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A formalization of one of the main claims of "Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes" by Schubert et al. 2014[1]

Valentin Grouès [a,*], Carlos Vega Moreno [b] and Venkata Pardhasaradhi Satagopam [c]

[a] *Luxembourg Centre for Systems Biomedicine, Université du Luxembourg, Luxemburg*
E-mail: valentin.groues@uni.lu; ORCID: https://orcid.org/0000-0001-6501-0806
[b] *Luxembourg Centre for Systems Biomedicine, Université du Luxembourg, Luxemburg*
E-mail: carlos.vega@uni.lu; ORCID: https://orcid.org/0000-0002-7979-9921
[c] *Luxembourg Centre for Systems Biomedicine, Université du Luxembourg, Luxemburg*
E-mail: venkata.satagopam@uni.lu; ORCID: https://orcid.org/0000-0002-6532-5880

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Abstract. Schubert et al. claimed in previous work that mutations in STX1B are associated with epilepsy. We present here a formalization of that claim, stating that all things of class "STX1B mutation" that are in the context of a thing of class "human" frequently have a relation of type "co-occurs with" to a thing of class "epilepsy" in the same context.

Keywords: Human, STX1B mutation, epilepsy

1. Introduction

Schubert et al. [2] state that "Our results thus implicate STX1B and the presynaptic release machinery in fever-associated epilepsy syndromes.". We present here a formalization of the main scientific claim from this quote by using a semantic template called the super-pattern [1].

[1] As RDF/nanopublication: http://purl.org/np/RAeRSya2qIYymsBxiqOZP_oaQpHXUVXiydKvPCFM-7DDQ
[*] Corresponding author. E-mail: valentin.groues@uni.lu.

2. Formalization

Our formalization looks as follows:

CONTEXT-CLASS (“in the context of all ...”):	human
SUBJECT-CLASS (“things of type ...”):	STX1B mutation
QUALIFIER:	frequently
RELATION-TYPE (“have a relation of type...”):	co-occurs with
OBJECT-CLASS (“to things of type...”):	epilepsy

In the context class we use the class “human” (Q5) from Wikidata. In the subject class, we use a new minted class “STX1B mutation” that is a subclass of “mutation” (Q42918) from Wikidata and is related to the class “STX1B” (Q18048867) from Wikidata. In the object class we use the class “epilepsy” (Q41571) from Wikidata.

3. RDF code

This is our formalization as a nanopublication in TriG format:

```
@prefix this: <http://purl.org/np/RAeRSya2qIYymsBxiqOZP_oaQpHXUVXiydKvPCFM-7DDQ> .
@prefix sub: <http://purl.org/np/RAeRSya2qIYymsBxiqOZP_oaQpHXUVXiydKvPCFM-7DDQ#> .
@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:spi a sp:SuperPatternInstance ;
    rdfs:label "Mutations in STX1B are associated with epilepsy" ;
    sp:hasContextClass <http://www.wikidata.org/entity/Q5> ;
    sp:hasSubjectClass <http://purl.org/np/RAPVWYH0x-xyDa9PfBcGUFly3m1FNEO43KG9s0uH-y6yo#STX1B-mutation> ;
    sp:hasQualifier sp:frequentlyQualifier ;
    sp:hasRelation sp:cooccursWith ;
    sp:hasObjectClass <http://www.wikidata.org/entity/Q41571> .
}
sub:provenance {
  sub:activity a sp:FormalizationActivity ;
    prov:used <http://doi.org/10.1038/ng.3130> , sub:quote ;
    prov:wasAssociatedWith orcid:0000-0001-6501-0806 , orcid:0000-0002-6532-5880 , orcid:0000-0002-7979-9921 .
  sub:assertion prov:wasGeneratedBy sub:activity .
  sub:quote prov:value "Our results thus implicate STX1B and the presynaptic release machinery in fever-associated epilepsy
  syndromes" ;
    prov:wasQuotedFrom <http://doi.org/10.1038/ng.3130> .
}
sub:pubinfo {
  sub:sig npx:hasAlgorithm "RSA" ;
    npx:hasPublicKey
"MIGfMA0GCSqGSIb3DQEBAQUAA4GNADCBiQKBgQCY36SLWPLee0SZGM108+7dyjGzKFYg9t09XuL3js13jO3CDzqAZygcrwbJsbLQMRHYvWf0Mkly1ePLgdb43NqEb
XiDHC4o49nHjhi2bSWeRDJ4jFicvhpL1Sjv5N0oFU3wTA8KGN+rYvQ10xhZXcjuuiCMb6yInj6wJ2S+wI14fwIDAQAB" ;
    npx:hasSignature
"QF+C9lXmczrn9cJWuimwLG45Mpntk2CcRIWbeWmKvfE9gmQ6MPKa/x6AfNgVQRnPWppJdDoWepK6m/+m8tWY1WQsXn0KZ8sER+graEHQYuE70Mz9JzuBTyYu0vpWj
5jteoCve5fyvFkhkYVjoRK9tV40GDW6zh+bHqH5tBwrc/0=" ;
    npx:hasSignatureTarget this: .
  this: dct:created "2021-10-29T10:35:33.912+02:00"^^xsd:dateTime ;
    dct:creator orcid:0000-0001-6501-0806 ;
    npx:introduces sub:spi ;
```

```
    <https://w3id.org/linkflows/reviews/isUpdateOf> <http://purl.org/np/RAGo62Hb_Bx1klF4pn1q1Ty40860e3A7Sz4hr2vojZ2wA> ;
    nt:wasCreatedFromProvenanceTemplate <http://purl.org/np/RAB_oy10D3XUP-zYlqGz7Uj58AsUXhEKeGqmRFg5LSgDM> ;
    nt:wasCreatedFromPubinfoTemplate <http://purl.org/np/RAA2MfqdBCzmz9yVWjKLXNbyfBNcwsMmOqcNUxkk1maIM> ,
<http://purl.org/np/RAOGu9Lh0BD4tbIRB9RG6RGRA_ObDh75NTbIqaWgxxs8M> ;
    nt:wasCreatedFromTemplate <http://purl.org/np/RAv68imZrEjfcp2rnEg1hzoBqEVc0cQMtp9_1Za0BxNM4> .
}
```

