

Title: A formalization of one of the main claims of “Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum–associated degradation pathway” by Enns et al. 2014

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http://purl.org/np/RA12IVwEtmddK9OwDkZQZlgJaOD2-0NXtAtO_jDaG-3VQ

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Abstract:

Enns et al. claimed in previous work that NGLY1 deficiency is a novel autosomal recessive disorder of the ERAD pathway. We present here a formalization of that claim, stating that all things of class “NGLY1 deficiency” that are in the context of a thing of class “human” always have a relation of type “is caused by” to a thing of class “dysfunction of ERAD pathway” in the same context.

1. Introduction

Enns et al. [1] state that “NGLY1 deficiency is a novel autosomal recessive disorder of the endoplasmic reticulum–associated degradation pathway.”. We present here a formalization of the main scientific claim from this quote by using a semantic template called the super-pattern [2].

2. Formalization

Our formalization looks as follows:

CONTEXT-CLASS (“in the context of all ...”): [human](#)

SUBJECT-CLASS (“things of type ...”): [NGLY1 deficiency](#)

QUALIFIER: [always](#)

RELATION-TYPE (“have a relation of [is caused by](#) type...”):

OBJECT-CLASS (“to things of type...”): [dysfunction of ERAD pathway](#)

In the context class we use the class “human” (Q5) from Wikidata. In the subject class, we use the class “NGLY1 deficiency” from Mondo Disease Ontology. In the object class we minted a new class “dysfunction of ERAD pathway” that is related to the class “ERAD pathway” (Q21101062) from Wikidata.

3. RDF Code

This is our formalization as a nanopublication in TriG format:

```
@prefix this: <http://purl.org/np/RA12lVwEtmdK9OwDkZQZlgJaOD2-0NXtAtO_jDaG-3VQ> .
@prefix sub: <http://purl.org/np/RA12lVwEtmdK9OwDkZQZlgJaOD2-0NXtAtO_jDaG-3VQ#> .
@prefix np: <http://www.nanopub.org/nschema#> .
@prefix dct: <http://purl.org/dc/terms/> .
@prefix nt: <https://w3id.org/np/o/ntemplate/> .
@prefix npx: <http://purl.org/nanopub/x/> .
@prefix xsd: <http://www.w3.org/2001/XMLSchema#> .
@prefix rdfs: <http://www.w3.org/2000/01/rdf-schema#> .
@prefix orcid: <https://orcid.org/> .
@prefix prov: <http://www.w3.org/ns/prov#> .
@prefix sp: <https://w3id.org/linkflows/superpattern/terms/> .

sub:Head {
  this: np:hasAssertion sub:assertion ;
  np:hasProvenance sub:provenance ;
  np:hasPublicationInfo sub:pubinfo ;
  a np:Nanopublication .
}
sub:assertion {
  sub:sp1 a sp:SuperPatternInstance ;
  rdfs:label "NGLY1 deficiency is a novel autosomal recessive disorder of the ERAD pathway." ;
  sp:hasContextClass <http://www.wikidata.org/entity/Q5> ;
  sp:hasSubjectClass <http://purl.obolibrary.org/obo/MONDO_0014109> ;
  sp:hasQualifier <https://w3id.org/linkflows/superpattern/terms/alwaysQualifier> ;
  sp:hasRelation <https://w3id.org/linkflows/superpattern/terms/isCausedBy> ;
  sp:hasObjectClass <http://purl.org/np/RAZVLqlkbwiX40n0GNxcxJany2Cw3oxMcRnuZtjBCLryU#Dysfunction_of_ERAD_pathway> .
}
sub:provenance {
  sub:activity a sp:FormalizationActivity ;
  prov:used sub:quote , <https://dx.doi.org/10.1038%2Fgim.2014.22> ;
  prov:wasAssociatedWith orcid:0000-0003-0169-8159 .
  sub:assertion prov:wasGeneratedBy sub:activity .
  sub:quote prov:value "NGLY1 deficiency is a novel autosomal recessive disorder of the endoplasmic reticulum-associated degradation pathway" ;
  prov:wasQuotedFrom <https://dx.doi.org/10.1038%2Fgim.2014.22> .
}
sub:pubinfo {
  sub:sig npx:hasAlgorithm "RSA" ;
  npx:hasPublicKey
    "MIGfMA0GCSqGSIb3DQEBQUAA4GNADCBiQKBgQDOWrvo3T4o7/c9+a16pWVu3tb32/oqIpuNK/bZlFUGR6bnQRBl+7ub5H++34CVFUpwEqb4RXsx39Q0fM1UPvJbZbX
    DYAsCEPzn0mzN+YAYdSZKAEyJM2cf4cqjQLwKBab0luVKfKetxTaKB/Jr/9U770dEc2LR3lOMFcbALPSXwIDAQAB" ;
  npx:hasSignature
    "hyYI2msaoxuiMomUFjxJ0K37Cko2r6FQsFper7fJQGC9Y5HMIR//VzDqrzXwz1BGdno3PG8WLV3dvy2vDj+ULXqXxFfp4IuSVekvt902OJXbQuCv40jTVPSgc0LsT0HA
    1l64tIu+nR8o2d8f9lr7mZSCitDc0FsEAcw2U9RUIF4=" ;
  npx:hasSignatureTarget this: .
  this: dct:created "2021-12-17T10:26:28.040+01:00"^^xsd:dateTime ;
  dct:creator orcid:0000-0003-0169-8159 ;
```

```

    npx:introduces sub:spi ;
    npx:supersedes <http://purl.org/np/RASpVPrnUq6S0sOkrg0sUvRbsfmC7dHd73FgUA6oRm1Gg> ;
    <https://w3id.org/linkflows/reviews/isUpdateOf> <http://purl.org/np/RAGgV-nRMWhmgtxx5n3yE29NaopuuDVKAuZC8IzCATag> ;
    nt:wasCreatedFromProvenanceTemplate <http://purl.org/np/RAE1wni0y0y039PlK9QkQ-wqbC3q-R2nXraP5huu8W39k> ;
    nt:wasCreatedFromPubinfoTemplate <http://purl.org/np/RA2vCBXZf-icEcVRGhulJXugTGxpsV5yVr9yqCI1bQh4A> ,
    <http://purl.org/np/RAA2MfqdBczmz9yVWjKLXNbyfBNcwsMmOqcNUxkk1maIM> ,
    <http://purl.org/np/RAjpbM1w3owYhJUBo3DtsuD1XsNAJ8cnGeWAutDVjuAuI> ;
    nt:wasCreatedFromTemplate <http://purl.org/np/RAv68imZrEjfcP2rnEglhzoBgEVc0cQMtp9_1Za0BxNM4> .
}

