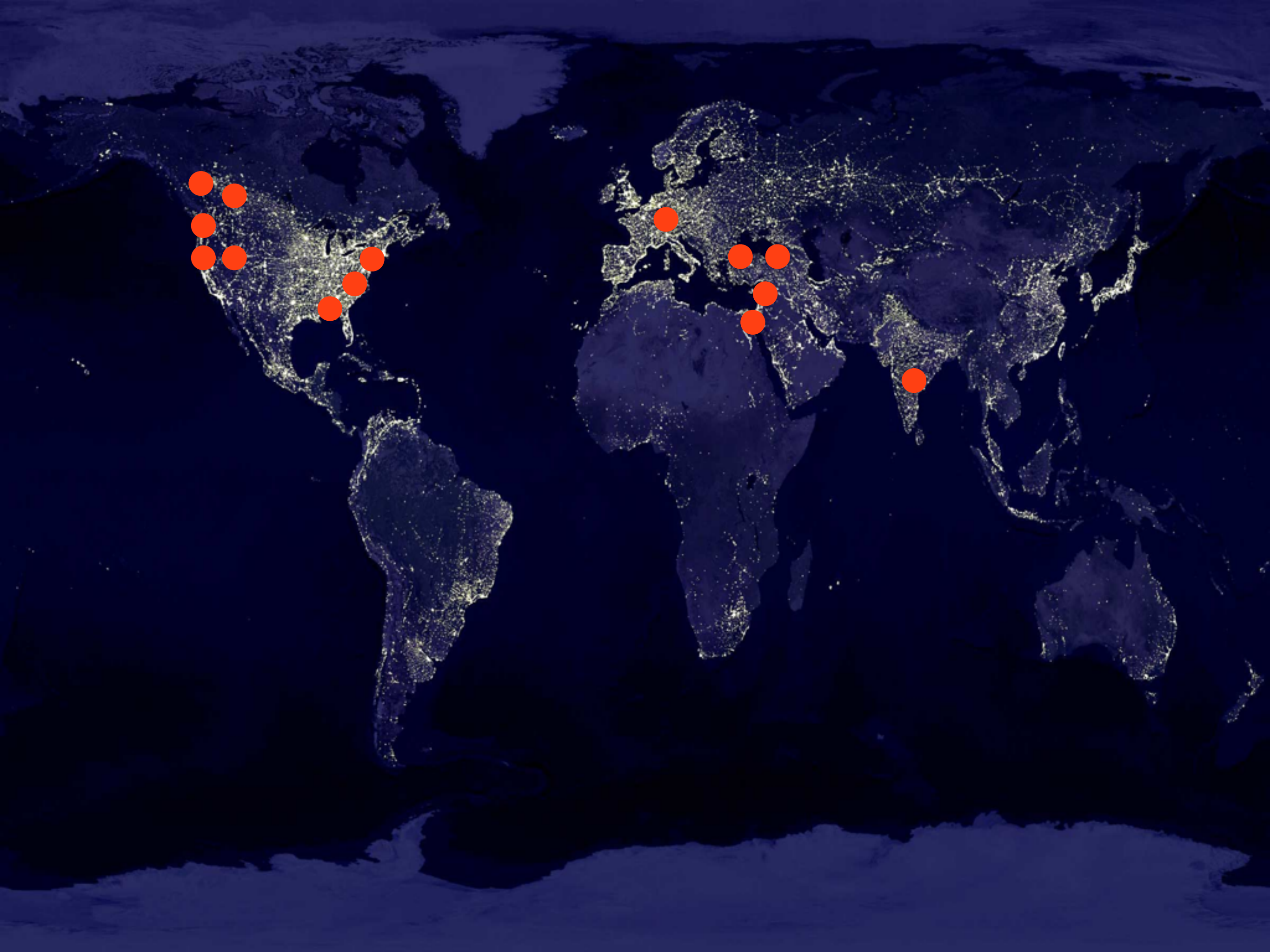
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