SYMPOSIUM

The Unified Medical Language System™: current research and development

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CONTENTS

The UMLS® project: making the conceptual connection between users and the information they need

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Conceptual connections between users and information sources depend on an accurate representation of the content of available information sources, an accurate representation of specific user information needs, and the ability to match the two. Establishing such connections is a principal function of medical librarians. The goal of the National Library of Medicine's Unified Medical Language System℠ (UMLS®) project is to facilitate the development of conceptual connections between users and relevant machine-readable information. The UMLS model involves a combination of three centrally developed Knowledge Sources (a Metathesaurus, a Semantic Network, and an Information Sources Map) and a variety of smart interface programs that make use of these Knowledge Sources to help users in different environments find machine-readable information relevant to their particular practice or research problems. The third experimental edition of the UMLS Knowledge Sources was issued in the fall of 1992. Current priorities for the UMLS project include developing applications that make use of the Knowledge Sources and using feedback from these applications to guide ongoing enhancement and expansion of the Knowledge Sources. Medical librarians are involved heavily in the direction of the UMLS project, in the development of the Knowledge Sources, and in their experimental application. The involvement of librarians in reviewing, testing, and providing feedback on UMLS products will increase the likelihood that the UMLS project will achieve its goal of improving access to machine-readable biomedical information.

INTRODUCTION

Access to information pertinent to clinical practice and health-related research depends on effective organizational, technical, and conceptual connections between users and information sources. In one sense, the goal of all health sciences librarians is to develop and strengthen these connections. By establishing the National Network of Libraries of Medicine (NN/LM), the National Library of Medicine (NLM) and other U.S. health sciences libraries have collaborated to build an organizational infrastructure that supports rapid delivery of biomedical information throughout the nation [1]. By working toward the implementation of Integrated Advanced Information Management Systems (IAIMS)*, health sciences librarians are removing organizational barriers that hamper information transfer within individual institutions [2–3]. Both inter- and intraorganizational arrangements benefit from, and also foster, the use of technology to speed access to biomedical information.

DOCLINE® [4], its supporting database SERHOLD® [5], and their regional precursors [6] are prime examples of the success that can be achieved by overlaying improved technical connections on an already effective organizational infrastructure. Conversely, projects to improve technical connections to infor-

* Formerly known as "Integrated Academic Information Management System."

mation within specific institutions, such as mounting a local version of MEDLINE® or setting up a CD-ROM network, can encourage collaboration between different parts of an organization and can lead to organizational ties that support effective information transfer. The rapidly expanding Internet and the effort to transform it into a full-fledged National Research and Education Network (NREN) are current examples of technical connections between users and information sources that also have the potential to promote organizational collaboration [7].

Although NLM and other health sciences libraries have been successful in developing new organizational and technical connections that enhance access to information, perhaps their most critical function is to establish conceptual connections between user questions and information relevant to these questions. Conceptual connections depend on an accurate representation of the content of existing information sources, an accurate interpretation of specific user information needs, and the ability to match the two. Organizational and technical connections can speed the delivery of information to users; a good conceptual connection ensures that the information delivered is relevant and useful.

For more than one hundred years, U.S. health sciences librarians have made use of NLM products to construct the conceptual connection between users and biomedical information found in the published literature. NLM is one of the primary producers of consistent and useful conceptual representations of available biomedical information sources. *Index Medicus*, the NLM catalogs, the Medical Subject Headings (MeSH®), and the MEDLARS® family of databases are all products of NLM's effort to provide usable abstractions of the content of published documents. For the past eighteen years, NLM has also collaborated with other federal agencies to create and to provide access to structured factual databases in the fields of toxicology and environmental health, cancer [8], and acquired immunodeficiency syndrome (AIDS) [9].

Of course, librarians in other health-related institutions also work on such conceptual representations by developing local catalogs (often based on NLM data) and by creating specialized indexes of local or national significance. Perhaps the majority of individual medical librarians focus on the user end of the conceptual connection, however—on translating user questions into a form more likely to elicit a useful response from available information sources, on selecting from among available sources those likely to have relevant information, and on culling the most useful items from the potentially relevant material retrieved. Reference interviews; mediated searches of print or online sources; user training in the use of MeSH, *Index Medicus*, MEDLINE in any of its manifestations, or the local online catalog—all of these activities help to translate user questions into a form more likely to receive useful answers from available information sources.

