



# Refined JST Thesaurus Extended with Data from other Open Life Science Data Sources

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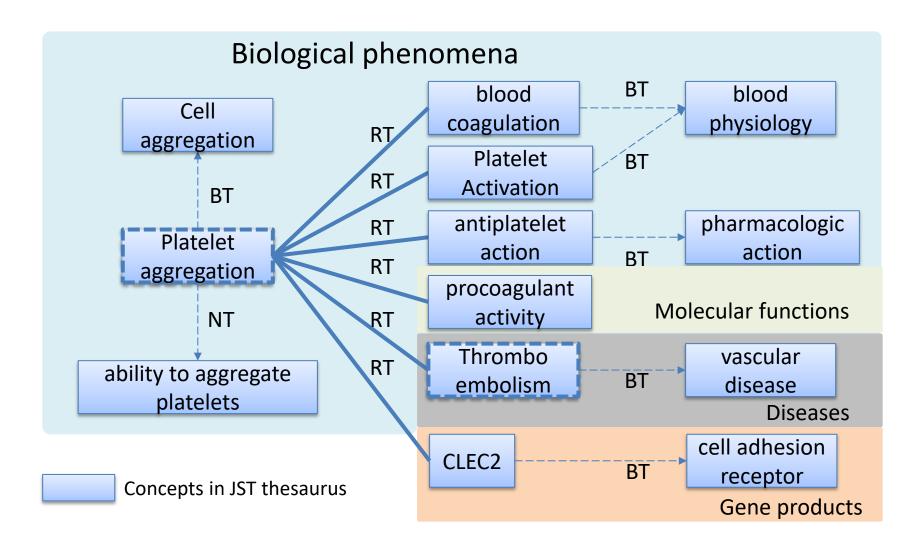
JIST2017, 10 - 12 November 2017 Gold Coast, Australia

## Japan Science and Technology (JST) thesaurus

- It is developed and provided by Japan Science and Technology Agency.
- It is mainly used for indexing scientific literatures.
- All of the terms are written in both of English and Japanese languages.
- It contains approximately 245,000 concepts including 90,000 life science concepts.
  - Gene products (e.g. CLEC2)
  - Drugs (e.g. Gefitinib)
  - Biological phenomena (e.g. platelet aggregation)
  - Diseases (e.g. Thromboembolism)
  - Anatomy (e.g. cartilage)
  - ...



## Structure of JST thesaurus

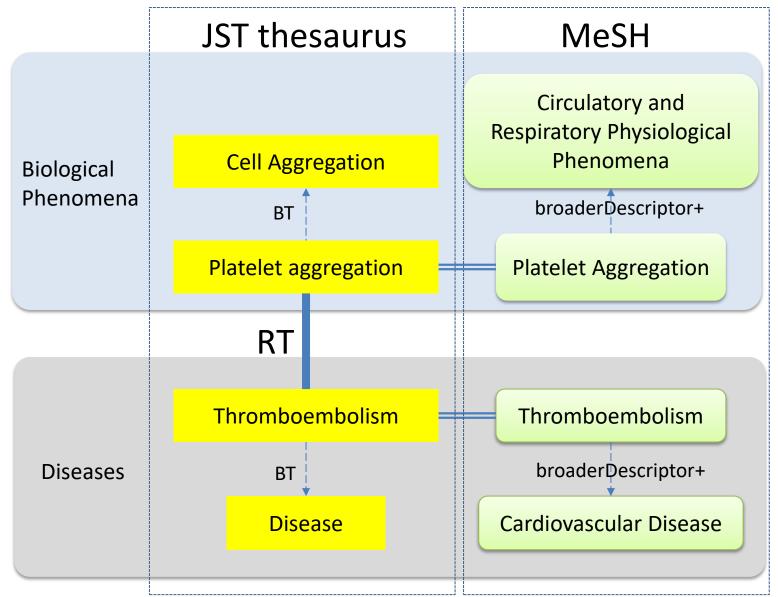


BT: Broader term NT: Narrower term RT: Related term



### Comparison between JST thesaurus and MeSH

The advantages of JST thesaurus





# Problems of the original JST thesaurus (1/2)

#### Problem #1

The JST thesaurus has only three kinds of simple relations (BT, NT, and RT).

