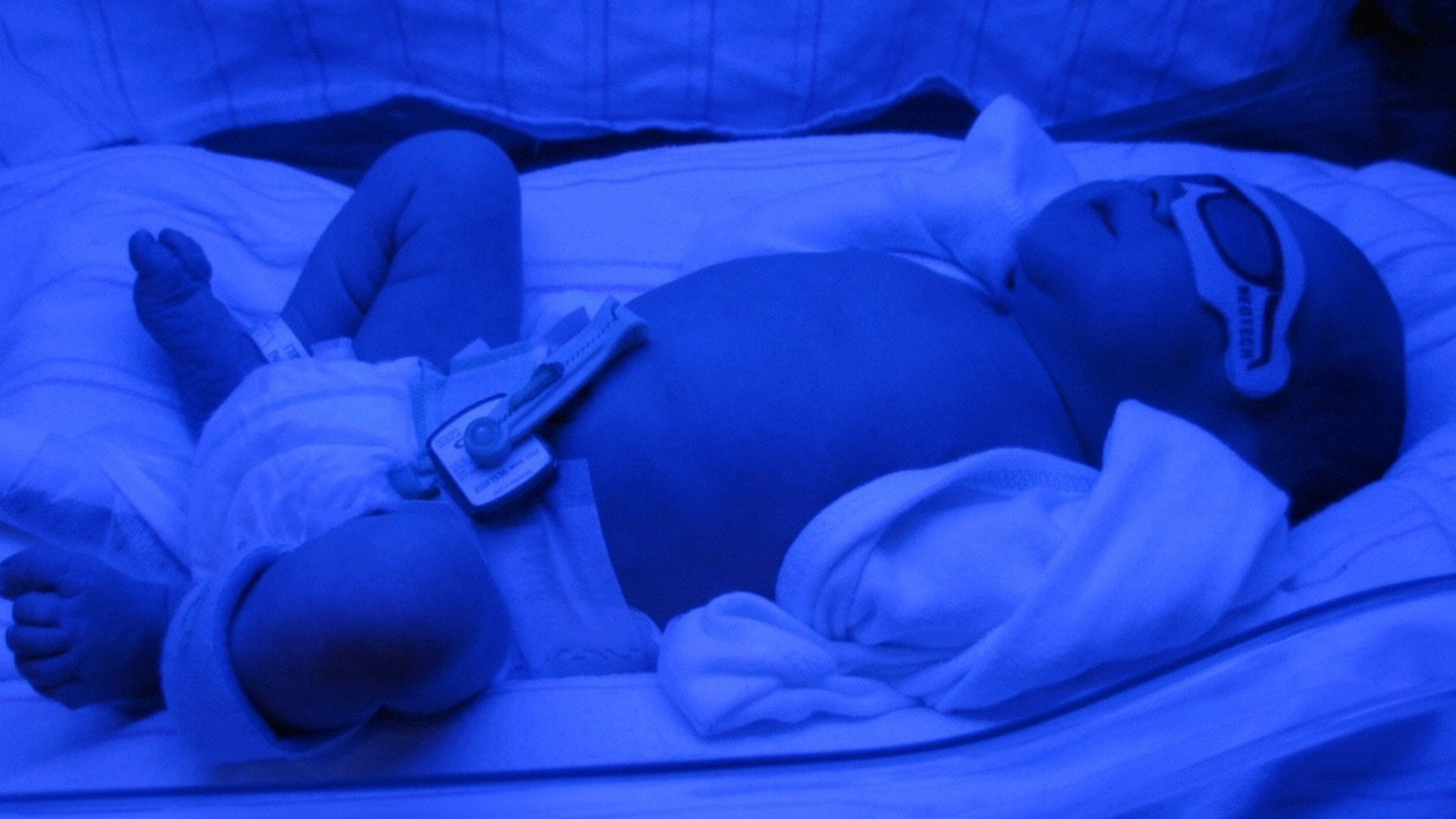
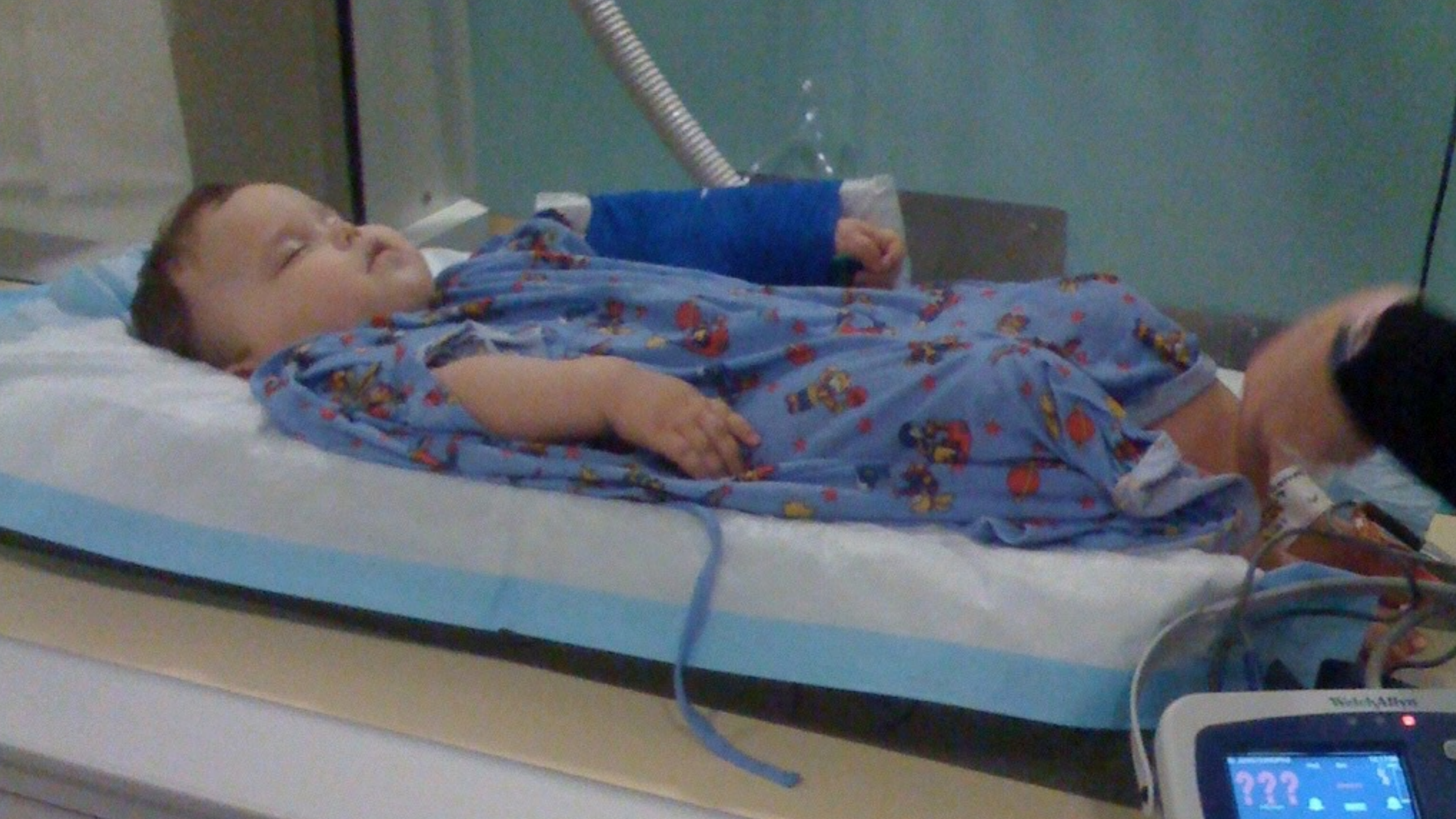
SANFORD-BURNHAM'S

