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Using UMLS metathesaurus concepts to describe medical images: dermatology vocabulary

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Abstract

Web servers at the National Library of Medicine (NLM) displayed images of ten skin lesions to practicing dermatologists and provided an online form for capturing text they used to describe the pictures. The terms were submitted to the UMLS Metathesaurus (Meta). Concepts retrieved, their semantic types, definitions and synonyms, were returned to each subject in a second web-based form. Subjects rated the concepts against their own descriptive terms.

They submitted 825 terms, 346 of which were unique and 300 mapped to UMLS concepts. The dermatologists rated 295 concepts as ‘Exact Match’ and they accomplished both tasks in about 30 min.

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1. Introduction

Faster connections, compression algorithms, and more sophisticated web software has made multimedia objects (still images, sound recordings, motion videos) practical to store and retrieve via the Internet.

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Users need better methods to selectively retrieve multimedia objects. Image pattern recognition is still in its infancy and controlled indexing vocabularies are usually limited to narrow domains. The National Library of Medicine (NLM) has developed the Unified Medical Language System (UMLS) Metathesaurus (Meta) which is being continuously enlarged and refined. It affords a new approach to semantic indexing of medical images based on robust mapping of approximately 100 controlled biomedical vocabularies.

The version (2002) of Meta used in this study provided nearly 800,000 concepts and 1.92 million names (strings) from 110 maintained controlled vocabularies in biomedicine and a web-based query tool, the Knowledge Source Server. The goals of this study were to determine: (1) the extent to which terms used by domain experts to describe medical images can be mapped to concepts contained in Meta; (2) whether such concepts are acceptable to the experts as descriptors of the images; and (3) to explore use of the internet to conduct distributed research when the subjects are remotely located and not highly sophisticated in the use of computers or the internet.

2. Background

Several investigators have addressed the issue of using controlled vocabularies to index and retrieve medical images. Macura and Macura [1] built a 'Radiologic Pictionary' using a picture-based controlled vocabulary to permit query of a radiology image database.

Duvaufferier et al. [2] devised a system for semi-automatic indexing of radiology images with concepts from ADM, a diagnostic knowledge base. Lowe et al. [3] used their SAPHIRE software linked to the UMLS for automated indexing of the text of radiology imaging reports as a proxy for indexing the associated images. They compared this automated indexing to manual entry of terms from the image reports by individuals with in-depth knowledge of the UMLS. They found that human indexers identified appropriate Metathesaurus (Meta) concepts for 81% of the terms they had previously identified in 50 radiology imaging reports. The automated procedure (SAPHIRE) identified UMLS concepts for only 64% of the test terms. They used the 1998 version of the Metathesaurus in their study; it contains only 61% of the concepts available in the version (2002) used in the present study. Tagare et al. [4] discussed the characteristics of medical image databases and proposed criteria for indexing and retrieval. Bidgood et al. [5] developed a descriptive system for clinically relevant image indexing called 'image acquisition context' which combined elements of a procedural standard for the image acquisition (DICOM) with a subset of clinical controlled vocabulary from the Systematized Nomenclature of Medicine (SNOMED). Diepgen and Eysenbach [6] put a large dermatology image database that was indexed by diagnosis and body region on a web server. Other investigators have also reported on use of the Internet to conduct collaborative research [7–9] using investigators and subjects who were computer and Internet sophisticated and for the most part with training in informatics. Humphreys et al. [10] carried out a highly successful Internet-based Large-Scale Vocabulary Test as the basis for additions to the UMLS Metathesaurus.

In the early 1990s the Board of Scientific Counselors (BoSC) of the Lister Hill National Center for Biomedical Communications suggested [11] that it would be interesting to determine whether UMLS Metathesaurus concepts could be used as a tool for indexing a set of gross and microscopic pathology images that had been collected during the production of a series of videodiscs intended for use in undergraduate pathology education [12].

A preliminary study (unpublished) was carried out between 1991 and 1994. Six urologists were asked to submit lists of terms they would use to describe a set of 12 prostate photomicrographs that had been

recorded on videodiscs. These terms were submitted to the Metathesaurus as it existed in 1994. Of the 197 unique terms they submitted, only 85 (57%) retrieved concepts from Meta. Most of the lookup failures were attributable to a lack of morphological concepts. At that time the Metathesaurus contained only 27 vocabularies, 190,863 concepts, and 336,359 names.