Therefore, we cannot describe the following relationships,

- Diseases, and the preceding biological phenomena
- Disease states, and the succeeding ones
- Diseases, and gene products regulating them
- etc.

#### Solution to Problem #1

We have **sub-classified RTs** by using thirty-one kinds of relations such as "has function", and we name the thesaurus "**Refined JST thesaurus**".



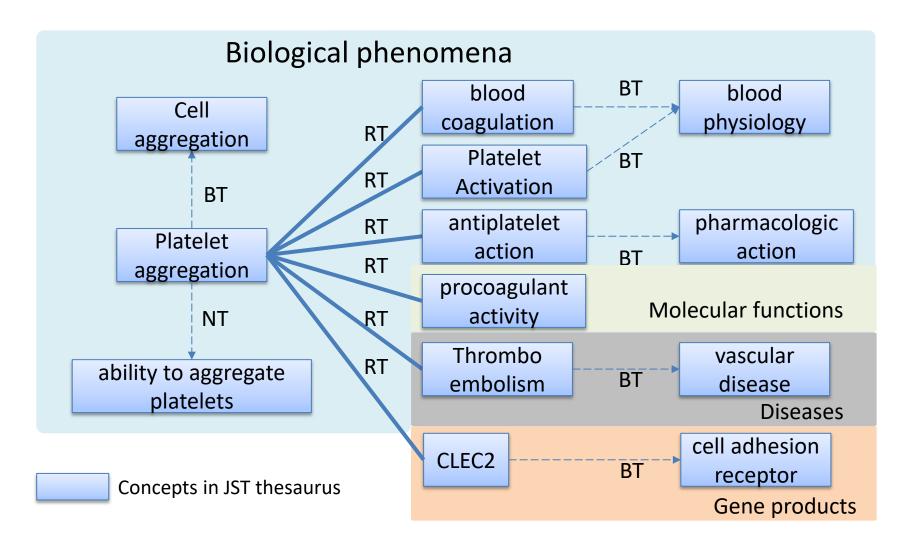
# Thirty-one relations used for the RT sub-classifying

```
rdfs:subClassOf
                                                xkos:succeeds
skos:narrower (has narrower)
                                                xkos:precedes
sio:SIO_000028 (has part)
                                                 sio:SIO 000657(is transformed from)
                                                 sio:SIO 000655 (transforms into)
sio:SIO 000068 (is part of)
sio:SIO 000218 (is quality of)
                                                 sio:SIO 000203 (is connected to)
                                                 sio:SIO 000365 (is creator of)
sio:SIO 000217 (has quality)
                                                 sio:SIO 000364 (has creator)
sio:SIO 000226 (is function of)
                                                 sio:SIO 000145 (is location of)
sio:SIO 000225 (has function)
                                                 sio:SIO 000061 (is located in)
sio:SIO 000123 (antonym)
                                                 obo:RO 0002234 (has output)
sio:SIO 000228 (has role)
                                                 obo:RO 0002353 (output of)
sio:SIO 000227 (is role of)
                                                 sio:SIO 000064 (is provider of)
sio:SIO 001279 (has phenotype)
                                                 sio:SIO 000066 (has provider)
nbdc:isPhenotypeOf
                                                 sio:SIO 000122 (synonym)
sio:SIO_001154 (regulates)
                                                 sio:SIO 000283 (is similar to)
sio:SIO_001155 (is regulated by)
                                                 skos:related (RT)
```



Semanticscience Integrated Ontology (**SIO**) sio: http://semanticscience.org/resource/