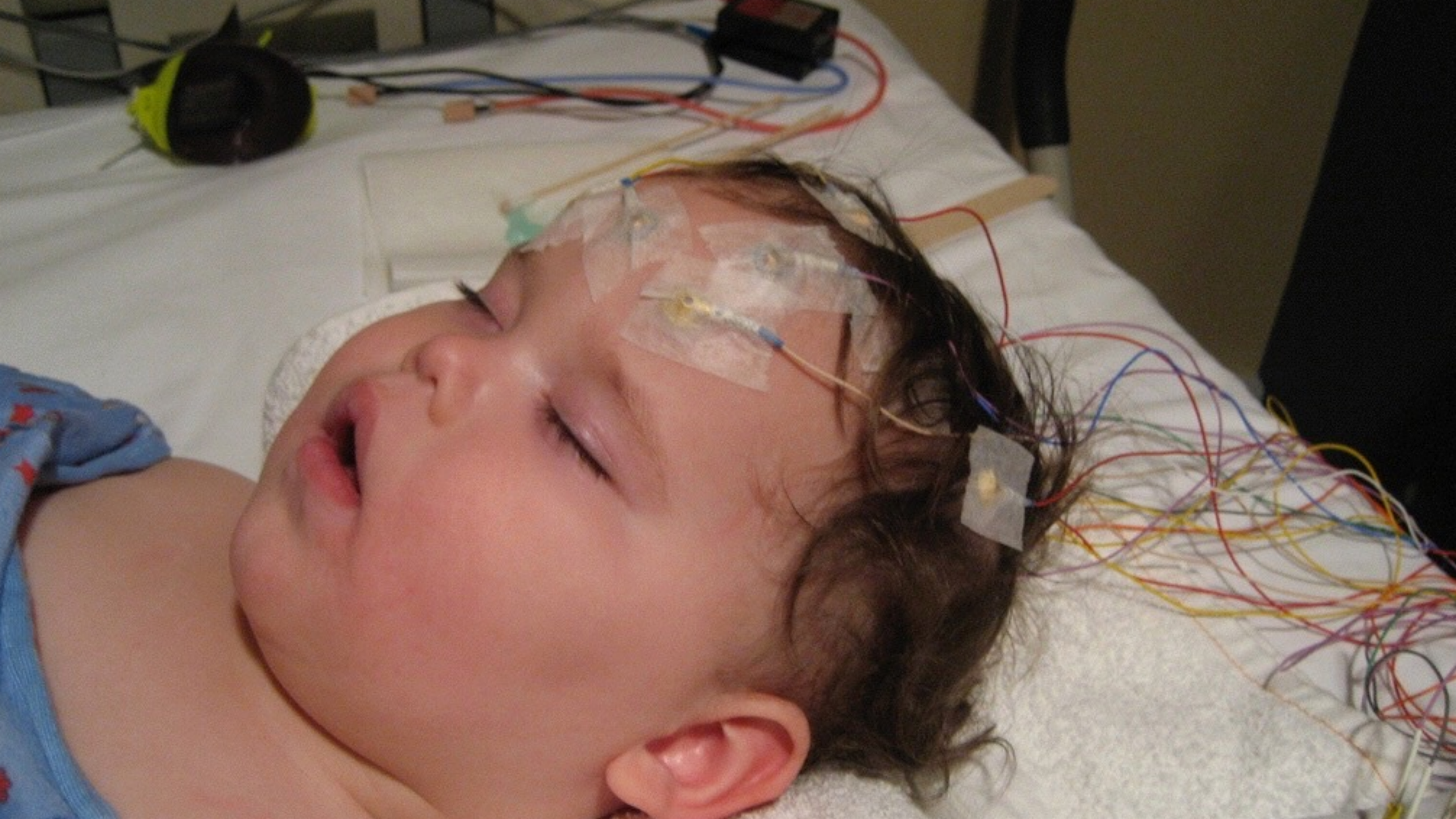
2014 ANNUAL GALA

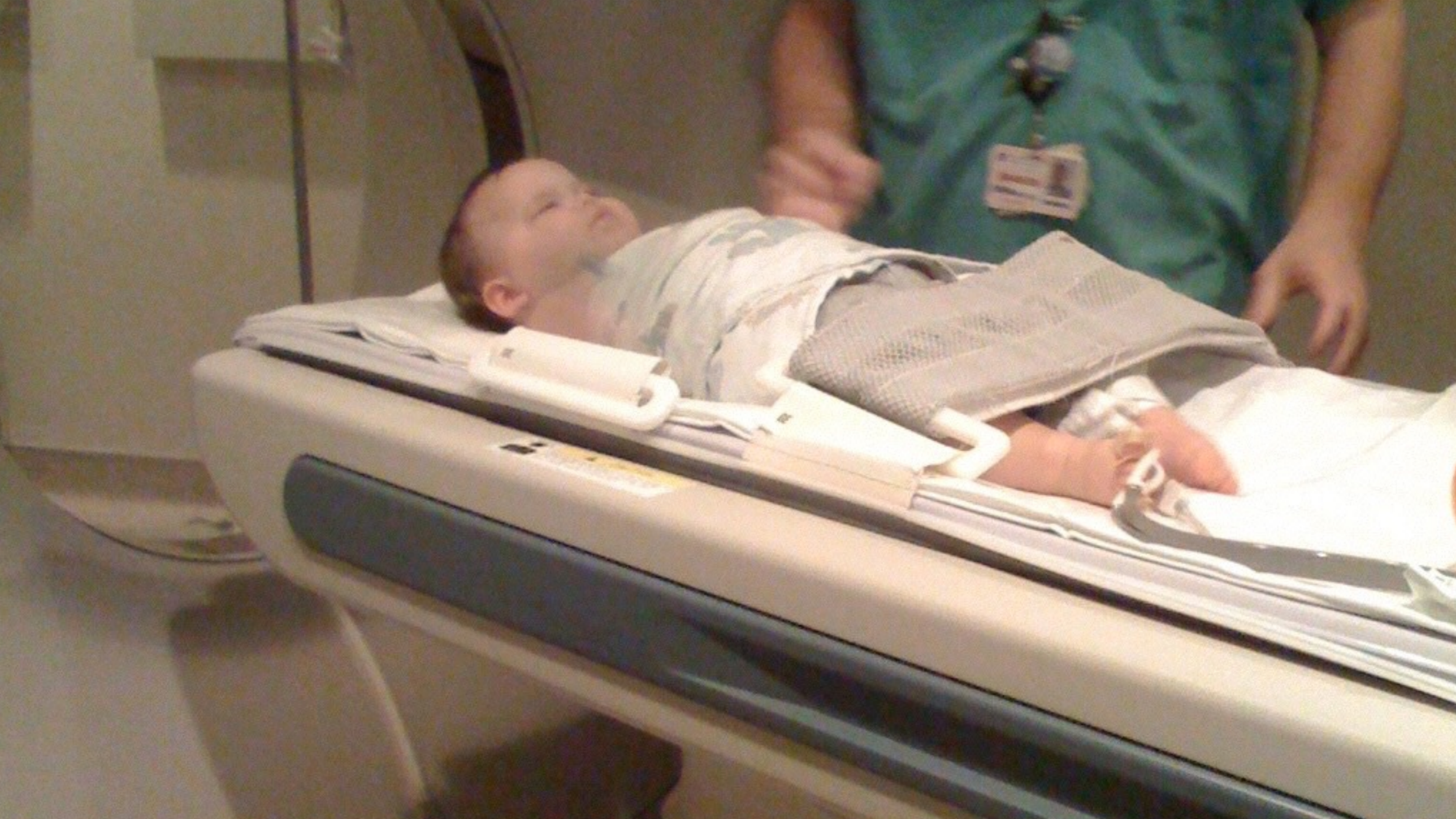
TOP HAT

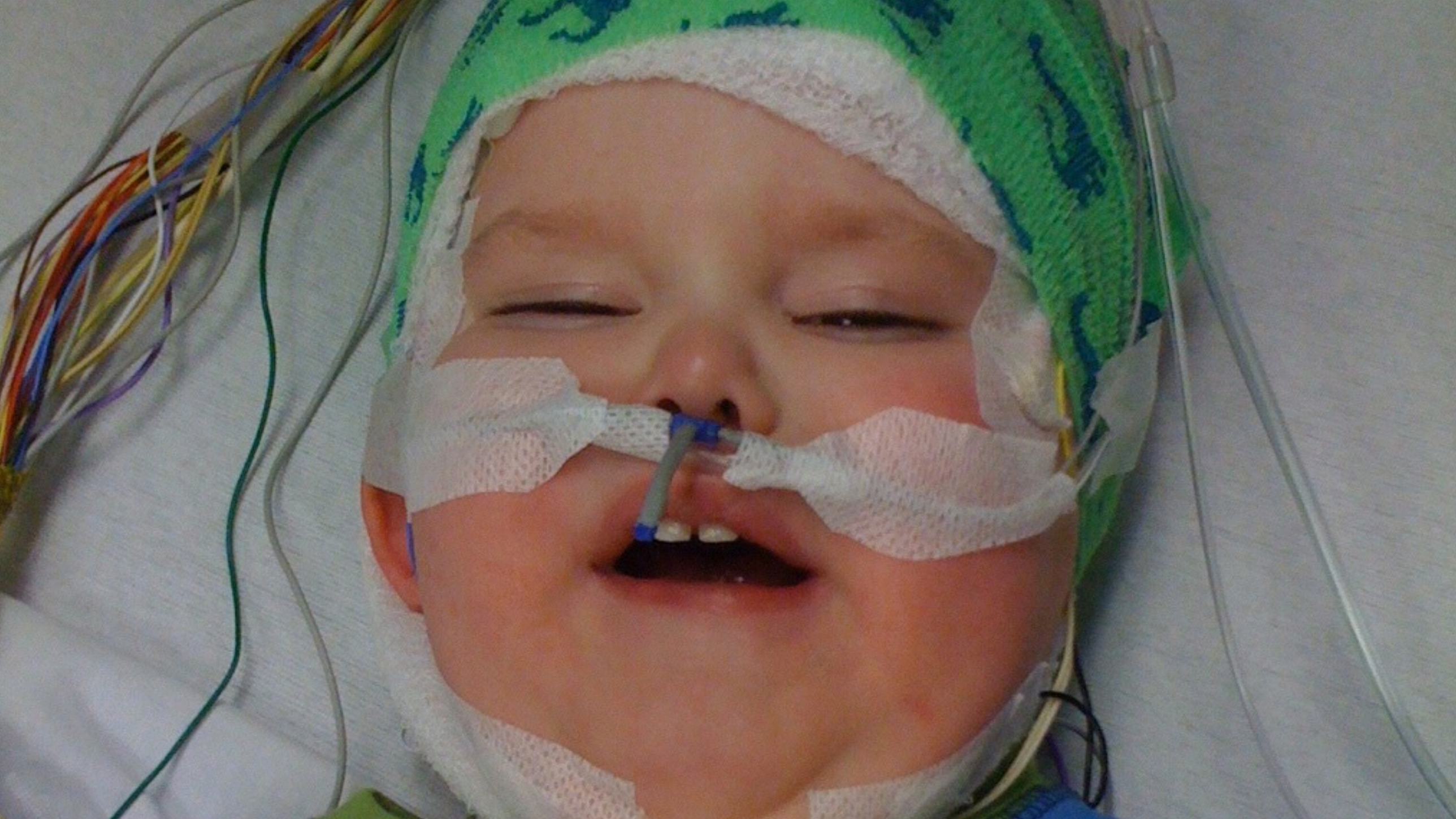

One of a Kind
