COMP90055 Computing Project Report

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Name of Degree Enrolled in: Master of Information Technology — Computing

Subject Code: COMP90055 Computing Project

Total credit points for entire project: 25

Type of Project: Research project

Semester in which project commenced: Semester 1, 2015

Semester in which project is expected to complete: Semester1, 2015

Project Title: Information Extraction of biomedical relationships in published colon cancer

literature

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Information Extraction of biomedical relationships in published colon cancer literature



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This dissertation is submitted for the degree of Master of Information Technology



Declaration

I certify that

- this thesis does not incorporate without acknowledgement any material previously submitted for a degree or diploma in any university; and that to the best of my knowledge and belief it does not contain any material previously published or written by another person where due reference is not made in the text.

- where necessary I have received clearance for this research from the University's Ethics Committee (Approval Number) and have submitted all required data to the Department

- the thesis is 8000 words in length (excluding text in images, table, bibliographies and appendices).

Ruichen Teng June 2015

Acknowledgements

And I would like to acknowledge ...

Abstract

This is where you write your abstract ...

Table of contents

Li	st of f	figures	xiii
Li	st of t	tables	XV
No	omen	clature	xvii
1	Intr	oduction	1
	1.1	Motivation	1
	1.2	Definitions and Assumptions	2
	1.3	Research Question	2
	1.4	Thesis Structure	3
2	Rela	ated Work	5
	2.1	Relation Extraction	5
	2.2	Named Entity Recognition	5
	2.3	Pattern Based Methods	6
	2.4	Co-occurrence based methods	6
	2.5	Rule-Based Methods	7
	2.6	Kernel Based Methods	7
	2.7	Feature Based Methods	7
	2.8	Semi-Supervised Methods	7
3	Cor	e	9
	3.1	Data Collection	9
		3.1.1 Background	9
	3.2	Algorithm	10
	3.3	System Adaptation	10
		3 3 1 BioNLP Shared Tasks	10

••	TD 11 C
XII	Table of contents
All	Table of contents

	3.4	3.3.2 Variome Annotation Schema	
4	Con	clusion	13
	4.1	Conclusion	13
Re	feren	ices	15

List of figures

List of tables

3.1	A nice looking table	10
3.2	Events	11
3.3	Relations under Variome Annotation Schema	11

Nomenclature

Roman Symbols

```
F complex function
```

F complex function

Greek Symbols

```
\gamma a simply closed curve on a complex plane
```

 γ a simply closed curve on a complex plane

```
\iota unit imaginary number \sqrt{-1}
```

 ι unit imaginary number $\sqrt{-1}$

```
\pi \simeq 3.14...
```

 $\pi \simeq 3.14...$

Superscripts

```
j superscript index
```

j superscript index

Subscripts

0 subscript index

0 subscript index

Other Symbols

 \oint_{γ} integration around a curve γ

xviii Nomenclature

 \oint_{γ} integration around a curve γ

Acronyms / Abbreviations

CIF Cauchy's Integral Formula

CIF Cauchy's Integral Formula

Introduction

1.1 Motivation

Text mining is the process of searching for patterns in natural language text using methods in computer science, linguistics, and statistics. Despite being unstructured and only human-understandable, text is still our primary media for exchange of information[13]. The prevalence of textual data presents a big challenge to computer-driven natural language understanding. *Information extraction*, in particular, refers to the task of acquiring organized, structured and queryable format of data from the unstructured corpus.

While text mining is widely used in areas like marketing and document verifying, it has received increased attention for its application to biomedical literatures[1, 5, 6]. This trend stems from the direct need of biomedical workers and researchers to cope with information explosion in their field. For instance, MEDLINE(Medical Literature Analysis andRetrieval System Online), the online database of United States National Library of Medicine, has accumulated nearly 0.8 million citations and 2.7 billion searches in 2014 alone[7], with total citations reaching 25 million. Within these publications there are valuable research results that should add to human knowledge. In the meantime, our primary knowledge base in life science - the biomedical databases, are still mostly being populated manually by *biocurators* - the "museum catalogers of the Internet age"[12]. They are professional scientists who read biomedical articles, record relevant data and organize them according to the biomedical database schema. The sheer volume of publications has made this process increasingly unrealistic[4].

Not only does data overload make knowledge discovery demanding, it also leads to a decline in literature quality. Nowadays biomedical workers and researchers are more prone to drawing wrong conclusions because they simply can not read all the relevant publications, among which oftentimes contradicting results are reported. Needless to say, we are

2 Introduction

in desperate need of automatic tools for systematically analyzing documents and extracting information. In fact, it has been argued that text mining is required to improve the coverage of databases[3].