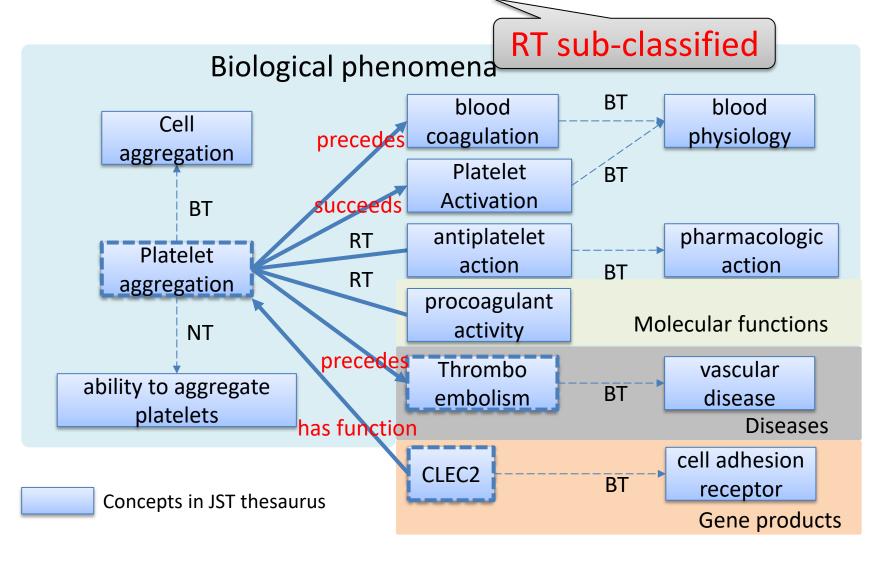
# Structure of original JST thesaurus



BT: Broader term NT: Narrower term RT: Related term



# Structure of refined JST thesaurus



BT: Broader term NT: Narrower term RT: Related term



# Problems of the original JST thesaurus (2/2)

#### Problem #2

The JST thesaurus is designed to collect concepts broadly and shallowly.

For example, the JST thesaurus includes **two** "fibrinolysis" related gene products. On the other hand, Gene Ontology includes the **twenty-seven** related gene products.

#### Solution to Problem #2

We extend the refined JST thesaurus by incorporating information from other ontologies, thesauri, databases and LOD.



# Aim of this study

#### **Aim**

- To update the refined JST thesaurus using thirtyfive relations including new relations
- To evaluate the effectiveness of using sub-classified relations (refined JST thesaurus) by comparing networks (graphs) constructed from the refined JST thesaurus with those from the original JST thesaurus
- To integrate the refined JST thesaurus with other ontologies, thesauri, and LOD.



# Updating the refined JST thesaurus using thirtyfive relations including new relations



# List of newly added relations

Relations	Definitions
sio:SIO_000230 (has input)	Relation between a material and a process related to it as the input to the molecular mechanism
sio:SIO_000231 (is input in)	Reverse relation of "has input"
sio:SIO_000132 (has participant)	Relation between a <u>material</u> or <u>process</u> and a <u>process</u> related to it in the <b>molecular mechanism</b>
sio:SIO_000062 (is participant in)	Reverse relation of "has participant"

<u>Red bold underlined letter</u>: Domain of the relation <u>Blue Italic underlined letter</u>: Range of the relation



Evaluating the effectiveness of using subclassified relations by comparing networks (graphs)

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Example: Classifying diseases using subclassified relations (refined JST thesaurus)





Storing the RDF data in the triple store, and constructing knowledge graphs

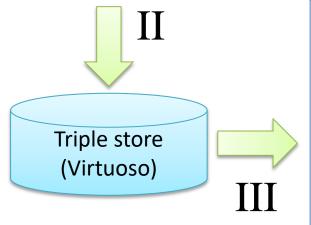
Convert to RDF (Turtle)

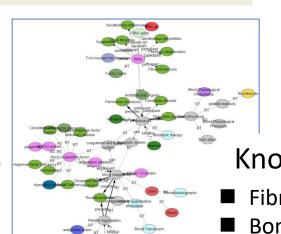


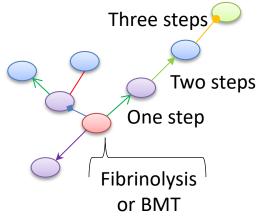


## Example ## jstid:201006070470021247

sio:SIO\_000225 jstid:200906034198027470







Knowledge graph

- Fibrinolysis network
- Bone metabolism turnover (BMT) network
- etc.

Performing SPARQL queries



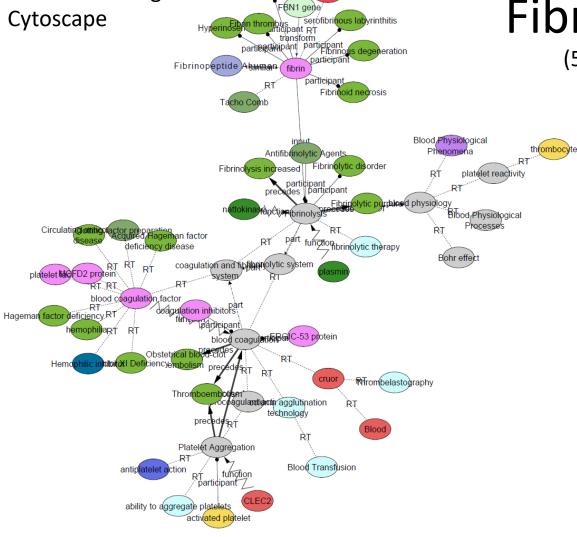
Visualized using

# Fibrinolysis network

(54 concepts, 57 relations)

