Wikipedia:
"*n*++ made easy"

Matt Might
University of Utah / NGLY1.org
matt.might.net

What is Wikipedia?

What is Wikipedia?

NGLY1

From Wikipedia, the free encyclopedia

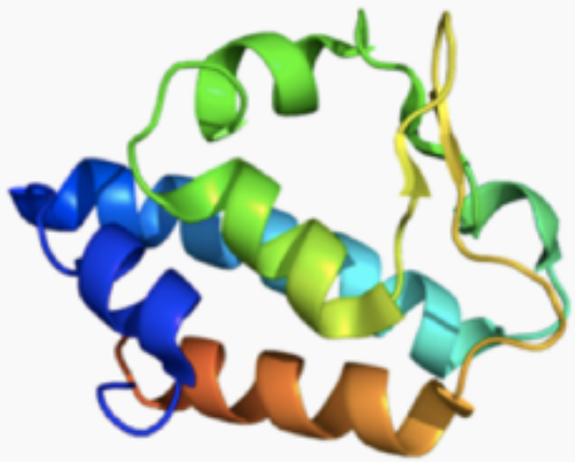
Peptide-N(4)-(N-acetyl-beta-glucosaminyl)asparagine amidase is an enzyme that in humans is encoded by the *NGLY1* gene.[1]

Contents [hide]

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-glucosaminylamine and a peptide containing an aspartate

N-glycanase 1



PDB rendering based on 2ccq.

Available structures

PDB Ortholog search: PDBe RCSB

Anyone can view.

Anyone can edit.

Wikipedia is the world’s database.

It’s what *parents* and *patients* search.

“M.D. by Wikipedia”

You do too.

San Francisco Chronicle

And that's how things came back around to the Wilseys. At Baylor, geneticist Matthew Bainbridge had decided to look for further information on NGLY1 as a source of Grace's disease, and he found his answers in two unlikely places: Matthew Might's blog and Wikipedia.

"I feel silly saying it," Bainbridge said about the online encyclopedia. He doesn't typically use Wikipedia for research, but in this case, as he was skimming for basic information on NGLY1, he found a link to the Duke paper.

And when he read Matthew Might's blog, one particular, very unusual symptom stood out: Bertrand didn't produce tears.

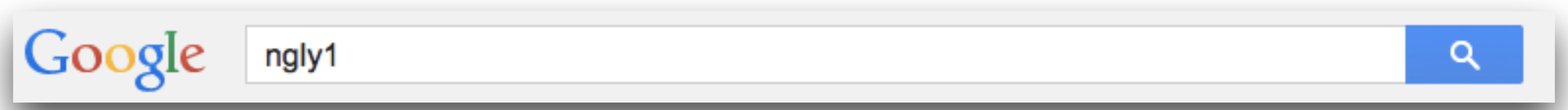
"I wrote to the Wilseys and asked, 'Does Grace make tears?' " Bainbridge said. The Wilseys confirmed that no, she rarely did. "That was the 'eureka' moment. That was 'bingo!' "

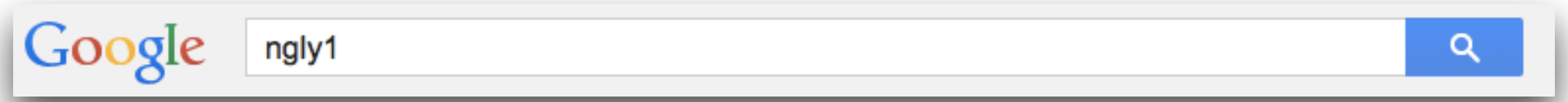
Research & Current Projects

The medical team working around the clock is the hero of this story. The group includes glycobiologists, cell biologists, biochemical geneticists, chemists, gene therapy innovators, animal experts, and even a Nobel Prize winner. Simply put, it's a world-class team.

Dr. Lars Steinmetz preparing samples for sequencing.

Why Wikipedia works


Google
ngly1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Grace Wilsey Foundation - gracewilsey.org
Ad www.gracewilsey.org/
Developing cures for **NGLY1** and other rare diseases in children.

