informatics: Identifying and Tracking Informatics Sub-Discipline Terms in the Literature

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Summary
Objective: To identify the breadth of informatics sub-discipline terms used in the literature for enabling subsequent organization and searching by sub-discipline.

Methods: Titles in five literature sources were analyzed to extract terms for informatics sub-disciplines: 1) United States (U.S.) Library of Congress Online Catalog, 2) English Wikipedia, 3) U.S. National Library of Medicine (NLM) Catalog, 4) PubMed, and 5) PubMed Central. The extracted terms were combined and standardized with those in four vocabulary sources to create an integrated list: 1) Library of Congress Subject Headings (LCSH), 2) Medical Subject Headings (MeSH), 3) U.S. National Cancer Institute Thesaurus (NCIt), and 4) EMBRACE Data and Methods (EDAM). Searches for terms in titles from each literature source were conducted to obtain frequency counts and start years for characterizing established and potentially emerging sub-disciplines.

Results: Analysis of 6,949 titles from literature sources and 67 terms from vocabulary sources resulted in an integrated list of 382 terms for informatics sub-disciplines mapped to 292 preferred terms. In the last five decades, “bioinformatics”, “medical informatics”, “health informatics”, “nursing informatics”, and “biomedical informatics” were associated with the most literature. In the current decade, potentially emerging sub-disciplines include “disability informatics”, “neonatal informatics”, and “nanoinformatics” based on literature from the last five years.

Conclusions: As the field of informatics continues to expand and advance, keeping up-to-date with historical and current trends will become increasingly challenging. The ability to track the accomplishments and evolution of a particular sub-discipline in the literature could be valuable for supporting informatics research, education, and training.

1. Introduction

The origins of informatics and biomedical informatics date back to the 1950s [1–3] (Figure 1). Related terms in other languages existed in the 1950s (e.g., “informatik” in German [4]) and 1960s (e.g., “informatique” in French [1, 5]) now referring to the discipline of computer science, which is defined as “the branch of knowledge concerned with the construction, programming, operation, and use of computers” [6]. In the 1970s, the formal English term “informatics” emerged with a definition of “the discipline of science which investigates the structure and properties (not specific context) of scientific information, as well as the regularities of scientific information activity, its theory, history, methodology, and organization” [1, 3]. This definition has since been revised to “the branch of study that deals with the structure, properties, and communication of information and with means of storing or processing information” [7].

The terms “bioinformatics” and “medical informatics” also appeared in the 1970s [1, 8, 9] and their parallel development over the next two decades eventually led to broader acceptance of the term “biomedical informatics” in the 1990s [10–12]. This umbrella discipline with four major sub-disciplines (“bioinformatics”, “imaging informatics”, “clinical informatics”, and “public health informatics”) is defined as “the interdisciplinary field that studies and pursues the effective uses of biomedical data, information, and knowledge for scientific inquiry, problem solving, and decision making, driven by efforts to improve human health” [11–13]. As reflected in MEDLINE, myriad other sub-disciplines

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have appeared in the literature such as “nursing informatics” [14] and “dental informatics” [15] in the 1980s and “translational bioinformatics” [16] and “clinical research informatics” [17] in the 2000s.

In the context of biomedicine and health care, the continuous emergence of informatics sub-disciplines has led to the “adjective problem” or variability in words preceding the term “informatics” [18, 19]. For example, the terms “biomedical informatics” and “health informatics” could be considered components of the broader terms “biomedical and health informatics” and “health and biomedical informatics,” while “medical informatics” and “bio-informatics” may be viewed as narrower terms [18]. Efforts to define the fields of medical informatics, bioinformatics, and biomedical informatics have involved analyzing the scientific literature to characterize and visualize trends for journals, articles, authors, and research topics [20–35]. Many of these bibliometric studies have focused on citations in MEDLINE/PubMed that are either indexed with a particular MeSH descriptor (e.g., “medical informatics” [22] or “bioinformatics” [28]) or associated with a specific publication venue (e.g., Proceedings of the American Medical Informatics Association [AMIA] Annual Symposium [24] or Methods of Information in Medicine [36]). As the field of informatics as a whole grows, there will be an increasing need to organize both the terms and literature associated with its sub-disciplines to support specialized informatics research, education, and training.

2. Objectives

The goal of this study was to develop a semi-automated approach for identifying terms reflecting established and potentially emerging informatics sub-disciplines from existing literature and vocabulary sources. This work represents a first step towards creating a formal and standardized representation of informatics specializations that may be used to facilitate the search and retrieval of literature by sub-discipline for subsequent analysis.