The following nanopublications introduce the newly minted classes in TriG format. This is the class definition of “STX1B mutation”:

```
@prefix this: <http://purl.org/np/RAPVWYH0x-xyDa9PfBcGUFly3m1FNEO43KG9s0uH-y6yo> .
@prefix sub: <http://purl.org/np/RAPVWYH0x-xyDa9PfBcGUFly3m1FNEO43KG9s0uH-y6yo#> .
@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:STX1B-mutation a <http://www.w3.org/2002/07/owl#Class> ;
    rdfs:label "STX1B mutation" ;
    rdfs:subClassOf <http://www.wikidata.org/entity/Q42918> ;
    skos:definition "mutation in STX1B" ;
    skos:relatedMatch <http://www.wikidata.org/entity/Q18048867> .
}
sub:provenance {
  sub:assertion prov:wasAttributedTo orcid:0000-0001-6501-0806 .
}
sub:pubinfo {
  sub:sig npx:hasAlgorithm "RSA" ;
    npx:hasPublicKey
"MIGfMA0GCSqGSIb3DQEBAQUAA4GNADCBiQKBgQCY36SLWPLee0SZGM108+7dyjGzKFYg9t09XuL3js13jO3CDzqAZygcrwbJsbLQMRHYvWf0Mkly1ePLgdb43NqEb
XiDHC4o49nHjhi2bSWeRDJ4jFicvhpL1Sjv5N0oFU3wTA8KGN+rYvQ10xhZXcjuuiCMb6yInj6wJ2S+wI14fwIDAQAB" ;
    npx:hasSignature
"EVu/+D116w+WZYRCxa5Q4AgDxW7pGuClv6tHouXYSGTCgGu9nZo2pjEqDBmiUyWY8iYyr4FMxGJz2E0bwMU9Ui5svH0EBZPiJQHT6tvfrSRYOP7txHN/LMaxn+IWR
p4Ed1kN9/q5J+BUnnD7x6UghL2s/VlnpSWjVq/EJocnDbU=" ;
    npx:hasSignatureTarget this: .
  this: dct:created "2021-06-21T14:08:44.937+02:00"^^xsd:dateTime ;
    dct:creator orcid:0000-0001-6501-0806 ;
    npx:introduces sub:STX1B-mutation ;
    nt:wasCreatedFromProvenanceTemplate <http://purl.org/np/RANwQa4ICWS5SOjw7gp99nBpXBasapwtZF1fIM3H2gYTM> ;
    nt:wasCreatedFromPubinfoTemplate <http://purl.org/np/RAA2MfqdBCzmz9yVWjKLXNbyfBNcwsMmOqcNUxkk1maIM> ;
    nt:wasCreatedFromTemplate <http://purl.org/np/RAdpgRpigXtt8iPV9uOPf3wIT3qzOI8Sg2Q72CNV8g-Yo> .
}
```

References

[1] C.I. Bucur, T. Kuhn, D. Ceolin and J. van Ossenbruggen, Expressing high-level scientific claims with formal semantics, in: *Proceedings of the 11th Knowledge Capture Conference 2021*. doi:10.1145/3460210.3493561.

[2] J. Schubert, A. Siekierska, M. Langlois et al., Mutations in STX1B, encoding a presynaptic protein, cause fever-associated epilepsy syndromes, *Nat Genet* **46** (2014), 1327–1332. doi:10.1038/ng.3130.