Between 1993 and 1999, in collaboration with Michael Becich and Charlie Hatton at the University of Pittsburgh Medical Center, a distributed digital image database was created that now resides on servers located in Bethesda, MD, Pittsburgh, PA and Urbana, IL. The institutions that contributed images to the database were: The University of Pittsburgh, Allegheny General Hospital, the University of Illinois at Urbana-Champaign, the University of Medicine and Dentistry of New Jersey, the University of Vermont and the Aga Khan University (Karachi). We then built a system to test the hypothesis suggested in the BoSC report [13]. It runs on a web server located at NLM and has three modes:

- Index (on-line lookup of Metathesaurus concepts and associating selected ones with images in an Oracle table);
- Search (on-line lookup of concepts and using them to retrieve images);
- Upload (uploading either physical image files or URLs).

The system's database currently contains over 1000 images. Domain experts indexed images except those from the University of Illinois at Urbana-Champaign which were indexed by one of the authors using image descriptions provided by domain experts. We now report on an effort to use this system to determine to what extent the terms used by domain experts to describe medical images can be mapped to Metathesaurus concepts.

3. Methods

We asked Dr. Robert Schosser, Chairman of Department of Dermatology, University of Kentucky, to select ten images from an unpublished collection of photographs and photomicrographs of common dermatoses. These images were uploaded to the web server where they could be viewed at a resolution of 768×512 pixels in 24-bit color. We then recruited volunteer dermatologists to provide us with up to ten terms they would use to describe each image and then to judge the relevance of the Metathesaurus concepts to which each term is mapped. The Sulzberger Institute for Dermatology Education of the American Academy of Dermatology assisted in the recruitment of volunteers. Nine of 19 dermatologists who agreed to take part in the experiment completed both tasks.

The experiment was conducted entirely via password protected web sites maintained at NLM. We separated the process into two tasks and used two forms to collect and analyze the data. In the first task, domain experts saw a divided screen with thumbnail pictures of the test images in one frame and a larger size of the selected image in the other frame together with ten data (term) entry blocks (Fig. 1). Note that subjects were given access to an online medical dictionary since neither the Knowledge Source Server nor our software has a built-in spelling checker. When the user clicked the 'Submit' button, the form was returned showing all of the user's inputs. The user was then given an opportunity to make corrections, additions and/or deletions prior to sending the data to the server. Terms submitted by each volunteer were collected in a comma separated flat file and later imported into an Oracle database.

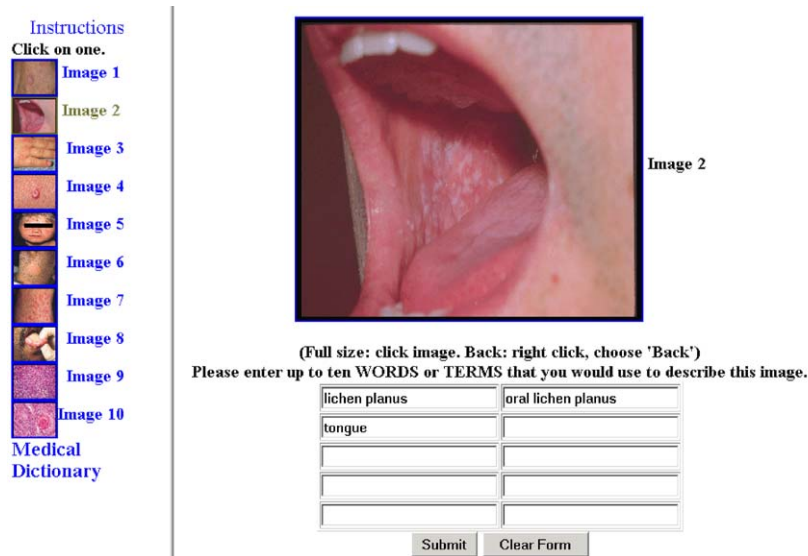


Fig. 1. Screen capture of term input page on the web site.