THE CURRENT INFORMATION ENVIRONMENT

A number of factors are having profound effects on the delivery of biomedical information and, therefore, on the operations of health sciences libraries. These include low-cost, powerful computers; high-capacity local, regional, national, and international telecommunications networks; high-quality public biomedical databases; extensive local database-building efforts; and more and better information processing and retrieval software. As hardware and software capacities increase, the potential for using computers to improve the conceptual connection between users and the information they need also increases. The inevitable result is to blur the distinctions between representing or establishing the content of information sources and identifying the specific information that users actually need.

As technology made it possible, database producers and providers, including NLM, have built systems that are able to guide users in constructing queries that are likely to retrieve relevant information. Since the advent of PaperChase at Beth Israel Hospital in the 1970s [10], there has been a steady growth in the number of user-friendly front-ends to MEDLINE; GRATEFUL MED® [11], miniMEDLINE [12], and a range of CD-ROM products [13] are some examples. While these systems fall far short of the capabilities of expert reference librarians, they are improving steadily. Because end-user searching allows access to information at times and in places where obtaining assistance from librarians is impossible or prohibitively inconvenient, it is safe to assume that an increasing number of individuals will use these systems and that the products will continue to improve.

At the same time that traditional database producers and providers have been working on interfaces for direct use by those without special search training, health science librarians themselves have become increasingly involved in database production and distribution. Integrated access to online catalogs and MEDLINE [14], establishment of institutional CD-ROM networks, and development of specialized databases are just some of the evidences of this parallel development, which has been encouraged by the IAIMS concept and by the vision of the "library without walls." In their institutional environments, health science librarians are also engaged in developing or selecting interfaces that can be used directly by end users. The current emphasis on establishing powerful computer-based patient record systems [15] and the

recognition that these systems should be linked to "outside" information sources, such as MEDLINE, mean that the medical librarian's expertise is becoming increasingly relevant to local efforts to develop clinical information systems.

THE UMLS AND THE CONCEPTUAL CONNECTION

NLM's Unified Medical Language System℠ (UMLS®) project is an effort to exploit current and emerging information technologies to aid the establishment of effective conceptual connections between user inquiries and relevant machine-readable biomedical information [16]. While the ultimate beneficiaries of the UMLS effort are health professionals and biomedical researchers, the initial UMLS products are designed for system developers. The UMLS development strategy assumes that information relevant to particular questions will continue to be distributed across many disparate databases, including bibliographic databases, local patient record systems, factual databases, knowledge bases, and expert systems. This is also a key IAIMS premise. A second UMLS assumption is that there will continue to be great variety in the terms used by different databases and by different users to express the same or similar biomedical concepts. In many cases, these differences in terminology reflect underlying differences in purpose and perspective. MeSH, for example, was developed to reflect the concepts appearing in the medical literature. The International Classification of Diseases was developed to promote consistent worldwide reporting of the causes of death and the incidence of disease. The unsurprising result is that the two are very different—and that neither is well suited to describing the patient signs and symptoms or the routine procedures that are critical components of clinical care.

As librarians know too well, the distribution of relevant information among many different sources and the variety of vocabularies and classifications used in these sources work against the establishment of useful conceptual connections between users and machine-readable information. These barriers prevent end users from retrieving and integrating information from multiple sources. They also frustrate the efforts of librarians and system developers to help these users. Meanwhile, the demand for "transparent" integrated access to information increases. As individuals become more frequent users of automated systems in their daily work, their interest in using information from these systems as the basis for searches in other automated information sources also increases. In the health care arena, the obvious example of this kind of searching is the use of data in a computer-based patient record to retrieve related information from a bibliographic database, an expert system, and the like.

The UMLS hypothesis is that a combination of new machine-readable knowledge sources and smart interface programs will enable better conceptual connections between user questions and relevant machine-readable information. Three Knowledge Sources have been identified as essential: a Metathesaurus of concept names derived from a variety of existing biomedical vocabularies and classifications [17–18], a Semantic Network representing sensible relationships among the broad categories or "semantic types" to which all Metathesaurus concepts have been assigned [19–20], and an Information Sources Map that describes the content of biomedical databases in terms of MeSH descriptors, semantic types, and semantic type relationships and provides other aids to automatic selection of and connection to relevant databases [21].