Examples of subjects of concepts in Fibrinolysis network

Subject	Number of concepts				
Diseases, pathology, and symptoms	16				
Hematology	12				
Protein and peptide	7				
Medicine and medical cares	5				
Drugs	3				
Tissue and organ	3				
Cytology	2				
Enzyme	2				
Genetics	2				
Immunology	1				
Organic compounds	1				
Pharmacology	1				



#### Results of RT sub-classification in Fibrinolysis network

Relations	Number of relations
Specified relations (e.g., has function)	28
Unspecified relations (RTs)	29



## **Bone Metabolic Turnover**

(BMT) network (139 concepts, 140 relations)

#### Results of RT sub-classification in BMT network

Relations	Number of relations
Specified relations (e.g., has function)	78
Unspecified relations (RTs)	62

Examples of subjects of concepts in the BMT network

Subject	Number of concepts
Diseases, pathology, and symptoms	28
Tissue and organ	20
Protein and peptide	17
Cytology	15
Development and growth	14
Drugs	7
Medicine and medical cares	7
Genetics	6
Hormone	5
Immunology	4
Enzyme	3
Pharmacology	2



Cytoscape

Visualized using

calcium binding protein

bone metabolic marker

Classification of diseases connected to fibrinolysis within three steps (relations)

Group	Concept L	Relation L	Concept L	Relation	$\vdash$	Concept	⊢> Relation →	Concept
F1		precedes	Fibrinolysis increased		•			
11		precedes	Fibrinolytic purpura					
F2		is participant in	Fibrinolytic disorder					

fibrin

blood coagulation

coagulation and

fibrinolytic system

blood coagulation

has input

RT

(sibling?)

is part of

RT

(sibling?)

F3

F4

NBDC

fibrinolysis

Group	Concept □	Relation	Concept	Relation	Concept	Relation	Concept
F1		precedes	Fibrinolysis increased				
11		precedes	Fibrinolytic purpura				
F2		is participant in	Fibrinolytic disorder				

is participant

in

precedes

RT

succeeds

is function of

Fibrin thrombus

**Fibrinous** degeneration

Hyperinosemia

**Fibrinoid** 

**Necrosis** 

Serofibrinous inflammation

serofibrinous labyrinthitis

Obstetrical

blood-clot

embolism blood

coagulation

factor platelet

aggregation

blood

coagulation factor

RT

precedes

RT

Factor XI Deficiency

Thromboembolism

Hageman factor deficiency

hemophilia

Acquired Hageman factor deficiency disease

Circulating anticoagulant disease

#### Classification of diseases connected to BMT within three steps (relations)

Group	Concept $\sqsubset$	⇒Relation 🛱	Concept $\vdash$	Relation	Concept $\subseteq$	Relation 🗖	Concept
B1				is participant in	Osteolysis, Essential		
	1	1	1		osteolysis	1	
	1	1	1	!	Osteolytic lesion	†	
B2	1	1	1	precedes	Alveolar Bone Loss	1	
	1	1	Bone	!	Osteitis Fibrosa	1	
	1	1	Resorption		Cystica		
В3				has participant	Calcitonin	is participant	Disorders of thyrocalcitonin secretion
B4				is function of	parathyroid hormone	in '	Ectopic parathormone production
B5		has part		is participant in	Osteoplastic sarcoma		production
	bone metabolic				Lipoma ossificans	†	
	turnover (BMT)	1	1		Bony cataract		
В6	1		precedes	Osteoplastica	†		
Do	1	1	1	preceass	Enostosis	<u> </u>	
	1	1	1		tuberculous		
B7	1	1	osteogenesis		dactylitis PTHRP	has function	Hypercalcemia
י ע	1	1	1		FILIKE	nas jancion	Disorders of
В8				is function of	Calcitonin	is participant in	thyrocalcitonin secretion
	1	1	1		1		Hypocalcitoninemia
В9				is nautiain aut in	parathyroid hormone		Ectopic parathormone production
B10		synonym	bone metabolism	is participant in	Abnormal bone metabolism		
		1		1	Acro-Osteolysis		