We then submitted the terms to the Metathesaurus (2002 Release) using the online interface to the UMLS Knowledge Source Server. We first submitted the terms using the default search routine, which utilizes a normalized string index (-ns). If no concept was returned or if the semantic type of the concept was obviously not related to the picture, we resubmitted it using the approximate matching (-am) routine. (Examples: (a) ‘malar’ maps only to ‘zygomatic bone’ using the default but it also maps to ‘cheek’, ‘butterfly rash’ and ‘butterfly malar facial rash’ using -am. (b) ‘well circumscribed’ is not mapped to a concept using ‘-ns’, but using ‘-am’ it maps to ‘circumscribed alopecia’ which is exactly what was shown in the slide.) When we used the -am routine, the ‘order by semantic group’ option was always selected since approximate matching can return up to 50 concepts. The concept(s) retrieved, semantic type(s), synonyms and definitions (when available) were returned to the expert in the second form (Fig. 2).

Although online instructions emphasized that the Metathesaurus maps *terms* that may be made up of more than one word, after a trial run with one dermatologist we decided to modify user input in the following ways:

(a) If user submitted single words in successive blocks and the words taken together mapped to a concept, then the multiword term was submitted to the server. (Example: If ‘basal’ and ‘cell’ and ‘carcinoma’ were submitted in three successive blocks, ‘basal cell carcinoma’ was looked up in the Metathesaurus.)

(b) If user submitted multiple words in individual input blocks and the term did not map to a concept, then the individual words were looked up. (Example: ‘fine erythematous papules’ submitted in one block and no concept was returned by the Metathesaurus, ‘fine’ and ‘erythematous papules’ were submitted as separate terms and both are mapped to meta concepts.)

(c) In ten instances substitutions were suggested for words that were not mapped to a concept. (Examples: The term ‘black skin type’ is not mapped to a concept; ‘black race’ was suggested as a substitute. ‘Lavender’ is mapped only to the plant and its extracts; ‘violaceous’ was suggested as a substitute.)

Your term/words	Meta concept	Semantic Type	Definition/Synonyms/Notes	Ratings
1. lichen planus <u>Multiple Concepts</u>	a. Lichen Planus	Disease or Syndrome	DEF: An inflammatory, pruritic disease of the skin and mucous membranes, which can be either generalized or localized. The lesions may be discrete or coalesce to form plaques.	<p>a. The concept</p> <input type="text" value="is broader than"/> <input type="button" value="v"/> what I had in mind. -----
	b. Lichen Planus, Oral	Disease or Syndrome	DEF: Oral lesions accompanying cutaneous lichen planus or often occurring alone. The buccal mucosa, lips, gingivae, floor of the mouth, and palate are usually affected (in a descending order of frequency).	<p>b. The concept</p> <input type="text" value="Click here to select"/> <input type="button" value="v"/> <input type="text" value="Click here to select"/> <input type="button" value="v"/> is exactly is broader than is narrower than is related to is not related to <p>c. The concept</p> <input type="text" value="is not related to"/> <input type="button" value="v"/> what I had in mind.
	c. Lichen-organism	Organism	DEF: Any of a group of plants formed by a mutual combination of an alga and a fungus. (MeSH)	<p>c. The concept</p> <input type="text" value="is not related to"/> <input type="button" value="v"/> what I had in mind.
2. oral mucosal disease	None		"Oral mucosal (Spatial Concept)" and "Disease (Disease or Syndrome)" are Meta concepts. Since indexers can combine concepts, please rate as if your term were a Meta concept. SYN: OLP; DEF: Oral lesions accompanying	<p>The concept</p> <input type="text" value="Click here to select"/> <input type="button" value="v"/> what I had in mind.

Fig. 2. Screen capture of form used for rating of UMLS concepts.

Concepts found in the Metathesaurus using the default (normalized string index), that were obviously not related to the image were not returned to the dermatologist. (Example: ‘patch’ is mapped to six concepts using default matching. Only ‘patch formation’ and ‘plaque (lesion)’ were returned to the dermatologist since the semantic types of the other four concepts (Patch repair, Patch surgical material, PATCHED and Patch drug form) returned ruled them out as possibilities for description of the image. The domain experts were asked to rate each concept as ‘exactly’, ‘broader than’, ‘narrower than’, ‘related to’ or ‘not related to’ what they had in mind when they looked at the slide. Their ratings for each concept were recorded in a flat file and later imported into the Oracle database. The terms collected, concepts retrieved, and ratings of the concepts were all imported into one Oracle table. Each table row also contains the identification of each participant, the type of search used (-ns or -am), and information regarding whether input terms were combined or split. Tool for Oracle Application Development (TOAD) was used extensively for data analysis.