The expectation is that interface programs that use these UMLS Knowledge Sources will be able to emulate some of the functions of an expert reference librarian, while accessing a broader range of information sources. The scenario envisioned is as follows: a user generates a question, either by marking concepts of interest in one automated source, such as a patient record, by responding to a system alert about the availability of information that is potentially relevant to the problem at hand, or by directly entering a search inquiry. An interface program will use information in the Metathesaurus and the Semantic Network to interpret the user's question at some useful level; that is, to identify its key concepts and the relationships between them. The interface will display selected information from the Knowledge Sources to assist the user in clarifying or refocusing the question as necessary. Having identified a set of concepts, semantic types, and relationships relevant to the user query, the interface program will match these with the descriptions of available databases in the Information Sources Map, select potentially relevant databases, convert the query into terms likely to be recognized by these databases, run the searches, and then merge and rank the output for display to the user. This is the general target toward which the UMLS project is aiming. Significant research and development is still needed to achieve systems that can perform all of these functions.

As this scenario illustrates, both the structured Knowledge Sources and the smart interface programs are essential to the UMLS model. By themselves, the Knowledge Sources will have utility as reference tools for librarians, but they are unlikely to be of direct interest to the majority of end users. An effective interface program will capture and perhaps constructively constrain the user's question and limit the user's exposure to the UMLS Knowledge Sources to the spe-

cific information needed to clarify an ambiguous question, to suggest related topics of potential interest, or to focus or rank retrieval. On the other hand, use of the UMLS Knowledge Sources should increase the ability of an interface program to suggest potentially useful searches to the user, to interpret user questions correctly, to select appropriate information sources, and to evaluate and format retrieval.

UMLS DEVELOPMENT HISTORY

From its inception, the UMLS project has been directed by a team of NLM staff, supported by research and development contracts with medical informatics groups at a number of universities and one private company† and advised by a range of potential and then actual users of UMLS products. Participants represent a broad range of relevant expertise, including medical librarianship, medical informatics, computer science, and linguistics, as well as medicine, basic biomedical science, and other health-related disciplines. Experience to date indicates that multidisciplinary teams are essential both to the development of the UMLS Knowledge Sources and to their successful application.

In the initial phase of the project (1986–1988), the emphasis was on assessing needs, defining the components likely to be needed for a fully functioning UMLS, exploring several potential strategies for building these components, and producing tools for the research effort. From 1989 to 1991, the priority shifted to rapid development and distribution of the initial versions of the three UMLS Knowledge Sources, although work on studying user information needs and on developing prototypes of UMLS functionality also continued. The plan was to produce Knowledge Sources that represented useful steps toward the full capabilities ultimately desired and to get them into the hands of a wide range of system developers as soon as possible [22]. Accordingly, NLM issued the first experimental edition of the UMLS Knowledge Sources in the fall of 1990 and has released updated editions in 1991 and 1992.

The experimental editions of the UMLS Knowledge Sources are available free of charge under the terms of an experimental agreement which requires recipients to provide feedback to NLM. This initial broad distribution has the dual effect of generating feedback on needed changes and enhancements to the Knowledge Sources based on experiments applying them to real information problems [23], and promoting the development of a variety of prototype interface programs that make use of the Knowledge Sources. The Knowledge Sources are distributed in multiple machine-readable formats to facilitate experimentation in different hardware and software environments and structured to allow the addition of locally significant information. While software developers will probably always have to add some information to support their specific applications, such as Information Sources Map records for local patient record databases, the UMLS Knowledge Sources should eventually encompass most of the information about health-related terminology, concepts, and databases needed to facilitate retrieving and integrating of biomedical information from multiple machine-readable sources. In the future, the UMLS Knowledge Sources are likely to be accompanied by a set of utility programs that will help software developers to select and manipulate UMLS data. Eventually, there may also be a suite of re-usable program modules, such as term look-up routines and source selection algorithms, that use the UMLS Knowledge Sources to perform specific operations. At present, however, the UMLS Knowledge Sources are issued without accompanying programs, except for Macintosh-based browsers for the Metathesaurus [24] and the Semantic Network. These browsers are also intended for use by system developers—to enable them to gain an understanding of these Knowledge Sources before they begin extracting information for use in their applications.

In 1992, the UMLS project entered its third and current phase. The primary objective for the 1992–1994 period is to develop a range of useful applications that rely on the information available in the UMLS Knowledge Sources. Other important objectives are to build robust production systems for creating and updating the UMLS Knowledge Sources, to issue annual editions of the Knowledge Sources with modifications and enhancements that reflect feedback from those developing UMLS applications, to develop policies and procedures for routine distribution of the Knowledge Sources as they emerge from the experimental phase into regular production mode, and to define and develop UMLS utility programs, again based on feedback from those attempting to apply the UMLS Knowledge Sources.