# Extending the refined JST thesaurus using other ontologies, thesauri, and LOD



# No. of JST thesaurus concepts having links to concepts of other ontologies in Fibrinolysis, and BMT networks

Ontologies	Fibrinolysis network (54)*	BMT network (139)*
MeSH	21 [38.9%]	56 [40.3%]
ChEBI	2 [3.7%]	4 [2.9%]
Gene Ontology	3 [5.6%]	9 [6.5%]
MEDDRA	7 [13.0%]	11 [7.9%]
SNOMEDCT	20 [37.0%]	36 [25.9%]
NCIT	10 [18.5%]	53 [38.1%]
NDFRT	6 [11.1%]	20 [14.3%]
ICD10	1 [1.9%]	2 [1.4%]
Protein ontology	1 [1.9%]	9 [6.5%]
ICD9	0 [0.0%]	2 [1.4%]
OMIM	4 [7.4%]	16 [11.5%]
FMA	1 [1.9%]	6 [4.3%]

<sup>\*:</sup> The numbers in parentheses donates the numbers of concepts



# JST thesaurus concepts corresponding to Gene Ontology (GO) terms in the Fibrinolysis network, with the number of the related human gene products

JST thesaurus concepts in Fibrinolysis network			Gene Ontology (GO) terms		
Label of JST thesaurus concepts	No. of gene products		Label of GO terms	No. of gene products	
Fibrinolysis	2		fibrinolysis	27	
blood coagulation	0 _ 9		blood coagulation	338 - 422	
platelet aggregation	0	9	platelet aggregation	57	
others	7_		-	-	



## Conclusion

- We can handle information regarding relationships among the concepts of various levels and categories by using thirty-five sub-classified relations having meaning.
- Using the sub-classified relations, we classified the sixteen fibrinolysis-related diseases into four groups.
- We can integrate the JST thesaurus with other ontologies, and thesauri such as Gene Ontology by matching the labels and synonyms.
- For example, we extended the fibrinolysis network with 422 human gene products derived from Gene Ontology.
- We consider that it would be a benefit and a contribution for the semantic web research community to provide a knowledge base for reasoning, such as DL reasoning.



## **Future** work

- To update and extend the refined JST thesaurus, and the knowledge graph.
- To carefully design the structure of the relationship between concepts to convert the thesaurus into a more rigorous and solid ontology.
- To consider removing the existing relations in order to avoid duplication and conflict.
- To establish a general methodology for extending/refining an existing thesaurus or ontology.
- To extend the methodology to other science and technology fields.



## **Publication**

 At present, the refined (RT sub-classified) JST thesaurus is published by a tentative public SPARQL endpoint with CC BY-NC license (http://lod.hozo.jp/repositories/JstNbcdOnt).



# Acknowledgement

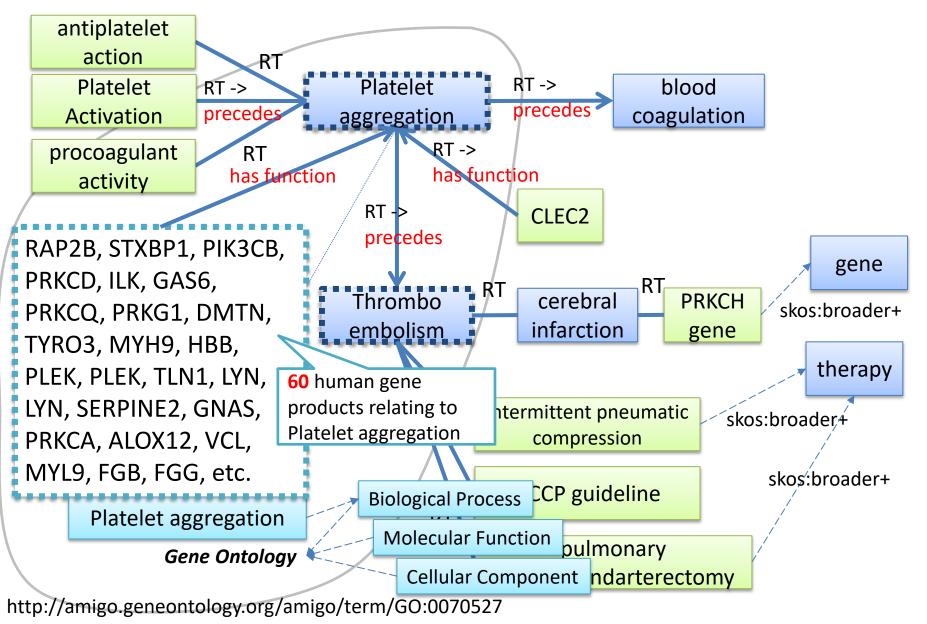
 This work was supported by an operating grant from the Japan Science and Technology Agency and JSPS KAKENHI Grant Number JP17H01789.



