Measuring the Similarity and Relatedness of Concepts in the Medical Domain : IHI 2012 Tutorial

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- The contents of this tutorial are solely the responsibility of the authors and do not necessarily represent the official views of the National Institutes of Health.

What (we hope) you will learn!

- The distinction between semantic similarity and relatedness (and why both are useful)
- How to measure using information from ontologies, definitions, and corpora
- How to use the freely available software UMLS::Similarity and UMLS::Interface
- How to conduct experiments using freely available reference standards
- How to integrate these measures into clinical NLP applications

Outline

- Introduction to the measures
 - Pedersen, 30 minutes
- Using path and information content measures

- McInnes, 45 minutes
- Using vector and lesk measures
 - Liu, 15 minutes
- Evaluating measures and deploying
 - Pakhomov, 30 minutes

Logistics

- Questions? Just ask!
 - We've planned for ~5 minutes of questions each half hour, but if yours are more extensive or specific to your situation please consider asking after tutorial or via email
- Mailing list, software, data, web interfaces, TUTORIAL SLIDES, and more information :

- http://umls-similarity.sourceforge.net

 Need a break? Feel free, but on your own (and be quick about it!;)

- we'll keep going til end of session (5:00 pm)

Introducing Measures of Semantic Similarity and Relatedness (without tears)

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What are we measuring?

- Concept pairs
 - Assign a numeric value that quantifies how similar or related two concepts are
- Not words
 - Must know concept underlying a word form
 - Cold may be *temperature* or *illness*
 - Concept Mapping
 - Word Sense Disambiguation
 - This tutorial assumes that's been resolved

Why?

- Being able to organize concepts by their similarity or relatedness to each other is a fundamental operation in the human mind, and in many problems in Natural Language Processing and Artificial Intelligence
- If we know a lot about X, and if we know Y is similar to X, then a lot of what we know about X may apply to Y
 - Use X to explain or categorize Y

Similar or Related?

- Similarity based on is-a relations
 - How much is X like Y?
 - Share ancestor in is-a hierarchy
 - LCS : least common subsumer
 - Closer / deeper the ancestor the more similar
- Tetanus and strep throat are similar
 - both are kinds-of bacterial infections

Least Common Subsumer (LCS)



Similar or <u>Related</u>?

- Relatedness more general
 - How much is X related to Y?
 - Many ways to be related
 - is-a, part-of, treats, affects, symptom-of, ...
- Tetanus and deep cuts are related but they really aren't similar
 - (deep cuts can cause tetanus)
- All similar concepts are related, but not all related concepts are similar

Measures of Similarity (all available in UMLS::Similarity)

- Path Based
 - Rada et al., 1989 (path)
 - Caviedes & Cimino, 2004 (cdist)
- Path + Depth
 - Wu & Palmer, 1994 (wup)
 - Leacock & Chodorow, 1998 (lch)
 - Zhong et al., 2002 (zhong)
 - Nguyen & Al-Mubaid, 2006 (nam)

Measures of Similarity (all available in UMLS::Similarity)

- Path + Information Content
 - Resnik, 1995 (res)
 - Jiang & Conrath, 1997 (jcn)
 - Lin, 1998 (lin)

Path Based Measures

- Distance between concepts (nodes) in tree intuitively appealing
- Spatial orientation, good for networks or maps but not is-a hierarchies
 - Reasonable approximation sometimes
 - Assumes all paths have same "weight"
 - But, more specific (deeper) paths tend to travel less semantic distance
- Shortest path a good start, but needs corrections



We count nodes...

- Maximum = 1
 - self similarity

- path(tetanus,tetanus) = 1

- Minimum = 1 / (longest path in isa tree)
 - path(typhoid, oral thrush) = 1/7
 - path(food poisoning, strep throat) = 1/7
 - etc...







- Are bacterial infections and yeast infections similar to the same degree as are tetanus and strep throat ?
- The path measure says "yes, they are."

Path + Depth

- Path only doesn't account for specificity
- Deeper concepts more specific
- Paths between deeper concepts travel less semantic distance

Wu and Palmer, 1994

2 * depth (LCS (a,b))

• wup(a,b) =

depth (a) + depth (b)

depth(x) = shortest is-a path(root,x)

wup(strep throat, tetanus) = (2*2)/(4+3) = .57



wup (bacterial infections, yeast infections) = (2*1)/(2+3) = .4



?

- Wu and Palmer say that strep throat and tetanus (.57) are more similar than are bacterial infections and yeast infections (.4)
- Path says that strep throat and tetanus (.25) are equally similar as are bacterial infections and yeast infections (.25)

Information Content

- ic(concept) = -log p(concept) [Resnik 1995]
 - Need to count concepts
 - Term frequency +Inherited frequency
 - p(concept) = tf + if / N
- Depth shows specificity but not frequency
- Low frequency concepts often much more specific than high frequency ones

Information Content term frequency (tf)



Information Content inherited frequency (if)



Information Content (IC = $-\log (f/N)$ final count (f = tf + if, N = 365,820)



Lin, 1998

2 * IC (LCS (a,b))

• lin(a,b) = ------

IC (a) + IC (b)

• Look familiar?

2* depth (LCS (a,b))

• wup(a,b) =

depth(a) + depth (b)









- Lin says that strep throat and tetanus (.49) are more similar than are bacterial infection and yeast infection (.28)
- Wu and Palmer say that *strep throat* and *tetanus* (.57) are more similar than are *bacterial infection* and *yeast infection* (.4)
- Path says that strep throat and tetanus (.25) are equally similar as are bacterial infection and yeast infection (.25)

What about concepts not connected via is-a relations?