3. Methods

The overall approach implemented in this study involved four major phases: 1) extracting titles from literature sources, 2) extracting terms for informatics sub-disciplines within these titles, 3) extracting relevant terms from vocabulary sources, and 4) creating an integrated list of informatics sub-discipline terms based on the literature and vocabulary sources (Figure 2).

3.1 Extracting Titles from Literature Sources

Titles were obtained from two general and three biomedical literature sources: 1) Library of Congress Online Catalog, 2) English Wikipedia, 3) National Library of
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The Library of Congress (LC) is the oldest federal cultural institution in the United States (U.S.) and largest library in the world with collections containing over 158 million books, periodicals, manuscripts, maps, music, recordings, images, and electronic resources [39]. The LC Online Catalog includes over 18 million records dating back to the 1890s describing these collections and Search/Retrieval via URL (SRU) Web services are available for performing automated searches that return the number of results as well as full records in the specified format (e.g., MAAchine-Readable Cataloging [MARC], Metadata Object Description Schema [MODS], or Dublin Core [DC]) [40–42]. The SRU Web services were used to obtain records in MODS XML format resulting from a search for the keyword “informatics” in titles in the LC Online Catalog. Each record was processed to extract: Library of Congress Control Number (LCCN), publication year (e.g., “1968-”, “[1983?”], or “c2005-c2007”), publication type (e.g., “text” or “software, multimedia”), title, and any alternate titles.

Wikipedia is a free, community-driven encyclopedia that was created in 2001 [43]. There are almost 300 different language editions of Wikipedia; English Wikipedia contains over 4.5 million articles [44]. Using the MediaWiki Web service Application Programming Interface (API), a search for “*informatics” in titles was performed, which returned a listing of matching titles [45]. For each title, the MediaWiki API was then used to obtain the oldest entry in the revision history from which the unique page identifier and creation year were extracted.

The U.S. National Library of Medicine (NLM) is the world’s largest biomedical library that provides electronic information services and resources to scientists, health professionals, and the public around the globe [46]. Among these resources are: 1) NLM Catalog containing bibliographic data for over 1.4 million journals, books, audiovisuals, computer software, electronic resources, and other materials [47, 48], 2) PubMed comprising over 24 million citations for biomedical literature including those from MEDLINE [49], and 3) PubMed Central providing access to over 3 million full-text articles [50]. For each of the three resources, a query for “informatics” in titles (i.e., “informatics”[ti]) was used to identify records of interest. Relevant records were obtained in XML format using the National Center for Biotechnology Information Entrez Programming Utilities, ESearch and EFetch, respectively [51]. Each retrieved record was then processed to extract: unique identifier (NLM identifier [NLM ID], PubMed identifier [PMID], or PubMed Central identi-
fier [PMCID]), publication year, and title. In addition, publication type (e.g., "Serial", "Book", or "Book Chapter") and alternate titles were extracted for NLM Catalog. To address overlap in literature sources, NLM Catalog records containing a LCCN and PubMed Central records containing a PMID were excluded.

3.2 Extracting Terms from Literature Titles

A semi-automated process was used to identify terms for informatics sub-disciplines in literature titles. First, a Ruby script was developed for the automated pre-processing of titles and extraction of terms from within these titles. A manual evaluation of a subset of titles and extracted terms was then conducted for assessing the performance of the automated algorithm. Finally, a manual review of all informatics sub-discipline terms was performed to resolve any issues.

Pre-processing of titles involved removing HTML tags and entities (e.g., "&quot; and "&amp;"); Unicode encodings (e.g., “u201c” for left quotation mark), and punctuation at the end or beginning of a title (e.g., “,” and “”). Other pre-processing tasks included removing punctuation at the end of words within a title, adding spaces around certain punctuation for use as “stop symbols” in the next step (e.g., “)” → “”), and lowercasing the entire title.