1.2 Definitions and Assumptions

Below are a list of terms which I will use throughout this thesis, detailed examples will be given in the relevant sections, but a general definition is first given here to avoid any confusions that might arise when reading the following paragraphs.

• Relation Extraction: In general, relation extraction refers two the process of discovering a relationship between entities in text. In the domain of natural language processing, the relation can be semantic, syntactic, etc, with semantic relations being the most important for knowledge discovery. Relations can be unary, binary and complex with complex relations sometimes intermingled with the concept of events, In this project we are mainly concerned with binary relations as shown. Relation extraction is a form of information extraction where the semantic relations between entities are extracted. Specifically, in this project we focus on the relation extraction among other informations. Biomedical relations covers a wide range of knowledge in this field.

• Event Extraction: to be added

1.3 Research Question

This project looks to investigate the application of information extraction system Approximate Subgraph Matching (ASM)[?], on relation extraction tasks, specifically regarding the relation extraction on the variome corpus. In this project we focus on the relation extraction among other informations. Biomedical relations covers a wide range of knowledge in this field. As it turns out, this is not a trivial process, The relation extraction tool has a very promising applications for researchers and medical field workers, pharmaceutical companies, and the general public.

Relation extraction can be helpful in information search, knowledge discovery and hypothesis generation.

1.4 Thesis Structure 3

1.4 Thesis Structure

The thesis is organized as follows: chapter 1 will

therefore to allow researchers to identify needed information more efficiently, quote Based on the information in 1.1 and 1.2, the aim of this project is to develop tools that can assist the human bio-curation process.

Related Work

This chapter will explore the research related to this thesis.

2.1 Relation Extraction

In general, relation extraction refers two the process of discovering a relationship between entities in text. In the domain of natural language processing, the relation can be semantic, syntactic, etc, with semantic relations being the most important for knowledge discovery. Relations can be uniary, binary and complext with complex relations sometimes intermingled with the concept of events, In this project we are mainly concerned with binary relations as shown

2.2 Named Entity Recognition

Named Entity Recognition, or NER, is the task of identifying elements in text that belong to pre-defined categories like person, organization, etc. Specifically, NER in biomedical text mining aims at identifying thing like proteins, diseases, genes, etc. There is extra difficulty for NER in the biomedical domain mainly because of the following reasons.

quote: A survey of current work in biomedical text mining This task has been challenging for several reasons. First, there does not exist a complete dictionary for most types of biological named entities, so simple text- matching algorithms do not suffice. In addition, the same word or phrase can Recognising biological entities in text allows for further extraction of relationships and other information by identifying the key concepts of interest refer to a different thing depending upon context (eg ferritin can be a biological substance or a laboratory test). Conversely, many biological entities have several names (eg PTEN and MMAC1 refer the same gene). Biological entities may also have multi-word names (eg carotid artery), so the problem is additionally complicated by the need to determine name boundaries and resolve overlap of candidate names. Because of the potential utility and

6 Related Work

complexity of the problem, NER has attracted the interest of many researchers, and there is a tremendous amount of published research in this topic. With the large amount of genomic information being generated by biomedical researchers, it should not be surprising that in the genomics era, much of the work in biomedical NER has focused on recognising gene and protein names in free text. quote: A survey of current work in biomedical text mining

2.3 Pattern Based Methods

Relation extraction started from the task of extracting protein protein interactions from text by pattern matching. First, a set of part-of-speech rules are applied to split the sentence into simple sentences, e.g. (P1 VB1 P2 VB2 CC P3) is splitted to P1 VB1 P2 and P1 VB2 P3. Next, a set of word patterns is applied to extract relations from these sentences. In addition, citeDiscovering patterns to extract protein—protein interactions from the literature: Part II proposed an idea of minimal description length, as in finding a a pattern set that has the most balance between high presion, short rule length/lower rule complexity by dynamic programming to optimize the rule set. Pattern based methods has achieved quite respectable performances, but it has the limitations that it would have a tough time capturing the richness in expressions, such as the anaphora terms like pronouns

2.4 Co-occurrence based methods

Many approaches though extract information about genes from scientific texts using only information about the co-occurrence of terms in a sentence or abstract (Chaussabel and Sher, 2002; Becker et al., 2003; Tanabe et al., 1999; Stapley and Benoit, 2000; Jenssen et al., 2001; Wren et al., 2004). Simply ways of indicating co-occurrence measure include point-wise mutual information, log likelihood ratio. More advanced measures include the associative concept space, The main advantage of co-occurrence based methods is their simple implementation and low computational complexity.

citeCo-occurrence based meta-analysis of scientific texts: retrieving biological relationships between genes,

2.5 Rule-Based Methods

2.6 Kernel Based Methods

Kernels provide a similarity measure between two objects in some complex feature space. In contrast to feature based methods, kernel-based methods allow the original representation of the object to be retained and the kernel function will work out the similarity measure. For instance, a sentence maybe represented as a dependency graph, the feature based methods would want to select features such as number of nodes, edges, directions, etc, whereas kernel based methods allows feeding the two entire graph representations into a kernel function and output the distance measure.