NGLY1 Rare Disease - ngly1.org
Ad www.**ngly1**.org/
Info and support for kids diagnosed with N-glycanase deficiency.
Mission & Vision - Family Support - Equipment Program - Upcoming Events

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.org
www.**ngly1**.org/
The Grace Wilsey Foundation has created an informational video to answer the question: "What is **NGLY1** Deficiency?" Please share with all those whom might ...

NGLY1 N-glycanase 1 [Homo sapiens (human)]
www.ncbi.nlm.nih.gov/... National Center for Biotechnology Information
Jan 13, 2015 - This gene encodes an enzyme that catalyzes hydrolysis of an N(4)-(acetyl-beta-D-glucosaminyl) asparagine residue to ...

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.

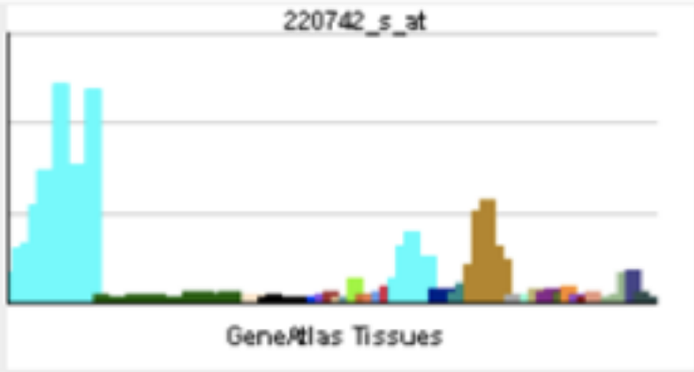
References [edit]

1. ^ *a* *b* "Entrez Gene: NGLY1 N-glycanase 1".
2. ^ Need AC, Shashi V, Hitomi Y, Schoch K, Shianna KV, McDonald MT, Meisler MH, Goldstein DB (May 2012). "Clinical application of exome sequencing in undiagnosed genetic conditions". *J Med Genet* **49** (6):

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



More reference expression data

Orthologs

Species	Human	Mouse
Entrez	55768	59007
Ensembl	ENSG00000151092	ENSMUSG00000021785
UniProt	Q96IV0	Q9JI78
RefSeq	NM_001145293	NM_021504

Reaching n=2?

Edit Wikipedia

Gene name, rare feature

“NGLY1” or “alacrima”

Wikipedia does not have an article with this exact name.

Alacrima

From Wikipedia, the free encyclopedia

Alacrima refers to an abnormality in tear production that could mean reduced tear production or absent tear production. Because a lack of tears presents in only in a few rare disorders, it aids in diagnosis of these disorders, including Triple-A syndrome and NGLY1 deficiency. [1][2][3]

Alacrima can be formally diagnosed through a Schirmer's test.

1. ^ "NGLY1 Foundation". *NGLY1 deficiency*.
2. ^ Need AC, Shashi V, Hitomi Y, Schoch K, Shianna KV, McDonald MT, Meisler MH, Goldstein DB (May 2012). "Clinical application of exome sequencing in undiagnosed genetic conditions". *J Med Genet* **49** (6): 353–61. doi:10.1136/jmedgenet-2012-100819. PMC 3375064. PMID 22581936.
3. ^ Enns GM, Shashi V, Bainbridge M, et al. (March 2014). "Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum-associated degradation pathway". *Genet. Med.* **16** (10): 751–8. doi:10.1038/gim.2014.22. PMID 24651605.

Possible clinical significance

Possible clinical significance

The Undiagnosed Disease Network has identified a patient presenting with *phenotype* with a putative causal mutation (*c.????*, *p.????*) in gene X. [*n*]

[*n*] http://undiagnosed.gov/case/1234

Case description

- Symptoms/features (HPO and “PPO”)
- Prior plausible/tentative diagnoses
- Attempted treatments / therapies
- Any and all variants of interest
- Pictures of patient (if possible)
- UDN contact: *Phone*, email, web form

Three pitfalls

1: More than just English

(Arabic may be good too.)