CURRENT UMLS APPLICATIONS RESEARCH

NLM is pursuing several different strategies for ensuring the development of useful UMLS applications; that is, the smart programs needed to take advantage of the UMLS Knowledge Sources. These include in-house research and development, work under con-

† The current UMLS contractors are Brigham and Women's Hospital (PI: Robert Greenes, M.D., Ph.D); Columbia University (James Cimino, M.D.); Lexical Technology (PI: Mark Tuttle); Massachusetts General Hospital (PI: G. Octo Barnett, M.D.); University of Pittsburgh (PI: Randolph Miller, M.D.), with subcontractor University of Utah (PI: Homer Warner, M.D., Ph.D.); and Yale School of Medicine (PI: Perry Miller, M.D., Ph.D.).

tract and purchase order, grant support for UMLS-related projects, as well as the free distribution of the early versions of the UMLS Knowledge Sources to anyone interested in applying them to biomedical information problems. NLM's own UMLS applications research is focused in three areas, two of which are described in articles in this symposium. Kingsland et al. provide a look at the current capabilities of the Coach℠ search assistant designed to assist GRATEFUL MED users [25]. McCray and her coauthors report on experiments in applying the UMLS Knowledge Sources to natural language processing [26]. Humphrey is also experimenting with the use of the Metathesaurus and Semantic Network as part of the Medindex expert indexing system project [27].

NLM's UMLS contractors are working on a variety of promising applications. Two representative projects at different stages of development are described in this symposium. Cimino et al. discuss the initial phase of a multiyear project at Columbia University to test the usefulness of "generic queries" and of the UMLS Knowledge Sources in retrieving external information pertinent to diagnoses and procedures recorded in a clinical information system [28]. Clyman et al. report on the development of a prototype Information Sources Map application currently being implemented at Yale School of Medicine and Yale New Haven Hospital [29]. Both of these efforts are integrated with IAIMS projects. Other interesting applications projects directed by UMLS contractors include the Interactive Query Workstation (IQW) project at Massachusetts General Hospital [30], the CHARTLINE project at the University of Pittsburgh [31], and the Physician's Information Assistant (PIA) project at Lexical Technology [32].

Recently, NLM awarded five small purchase orders to support the incorporation of the UMLS Knowledge Sources into existing applications. The recipients of the awards for these eighteen-month projects are Chicago Medical School/University of the Health Sciences (David Trace, M.D., and Frank Naeymi-Rad, Ph.D.), for the application of the UMLS Knowledge Sources in a medical informatics training program; Georgetown University (Naomi Broering, M.L.S.), to explore the use of the Metathesaurus in indexing images; State University of New York at Buffalo (John Eisner, D.D.S.), for a curriculum analysis project involving a number of U.S. dental schools; the University of Washington (Sherrilynne Fuller, Ph.D.), to provide access to the UMLS Knowledge Sources via the WILLOW database interface [33]; and the American Lake VA Medical Center (Kenric Hammond, M.D.), to link the VA clinical information system to MEDLINE.

NLM's grant program is another potential source of funding for UMLS-related applications. In 1991, the University of Maryland at Baltimore (Gary Freiburger, M.L.S.) received an NLM Information Systems Grant for a project to incorporate the UMLS Knowledge Sources into a campuswide interface to a variety of bibliographic and full-text databases. Funding for more basic information science research using the UMLS Knowledge Sources is available under the National Science Foundation's Program in Database Activities in Biological, Behavioral, and Social Sciences.

As of September 1992, nearly three hundred individuals and institutions had signed the experimental agreement for use of the UMLS Knowledge Sources and received copies of the complete databases on CD-ROM. Feedback from many of these users indicates that they are applying the UMLS Knowledge Sources to a wide range of information problems, including automated indexing, natural language processing, indexing patient data, and linking clinical and bibliographic information. There are published reports on a number of these projects [34–36].

CURRENT STATUS OF THE UMLS KNOWLEDGE SOURCES

In contrast to the distributed, pluralistic approach being used to develop useful UMLS applications, the UMLS Knowledge Sources are being developed centrally. NLM is directing the creation, maintenance, and distribution of the Knowledge Sources with significant assistance from the UMLS contractors. Lexical Technology has been instrumental in designing and producing the UMLS Metathesaurus®, including preprocessing its source vocabularies, building the connections between different names for the same concepts, and computing the occurrence and co-occurrence of concepts in MEDLINE and other information sources [37]. Current and former UMLS collaborators at Brigham and Women's Hospital, Massachusetts General Hospital, Columbia University, the University of Pittsburgh, the University of Utah, Yale School of Medicine, and Carnegie-Mellon University have contributed to defining the structure and attributes of the UMLS Knowledge Sources and to developing their content.

