Wikipedia: “n++ made easy”

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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]

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#### 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.

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N-glycanase 1

PDB rendering based on 2ccq.

**Available structures**

- PDB
  - Ortholog search: PDBs
- 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.
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.
Why Wikipedia works
Clinical significance

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 ngly.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

1. ^a b "Entrez Gene: NGLY1 N-glycanase 1".
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.
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]

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
Peptide-N4-(N-acetyl-beta-glucosaminy1)asparagine amidase

From Wikipedia, the free encyclopedia

In enzymology, a peptide-N4-(N-acetyl-beta-glucosaminy1)asparagine amidase (EC 3.5.1.52) is an enzyme that catalyzes a chemical reaction that cleaves a N\textsubscript{4}-(acetyl-beta-D-glucosaminy1)asparagine residue in which the glucosamine residue may be further glycosylated, to yield a (substituted) N-acetyl-beta-D-glucosaminy1amine 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.

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1 Nomenclature
2 Structural studies
3 References
CDG4
CDGIV
“CDG1V”
3: Don’t fire and forget!
Alerts
Monitor the web for interesting new content

Search for: 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, ...
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?
http://foo.com/ngly1
Or, buy AdWords!
This internet will find them.

- 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.