Sanford-Burnham: Making the impossible possible

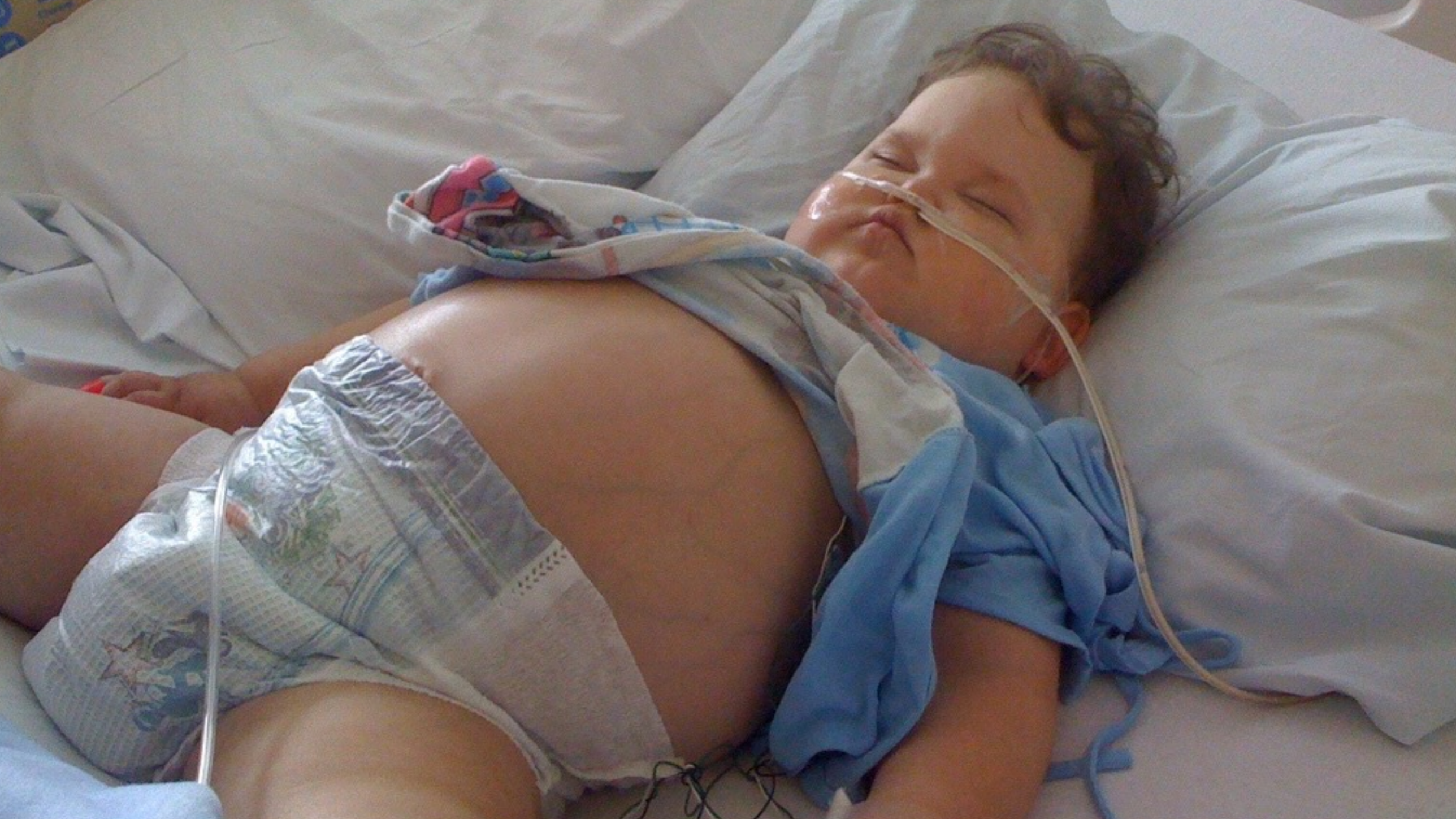


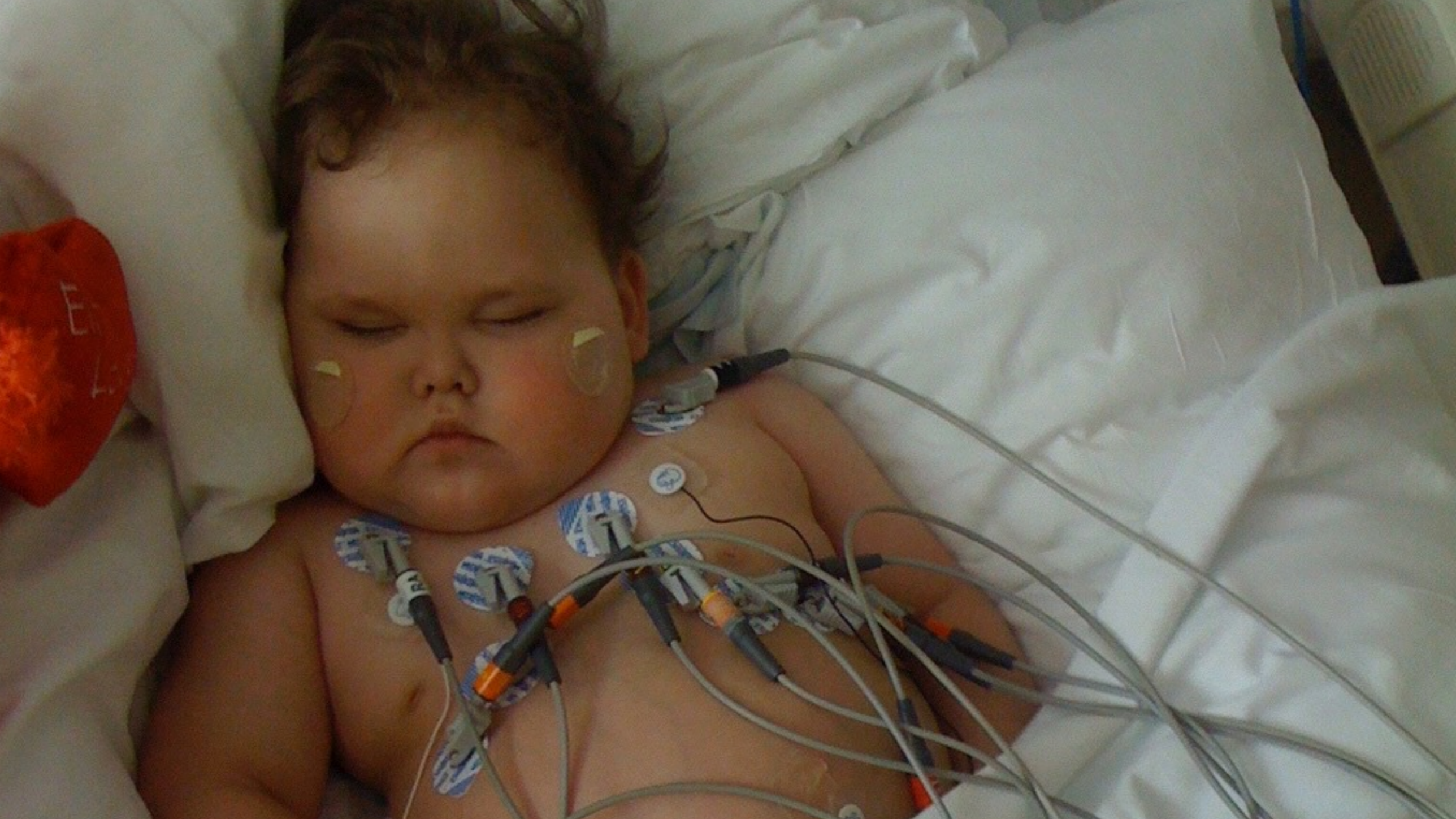


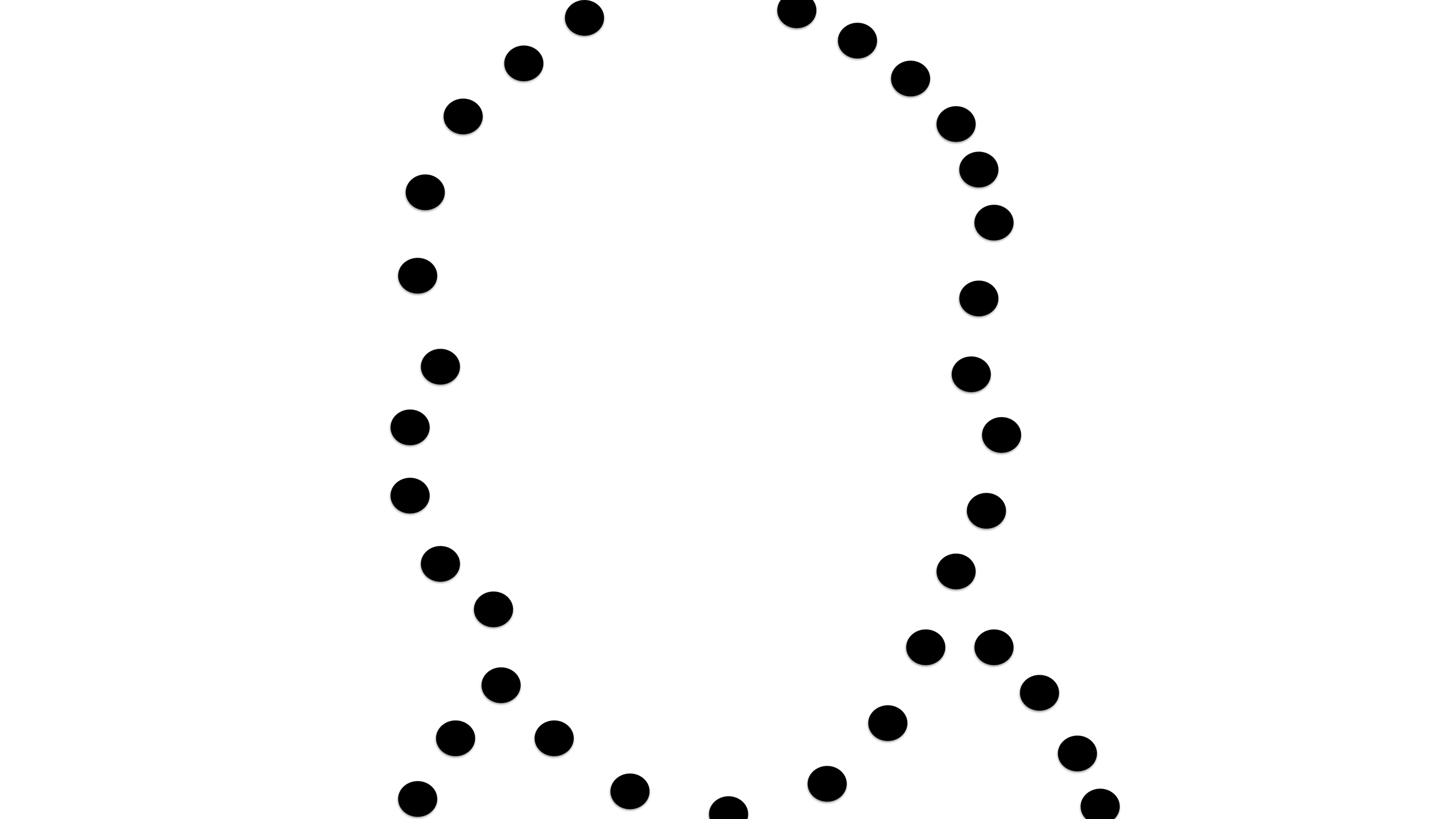