4. Results

Nine dermatologists used 825 terms, 346 of which were unique, to describe the ten images. Of the 300 terms that mapped to semantically reasonable UMLS concepts, 242 (81%) did so with the default (normalized string) routine. Fifty-eight required use of approximate matching. Forty-six (16%) of the unique terms did not map to a concept. Of the terms that did not map to a concept, only four (‘eroded’, ‘flesh colored’, ‘hairless’ and ‘non-scaling plaque’) were used by more than one dermatologist. Seventeen (7%) of the 242 terms that retrieved concepts using the default (-ns) mapped to multiple concepts; in every case one of the concepts was rated ‘exact match’. In all, 326 concepts were retrieved. Of the 326 concepts 286 (88%) were rated as ‘exact match’ by the dermatologists. Twenty-four (7%) were rated as ‘not related’. Of the 300 unique terms that mapped to a concept, 295 (98%) retrieved a concept that was rated as an ‘Exact Match’ (Table 1); 17 (5%) retrieved concepts rated as ‘not related to’. Table 2

Table 1
Dermatologists' rating of relevance of UMLS concepts to which their terms were mapped

# Unique terms	# Uses	% Exact match ^a
187	1	97
46	2	100
21	3	100
15	4	100
14	5	100
3	6	100
3	7	100
7	8	100
2	9	100
1	20	100
1	26	100

^aAt least one of the concepts retrieved rated 'EM' by a majority of raters.

Table 2
Relationship between number of dermatologists using term and ratings of concepts retrieved

Image	Terms	Number of dermatologists using term											
		1		2		3		4		5		6 or more	
		Terms	%EM ^a	Terms	%EM ^a	Terms	%EM ^a	Terms	%EM ^a	Terms	%EM ^a	Terms	%EM ^a
1	37	13	85	11	100	4	100	2	100	2	100	5	100
2	31	13	100	6	100	4	100	3	100	2	100	3	100
3	38	18	100	5	100	4	100	3	100	5	100	3	100
4	35	20	95	6	100	1	100	3	100	2	100	3	100
5	40	18	94	10	100	3	100	4	100	1	100	4	100
6	39	23	100	7	100	2	100	0	—	1	100	6	100
7	49	28	100	10	100	1	100	1	100	4	100	5	100
8	32	18	100	6	100	1	100	2	100	2	100	3	100
9	47	28	96	12	100	3	66 ^b	2	—	2	—	0	—
10	49	32	97	12	100	3	—	1	100	1	—	0	—

^aIf multiple concepts retrieved, at least one concept was rated 'EM' by all raters.

^bTerm was 'horn cyst'; concept is 'epidermal cyst'. One dermatologist rated it 'nr' for image 9 and 'nt' for image 10.

shows the relationship between the number of dermatologists using a given term and the relevance of the concept retrieved to the image being described. Except for the case of the data on image 9, if two or more dermatologists used a term, then at least one concept was retrieved that received an exact match rating. In 12 instances out of 330, different ratings were obtained from two or more dermatologists. These ratings are summarized in Table 3. Table 4 shows the semantic types of concepts that domain experts associated with clinical dermatology images.

As noted in the Background section, experience with the earliest version of the Metathesaurus was unrewarding due to a large extent to the lack of robust morphological concepts. The 197 terms collected in the 1994 study of prostate images were resubmitted as a part of the Large-Scale Vocabulary Test (LSVT)

Table 3
Disagreements in ratings of UMLS concepts

Term (search method)	Concept	Image	Ratings
Buccal mucosa (-ns)	Mouth mucosa	2	EM, EM, EM, EM, BT
Reticulate (-ns)	Reticular tissue Reticular	2	RT, NR, NR EM, EM, EM
Periungual (-am)	Periungual skin of finger	3	EM, EM, EM, NT
Areata (-am)	Alopecia areata Alopecia areata, guttate Concentric alopecia areata Other alopecia areata	6	EM, EM, EM, EM RT, NT, NT NT, NT, RT BT, RT
Nonscarring (-am)	Non-scarring hair loss Other non-scarring alopecia	6	EM, EM, RT, RT EM, RT
Patch (-ns)	Patch formation Plaque (lesion) Patchy distribution	6	EM, RT, NR, NR RT, NR, NR EM, EM, RT, NR
Leg (ns)	Lower leg Lower extremity	7	EM, RT EM, EM
Tongue (-ns)	Tongue Benign neoplasm of tongue Procedures on the tongue	8	EM, EM, EM EM, RT, NT NR, NR, NR
Verruca (-ns)	Common wart Human papilloma virus infection Plantar wart	8	EM, EM EM, EM EM, NR
Horn cyst (-ns)	Epidermal cyst	9&10	EM, EM, EM, NR, NT
Pearly (-am)	Keratin pearl formation	9&10	EM, EM, EM, NR
Squamous (am)	Squamous epithelial cell Squamous epithelium, NOS	9&10	EM, NR EM, EM, EM, EM

Note: Images 9 and 10 are different magnifications of the same photomicrograph.