For each pre-processed title, the algorithm for extracting informatics sub-discipline terms involved: 1) identifying each occurrence of a word ending with “informatics” in the title (thus excluding words like “informatics-based” or “informatics-driven”), 2) looking at a window of two words preceding each occurrence, and 3) determining if this window contains a defined stop symbol (e.g., “,” “;” “,” “&”, and “”), pattern (e.g., numbers ending with “th” and words ending with “s”), or word from the list of 132 PubMed stopwords [52] or customized subset of the SPECIALIST Lexicon that includes commonly occurring English words and biomedical vocabulary [53, 54]. This customized subset of stopwords was generated from four tables in the SPECIALIST Lexicon 2014 Release: (1) LRAGR (Agreement and In-
and National Center for Biomedical Ontology (NCBO) BioPortal [65, 66], which include hundreds of source vocabularies or ontologies.

For LCSH, MeSH, and NCIt, available Web-based browsers were used to search for informatics sub-discipline terms (e.g., “informatics” or “informatic”). For EDAM, navigating the hierarchy in the available browser for terms was supplemented by searching the ontology in Web Ontology Language (OWL) format [61]. Where available, associated information such as unique identifier (e.g., MeSH ID or NCIt Code), year introduced, synonyms or variants, and UMLS semantic type were obtained. Combining the list of terms from the four vocabulary sources resulted in a seed list of preferred and variant terms.

### 3.4 Integrating Terms from Literature and Vocabulary Sources

The five lists of terms from the literature sources were combined with the seed list of terms from the four vocabulary sources to create a compiled list of informatics sub-discipline terms. For each entry in this list, a manual process was used to identify a “preferred term” for grouping variants and synonyms. These preferred terms were either those identified in the vocabulary sources or newly created. For example, the preferred term “cheminformatics” from the vocabulary sources had four variants in LCSH and EDAM (“chemical informatics”, “cheminformatics”, “chemoinformatics”, and “chemistry informatics”) and four additional variants in the literature sources (“chemi-informatics”, “chemo-informatics”, “chem(o)informatics”, and “chemocentric informatics”). For cases involving creation of preferred terms, initial guidelines included identifying a single word (not including the “.” character) as the preferred term where possible, which was a pattern observed in the vocabulary sources (e.g., “agroinformatics” vs. “agro-informatics” or “agricultural informatics”). As an example, the preferred term “geroinformatics” was created for “gero-informatics” and “gerontological informatics” that appeared in the literature titles.

Supplementary Appendix A includes the integrated list of informatics sub-discipline terms with mappings to preferred terms.

For each informatics sub-discipline, titles in the five literature sources (LC Online Catalog, English Wikipedia, NLM Catalog, PubMed, and PubMed Central) were searched for the preferred and any variant terms to obtain frequency counts and start years using the same Web services from the first phase for extracting literature titles. Analysis of the results included identifying the most frequent sub-disciplines and potential emergence of sub-disciplines by decade across literature sources.

### 4. Results

For each literature source, Table 2 summarizes the number of retrieved records, number of titles extracted from these records, and year of the earliest record (Table 2A); number of records in the sample used for evaluation and performance of the term extraction algorithm for this sample (Table 2B); and, the number of informatics sub-discipline terms before and after manual review (Table 2C). The five literature sources provided a total of 6,949 informatics-related terms from 5,349 records (as of April 18, 2014), with 1967 as the year associated with the earliest record from the NLM Catalog. Evaluation of the term extraction algorithm for a sample of records from each source found that on average, there were 93.1 ± 3.0% exact matches and 6.9 ± 3.0% partial matches. Manually resolving the partial matches reduced the total number of terms from 774 (620 unique) to 508 (371 unique) and involved an average of 29.1 ± 12.7% fixes.

A total of 67 informatics-related terms were identified across the four vocabulary sources (as of March 2014). The LCSH search returned 21 results (15 topic and 6 complex subject [excluded]); MeSH search returned 18 results (8 descriptors and 10 entry terms); NCIt search returned 18 results where only those with a UMLS semantic type of “Occupation or Discipline”...
or “Biomedical Occupation or Discipline” were included; and, EDAM provided 10 results. Combining the results resulted in a seed list of 44 unique terms mapped to 23 preferred terms for informatics sub-disciplines (Table 3). Of these, four were common to all vocabulary sources (“bioinformatics,” “clinical informatics,” “informatics,” and “medical informatics”) and 11 were unique to a source (e.g., “agroinformatics” in LCSH, “public health informatics” in MeSH, “resource informatics” in NCIt, and “toxicoinformatics” in EDAM).