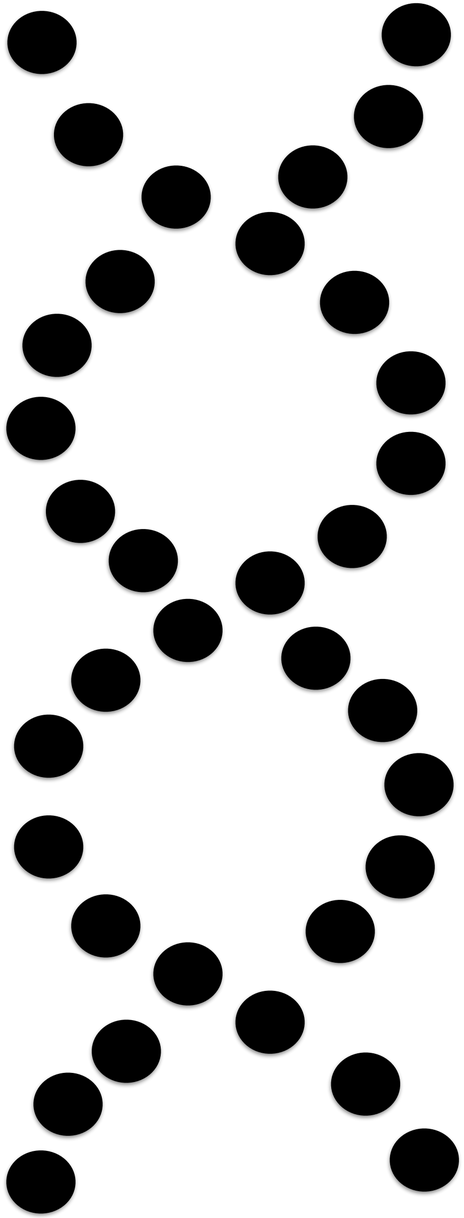


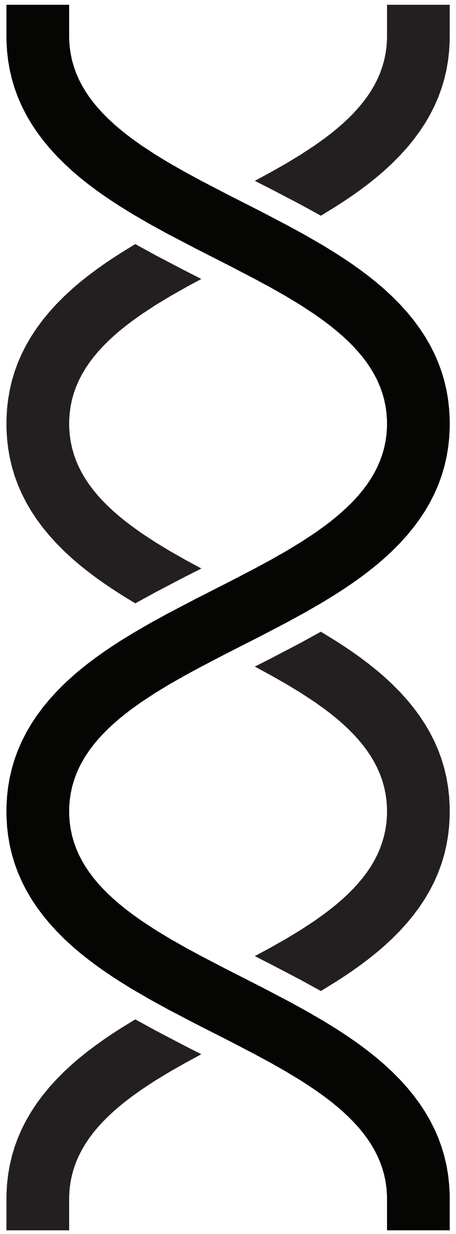


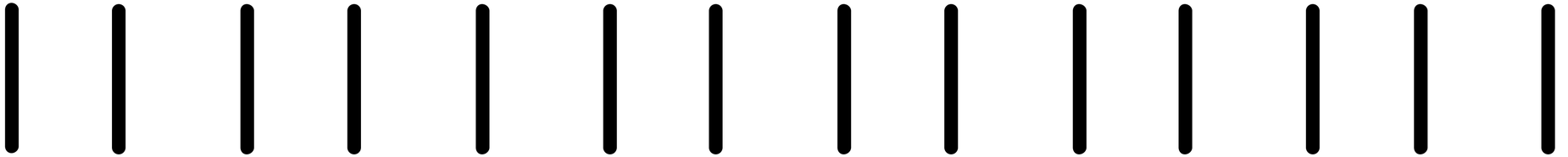


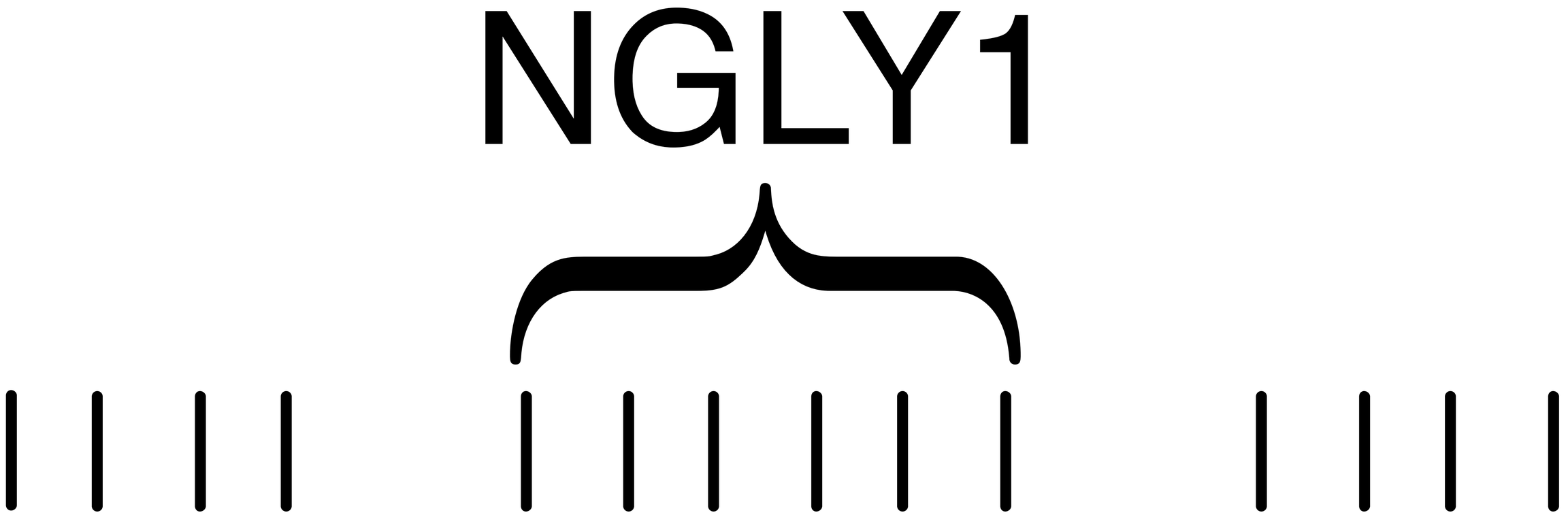