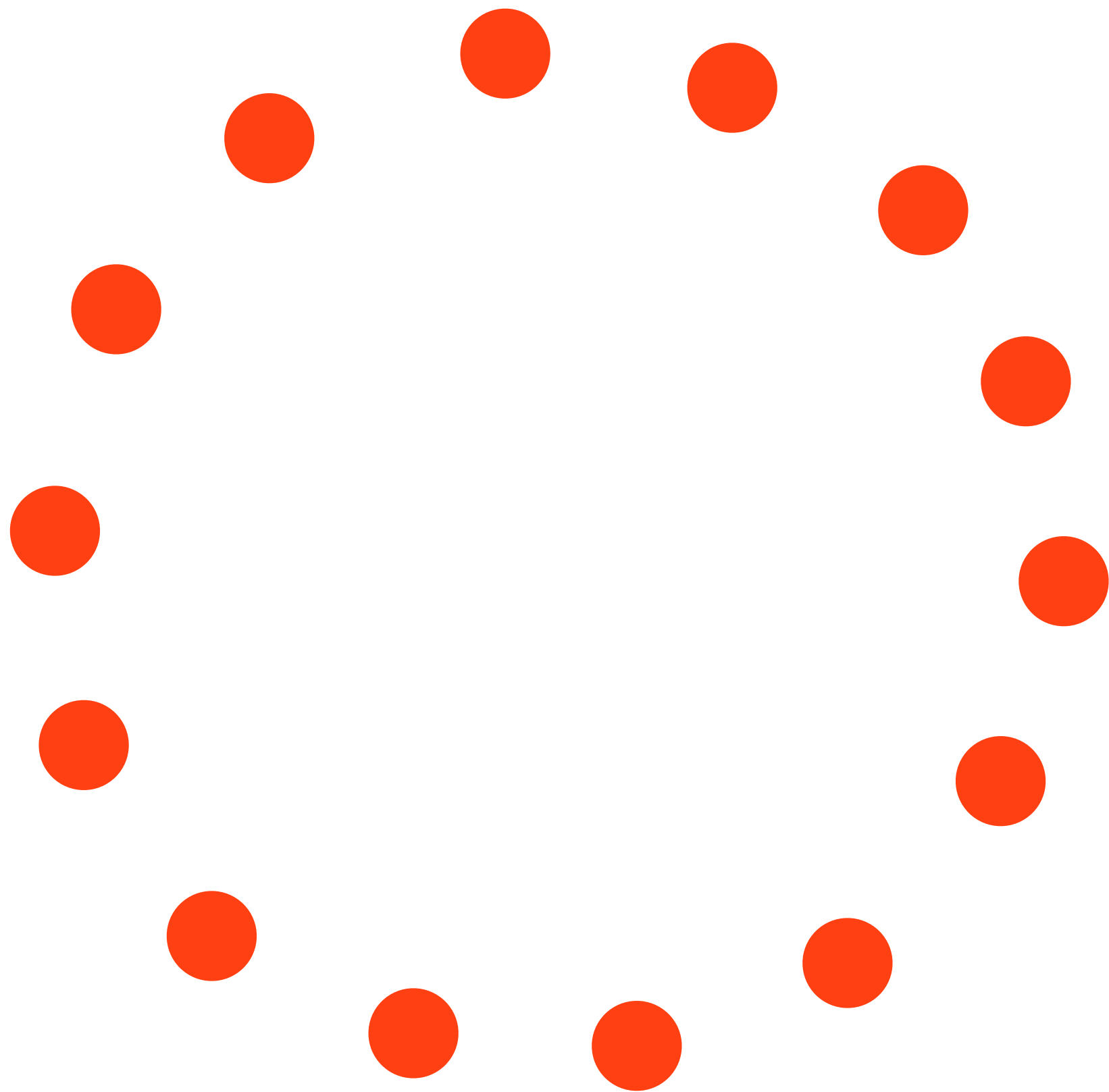
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THE RAREST DISEASE