2.7 Feature Based Methods

2.8 Semi-Supervised Methods

Core

3.1 Data Collection

Our dataset is the Variome Corpus[11], which is openly accessible. ¹ Verspoor et al. [11] gave a detailed illustration of the document selection and annotation process. I will summarize the main points here.

3.1.1 Background

A major part of the current biomedical research lies in understanding the relations between human genetic variation and disease phenotypes. The *Human Variome Project*, or *HVP*, is a global initiative to collect all genetic variation information affecting human health[9]. In particular, it acts as a liaison between individuals and organizations to integrate the genetic variants into databases that are open to the general public[11]. The *International Society for Gastrointestinal Hereditary Tumours (InSiGHT)*, is an international organization which aims to benefit patients with hereditary gastrointestinal(GI) tumours by research, education and personal assistance. In 2008, InSiGHT and HVP began a collaboration which propels InSiGHT to refine its process in the integration and interpretation of genetic variants. Consequently, a substantial effort was made to understand the mutation of mismatch repair(MMR) genes, the cause of Lynch Syndrome - one of the main syndromes of GI cancer[10]. A total of 10 full-text articles were selected from PubMed Central®by searching the common Lynch syndrome genes. These documents are mostly about inherited colon cancer. The annotation schema, also known as the Variome Annotation Schema[11], include 11 entity types and 13 relation types. as can be seen in the table here

In short, the corpus is Inspired by needs of inSIGHT database, but intended for broader applications. Documents relevant to the genetics of Lynch syndrome, which covers inherited colon cancer as well as certain other cancers. Selected with PubMed Central To train tools

¹http://www.opennicta.com.au/home/health/variome

10 Core

Dental measurement	Species I		Species II	
Dentai measurement	mean	SD	mean	SD
I1MD	6.23	0.91	5.2	0.7
I1LL	7.48	0.56	8.7	0.71
I2MD	3.99	0.63	4.22	0.54
I2LL	6.81	0.02	6.66	0.01
CMD	13.47	0.09	10.55	0.05
CBL	11.88	0.05	13.11	0.04

Table 3.1 A nice looking table

for mining genetic variation and its relationship to disease Here in this information extraction task, we treat the manually annotated data as the *gold data*,

3.2 Algorithm

To separate Named Entity Recognition from Relation Extraction problem, the named entity annotations are provided in training, development and test sets. Effectively, this is just asking computer to extract possible relations between these entities without worrying how to recognize them accurately. Of course, named entity recognition is an important step in a real relation extraction task as cite cite, because biological named entity recognition is such a complex problem and sometimes the relation extraction success would depend on the accuracy of NER. Named entity recognition is the process of Iis project cite the performance of the subgraph matching method, as an instance-based learning strategy (Alpaydin, 2004), is dependent on having good training examples that express the events in a range of syntactic structures, cite

3.3 System Adaptation

As mentioned in 2.7, the project aims at adapting an existing system used for event-retrieval tasks on relation extraction tasks.

3.3.1 BioNLP Shared Tasks

BioNLP shared Task series is a community-wide text mining challenge specifically for biomedical literatures. The GE task in shared task 2013 aims at retireving events of the following format:

3.4 Results

Table 3.2 Events

Dental measurement	Species I		Species II	
Dentai measurement	mean	SD	mean	SD
I1MD	6.23	0.91	5.2	0.7
I1LL	7.48	0.56	8.7	0.71
I2MD	3.99	0.63	4.22	0.54
I2LL	6.81	0.02	6.66	0.01
CMD	13.47	0.09	10.55	0.05
CBL	11.88	0.05	13.11	0.04

Table 3.3 Relations under Variome Annotation Schema

Dental measurement	Species I		Species II	
Dentai measurement	mean	SD	mean	SD
I1MD	6.23	0.91	5.2	0.7
I1LL	7.48	0.56	8.7	0.71
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I2LL	6.81	0.02	6.66	0.01
CMD	13.47	0.09	10.55	0.05
CBL	11.88	0.05	13.11	0.04

The a1 files does (to be added), the a2 files does (to be added),

3.3.2 Variome Annotation Schema

During the duration of this project, most of my attempts to fully adapt the system to relation extraction tasks have failed. In essence, the differentiation in the retrieval process lies in the event retrieval relies on the detection and prediction of a trigger word, where as the relation extraction does not.

3.4 Results

Conclusion

4.1 Conclusion

General literature mining at the semantic level.

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