- Connected via other relations?
 - Part-of, treatment-of, causes, etc.
- Not connected at all?
 - In different sections (axes) of an ontology (infections and treatments)
 - In different ontologies entirely (SNOMEDCT and FMA)
- Relatedness!
 - Use definition information
 - No is-a relations so can't be similarity

Measures of relatedness

- Path based
 - Hirst & St-Onge, 1998 (hso)
- Definition based
 - Lesk, 1986
 - Adapted lesk (lesk)
 - Banerjee & Pedersen, 2003
- Definition + corpus
 - Gloss Vector (vector)
 - Patwardhan & Pedersen, 2006

Path based relatedness

- Ontologies include relations other than is-a
- These can be used to find shortest paths between concepts
 - However, a path made up of different kinds of relations can lead to big semantic jumps
 - Aspirin treats headaches which are a symptom of the flu which can be prevented by a flu vaccine which is recommend for children

• so aspirin and children are related ??
Measuring relatedness with definitions

- Related concepts defined using many of the same terms
- But, definitions are short, inconsistent
- Concepts don't need to be connected via relations or paths to measure them
 - Lesk, 1986
 - Adapted Lesk, Banerjee & Pedersen, 2003



Could join them together ... ?



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Each concept has definition



Find overlaps in definitions...



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Overlaps

- Oral Thrush and Alopecia
 - side effect of chemotherapy
 - Can't see this in structure of is-a hierarchies
 - Oral thrush and folliculitis just as similar
- Alopecia and Folliculitis
 - hair disorder & hair
 - Reflects structure of is-a hierarchies
 - If you start with text like this maybe you can build is-a hierarchies automatically!
 - Another tutorial...

Lesk and Adapted Lesk

- Lesk, 1986 : measure overlaps in definitions to assign senses to words
 - The more overlaps between two senses (concepts), the more related

Banerjee & Pedersen, 2003, Adapted Lesk

- Augment definition of each concept with definitions of related concepts
 - Build a super gloss
- Increase chance of finding overlaps
- Implemented in UMLS::Similarity as lesk

The problem with definitions ...

- Definitions contain variations of terminology that make it impossible to find exact overlaps
- Alopecia : ... a result of <u>cancer treatment</u>
- Thrush : ... a side effect of <u>chemotherapy</u>
 - Real life example, I modified the alopecia definition to work better with Lesk!!!
 - NO MATCHES!!
- How can we see that "result" and "side effect" are similar, as are "cancer treatment" and "chemotherapy" ?

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Gloss Vector Measure of Semantic Relatedness

- Rely on co-occurrences of terms
 - Terms that occur within some given number of terms of each other other
- Allows for a fuzzier notion of matching
- Exploits second order co-occurrences
 - Friend of a friend relation
 - Suppose cancer_treatment and chemotherapy don't occur in text with each other. But, suppose that "survival" occurs with each.

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 cancer_treatment and chemotherapy are second order co-occurrences via "survival" Gloss Vector Measure of Semantic Relatedness

- Replace words or terms in definitions with vector of co-occurrences observed in corpus
- Defined concept now represented by an averaged vector of co-occurrences
- Measure relatedness of concepts via cosine between their respective vectors
- Patwardhan and Pedersen, 2006

- Inspired by Schutze, 1998

Implemented in UMLS::Similarity as vector

Thank you!

- http://umls-similarity.sourceforge.net
 - Tutorial slides
 - Links to web interfaces
 - Software downloads
 - Mailing list (join!!)
- Next Using UMLS::Similarity software !

References

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- J. Zhong, H. Zhu, J. Li, and Y. Yu. Conceptual graph matching for semantic search. Proceedings of the 10th International Conference on Conceptual Structures, pages 92-106, 2002

Supplemental Materials

- Semantic Similarity for the Gene Ontology
 - Various measures for GO :
 - http://www.geneontology.org/GO.tools_by_typ e.semantic_similarity.shtml

Supplemental Materials

- WordNet::Similarity
 - Predecessor of UMLS::Similarity
 - Developed at University of Minnesota, Duluth 2001-2006
 - Based on English lexical database WordNet
 - http://wordnet.princeton.edu
 - http://wn-similarity.sourceforge.net

Supplemental Materials

- WebServices::UMLSKS::Similarity
 - HSO implementation for UMLS
 - Developed at University of Minnesota, Duluth 2010-2012, ongoing
 - http://search.cpan.org/dist/WebService-UMLSKS-Similarity/

Introducing UMLS::Interface and UMLS::Similarity (without tears)

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Just in case

Kleenex is still available in the back of the room

Outline

Unified Medical Language System

- UMLS::Interface
 - Backbone of UMLS::Similarity

UMLS::Similarity

Unified Medical Language System

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• UMLS

- Metathesaurus
- Semantic Network
- SPECIALIST LEXICON

Metathesaurus

- ~1.7 million biomedical and clinical concepts; integrated semi-automatically
 - CUIs (Concept Unique Identifiers)
 - Hierarchical Relations
 - PAR/CHD (parent/child)
 - RB/RN (broader/narrower)
 - Non-hierarchical Relations
 - SIB (sibling)
 - RO (other relation)
 - Definitional information

Metathesaurus Sources

Foundational Model of Anatomy (FMA)

Medical Subject Headings (MSH)

SNOMED Clinical Terms (SNOMEDCT)

UMLS::Interface

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UMLS::Interface

 Perl interface to the UMLS present locally in a MySQL database.

• Backbone to UMLS::Similarity

- Main purpose is to return information about CUIs
 - Path information
 - Definitional information

- Two ways to interact with UMLS::Interface
 - API
 - command line programs

Two ways to interact with UMLS::Interface
API
command line interface programs

Nice API examples in the UMLS::Similarity package

Two ways to interact with UMLS::Interface



- Two ways to interact with UMLS::Interface
 - API
 - command line interface programs: 26
 - findPathToRoot.pl
 - findShortestPath.pl
 - getCuiDef.pl
 - findLeastCommonSubsumer.pl
 - getChildren.pl
 - getParents.pl
 - getRelated.pl
 - Ect

- Two ways to interact with UMLS::Interface
 - API
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 - Ect

findPathToRoot.pl <CUI | Term>

findPathToRoot.pl "bacterial infection"

findPathToRoot.pl "bacterial infection"

The paths between bacterial infection (C0004623) and the root: => C0000000 (**UMLS ROOT**) C1135584 (msh) C1256739 (mesh descriptors) C1256741 (index medicus descriptor) C0012674 (diseases) C0004623 (bacterial infections and mycoses) C0004623 (bacterial infection)



findPathToRoot.pl "bacterial infection"