Table 4
Semantic types of UMLS concepts used by dermatologists

Semantic type	No. of concepts	Semantic type	No. of concepts
Finding	60	Age group	3
Disease or syndrome	49	Quantitative concept	3
Qualitative concept	36	Therapeutic or preventive procedure	3
Spatial concept	33	Body space or junction	2
Functional concept	25	Body substance	2
Body part, organ, or organ component	21	Organism function	2
Pathologic function	20	Plant	2
Body location or region	17	Genetic function	1
Acquired abnormality	12	Fungus	1
Neoplastic process	12	Intellectual produce	1
Tissue	10	Laboratory procedure	1
Sign or symptom	7	Laboratory test or result	1
Cell	5	Organic chemical	1
Cell or molecular dysfunction	5	Organism attribute	1
Cell component	4	Temporal concept	1
Population group	4	Virus	1

Table 5
Metathesaurus sources of 330 UMLS concepts

Source ^a	Concepts	%	Unique	%	Source	Concepts	%	Unique	%
SNMI	241	73.0	57	17.3	UWDA	37	11.2	6	1.8
RCD	195	59.1	33	10.0	LOINC	21	6.1	1	0.3
MSH	89	27.0	0		PSY	21	6.1	0	
MDRAE	83	25.2	5	1.5	BI	18	5.5	0	
AOD	75	22.7	0		PDQ	17	5.2	2	0.6
MTHN	75	22.7	0		AIR	8	2.4	1	0.3
CRSP	69	20.9	1	0.3	HL7	5	1.5	0	
CCPS	62	18.8	9	2.7	CCS	4	1.2	0	
DXP	55	16.7	16	4.8	NCBI	2	0.6	0	
ICD	55	16.7	3	0.9	PCD	1	0.3	0	
LCSH	55	16.7	0		MIM	1	0.3	0	
COSTAR	48	14.5	0		MMSL	1	0.3	0	
COSTART	41	12.4	2	0.6	NOC	1	0.3	0	
WHO A	40	12.1	0		OS	1	0.3	0	
ICPC	39	11.8	1	0.3					

^aA list of all UMLS source vocabularies is at: <http://www.nlm.nih.gov/research/umls/METAB2.HTML>.

[9] and concept retrieval rate was much improved. It should be noted that SNOMED International, Logical Observations Identifiers, Names and Codes (LOINC), and the Read Clinical Classification System were the vocabularies studied in the LSVT experiment and added to the UMLS sources. We, therefore, decided to determine which Metathesaurus sources were responsible for the results of the current study. Those results are shown in Table 5. Table 6 summarizes our results on the basis of individual ‘indexers’. The

Table 6
Comparison of input from nine dermatologists

Indexer	Images indexed	Terms submitted	Terms found in meta	Exact match	%EM ^a	Time on tasks (min)
1	7	54	50	47	94	22
2	10	48	43	43	100	35
3	10	93	88	88	100	24
4	10	64	59	56	95	26
5	7	35	34	33	97	20
6	9	63	58	58	100	29
7	10	66	61	60	98	27
8	10	115	93	93	100	35
9	10	56	54	52	96	24

^aIf multiple concepts retrieved, at least one was rated exact match.

average time spent by the nine dermatologists in accomplishing Task 1 was 13 min; the average for Task 2 was 14 min (Table 6).

5. Discussion

Our results (Table 1) suggest that the probability of retrieving one or more UMLS concepts rated as an exact match is extremely high. That probability (Table 2) is virtually independent of the frequency of use by the domain experts either between images or between individual indexers. The ‘hit rates’ shown in Table 1 are considerably higher than those found earlier using prostate photomicrographs and than those of Lowe and his colleagues [3] using both human and automated methods to identify UMLS concepts to apply to radiology images. The information in Table 7 may help explain these differences; the growth of Meta increases the probability of a matching concept in one or more component vocabularies.