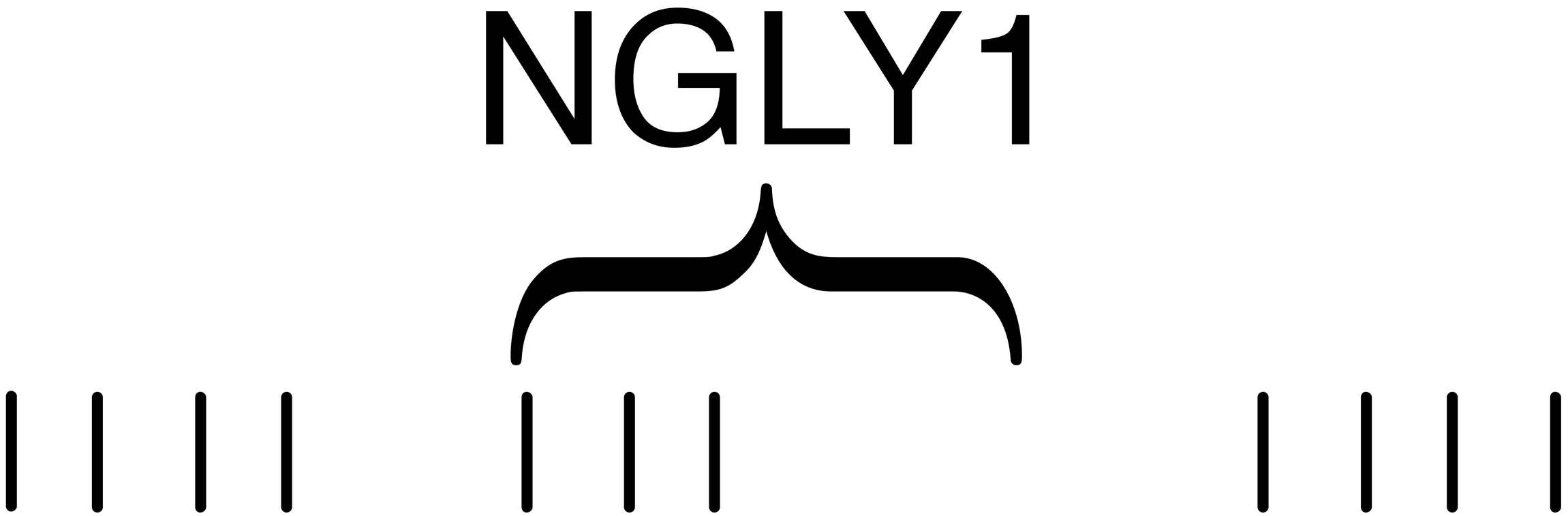









NGLY1


NGLY1

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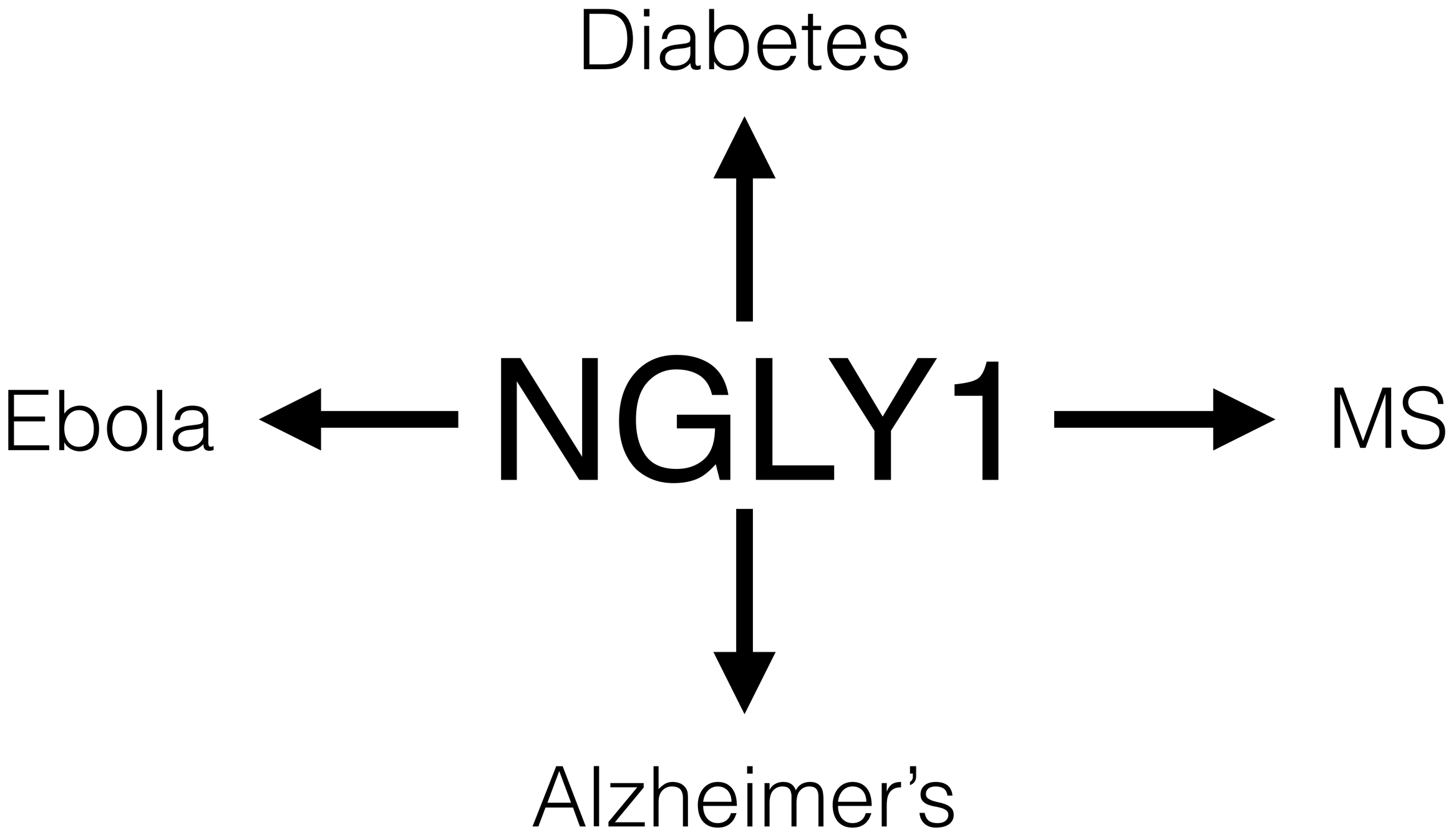