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Patient Bertrand Might (r.), Familie

Die Kinder ohne Tränen

Medizin Überall auf der Welt leiden Kinder an mysteriösen Krankheiten, die so selten sind, dass kein Arzt die richtige Diagnose stellt. Nun ist es Genforschern gelungen, ein solches Erbleiden zu entschlüsseln – weil die Eltern nicht aufgaben.

„The Genome is a language we are just learning“ (Inchrift auf dem Uni-Campus in Stanford).

Kinder sind meist keine gern gesehenen Gäste auf medizinischen Fachtagungen. Doch beim Symposium für seltene Krankheiten des Sanford-Burnham-Forschungszentrums im kalifornischen La Jolla in diesem Februar saßen sie in der ersten Reihe: Bertrand aus Salt Lake City, Grace aus Menlo Park und Tim aus

Deutschland. Bertrands Schwester hatte einen Ball mitgebracht. Tim robbte ihm auf dem Boden vor dem Podium hinterher.

Während die Kinder spielten, hielten die Forscher ihre Vorträge. Es waren Kliniker, Stoffwechselexperten und Genetiker von Weltrang. Statt vom Kinderlärm genervt zu sein, waren viele den Tränen nahe. „Das war ein magischer Moment“, erinnert sich Hudson Freeze, Direktor am Sanford Children's Health Research Center in La Jolla, „einer der besten in meiner Karriere.“

Was Freeze und seine Kollegen so rührte, war das Zusammentreffen von Kindern, die das gleiche Schicksal teilen. Sie begegneten sich am Ende einer langen Reise „durch die diagnostische Wüste“, wie es Freeze beschreibt.