The integrated list of informatics sub-discipline terms included a total of 382 terms mapped to 292 preferred terms (including “informatics”). Based on searching titles in the literature sources for each sub-discipline, 22 (7.5%) were found to be common across all sources and 141 (48.3%) were unique to a particular source (three terms originating from the vocabulary sources were not found in any of the literature sources). Figure 3 depicts the most frequent sub-disciplines as reflected by presence of preferred or variant terms in titles for each literature source (those with more than one title in English Wikipedia and top 25 for the other four sources as of August 6, 2014). Supplementary Appendix A contains the counts, start years, and queries for all sub-disciplines across the five literature sources. Table 4 highlights potentially emerging sub-disciplines by decade based on titles in the biomedical literature sources (NLM Catalog, PubMed, and PubMed Central) from the 1960s to 2010s (as of August 6, 2014).

### 5. Discussion

In this study, a semi-automated approach was used for creating an integrated list of informatics sub-discipline terms from titles in literature sources and terms in vocabulary sources. The results highlight the complementary nature of these sources and distribution of terms across the literature sources. Over the last five decades, there has been a continuous growth in the number of sub-discipline terms that is reflective of the expansion and advancement of the field of informatics.

### 5.1 Limitations

The main title and alternate titles from five literature sources were used to identify terms and generate basic statistics (frequency counts and start years) using available Web services. While LC Online Catalog, NLM Catalog, PubMed, and PubMed Central include English and non-English records [39, 48, 77, 78], term identification was limited to native English or translated titles. Potential next steps could involve expanding the techniques to support other languages and literature sources such as non-English language editions of Wikipedia (e.g., German and French Wikipedia) and WorldCat, a global catalog of library collections containing over 332 million records for more than 2.2 billion items representing over 485 languages [79]. Future plans also may in-
The automated identification of informatics sub-discipline terms in literature titles was restricted to terms with a maximum length of three where the last term could either be “informatics” (e.g., “clinical research informatics”) or end with “informatics” (e.g., “translational bioinformatics”). Manual review of the extracted terms revealed several limitations to be addressed in future versions of the algorithm. First, there were cases of under-specified terms due to the length restriction such as “care testing informatics” that was manually expanded to “point of care testing informatics” and “consumer health informatics” within “pediatric consumer health informatics”. Second, a number of over-specified terms were extracted such as “internet-based informatics”, “undergraduate medical informatics”, and “international nursing informatics” where the first word in each of these was manually removed. Lastly, several titles were found to include patterns like “informatics [in|for] x” (e.g., “informatics in pathology” and “informatics for consumer health”), “x and informatics” (e.g., “neurology and informatics”), and “x and y informatics” (e.g., “archives and museum informatics” and “gastroenterology and medical informatics”) that could be transformed (e.g., “informatics in pathology” → “pathology informatics”) or considered variant terms.

5.2 Future Directions

The process developed and used in this study for integrating terms from the various sources highlights the need for a standardized representation of informatics specializations that formally defines preferred terms, variant terms, descriptions, and relationships. The integrated list generated in this study resulted in an initial flat list of preferred and variant terms. Subsequent studies may start with this list and be
more focused on refining and organizing these terms into a “taxonomy of informatics sub-disciplines” that could potentially inform or complement MeSH, LCSH, and other vocabulary sources. For example, how might “cancer informatics”, “cancer cell informatics”, “cancer imaging informatics”, and “bladder cancer informatics” be organized with respect to each other? At a broader level, what is the relationship between “informatics”, “applied informatics”, and “theoretical informatics” or “biomedical informatics”, “foundational biomedical informatics”, and “integrative biomedical informatics”?

There have been some efforts to define hierarchical relationships between informatics sub-disciplines related to biomedical and health care [11, 18], which could serve as the foundation for formalizing and organizing specializations in informatics. Other general and domain-specific sources could also be leveraged for guiding organization such as the Classification of Instructional Programs, originally developed by the U.S. Department of Education, that provides a taxonomic scheme for fields of study [81] and the Subject Matter Domain axis of the HL7/LOINC Clinical Document Ontology that provides a hierarchical organization of clinical specialties and sub-specialties [82, 83]. In addition to defining relationships, next steps would also require the identification or formulation of descriptions for each informatics sub-discipline that may exist in published literature (e.g., for medical informatics [84, 85], bioinformatics [86], and biomedical informatics [11, 12, 87]) or other knowledge sources (e.g., LCSH that includes definitions for some informatics terms and their sources such as MeSH).