Diabetes
Ebola
NGLY1
MS
Alzheimer's


NGLY1

Hunting down my son's killer

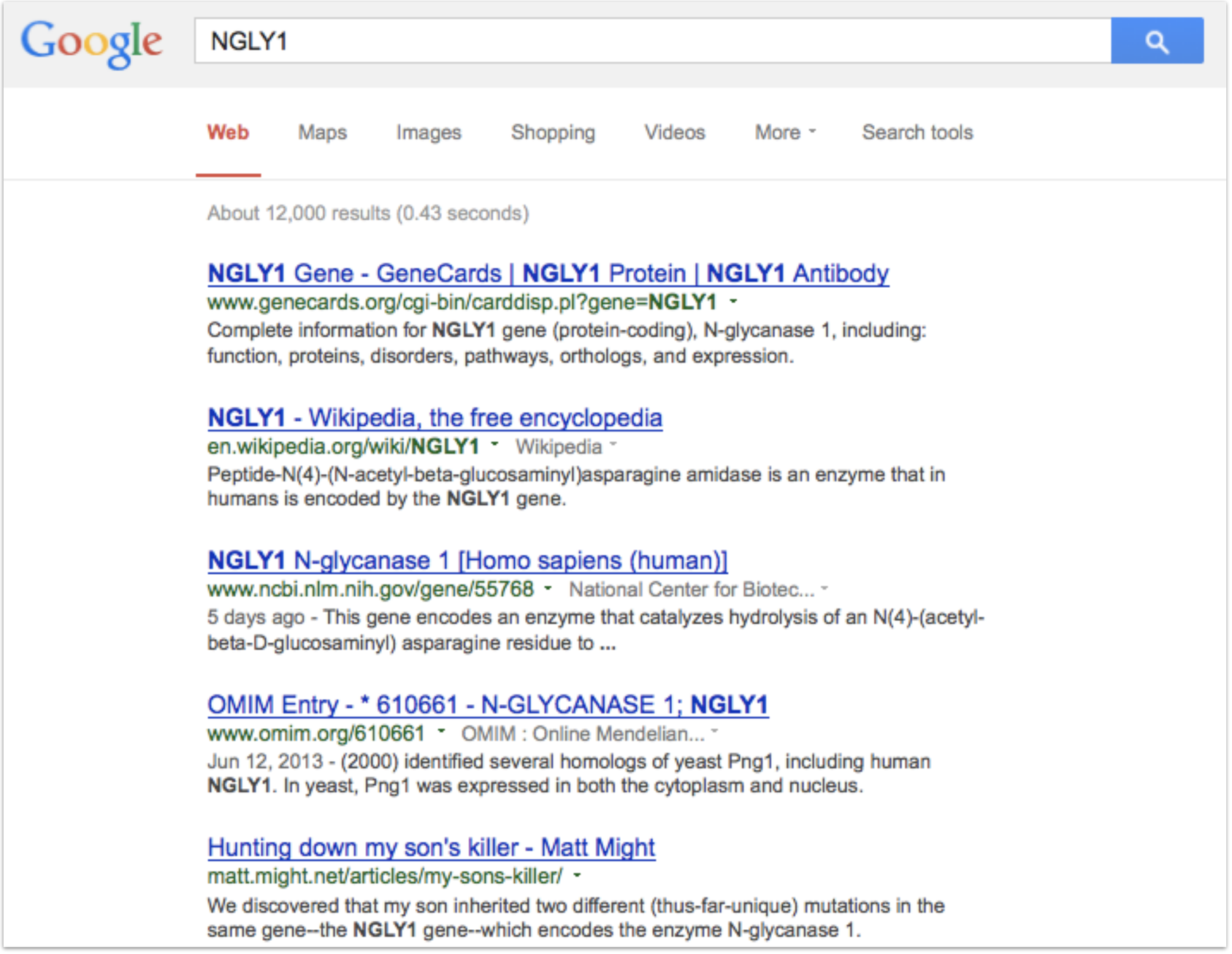
[article index] [email me] [@mattmight] [+mattmight] [rss]

I found my son's killer.

It took three years.

But we did it.