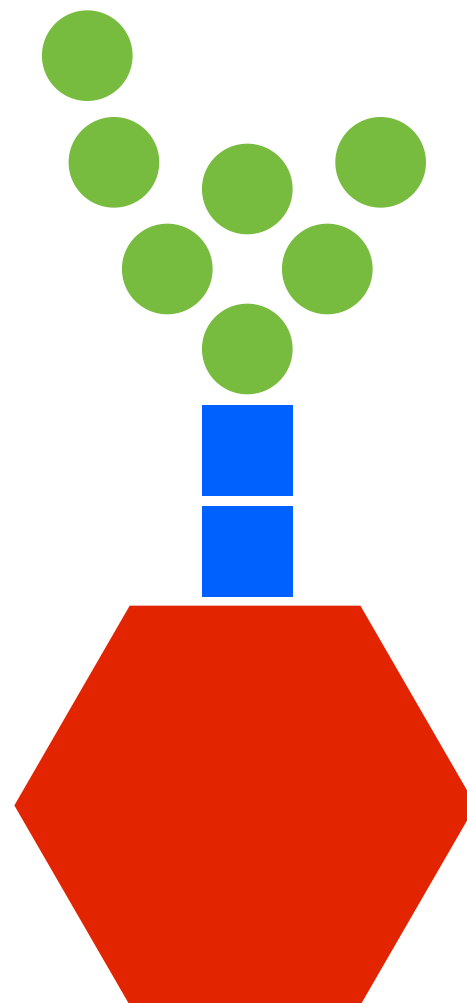
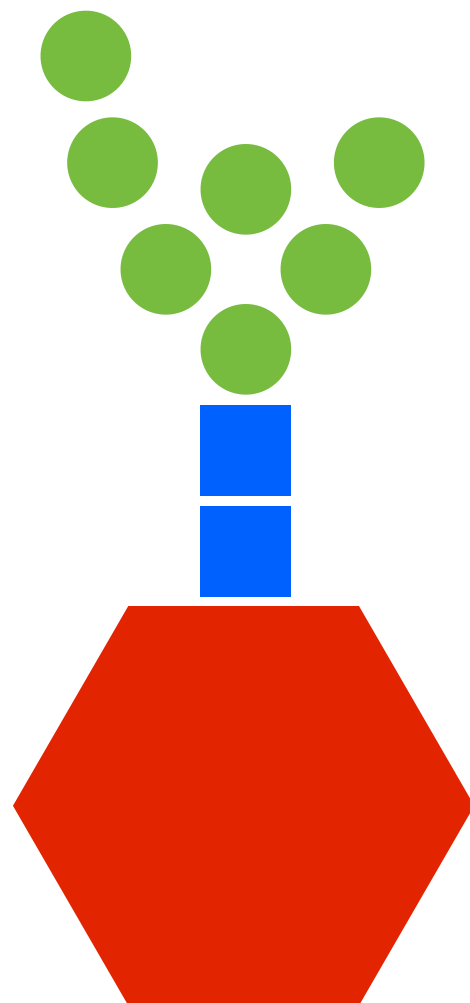
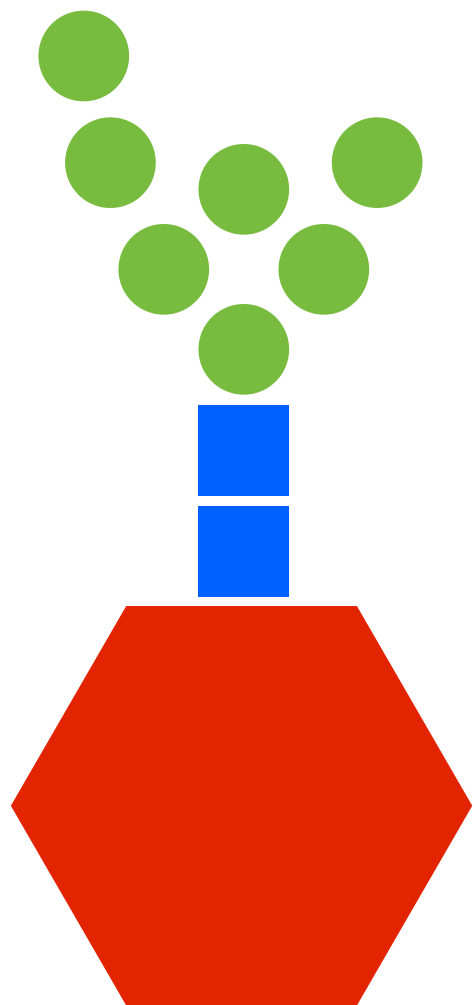
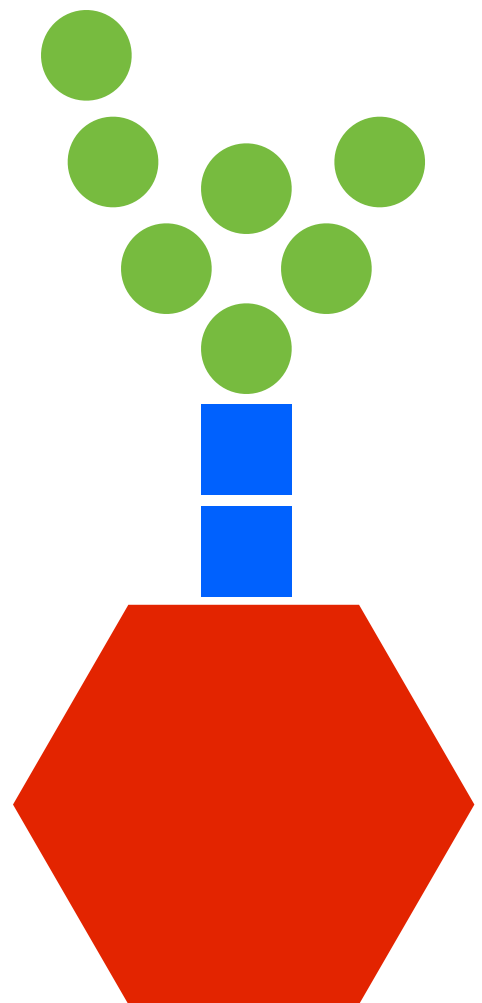
Bertrand, Grace und Tim leiden an einer sehr seltenen Krankheit, die bis vor Kurzem noch unbekannt war und erst mithilfe moderner molekularbiologischer Methoden entschlüsselt werden konnte. Ihre Geschichte erzählt vom Wandel, der sich ge-