A potentially valuable use of a taxonomy of informatics sub-disciplines is to inform strategies for searching and retrieving literature. For PubMed, informatics-specific queries could be developed, analogous to PubMed Special Queries [88] such as PubMed Clinical Queries [89] or the PubMed Cancer Subset [90], for identifying citations associated with a particular specialization (e.g., for neurology, an initial search would include the terms “neuroinformatics”, “neuro-informatics”, and “neurologics”). Records in the resulting subsets could be further indexed by topics using terms from relevant ontologies such as the Health Informatics Ontology (e.g., concepts related to networking and evaluation) [34], SNOMED CT (e.g., concepts for clinical findings) [91], HGNIC Database (e.g., concepts for human genes) [92], and ACM Computing Classification System (e.g., concepts related to information systems and applied computing) [93]. Analysis of these terms could reveal combinations that uniquely represent a particular specialization and be used to enhance searches to retrieve additional records.

### Table 4  Emergence of informatics sub-discipline terms in biomedical literature by decade (total terms and top 5)

<table>
<thead>
<tr>
<th>Decade</th>
<th>NLM Catalog</th>
<th>PubMed</th>
<th>PubMed Central</th>
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<tbody>
<tr>
<td>1960s</td>
<td>[Total: 1]</td>
<td>[Total: 0]</td>
<td>[Total: 0]</td>
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<td></td>
<td>medical informatics</td>
<td>N/A</td>
<td>N/A</td>
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<tr>
<td>1970s</td>
<td>[Total: 2]</td>
<td>[Total: 1]</td>
<td>[Total: 1]</td>
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<td></td>
<td>health informatics</td>
<td>medical informatics</td>
<td>clinical eeg informatics</td>
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<td>clinical eeg informatics</td>
<td>clinical eeg informatics</td>
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<tr>
<td>1980s</td>
<td>[Total: 6]</td>
<td>[Total: 8]</td>
<td>[Total: 2]</td>
</tr>
<tr>
<td></td>
<td>bioinformatics</td>
<td>health informatics</td>
<td>medical informatics</td>
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<td>nursing informatics</td>
<td>nursing informatics</td>
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<td>neuroinformatics</td>
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<td>protein bioinformatics</td>
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<td>archival informatics</td>
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<td>museum informatics</td>
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<tr>
<td>1990s</td>
<td>[Total: 16]</td>
<td>[Total: 48]</td>
<td>[Total: 20]</td>
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<td></td>
<td>biomedical informatics</td>
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<td>consumer health informatics</td>
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<td>2000s</td>
<td>[Total: 39]</td>
<td>[Total: 103]</td>
<td>[Total: 35]</td>
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<td>imaging informatics</td>
<td>structural bioinformatics</td>
<td>cheminformatics</td>
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<td>cheminformatics</td>
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<td>pathology informatics</td>
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<td>neuroinformatics</td>
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<td>personal health informatics</td>
<td>neonatal informatics</td>
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<td></td>
<td></td>
<td>translational biomedical informatics</td>
<td>neonatal informatics</td>
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*As of August 6, 2014
Topic, author, and other types of bibliometric analyses of these specialized subsets could then be performed to identify research trends and experts for defining the various informatics sub-disciplines. Such analyses could also be valuable for pinpointing areas with limited activity and potential opportunity for informatics innovations (e.g., in clinical sub-specialties like hematology or rheumatology).

5.3 Broader Implications

The findings of this study provide a broad perspective of the field of informatics and represent a first step towards characterizing its myriad specializations. Aside from bio-medicine and health care, informatics advances are being made in other domains (e.g., "socioinformatics", "geoinformatics", "communication informatics", "biodiversity informatics", "ecoinformatics", and "museum informatics") where there may be opportunities for trans-disciplinary synergies in methodological developments or applications. Internationally, there have been significant efforts to support and promote informatics education in biomedicine and health care by organizations such as AMIA [12, 94] and the International Medical Informatics Association (IMIA) [19, 95]. The contributions of this work support such efforts by: 1) highlighting the breadth of established and potentially emerging areas of informatics specialization and 2) enabling in-depth exploration and identification of competencies and needs associated with a particular specialization.

6. Conclusions

As the field of informatics continues to expand and advance, keeping up-to-date with historical and current trends will become increasingly challenging. The results of this study highlight the potential for identifying a comprehensive list of informatics sub-discipline terms and thus represent an essential step towards enabling the search and retrieval of literature by sub-discipline. The ability to track the accomplishments and evolution of a particular sub-discipline as reflected in the literature could be valuable for supporting informatics research, education, and training.

References


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