Thank you for your attention.







Mush-up of the information of the refined JST thesaurus and Gene Ontology.

28

Examples of reasoning using the inheritance approach "is-a," and "part-of composition."

#### Example 1.

**ABC** transporter

#### Example 2.

spliceosome 
[has part] splicing factor
splicing factor
[has function] RNA splicing



# Related works 1/2

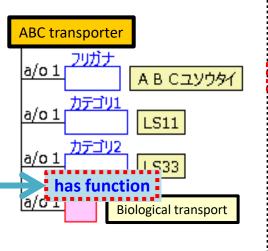
- Examples of the ontological development from thesauri
  - YAGO is constructed by unifying the categories and the infoboxes that are automatically extracted from Wikipedia with synsets of WordNet in a rule-based and heuristic method (Suchanek *et al.*, 2007).
  - AGROVOC is a thesaurus of agriculture and in the project, it is converted into the ontology by the refining RT in more specific relation, and the modeling using OWL is conducted (Soergel et al., 2004).



# Related works 2/2

- Examples of the ontological development by the life-sciences experts and the crowdsourcing
  - Mortensen et al. investigated crowdsourcing's performance for validating the relations among concepts in **SNOMED CT** (2015) and **Gene Ontology** (2016).
  - LEGO (http://geneontology.org/page/connectingannotations-lego-models ) is the Gene Ontology relating project where modeling semantic relations among biological processes, molecular functions, cellular components, and the related gene products is performed using expert crowdsourcing.





Convert to "BT"
Convert to "NT"
Convert to "has part"
Convert to "is part of"
Convert to "has function"
Convert to "is function of"
Convert to "has attribute"
Convert to "is attribute of"
Convert to "antonym"
Decide "RT"
Restore to "RT"

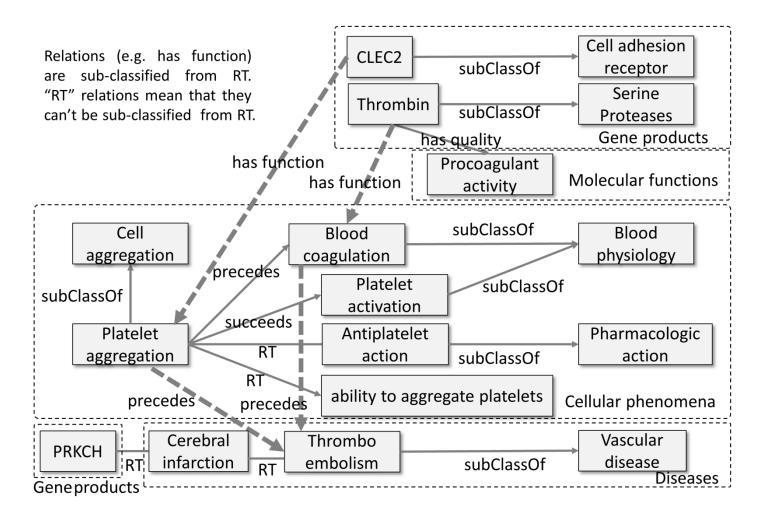
RT sub-classifying using the graphical ontology editor Hozo (in Japanese language)



Select the sub-classifying RT between "ABC transporter" and "Biological transport" with mouse left click, open the popup menu with mouse right click, and select "has function" on mouse left click in the popup window