The paths between bacterial infection (C0004623) and the root: => C0000000 (**UMLS ROOT**) C1135584 (msh) C1256739 (mesh descriptors) C1256741 (index medicus descriptor) C0012674 (diseases) C0004623 (bacterial infections and mycoses) C0004623 (bacterial infection)

UMLS ROOT C00000 s a MSH C1135584 s a bacterial infection and mycoses C004615 s a bacterial infection C0004623

--config option

CONFIG FILE NAMED 'config'

SAB :: include <source1>,<source2> REL :: include <relation1>,<relation2>

findPathToRoot.pl with --config

findPathToRoot.pl "bacterial infection" –config config

CONFIG FILE NAMED 'config'

SAB :: include SNOMEDCT REL :: include PAR, CHD
findPathToRoot.pl with --config

findPathToRoot.pl "bacterial infection" –config config

The paths between bacterial infection (C0004623) and the root: C0000000 (**UMLS ROOT**) C1623497 (SNOMEDCT) C0037088 (clinical findings) C0012634 (disorders) C0009450 (communicable diseases) C0004623 (bacterial infection)





findShortestPath.pl <CUI or Term> <CUI or Term>

findShortestPath.pl <CUI or Term> <CUI or Term>

DEFAULT: MSH using the PAR/CHD relations

findShortestPath.pl "salmonella food poisoning" "typhoid" –config config

CONFIG FILE NAMED 'config'

SAB :: include SNOMEDCT REL :: include PAR, CHD



findShortestPath.pl "salmonella food poisoning" "typhoid" –config config

The shortest path between salmonella food poisoning (C0036114) and typhoid (C0041466): => C0036114 (salmonella food poisoning) => C0036117 (salmonella infection) => C0041466 (fever, typhoid)



findShortestPath.pl "salmonella food poisoning" "typhoid" –config config – info

The shortest path between salmonella food poisoning (C0036114) and the tyhpoid (C0041466): => C0036114 (salmonella food poisoning) => PAR (SNOMEDCT) => C0036117 (salmonella infection) => CHD (SNOMEDCT) => C0041466 (fever, typhoid)



findLeastCommonSubsumer.pl

findLeastCommonSubsumer.pl "salmonella food poisoning" "typhoid" –config config

CONFIG FILE NAMED 'config'

SAB :: include SNOMEDCT REL :: include PAR, CHD



findLeastCommonSubsumer.pl

findLeastCommonSubsumer.pl "salmonella food poisoning" "typhoid" –config config

The least common subsumer between salmonella food poisoning (C0036114) and the tyhpoid (C0041466) is salmonella infection (C0036117)





getCuiDef.pl <CUI or Term>



getCuiDef.pl typhoid

The definition(s) of typhoid (C0041466):

1. acute systemic febrile infection caused by Salmonella typhi.

2. an acute systemic febrile infection caused by SALMONELLA TYPHI, a serotype of SALMONELLAENTERICA.

getCuiDef.pl with --sab

getCuiDef.pl typhoid --sab

The definition(s) of typhoid (C0041466):

1. CSP acute systemic febrile infection caused by Salmonella typhi.

2. MSH an acute systemic febrile infection caused by SALMONELLA TYPHI, a serotype of SALMONELLAENTERICA.

UMLS::Similarity

UMLS::Similarity

- A suite of perl modules: implement a number of semantic similarity and relatedness measures
- Similarity measures:
 - Use UMLS path information obtained by UMLS:Interface
- Relatedness measures:
 - The UMLS definition information obtained by UMLS::Interface

Measures

- Path-based similarity
 - Use only the path information between the two concepts
- Information content-based similarity
 - Incorporate the probability of the concept occurring in some text
- Relatedness measures
 - Use contextual information about a concept

Path-based Similarity Measures

- Conceptual Distance measure
 - Rada, et al (1989)
 - Caviedes and Cimino (2004)
- Wu and Palmer (1994)
- Leacock and Chodorow (1998)
- Zhong, et al (2002)
- Ngyuen and Al-Mubaid (2006)

Information-content based Similarity Measures

Resnik (1995)

• Lin (1997)

Jiang and Conrath (1997)

Relatedness Measures

Adapted Lesk

- Banerjee and Pedersen (2003)

Gloss Vector

- Patwardhan and Pedersen (2006)

Using UMLS::Similarity

Three ways to interact with UMLS::Similarity

- API

- command line
- web interface
 - http://atlas.ahc.umn.edu/
 - http://maraca.d.umn.edu/

Using UMLS::Similarity

Three ways to interact with UMLS::Similarity



- http://atlas.ahc.umn.edu/
- http://maraca.d.umn.edu/

umls-similarity.pl

- Main program in UMLS::Similarity
- At its most basic:
 - Takes two terms or CUIs as input
 - Returns similarity between them

umls-similarity.pl <CUI or Term> <CUI or Term>

umls-similarity.pl using defaults

umls-similarity.pl tetanus salmonella

umls-similarity.pl using defaults

umls-similarity.pl tetanus salmonella

0.0769<>tetanus(C0039614)<>salmonella(C0036111)

umls-similarity.pl using defaults

umls-similarity.pl tetanus salmonella

0.0769<>tetanus(C0039614)<>salmonella(C0036111)

Source: MSH Relations: PAR/CHD

Measure: path = 1 / length of the shortest path between the two concepts

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umls-similarity.pl with --config

umls-similarity.pl tetanus salmonella –config config

umls-similarity.pl with --config

umls-similarity.pl tetanus salmonella –config config

CONFIG FILE NAMED 'config'

SAB :: include SNOMEDCT REL :: include PAR, CHD

umls-similarity.pl with --config

umls-similarity.pl tetanus salmonella –config config

0.0714<>tetanus(C0039614)<>salmonella(C0036111)

CONFIG FILE NAMED 'config'

SAB :: include SNOMEDCT REL :: include PAR, CHD





umls-similarity.pl tetanus salmonella –config config

0.0714<>tetanus(C0039614)<>salmonella(C0036111)

CONFIG FILE NAMED 'config'

SAB :: include SNOMEDCT, MSH REL :: include PAR, CHD



umls-similarity.pl tetanus salmonella –config config

0.0714<>tetanus(C0039614)<>salmonella(C0036111)

CONFIG FILE NAMED 'config'

SAB :: include SNOMEDCT REL :: include PAR, CHD, RB, RN

Be careful

- Two reasons:
 - Sources were created for different purposes therefore have different granularity in their path information
 - UMLS::Similarity builds an index of the path to root information of all the CUIs in the source and relations specified in the config file

• This can get large!