The results also suggest (Table 2) that a single domain expert might index images with high likelihood that other experts in that domain would agree that the concept selected is appropriate to that image. Further, the results show that if two or more dermatologists used a term, then the probability of retrieving a concept that is rated ‘exact match’ approaches certainty. In only 12 out of 269 instances in which two or more dermatologists used the same term there were disagreements in ratings (Table 3).

The semantic type distribution shown in Table 4 differs from that described by Humphreys et al. [10] for the LSVT in that our semantic types have a lower proportion of Disorders (14%) and a higher proportion of Concepts (29%), Anatomy (22%) and Findings (20%). This distribution may reflect the more specialized task of clinical images indexing.

The data in Table 5 show that no single component vocabulary of the 2002 version of the Metathesaurus is sufficient to index the images independently. Only two vocabularies, SNMI (SNOMED) and RCD (Read Thesaurus) contained even a statistical majority of the concepts selected. The relative contributions of source vocabularies in our study suggest that the combined resources of SNOMED International and Read Clinical Descriptors contain 83% of the concepts used in this study. Although we cannot generalize beyond the clinical domain studied, this may imply that SNOMED CT (a new vocabulary based on the combination of SNOMED and the Read Thesaurus) [14] might be useful for indexing images associated with medical record text.

Table 7
Growth of the UMLS Metathesaurus

	1994	1996	1998	2002	2003AA
Concepts	152,444	252,892	476,313	776,940	875,255
Strings	137,259	543,108	1,051,901	1,920,170	2,343,341
Relationships	4,952,800	5,860,561	7,077,889	11,137,725	11,648,030
Sources	27	39	53	110	100

Current indexing of documents held by the National Library of Medicine (NLM) is performed by individuals with at least a baccalaureate in science and specialized training in the application of the Medical Subject Headings (MeSH) controlled vocabulary. NLM's only indexed online database of images, Images from the History of Medicine (IHM), is indexed by MeSH and Library of Congress subject headings (LCSH). The data in Table 5 also show that the use of MeSH alone for indexing would find matching concepts for about one-fourth of the terms in our experiment. These current results suggest that the UMLS Metathesaurus contains sufficient concepts to index images in a biomedical domain, dermatology, which is highly demanding of visual descriptors. The results (Table 6) also suggest that domain experts without prior training in indexing, may be able to index multimedia objects for a database using a controlled vocabulary and that the amount of professional time required to do so is probably within an acceptable range.

Only about half of the dermatologists who volunteered to take part in this study completed both tasks. Only six of these nine indexed all ten images. We made no effort to qualify our participants as did McCray et al. [9] and since all were volunteers, we did not attempt to determine the reasons for non-participation. Examination of the original term input data files suggests to us that some of our volunteers were unable to master use of the online forms. The Time-on-Tasks data in Table 6 suggests to us that non-participation was not due to that factor. In future efforts we will probably follow the example set by Humphreys and administer a pretest to qualify our volunteers.

6. Conclusions

- The UMLS Metathesaurus can serve as a controlled vocabulary useful in indexing clinical images.
- The utilities of the Knowledge Source Server can be adapted for use by domain experts relatively unfamiliar with library science or medical informatics.

Next: We intend to test the retrieval side of the equation by having potential users attempt to retrieve these (and perhaps other) images and track their efforts.

7. Summary

Nine dermatologists used Internet generated forms to (1) enter terms to describe pictures of skin lesions and (2) judge how well the Metathesaurus concepts to which their terms are mapped apply to those terms. They submitted 825 terms 346 of which were unique. Of the 346, 300 are mapped to 330 concepts.

Subjects were asked to rate the relevance of the concepts as ‘Exact Match’, ‘Broader than’, ‘Narrower than’, ‘Related to’ or ‘Not related’.

Forty-six terms were not mapped to any Meta concept. Three hundred terms retrieved one or more Meta concepts and 295 of those were rated ‘Exact Match’ by a majority of the dermatologists; 17 were rated as ‘Not related’ by at least one dermatologist.

The domain experts spent, on average, about 30 min to accomplish both tasks (term entry and concept judging). We conclude that the UMLS Metathesaurus can provide a controlled vocabulary for indexing of clinical dermatology images and that therein lays a realistic option for image libraries.

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