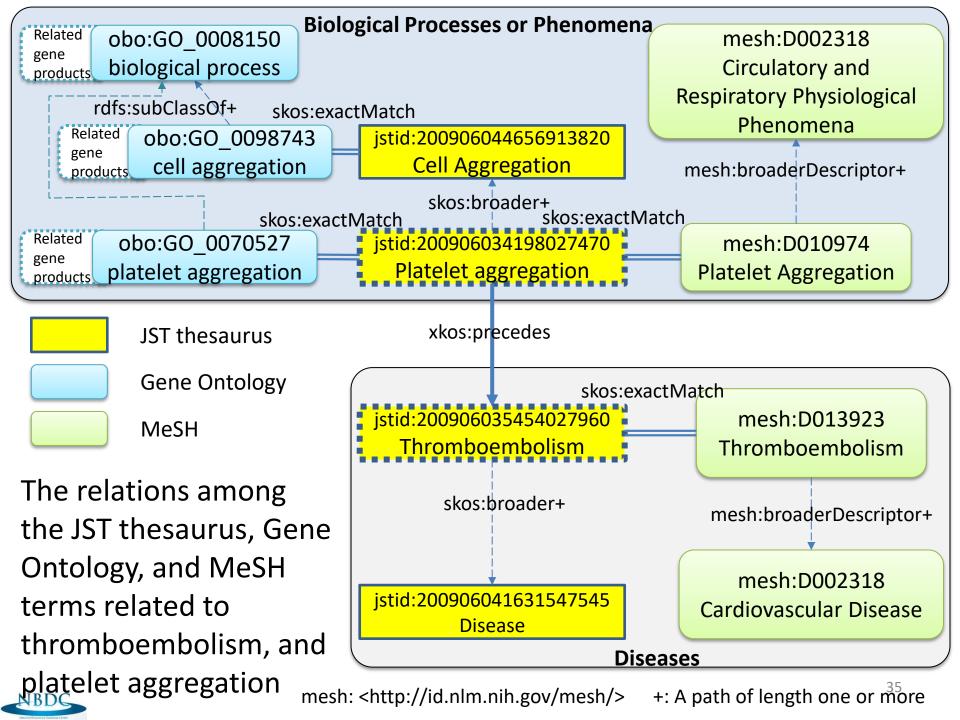
Biological transport

When the RT is converted to "has function", the inverse RT between "Biological transport" and "ABC transporter" is automatically converted to "is function of."



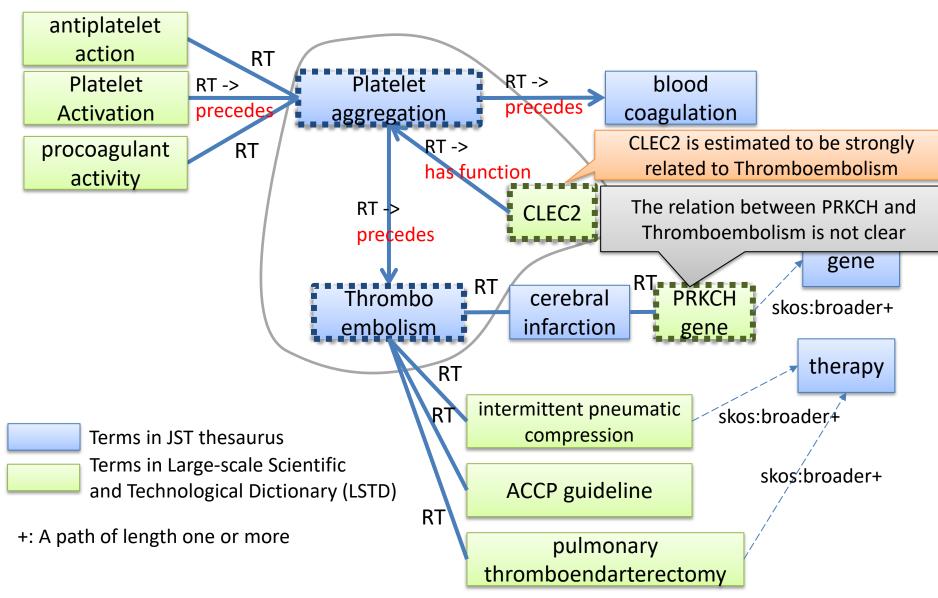






# Discovering diseases-related gene products from the RT sub-classified JST thesaurus graph





The graph of the relations among terms related to "Thromboembolism" in RT sub-classified JST thesaurus.