Using the –realtime option

umls-similarity.pl tetanus salmonella –config config – realtime

0.0714<>tetanus(C0039614)<>salmonella(C0036111)

CONFIG FILE NAMED 'config'

SAB :: include SNOMEDCT REL :: include PAR, CHD, RB, RN

For the brave – using the entire UMLS

umls-similarity.pl tetanus salmonella –config config – realtime

0.0714<>tetanus(C0039614)<>salmonella(C0036111)

CONFIG FILE NAMED 'config'

SAB :: include UMLS_ALL REL :: include PAR, CHD

umls-similarity.pl with –infile option

umls-similarity.pl --infile <input file>

input file

C0035078<>C0035078 C0018787<>C0027061 C0026269<>C0004238 tetanus<>salmonella typhoid<>salmonella strep throat<>impetigo C0004623<>tetanus typhoid<>C0018787 C0026269<>impetigo

umls-similarity.pl with --infile option

umls-similarity.pl --infile <input file>

output

1.0000<>C0035078(Failure, Kidney)<>C0035078(Failure, Kidney) 0.5000<>C0018787(Heart, NOS)<>C0027061(Myocardium, NOS) 0.2000<>C0026269(Stenosis)<>C0004238(Fibrillation, Atrial) 0.0769<>tetanus(C0039614)<>salmonella(C0036111) 0.0714<>typhoid(C0041466)<>salmonella(C0036111) -1.0000<>strep throat<>impetigo 0.2500<>Infections, Basterial(C0004623)<>tetanus(C0039614) 0.0909<>typhoid(C0041466)<>Heart, NOS(C0018787) 0.1000<>Stenosis(C0026269)<>impetigo(C0021099)

umls-similarity.pl with --infile option

umls-similarity.pl --infile <input file>

output

1.0000<>C0035078(Failure, Kidney)<>C0035078(Failure, Kidney) 0.5000<>C0018787(Heart, NOS)<>C0027061(Myocardium, NOS) 0.2000<>C0026269(Stenosis)<>C0004238(Fibrillation, Atrial) 0.0769<>tetanus(C0039614)<>salmonella(C0036111) 0.0714<>typhoid(C0041466)<>salmonella(C0036111) -1.0000<>strep throat<>impetigo 0.2500<>Infections, Bacterial(C0004623)<>tetanus(C0039614) 0.0909<>typhoid(C0041466)<>Heart, NOS(C0018787) 0.1000<>Stenosis(C0026269)<>impetigo(C0021099)
--measure options for path-based measures

- path: simple path
- cdist: conceptual distance
- wup: Wu and Palmer (1994)
- Ich: Leacock and Chodorow (1998)
- zhong: Zhong, et al (2002)
- nam: Ngyuen and Al-Mubaid (2006)

--measure options for path-based measures

- path: simple path
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- zhong: Zhong, et al (2002)
- nam: Ngyuen and Al-Mubaid (2006)

umls-similarity.pl <CUI or Term> <CU or Term> —measure <measures>

umls-similarity.pl with --measure

umls-similarity.pl tetanus salmonella –measure wup

0.3636<>tetanus(C0039614)<>salmonella(C0036111)

umls-similarity.pl with --measure

umls-similarity.pl tetanus salmonella –measure wup

0.3636<>tetanus(C0039614)<>salmonella(C0036111)

umls-similarity.pl tetanus salmonella –measure lch

1.1239<>tetanus(C0039614)<>salmonella(C0036111)

--measure options for IC-based measures

- res: Resnik (1996)
- lin: Lin (1997)
- jcn: Jiang and Conrath (1997)

umls-similarity.pl tetanus salmonella –measure lin

0.3636<>tetanus(C0039614)<>salmonella(C0036111)

Information Content

IC = -log(P(concept))

- P(concept) :
 - Calculated by summing the probability of the concept seen a corpus with the probability of the concept's descendants
 - Probabilities obtained from external corpus

Example: probability of hand



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Example: probability of hand

 $\begin{array}{l} \mathsf{P}(\mathsf{Bacterial Infection}) = \\ \mathsf{P}(\mathsf{C0004623}) + \\ \mathsf{P}(\mathsf{C0036117}) + \\ \mathsf{P}(\mathsf{C0038395}) + \\ \mathsf{P}(\mathsf{C0039614}) + \\ \mathsf{P}(\mathsf{C0036114}) + \\ \mathsf{P}(\mathsf{C0041466}) + \\ \mathsf{P}(\mathsf{C0021099}) + \\ \mathsf{P}(\mathsf{C0032269}) \end{array}$



Example: probability of hand



T.

IC-based measures: default

umls-similarity.pl tetanus salmonella –measure lin

- IC(concept) comes from UMLSonMedline
 - National Library of Medicine
 - Consists of concepts from 2009 AB UMLS and the frequency they occurred in medline using the Essie Search Engine (Ide et al 2007).
 - Medline: database of citations of biomedical and clinical articles.

Create your own IC file

- Two programs in utils/ directory
 - create-icfrequency.pl
 - create-icpropagation.pl

RAW TEXT

Background: The optimal femorotibial angle (FTA) after high tibial osteotomy (HTO) is still controversial. Our hypthesis was that FTA itself may not Be reliable because

. . .