2: Multiple gene names

“PNGase”

Peptide-N4-(N-acetyl-beta-glucosaminyl)asparagine amidase

From Wikipedia, the free encyclopedia

In enzymology, a **peptide-N4-(N-acetyl-beta-glucosaminyl)asparagine amidase** (EC 3.5.1.52) is an enzyme that catalyzes a chemical reaction that cleaves a N_4-(acetyl-beta-D-glucosaminyl)asparagine residue in which the glucosamine residue may be further glycosylated, to yield a (substituted) N-acetyl-beta-D-glucosaminylamine and a peptide containing an aspartate residue. This enzyme belongs to the family of hydrolases, specifically those acting on carbon-nitrogen bonds other than peptide bonds in linear amides.

The NGLY1 gene encodes the ortholog of this enzyme in humans.

Contents [hide]

peptide-N4-(N-acetyl-beta-glucosaminyl)asparagine amidase

Identifiers	
EC number	3.5.1.52
CAS number	83534-39-8
Databases	
IntEnz	IntEnz view
BRENDA	BRENDA entry
ExPASy	NiceZyme view
KEGG	KEGG entry
MetaCyc	metabolic pathway
PRIAM	profile

CDG4

CDGIV

“CDG1V”

3: Don’t fire and forget!

Google

+Matthew 3

Alerts

Monitor the web for interesting new content

ngly1

This will create an email alert for matt.might@gmail.com.

CREATE ALERT Show options

Alert preview

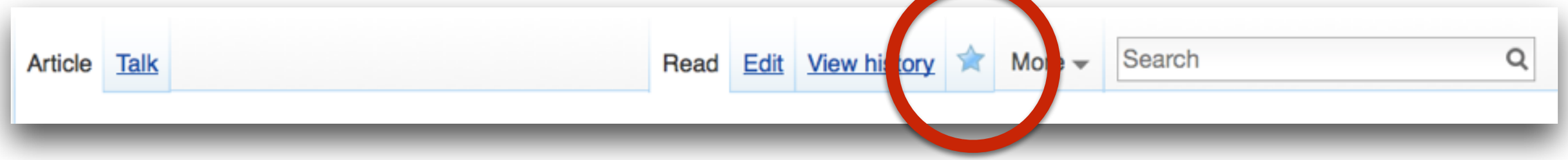
There are no recent results for your search query. Below is a sample of the type of results you will get.

NEWS

Insights into a rare genetic disease

Medical Xpress

(Left) In normal cells, misfolded proteins are deglycosylated predominantly by **Ngly1** and are degraded efficiently. (Right) In the absence of **Ngly1**, ...


Article
Talk
Read
Edit
View history
Search

If n goes to 2?

Edit Wikipedia!

