Patient Experience; Patient Engagement

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Patient Experience;
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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
NGLY1
NGLY1
NGLY1
NGLY1
NGLY1
What if $n = 1$?
Hudson Freeze, Sanford-Burnham
Undiagnosed

Patient Experience

Aftermath
Patient Experience
Logistics
What’s it like?
How to improve?
Exhaustion.
It takes two.
science lessons.
pseudo science lessons.
“Homework” for patients.
Undiagnosed

Patient Experience

Aftermath
Aftermath
“Actionable”
Patient-initiated science
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+
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
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 Biotech...
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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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 a biochemistry major, and Matt an electronics engineering major. Matt, however, was always in motion, 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 was to be in near-constant disaster. But Matt’s efforts to comfort him were not the only reason for his worry. Matt and Cristina decided to see if they could help Bertrand and to see if he was willing to help them.

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 — 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, treatable movement disorder called ataxia-telangiectasia. A subsequent genetic screen confirmed this diagnosis.

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

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

By Cristina on Saturday, October 25, 2014  in Columbia news  No comments
For patients, functional studies beat case-finding!
Animal models
NUTRITIONAL

INNOVATIVE QUALITY

Since 1932

KAL

YEAST Flakes

PREMIUM • UNSWEETENED • FORTIFIED

Gluten Free

Wonderful Nutty Flavor
Non-GMO

NET WT. 22 oz. (624 g)
Basic science
NGLY1
Enzyme replacement
PEG polymer
PEG polymer

TAT peptide
Replicable?
Yes!
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.

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
Gene X inhibitor
What do patients need?
“next step”
NGLY1

From Wikipedia, the free encyclopedia

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

### Contents [hide]
1. Function
2. Clinical significance
3. References
4. Further reading

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**Function** [edit]

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

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**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.
How to create a web site
What information to share
All variants
HPO
PPO
Parent Phenotyping Ontology
alacrima
why does my child have no tears?
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Only Fresh Coffee has been family-owned and operated since 1986, serving the freshest coffee, brewed from beans we roast ourselves. Enjoy fresh coffee at the neighborhood store and enjoy a cup today.

Local Fresh Coffee
Thank you!
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