Create your own IC file: step through RAW TEXT create-icfrequency.pl Background: The SAB :: include SNOMEDCT optimal femorotibial REL :: include PAR, CHD angle (FTA) after high tibial osteotomy C000000<>0 (HTO) is still C0000039<>9594 controversial. Our C0000052<>1518 hypthesis was that C0000097<>15978 FTA itself may not C0000102<>2149 Be reliable because C0000163<>1078

. . .

. . .















icfrequency and icpropagation

UMLS::Similarity utils/directory

create-icfrequency.pl

--term option (default)

--metamap option

create-icpropagation.pl--smooth

create-icfrequency.pl icfreq text --config config --metamap

CONFIG FILE: config

SAB :: include SNOMEDCT REL:: include PAR, CHD

RAW TEXT : text

Background: The optimal femorotibial angle (FTA) after high tibial osteotomy (HTO) is still controversial. Our hypthesis was that FTA itself may not be reliable because ...

create-icfrequency.pl/icfreq text --config config --metamap

CONFIG FILE: config

SAB :: include SNOMEDCT REL:: include PAR, CHD RAW TEXT : text

Background. The optimal femorotibial angle (FTA) after high tibial osteotomy (HTO) is still controversial. Our hypthesis was that FTA itself may not be reliable because ...

create-icfrequency.pl icfreq



CONFIG FILE: config

SAB :: include SNOMEDCT REL:: include PAR, CHD

RAW TEXT : text

Background: The optimal femorotibial angle (FTA) after high tibial osteotomy (HTO) is still controversial. Our hypthesis was that FTA itself may not be reliable because ...



CONFIG FILE: config

SAB :: include SNOMEDCT REL:: include PAR, CHD

RAW TEXT : text

Background: The optimal femorotibial angle (FTA) after high tibial osteotomy (HTO) is still controversial. Our hypthesis was that FTA itself may not be reliable because ...

create-icfrequency.p icfreq

--config config --metamap

ICFREQUENCY FILE : icfreq

SAB :: include SNOMEDCT REL :: include PAR, CHD

C0000000<>0 C0000039<>9594 C0000052<>1518 C0000097<>15978 C0000102<>2149 C0000163<>1078

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create-icpropagation.pl example

create-icpropagation.pl icprop icfreq --config config

CONFIG FILE: config

SAB :: include SNOMEDCT REL:: include PAR, CHD

ICFREQUENCY FILE : icfreq

SAB :: include SNOMEDCT REL :: include PAR, CHD

C0000000<>0 C0000039<>9594 C0000052<>1518 C0000097<>15978 C0000102<>2149 C0000163<>1078

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create-icpropagation.pl example

create-icfrequency.pl icprop icfreq --config config



create-icpropagation.pl example

create-icfrequency.pl icprop icfreq --config config

ICPROPAGATION FILE : icprop

SAB :: include SNOMEDCT REL :: include PAR, CHD

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Using icpropagation file

umls-similarity.pl tetanus salmonella

- config config
- icpropagation icprop

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– measure lin

CONFIG FILE: config

ICPROPAGATION FILE : icprop

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C000000<>1 C0000039<>2.3e^06 C0000052<>3.21e^07 C0000097<>3.38e^06 C0000102<>9.12e^07 C0000163<>4.78e^05

Using icpropagation file

umls-similarity.pl tetanus salmonella

- config config
- icpropagation icprop
- measure lin



Using icproapgation file

umls-similarity.pl tetanus salmonella

- config config
- icpropagation icprop
- measure lin

0.3636<>tetanus(C0039614)<>salmonella(C0036111)

Similarity overview

Similarity overview

CONFIG FILE

SAB :: include SNOMEDCT REL:: include PAR, CHD

umls-similarity.pl program
General options:
--config FILE
CONFIG FILE

SAB :: include SNOMEDCT REL:: include PAR, CHD

umls-similarity.pl program

- General options:
 - --config FILE
 - --measure MEASURE



CONFIG FILE

SAB :: include SNOMEDCT REL:: include PAR, CHD

umls-similarity.pl program

 General options:
 --config FILE
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CONFIG FILE

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umls-similarity.pl program

CONFIG FILE

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C0000097<>15978		C0000097<>3.38e^06				
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C0000163<>1078		C0000163<>1.07e^06	9			

UML::Similarity web interface

- Located:
 - http://atlas.ahc.umn.edu
 - http://maraca.d.umn.edu

UML::Similarity web interface	
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UMLS::Similarity is a freely available open source software package that can be used to obtain the similarity or relatedness between two biomedical terms from the Unified Medical Language System (UMLS).

DIRECTIONS: You may enter any two terms or Concept Unique Identifiers (CUIs) below. If terms are entered, then the relatedness or similarity of the possible CUIs will be computed and the pair with the highest score returned. The difference between similarity and relatedness is

Detailed instructions. About the Similarity Measures. About the Relatedness Measures.

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UML::Similarity web interface

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UMLS::Similarity Web Interface

UMLS::Similarity is a freely available open source software package that can be used to obtain the similarity or relatedness between two biomedical terms from the Unified Medical Language System (UMLS).

DIRECTIONS: You may enter any two terms or Concept Unique Identifiers (CUIs) below. If terms are entered, then the relatedness or similarity of the possible CUIs will be computed and the pair with the highest score returned. The difference between similarity and relatedness is

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View Definitions	
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DIRECTIONS: You may enter any two terms or Concept Unique Identifiers (CUIs) below. If terms are entered, then the relatedness or similarity of the possible CUIs will be computed and the pair with the highest score returned. The difference between similarity and relatedness is