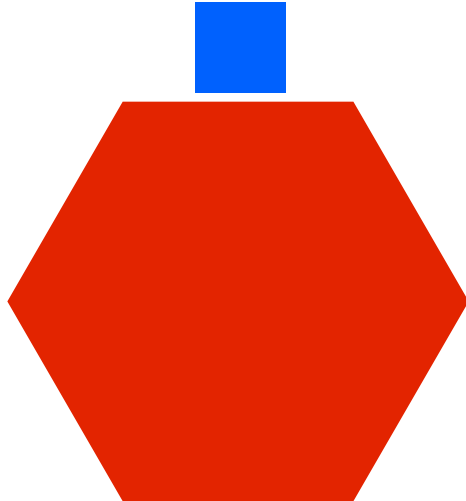
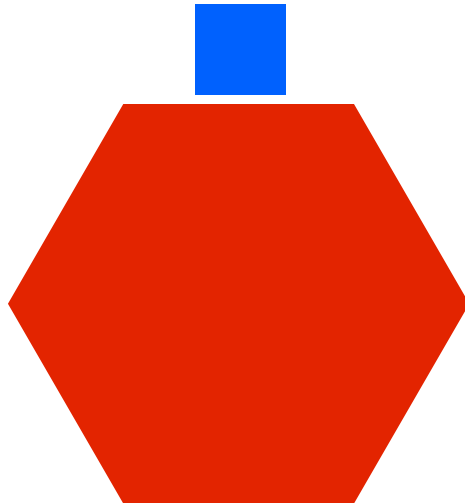
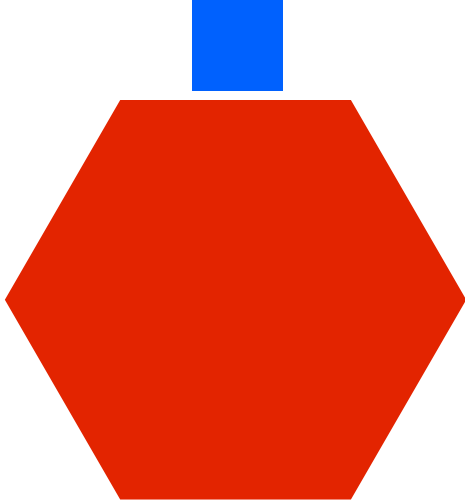
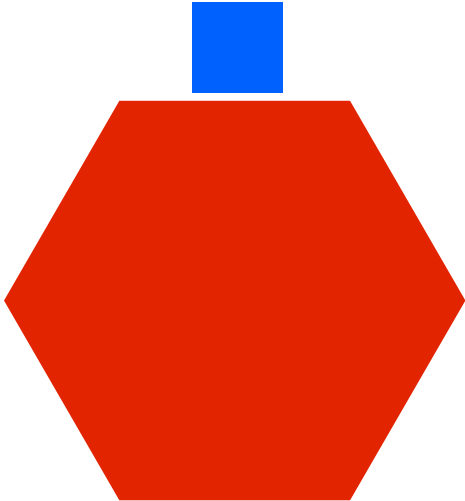
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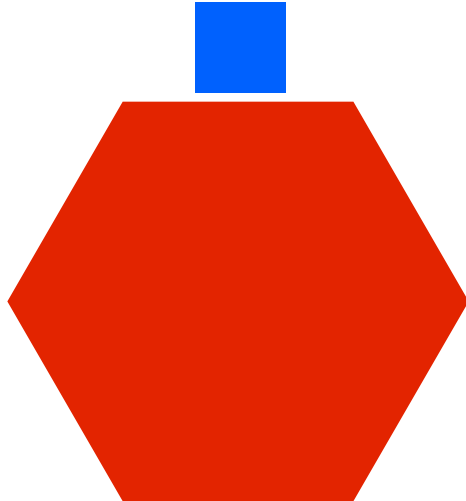
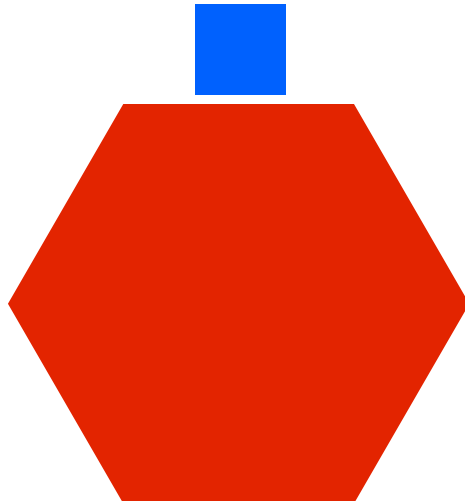
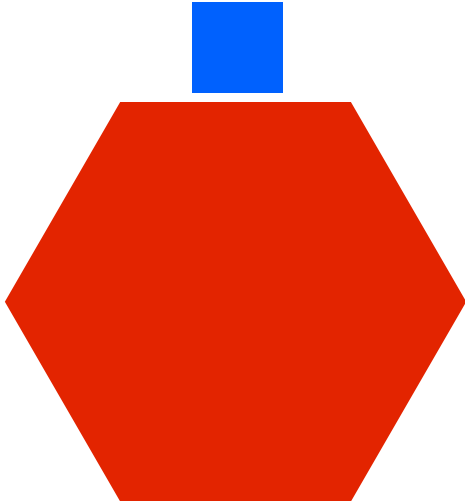
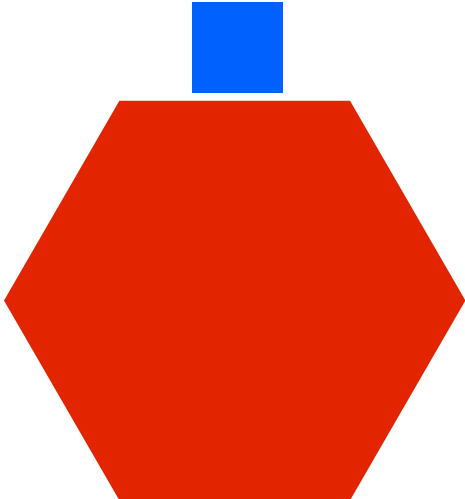
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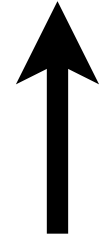








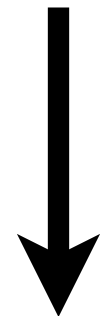
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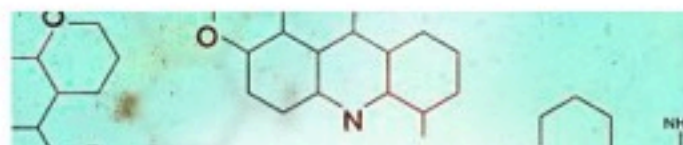
Is it possible to control cancer without killing it?

BY JEROME GROOPMAN



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For almost thirty years, William Kuhens worked on Staten Island as a basketball referee for the



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“One of a kind.” Seth Mnookin. *The New Yorker*.