In the fall of 1992, the third experimental edition of the UMLS Knowledge Sources was released. This edition includes the third versions of the Metathesaurus and the Semantic Network and the second version of the Information Sources Map. It incorporates a number of additions and changes recommended by the UMLS contractors and others who experimented with the previous editions. Significant improvements to the Metathesaurus include a revised and simplified relational structure, the addition of nearly 3,000 definitions from *Dorland's Illustrated Medical Dictionary, 27th edition* [38]; the incorporation of the French translation of MeSH main headings as

prepared by the Institut National de la Sante et Recherche Medicale, Paris [39]; the inclusion of concepts and terms from a number of additional controlled vocabularies; the expansion of coverage of terms and concepts from some of the vocabulary sources already represented in previous editions of the Metathesaurus; and the addition of information about the co-occurrence of diseases and findings in the AI/RHEUM knowledge base [40]. UMLS users are responsible for complying with any restrictions on use of copyrighted information, such as definitions from *Dorland's* and the French translation of MeSH, incorporated in the Metathesaurus. Schuyler et al. discuss the organization and the construction of the Metathesaurus in some detail in this symposium [41].

The 1992 version of the UMLS Semantic Network defines a total of 134 semantic types and 47 relationships that can be used to connect them. For 1992, the hierarchical connections between semantic types have been revised and many additional, nonhierarchical links between specific semantic types have been added. In the Semantic Network, relationships between semantic types are established at the highest possible level and inherited by descendant types. For example, the relationship "treats" is established between the semantic type "therapeutic or preventive procedure" and the semantic type "pathologic function." The semantic types that are children of "pathologic function," such as "disease or syndrome," inherit this relationship with "therapeutic or preventative procedure." In the 1992 edition of the Semantic Network, all the relationships between types are stated explicitly, including the inherited relationships. This new feature should reduce the amount of preprocessing required to use the UMLS Semantic Network for some applications. In this symposium, McCray et al. [42] and Cimino et al. [43] discuss uses of the Semantic Network.

The 1992 Information Sources Map has been expanded to include records for additional information sources in the fields of toxicology, the environment, and biology. A new "use" data element has been added, based on feedback from experimentation at Yale School of Medicine, some of which is described by Clyman et al. [44]. This data element identifies the likelihood that a particular database will be useful for an array of purposes, such as clinical care, library and information service, basic research, or emergency response and provides a useful adjunct to the subject scope information already present in the Information Sources Map. Some existing Information Sources Map data elements have been restructured for 1992 to facilitate quality control and maintenance.

Like the 1991 version, the 1992 Information Sources Map contains data elements useful in source selection, but does not yet include data useful in making automated connections to databases and in conducting actual searches of them. The addition of such data elements is a priority for 1993 to 1994. The initial step, most likely, will support automated connections to information sources accessible via the Internet. Mechanisms are also being developed to support efficient construction, submission, and update of Information Sources Map records by a number of database producers and providers. To ensure consistency, NLM will continue to be responsible for indexing the subject content of all information sources represented in the Map.

THE UMLS AND MEDICAL LIBRARIANS

Librarians at NLM have been instrumental in the planning and oversight of the UMLS project, in the design, construction, and quality control of the three UMLS Knowledge Sources, and in NLM's internal applications research involving the Knowledge Sources. Medical librarians are involved actively in the research and development being conducted by several UMLS contractors and, as previously indicated, are directing some of the UMLS work funded by purchase orders and grants.

The heavy involvement of medical librarians in the UMLS effort is a logical consequence of its primary goal—to enhance the conceptual connections between users and information sources. While the UMLS project addresses a wider range of information, including patient data, than has traditionally been the province of the medical librarian, the librarian's expertise is directly relevant to most of the problems the UMLS must address—including thesaurus construction; analysis, description, and selection of information sources; and interpretation of user needs.

Like the IAIMS initiative, the UMLS project offers an avenue for health sciences librarians to become more involved in the broad information issues facing their institutions. Possible roles for librarians range from bringing the UMLS project and its implications to the attention of those working on the development of local clinical or educational systems, to assessing the local usefulness of publicly available software products that make use of the UMLS Knowledge Sources as such products begin to emerge, to directing projects that test the utility of the UMLS Knowledge Sources in the library's current system building activities, as is already happening in several IAIMS sites.

If the UMLS project achieves its potential, medical librarians and their users will have powerful new tools for making conceptual connections between information needs and available machine-readable information. The more medical librarians are involved in reviewing, testing, and providing feedback on UMLS products, the greater the likelihood that this desirable outcome will become a reality.

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