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UMLS::Similarity is a freely available open source software package that can be used to obtain the similarity or relatedness betw	Language System (UMLS).	
View Definitions CLICK	Definitions	
View Shortest Path	tetanus (C0039614)	
Results:	MSH : A disease caused by tetanospasmin, a powerful protein toxin produced by CLOSTRIDIUM TETANI. Tetanus usually occurs after an acute injury, such as a	
The similarity of tetanus (C0039614) and salmonella (C0036111) using Path Length (path) is 0.0769.	characterized by tetanic muscular contractions and hyperreflexia. Localized tetanus	
Using:	presents itself as a mild condition with manifestations restricted to muscles near the wound. It may progress to the generalized form.	
SAB :: include MSH	CSP : disease caused by tetanospasmin, a powerful protein toxin produced by Clostridium tetani; tetanus usually occurs after an acute injury, such as a puncture	
REL :: include PAR/CHD	wound or laceration; generalized tetanus, the most common form, is characterized by tetanic muscular contractions and hyperreflexia; localized tetanus presents itself as a	
View relatedness of all possible senses	mild condition with manifestations restricted to muscles near the wound. MEDLINEPLUS :	
DIRECTIONS: You may enter any two terms or Concept Unique Identifiers (CUIs) below. If terms are entered, then the relate returned. The difference between similarity and relatedness is	Tetanus is a serious illness caused by tetanus bacteria. The bacteria live in soil, saliva, dust and manure. The bacteria usually enter the body through a deep cut, like those you might get from cutting yourself with a knife or stepping on a nail.	
Detailed instructions. About the Similarity Measures.	The infection causes painful tightening of the muscles, usually all over the body. It can	
About the Relatedness Measures.	lead to "locking" of the jaw, which makes it impossible to open your mouth or swallow. If this happens, you could die of suffocation.	
Term 1: tetanus Term 2: salmonella		
- Semantic Similarity		
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UMLS::Similarity is a freely available open source software package that can be used to obtain the similarity or relatedness betw	Language System (UMLS).
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Results: The similarity of tetanus (C0039614) and salmonella (C0036111) using Path Length (path) is 0.0769. Using: SAB :: include MSH REL :: include PAR/CHD View relatedness of all possible senses DIRECTIONS: You may enter any two terms or Concept Unique Identifiers (CUIs) below. If terms are entered, then the relater returned. The difference between similarity and relatedness is Detailed instructions. About the Similarity Measures. About the Similarity Measures.	The shortest path between C0039614 (Tetanus) and C0036111 (salmonellas) is: C0039614 (Tetanus) => C0009062 (Infections, Clostridium) => C0085426 (Infections, Gram-Positive Bacterial) => C0004623 (Infections, Bacterial) => C0004615 (Bacterial Infections and Mycoses) => C0012674 (Diseases (MeSH Category)) => C1256741 (Topical Descriptor) => C1256748 (Organisms (MeSH Category)) => C0004611 (Bacteria.) => C0018150 (bacterium gram-negative) => C0018152 (Gram Negative Facultatively Anaerobic Rods) => C0014346 (Enterobacteriaceae) => C0036111 (salmonellas) C0039614 (Tetanus) => C0009062 (Infections, Clostridium) => C0085426 (Infections, Gram-Positive Bacterial) => C0004623 (Infections, Bacterial) => C0004615 (Bacterial Infections and Mycoses) => C0012674 (Diseases (MeSH Category)) => C1256741 (Topical Descriptor) => C1256748 (Organisms (MeSH Category)) => C1256741 (Topical Descriptor) => C1256748 (Organisms (MeSH Category)) => C0004611 (Bacteria.) => C0751985 (Proteobacteria) => C0751988 (Gammaproteobacteria) => C0014346 (Enterobacteriaceae) => C0036111 (salmonellas)
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Questions?

Relatedness Measures Contained in the UMLS-Similarity Package

> Measuring the Similarity and Relatedness of Concepts in the Medical Domain : IHI 2012 Tutorial

> > Ying Liu

College of Pharmacy University of Minnesota Minneapolis, MN

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- Introduction of the semantic relatedness measures: lesk and vector
- How to use UMLS-Similarity to get the relatedness score

Ontology dependent and independent measures

- Ontology dependent measures
 - relay on the concept hierarchies or ontologies
 - is-a, has-part, and is-a-part-of...
 - path based: path, Wu&Palmer, and Leacock&Chodorow...
 - Information content (IC) based: Resnik, Lin and Jiang&Conrath
- Ontology independent measures
 - rely on the related concepts have a similar context
 - lesk : Adapted Lesk, Banerjee and Pedersen (2003)
 - gloss vector : Patwardhan and Pedersen (2006)

Lesk semantic relatedness measure

• Semantic relatedness is a function of the overlap between their definitions

$$rel_{lesk} = \sum_{i=1}^{n} freq_{overlap_i} * length_{overlap_i}^{2}$$

Example:

• influenza : infectious disease, fever, muscle pains, and general discomfort aspirin : relieve pains, reduce fever, an anti-inflammatory medication

$$rel_{lesk} = 1*1^{2} + 1*1^{2} = 2$$

Disadvantages of lesk method

- Based strictly on definitions and doesn't use any other knowledge source
- A word could have several forms example: Minnesota vs. MN
- Different words have the same semantic meaning example: utility vs. usage

Vector semantic relatedness measure

Definition

- **influenza** : infectious disease, fever, muscle pains, and general discomfort.
- **aspirin** : relieve pains, reduce fever, an anti-inflammatory medication
- Co-occurrence vector

Infectious: bacterial diagnoses fungi disease: behavior body cost feel research risks fever: attack body case fell health muscle: ache change exercise injury pains: nausea headache general: analysis appear body diet family discomfort: anger felt nausea stress relieve: chest drug time pains reduce: abnormal access asthma clinical error anti-inflammatory: drugs therapy medication: abuse choice diet expert patient



The procedure of the second-order context vector semantic relatedness method



Step1: A bi-gram example with window=3

move the window from left to right

Influenza is transmitted through the air by coughs or sneezes.

window=3

influenza<>is<> is<>through<> through<>the<> the<>by<> by<>coughs<> or<>sneezes is<>transmitted<> transmitted<>through<> through<>air<> air<>by<> by<>or<> coughs<>sneezes influenza<>transmitted<> transmitted<>the<> the<>air<> air<>coughs<> coughs<>or<>



move the window from left to right

Influenza is transmitted through the air by coughs or sneezes.

window=3

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influenza<>transmitted<> transmitted<>the<> the<>air<> air<>coughs<> coughs<>or<>