```

The following nanopublications introduce the newly minted classes in TriG format.

This is the class definition of “dysfunction of ERAD pathway”:

```

@prefix this: <http://purl.org/np/RAZVLq1kbwiX40n0GNxcxJany2Cw3oxMcrNuZtjBclryU> .
@prefix sub: <http://purl.org/np/RAZVLq1kbwiX40n0GNxcxJany2Cw3oxMcrNuZtjBclryU#> .
@prefix np: <http://www.nanopub.org/nschema#> .
@prefix dct: <http://purl.org/dc/terms/> .
@prefix nt: <https://w3id.org/np/o/ntemplate/> .
@prefix npx: <http://purl.org/nanopub/x/> .
@prefix xsd: <http://www.w3.org/2001/XMLSchema#> .
@prefix rdfs: <http://www.w3.org/2000/01/rdf-schema#> .
@prefix orcid: <https://orcid.org/> .
@prefix prov: <http://www.w3.org/ns/prov#> .
@prefix skos: <http://www.w3.org/2004/02/skos/core#> .

sub:Head {
  this: np:hasAssertion sub:assertion ;
  np:hasProvenance sub:provenance ;
  np:hasPublicationInfo sub:pubinfo ;
  a np:Nanopublication .
}

sub:assertion {
  sub:Dysfunction_of_ERAD_pathway a <http://www.w3.org/2002/07/owl#Class> ;
  rdfs:label "Dysfunction of ERAD pathway" ;
  skos:definition "Dysfunction of ERAD pathway" ;
  skos:relatedMatch <http://www.wikidata.org/entity/Q21101062> .
}

sub:provenance {
  sub:assertion prov:wasAttributedTo orcid:0000-0003-0169-8159 .
}

sub:pubinfo {
  sub:sig npx:hasAlgorithm "RSA" ;
  npx:hasPublicKey
    "MIGfMA0GCsgGSIb3DQEBAAQUAA4GNADCBiQKBgQDOWrvo3T4o7/c9+a16pWVu3tb32/oqIpuNK/bZlFUGR6bnQRB1+7ub5H++34CVFUIpWEQb4RXsx39Q0fM1UPvJb2bX
    DYAsCEPzn0mzN+YAYdSZKAEyjm2cf4cqjQLwKBab0luVKfKetxTaKB/Jr/9U770dEc2LR3lOMFcbALPSXwIDAQAB" ;
  npx:hasSignature
    "1HpXoxwEiDwRt0Snb8FuYflerV7zGddH0orZakfvel/t2mfo2YjppvSes+/zh1MjU2FxybUIV7KaitPVbxb1skOGyhb0mLAB6xNKwy+ia7TjX2NazJVVpHq4EPVXJY
    sjU3x80TjAWJce6sPbU9gtgHsRGC6fFFGp0rDq70ZP4=" ;
  npx:hasSignatureTarget this: .
  this: dct:created "2021-12-17T10:19:45.788+01:00"^^xsd:dateTime ;
  dct:creator orcid:0000-0003-0169-8159 ;
  npx:introduces sub:Dysfunction_of_ERAD_pathway ;
  npx:supersedes <http://purl.org/np/RAYXLV7qwfX03AGH7k5ZP6zeMj_vx2kj88z5UQ5OX3C90> ;
  nt:wasCreatedFromProvenanceTemplate <http://purl.org/np/RANwQa4ICWS5SOjw7gp99nBpXBasapwtZF1fIM3H2gYTM> ;
  nt:wasCreatedFromPubinfoTemplate <http://purl.org/np/RAA2MfqdBczmz9yVWjKLXNbyfBNcwsMmOqcNUxkk1maIM> ,
  <http://purl.org/np/RAjpbM1w3owYhJUBo3DtsuD1XsNAJ8cnGeWAutDVjuAuI> ;
  nt:wasCreatedFromTemplate <http://purl.org/np/RAdpgRpigXtt8iPV9uOpf3wIT3qzOI8Sg2Q72CNV8g-Yo> .
}

```

References

- [1] Enns, G., Shashi, V., Bainbridge, M. et al. Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulum–associated degradation pathway. *Genet Med* 16, 751–758 (2014). doi: 10.1038/gim.2014.22
- [2] Bucur, C.I., Kuhn, T., Ceolin, D., Ossenbruggen, J. van. Expressing high-level scientific claims with formal semantics. In: *Proceedings of the 11th Knowledge Capture Conference 2021*. doi: 10.1145/3460210.3493561.