Not quite like this.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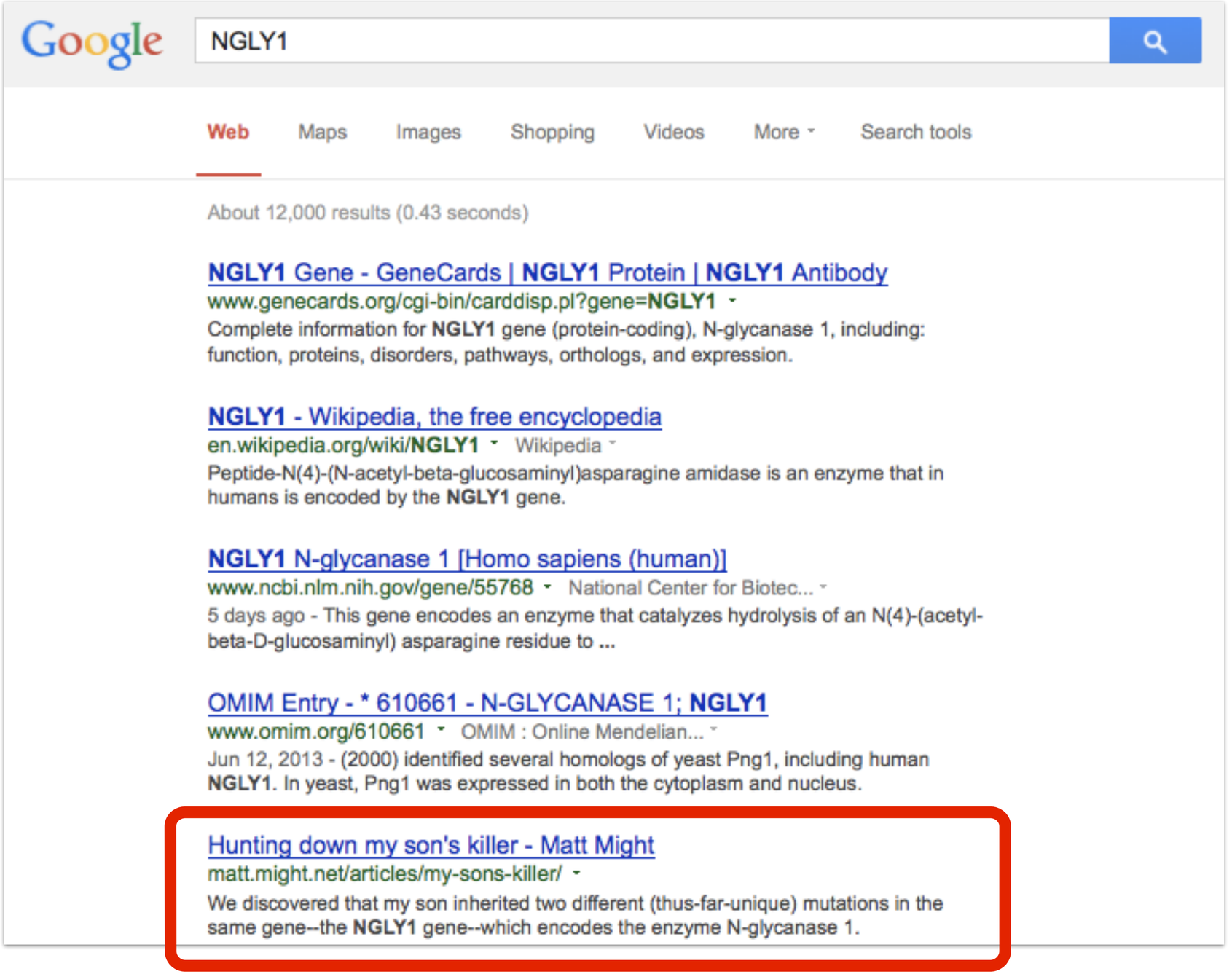
www.genecards.org/cgi-bin/carddisp.pl?gene=NGLY1
Complete information for NGLY1 gene (protein-coding), N-glycanase 1, including: function, proteins, disorders, pathways, orthologs, and expression.
NGLY1 - Wikipedia, the free encyclopedia
en.wikipedia.org/wiki/NGLY1 Wikipedia
Peptide-N(4)-(N-acetyl-beta-glucosaminyl)asparagine 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-glucosaminyl) 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.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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-glucosaminyl)asparagine amidase is an enzyme that in humans is encoded by the NGLY1 gene.
NGLY1 N-glycanase 1 [Homo sapiens (human)]
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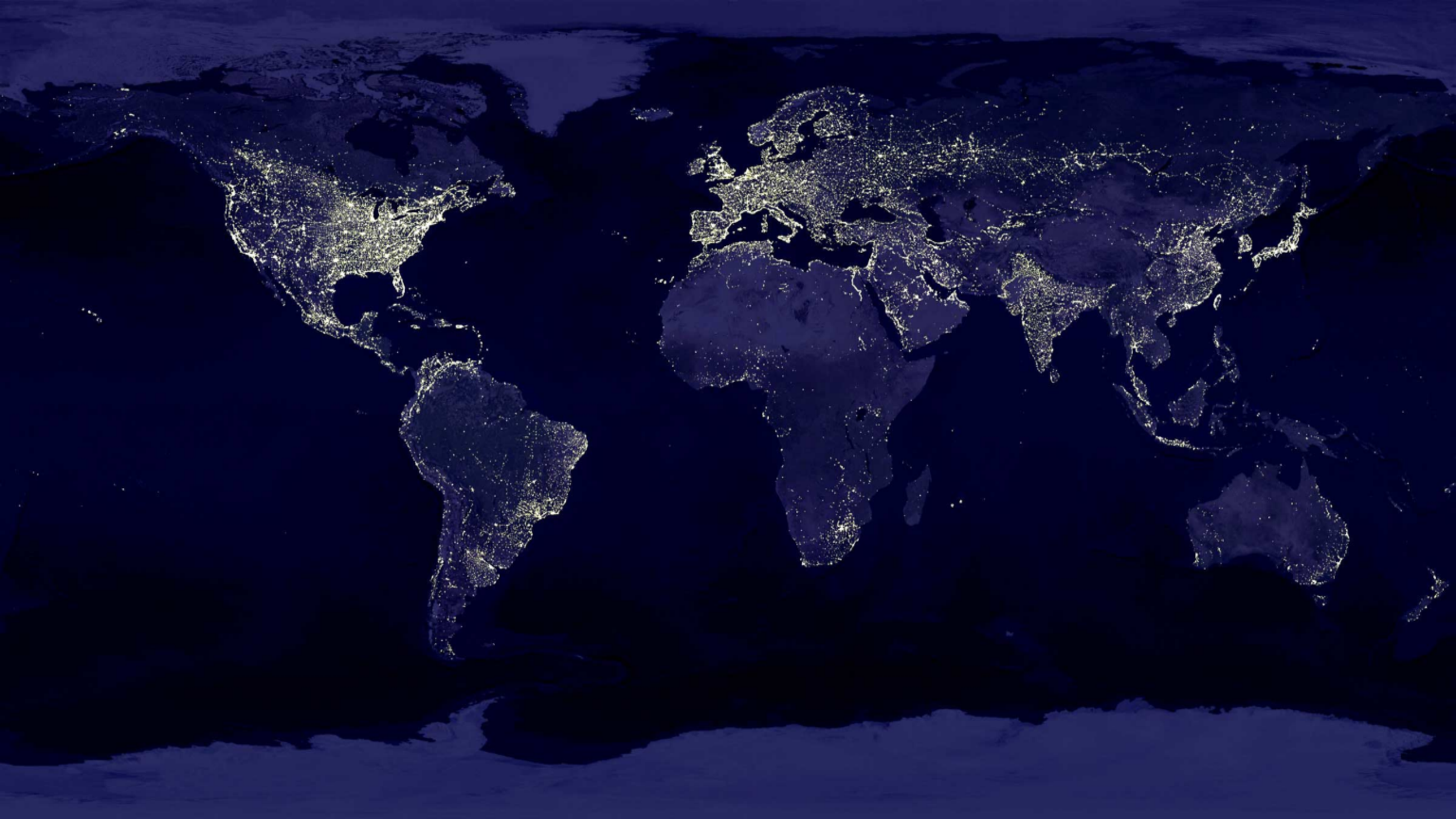