Step1: how to get the bi-gram list

Build the bi-gram list by Text-NSP

- Ngram Statistics Package
- download at : http://sourceforge.net/projects/ngram/
- count.pl or huge-count.pl
 - · generate the bi-gram list
- count2huge.pl
 - if use count.pl, need to convert the bi-gram order by count2huge.pl

The procedure of the second-order context vector semantic relatedness method



Sept 2: *n* x *m* co-occurrence matrix



Infectious: bacterial 2 diagnoses 1 fungi 3 disease: behavior 1 body 5 cost 3 feel 7 research 5 risks 2 fever: attack 3 body 5 case 10 fell 6 health 8 muscle: ache 2 change 5 exercise 9 injury 2 pains: nausea 8 headache 20 general: analysis 5 appear 6 body 9 diet 4 family 7 discomfort: anger 2 fell 4 nausea 6 stress 3 relieve: chest 6 drug 4 time 7 pains 9 reduce: abnormal 2 access 5 asthma 6 clinical 9 error 3 anti-inflammatory: drugs 4 therapy 7 medication: abuse 3 choice 6 diet 8 expert 4 patient 3

Sept 2: *n* x *m* co-occurrence matrix

- vector-input.pl of UMLS-Similarity
 - read the sorted bi-gram list
- co-occurrence matrix file has two parts
 - index file: record each vector's position and length
 - matrix file: record the vectors

The procedure of the second-order context vector semantic relatedness method



Step 3: Organization of a concept's definition



influenza : infectious disease, fever, muscle pains, and general discomfort. (CUI)

- + disease (parent) : an abnormal condition affecting the body of an organism.
- + **cold** (child) : running nose, sneeze.
- + cough (associate terms)
- + influenza is transmitted through the air by coughs and sneezes. (WordNet)

Step 3: Organization of a concept's definition

- --config option to define source and relations
- --dictfile option to import external definitions
 from WordNet or other sources

The procedure of the second-order context vector semantic relatedness method



Step 4: Geometric explanation of the semantic relatedness measure based on vector product



Influenza (C_1) : bacterial 2 diagnoses 1 fungi 3 behavior 1 body 19 cost 3 feel 7 research 5 risks 2 attack 3 case 10 fell 6 health 8 ache 2 change 5 exercise 9 injury 2 nausea 8 headache 20 analysis 5 appear 6 diet 4 family 7 anger 2 fell 4 nausea 6 stress 3 **aspirin** (C_2) : chest 6 drug 4 time 7 pains 9 nausea 8 headache 20 abnormal 2 access 5 asthma 6 clinical 9 error 3

attack 3 body 5 case 10 fell 6 health 8 drugs 4 therapy 7 abuse 3 choice 6 diet 8 expert 4 patient 3

The procedure of the second-order context vector semantic relatedness method



Command to get the vector relatedness

- umls-similarity.pl --measure vector --vectorindex index_file --vectormatrix matrix_file influenza aspirin
- Other options
 - · --config
 - · --dictfile
 - · --compoundfile
 - · --doubledef
 - · --stoplist
 - · --defraw
 - · --stem
 - · --debugfile

http://atlas.ahc.umn.edu/cgi-bin/umls_similarity.cgi

	UMLS::Similarity Web Interface
UMLS::Similarity is a freely available open source so	ftware package that can be used to obtain the similarity or relatedness between two biomedical terms from the Unified Medical Language System (UMLS). Please note, the link to the UMLS::Similarity package is severed at this time for the purpose of anonymity.
View Definitions Results: The relatedness of influenza (C0021400) and aspirin (The relatedness of influenza (C0021400) and aspirin (C0004057) using Vector Measure (vector) is 0.2601.
Jsing: SABDEF :: include UMLS ALL	
RELDEF :: include CUI/PAR/CHD/RB/RN	
DIRECTIONS: You may enter any two terms or Conce Detailed instructions. About the Similarity Measures Term 1: Influenza aspirin Semantic Similarity SAB: MSH REL: PARICHD Similarity: Path Length (path)	epi Unique Identifiers (CUIs) below. If terms are entered, then the relatedness or similarity of the possible CUIs will be computed and the pair with the highest score returned. The difference between similarity and relatedness is Term 1: influenza aspirin
Semantic Relatedness SABDEF: UMLS_ALL _ REL DEF: CLILDARICHDIRERN Compute Relatedness Clear how version info	1
his interface is based on the WordNet:Similarity web	interface L

Applications of UMLS-Similarity

- Used UMLS-Similarity to calculate the gene and disease similarity.
 "<u>Finding Disease Similarity Based on Implicit Semantic Similarity</u>" at Journal of Biomedical Informatics 2011 by Mathur and Dinakarpandian
- Used UMLS-Similarity to connect relevant users together in the conversation and also provide contextual recommendations relevant to the health information conversation system Cobot. "<u>Socio-Semantic Health</u> <u>Information Access</u>" at Association for the Advancement of Artificial Intelligence 2011 by Sahay and Ram
- Used UMLS-Similarity to improve the performance of the classifier OWCP (one word conjunct pairs). "<u>Coordination Resolution in Biomedi cal Texts</u>" Ph. D dissertation 2011 by Philip Ogren

The increase of registered UMLS-Similarity Users at Yahoo group



Sign up at : <u>http://tech.groups.yahoo.com/group/umls-similarity</u>
Anonymous users of the web interface are from 46 countries and territories

http://atlas.ahc.umn.edu/cgi-bin/umls_similarity.cgi



Summary

- Second order co-occurrence vector semantic relatedness method
 - Use proper relationship to construct the definition
 - Choice proper corpus to build the co-occurrence vector
 - Does not rely on the hierarchical structure
- http://atlas.ahc.umn.edu/cgi-bin/umls_similarity.cgi

Tomorrow...

- 10:30am-12:00pm Session D3-1A BioNLP (Vista Room)
- Semantic Relatedness Study Using Second Order Co-Occurrence Vector Computed by Biomedical Corpora, UMLS and WordNet

--Ying Liu, Bridget T. McInnes, Ted Pedersen, Serguei Pakhomov and Genevieve Melton-Meaux

Thank You !