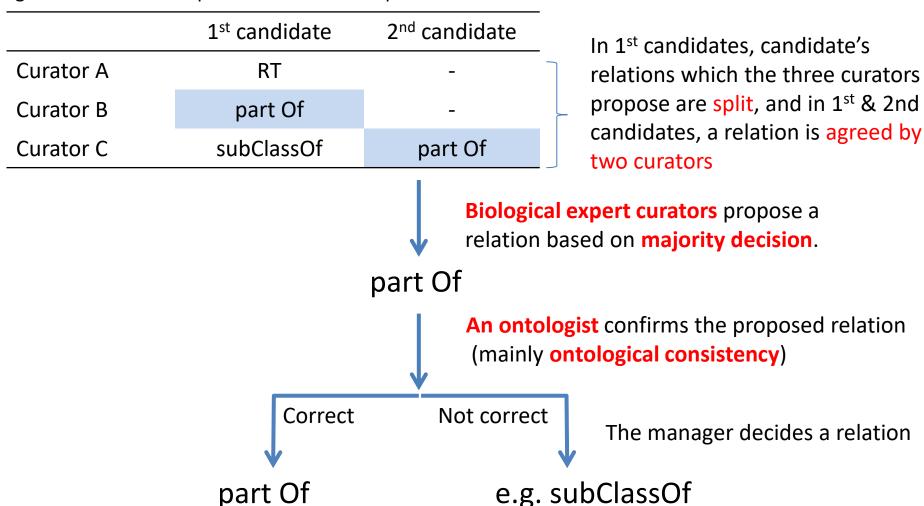


Improvement and evaluation of the method of RT sub-classification by using majority decision of a panel of life-sciences experts



# An example in RT sub-classification process based on majority decision

e.g. What is relationship between "blood corpuscle" and "blood"?

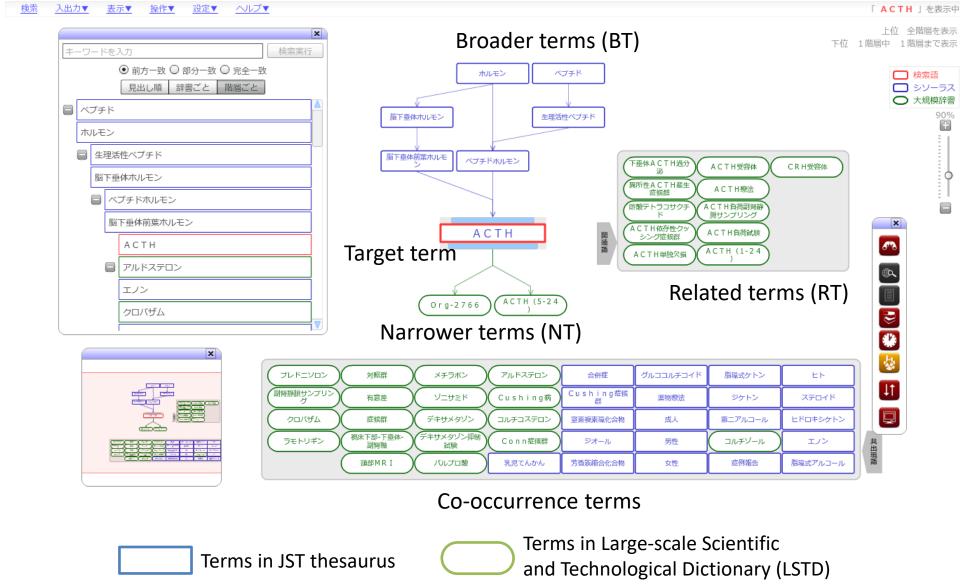




# Our previous works

- Discovering diseases-related gene products from the RT sub-classified JST thesaurus graph
  - T. Kushida, et al.,2016: "Refining JST Thesaurus and Discussing the Effectiveness in Life Science Research" (IESD 2016, co-located with ISWC 2016)
- Improvement and evaluation of the method of RT sub-classification by using majority decision of a panel of life-sciences experts
  - T. Kushida, et al. 2017: "Efficient construction of a new ontology for life sciences by subclassifying related terms in the Japan Science and Technology Agency thesaurus" (ICBO 2017)





# JST thesaurus map