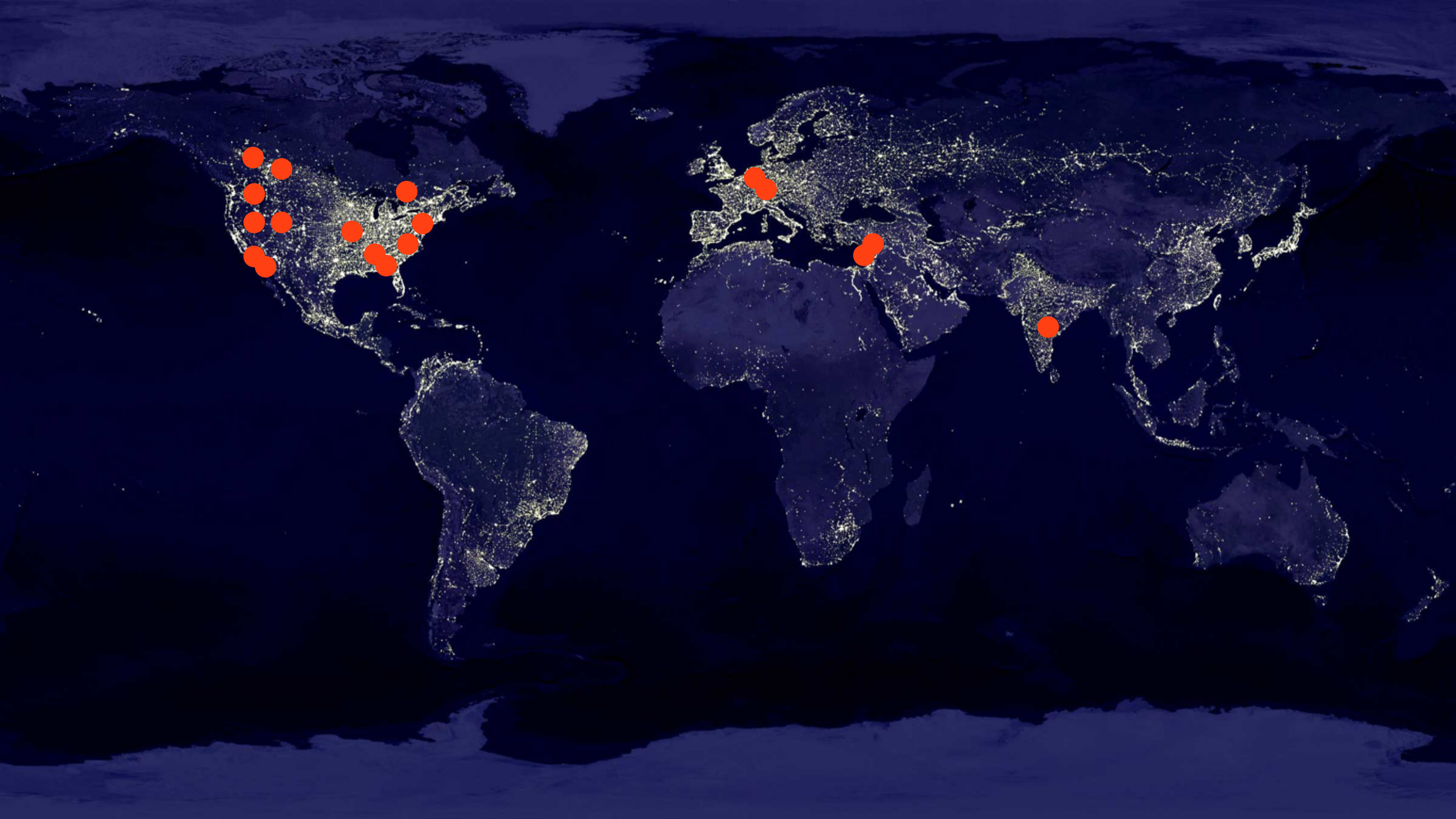

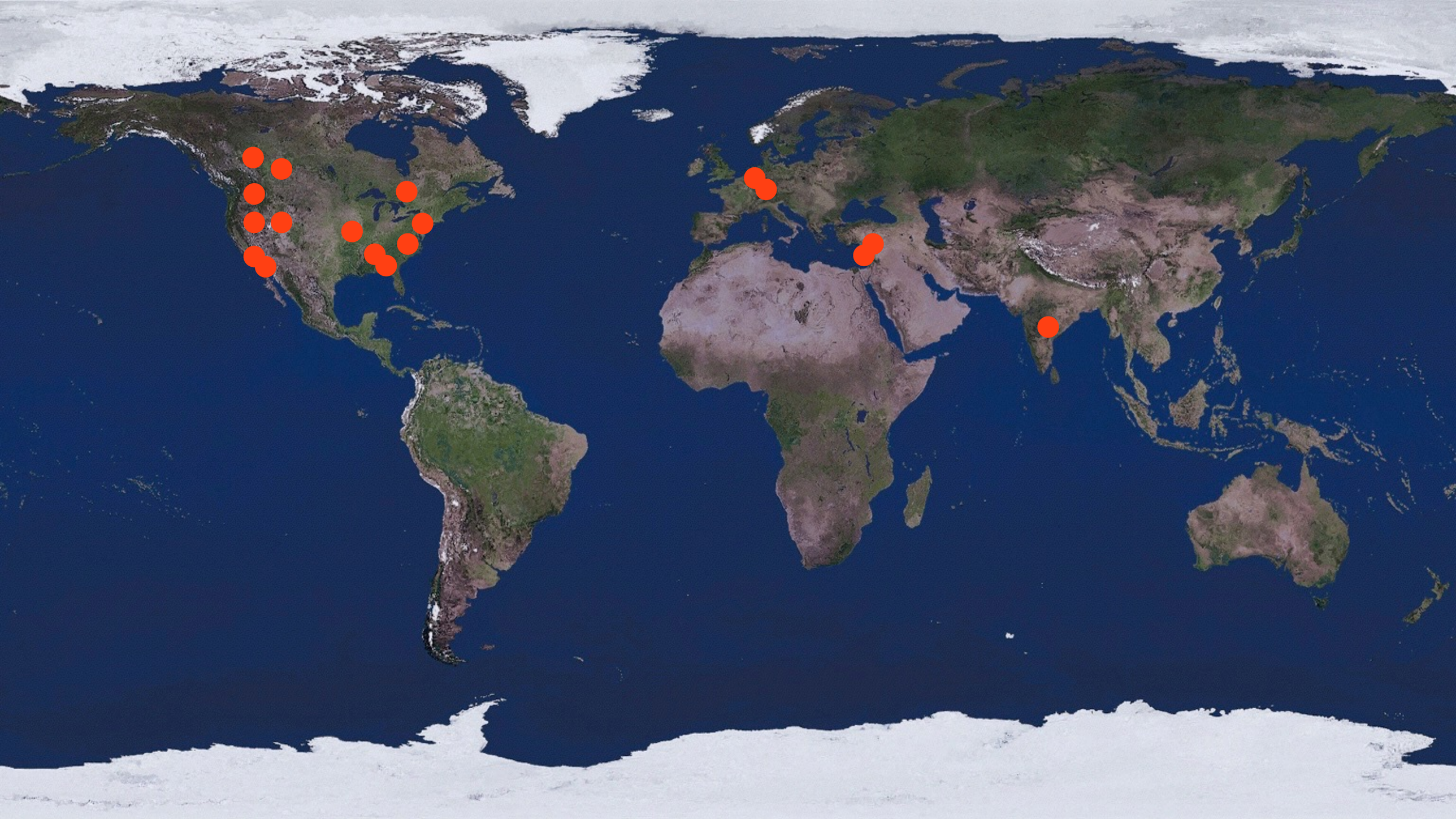

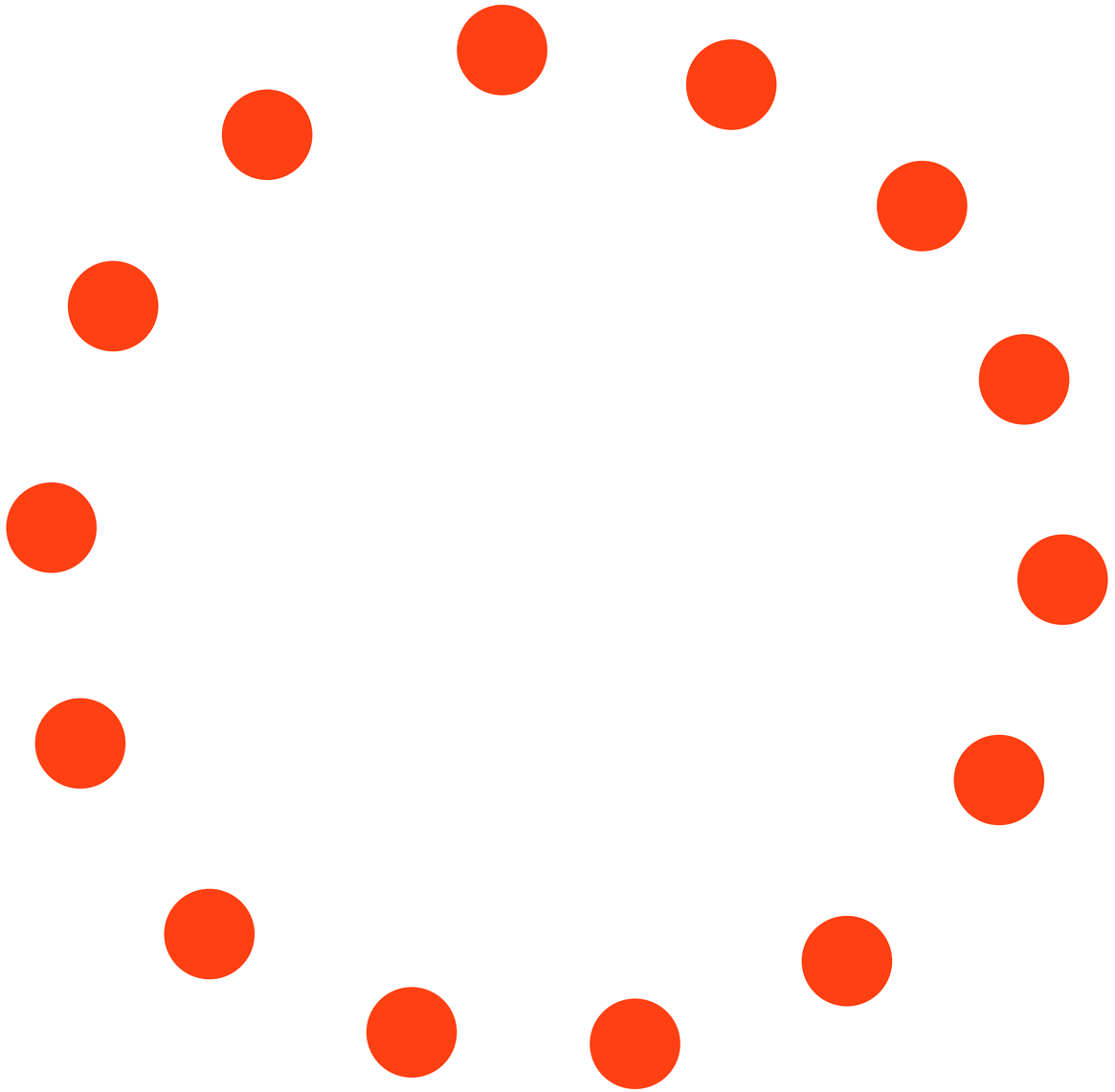

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EVERY ISSUE.
EVERY STORY
EVERY DEVICE.

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SCIENCE & TECH

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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" (Inschrift 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-

FOTO: STACI CUMMINGS

Kids who don't cry: New genetic disorder discovered

By **Jacque Wilson**, CNN
updated 2:53 PM EDT, Thu March 20, 2014

KATHERINE EMERY PHOTOGRAPHY/STANFORD CHILDREN'S HEALTH

Grace Wilsey was born with NGLY1 deficiency, which is caused by two mutations in the NGLY1 gene.

STORY HIGHLIGHTS

- A new genetic disorder called NGLY1 deficiency is identified in eight patients
- NGLY1 deficiency causes

(CNN) -- What do you do when your baby lies limp in your arms, staring blankly into the distance while never crying?

What do you do when tests show signs of liver damage and your baby's seizures won't stop, but doctors can't tell you what's wrong or


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