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Evaluating and deploying measures of semantic relatedness

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Outline

- Different types of evaluation (direct vs indirect)
- Creating a new reference standard
- Using existing resources
- Available reference standards (M&C, R&G, MayoSRS, MiniMayoSRS, UMNSRS)
- Evaluation metrics and statistical considerations (correlation, precision/recall, inter-rater agreement)
- Application to WSD

Evaluation

- Direct vs indirect approaches
 - Direct evaluation against a manually annotated corpus of word/term pairs
 - Indirect evaluation using measures to accomplish another task (e.g., WSD)

Evaluation

- Direct vs. Indirect approaches
 - Direct evaluation against a manually annotated corpus of word/term pairs
 - Indirect evaluation using measures to accomplish another task (e.g. WSD)

Pros and Cons

- Direct approaches
 - Pros:
 - relatively easy to implement and interpret
 - eliminate potential confounders present in indirect approaches
 - allow some control over the definition of relatedness/similarity
 - enable easy debugging and fine-tuning

– Cons:

- limited inter-annotator agreement due to individual variability
- limited generalizability

Pros and Cons

- Indirect Approaches
 - Pros:
 - assess the "real-world" impact of relatedness and similarity measures
 - less prone to reference standard reliability issues
 - Is not constrained by specific definition of relatedness/similarity – more generalizable

– Cons:

- relatively difficult to implement unless the dataset already exists (e.g. WSD)
- requires a study design that can isolate the effects of measures from other task variables

Direct Evaluation

- Creating a new reference standard
 - 1. select the data (word pairs) to be manually assessed
 - 2. determine the response variable (discrete, vs. continuous, nominal vs. numeric)
 - 3. determine the mode of presentation (paper, computer, spreadsheet, web-based, etc.)

- 4. select and train annotators
- 5. annotate
- 6. evaluate the reliability of the dataset

Direct Evaluation

Using existing resources as a reference

- Miller and Charles (General Eng)
- Rubenstein and Goodenough (General Eng)
- MayoSRS and MiniMayoSRS (Medical)
- UMNSRS (Medical)

Indirect Evaluation

- Creating a new reference standard
 - Too many variations to discuss
 - In general, follow standard guidelines for the specific task and determine the reliability of the reference standard
 - Example: WSD
 - manually label (or use already labeled) corpus of disambiguated text with known reliability

Indirect Evaluation

- Using existing resources
 - NLM WSD dataset
 - http://wsd.nlm.nih.gov/
 - 50 frequent UMLS concepts, each with 100 ambiguous instances
 - 11 annotators (majority vote)
 - Kappa statistic is discussed but not reported
 - http://wsd.nlm.nih.gov/info/AMIA2001weeber.pdf

Statistical Considerations

Reliability of reference standards

- Unadjusted for chance
 - Correlation
 - Percent agreement
- Adjusted for chance
 - Kappa
 - Intra-class correlation coefficient

Available Reference Standards

- Miller and Charles (M&C) and Rubenstein and Goodenough (R&G)
 - General English but...
 - May be used for annotator training purposes
 - Rubenstein and Goodenough (1965)
 - Synonymy judgements
 - 65 noun pairs
 - 51 subjects
 - Scale 0.0 4.0
 - R = 0.85 (repeated measures 2 weeks apart) 12

Miller and Charles Set

- 30 pairs of English nouns from R&G set

Word1	Word2	Mean score
Car	Automobile	3.92
Gem	Jewel	3.84
Coast	Shore	3.70
Monk	Oracle	1.10

- High correlation with R&G set r = 0.97
- Later reproduced by Resnik (1999) r = 0.90

Available Reference Standards

- Mayo Semantic Relatedness Standard (SRS)
 - Initially generated by a rheumatologist
 - 101 term pairs (originally 120) from four categories
 - Closely related, somewhat related, somewhat unrelated, completely unrelated
 - Not restricted to single word concepts
 - Annotated by 3 physicians and 12 medical coders
 - r = 0.51

Mayo SRS Format

Mean	CUI1	CUI2	TERM1	TERM2
6.69	C0311394	C0231685	difficulty walking	antalgic gait
2.38	C0035450	C0034079	rheumatoid nodule	lung nodule
1.00	C0409162	C0333286	hand splint	splinter hemorrhage
1.00	C0011849	C0032584	diabetes	polyp

Mini-Mayo-SRS

- 29 pairs with higher agreement
 - mean r (physicians) = 0.68
 - mean r (coders) = 0.78
- This set may be used in
 - system development (e.g., regression testing)
 - and to perform rough comparisons of relative performance of semantic relatedness measures

Available Reference Standards

UMN SRS

- result of a psycholinguistic study
- annotators: 8 medical residents (2 women, 6 men; mean age – 30 y.o.)
- continuous judgments using a touch screen
- 4 second time limit
- single word UMLS concepts from within three semantic types (drugs, diseases and symptoms) and across types
- total pairs: 724

Distribution of term pairs UMN SRS



Responses on relatedness task

587 pairs with ICC: 0.5



430 pairs with ICC of 0.73

Statistical Considerations

- Pearson's or Spearman's Correlation?
 - depends on the data
- Pearson assumptions
 - values are normally distributed
 - cases are independent
 - relationship between variables is linear
- Spearman assumptions:
 - values are monotonically distributed (important!)



UMN SRS characteristics

Frequency

Histogram of data\$Mean

Histogram of log(data\$Mean)





log(data\$Mean)





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Statistical Considerations

Direct Evaluation against Mayo or UMN SRS:

- Spearman rank correlation
- non-parametric methods
- consider non-linearity
- Indirect evaluations:
 - precision/recall
 - confidence intervals
 - testing for differences between means

An Example Application

 BioMEDICUS and SenseRelate for acronym disambiguation

> Biomedical Information Collection and Understanding System (BiomedICUS)

> > open source, UIMA-based

http://code.google.com/p/biomedicus/

SenseRelate

- broad-coverage word sense tagger
- uses semantic relatedenss
- http://www.d.umn.edu/~tpederse/senserelate.html



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