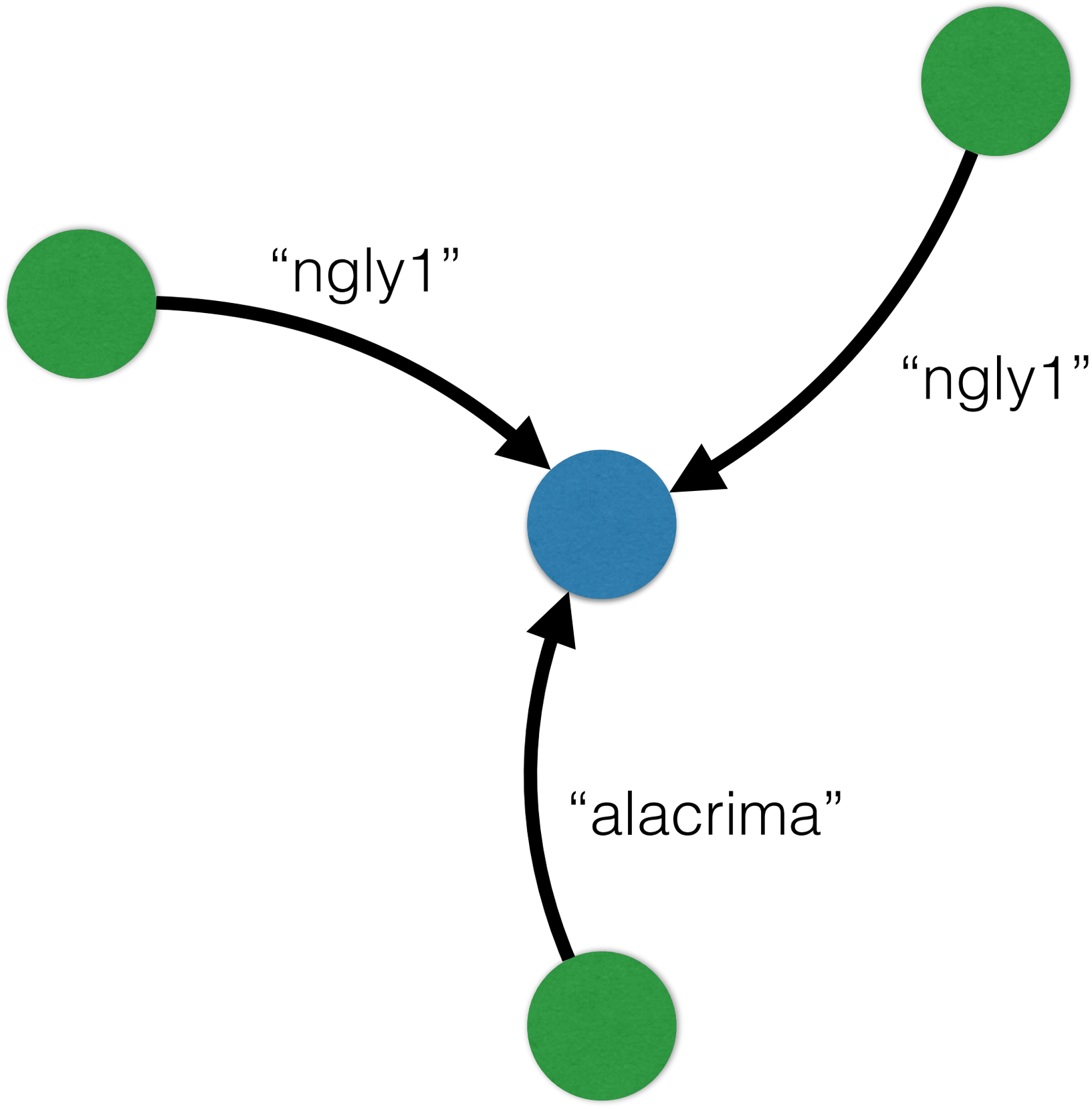
Help patients make sites.

Diagnosis is the *start*.

Teach patients SEO.

Search Engine Optimization

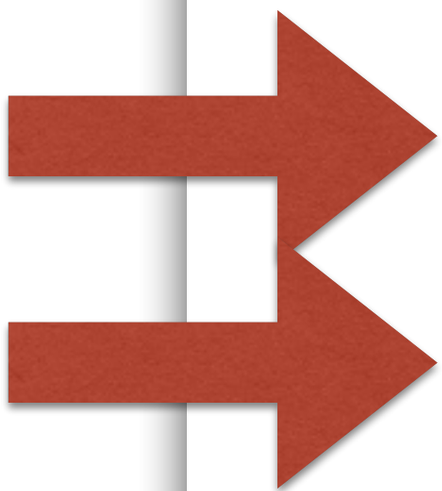
How does Google rank?


“ngly1”
“ngly1”
“alacrima”

http://foo.com/ngly1

http://ngly1.net/

Or, buy AdWords!



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This internet will find them.

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23

Action items: Wikipedia

- Do the same for other “pedias”: SNPedia
- Need *short* online case descriptions on .gov site.
- Need easy contact info: email, phone, web form.
- Need standard template for adding to Wikipedia.
- Need advice for patients on patient-run sites.