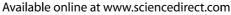
7 December 2005 Disk Used



ARTICLE IN PRESS





No. of Pages 8, Model 5+

Nathan (CE) / Karthikeyan (TE)

Journal of Biomedical Informatics

Journal of Biomedical Informatics xxx (2005) xxx-xxx

www.elsevier.com/locate/yjbin

# 2 In defense of the Desiderata 3 James J. Cimino \* 4 Departments of Biomedical Informatics and Medicine, Columbia University, New York, NY 10032, USA 5 Received 15 August 2005

### 7 Abstract

8 A 1998 paper that delineated desirable characteristics, or *desiderata* for controlled medical terminologies attempted to summarize 9 emerging consensus regarding structural issues of such terminologies. Among the Desiderata was a call for terminologies to be "concept 10 oriented." Since then, research has trended toward the extension of terminologies into ontologies. A paper by Smith, entitled "From 11 Concepts to Clinical Reality: An Essay on the Benchmarking of Biomedical Terminologies" urges a realist approach that seeks termi-12 nologies composed of universals, rather than concepts. The current paper addresses issues raised by Smith and attempts to extend the 13 Desiderata, not away from concepts, but towards recognition that concepts and universals must both be embraced and can coexist peace-14 ably in controlled terminologies. To that end, additional Desiderata are defined that deal with the purpose, rather than the structure, of 15 controlled medical terminologies.

16 © 2005 Published by Elsevier Inc.

17 *Keywords:* Controlled terminology; Ontology; Medical knowledge representation; Philosophy 18

### 19 1. Whence the Desiderata?

20 It would be a rare biomedical information system that 21 did not require some means for representing data and/or 22 knowledge in a formal, reproducible, and useful way. The 23 uses can be quite varied but they all require some capability 24 for symbolic manipulation. Otherwise, why bother with 25 controlled representation when one could just store raw 26 signals? At the simplest level, symbolic manipulation at 27 least requires a set of symbols (often referred to as *identifi*-28 ers) that distinguish the various elements of data and 29 knowledge. Human-understandable labels (often referred to as *terms*) for these elements are not always required,<sup>1</sup> 30 31 but they are usually deemed convenient, if for no other rea-32 son than the need to map human-collected data into sym-33 bols and to map symbols into human-usable results. Sets of these symbols and labels (or terms and identifiers) are usu-<br/>ally referred to as *controlled terminologies*.3435

The design and content of controlled terminologies are 36 quite diverse but, ultimately, they serve some common pur-37 poses: they must support the capture, storage, manipula-38 tion, and retrieval of the information they represent in 39 ways that faithfully preserve and communicate the original 40 information. The construction of a controlled terminology 41 does not always guarantee that successful representation 42 will occur; some approaches will predictably work better 43 than others. Biomedical informatics researchers have been 44 studying and describing these approaches for decades. One 45 attempt to synthesize and summarize that body of work, 46 written in the late 1990s, identified 12 desiderata that 47 reflected the ideas of many researchers of the time [1]. 48

One of the key points in the Desiderata was that termi-49 nologies should focus not only on the names of the data 50 and knowledge elements they intended to represent, but 51 also on their underlying meanings. The term "meaning" 52 has several definitions (or meanings!), including "the thing 53 one intends to convey especially by language" [2]. How one 54 conveys these "things" may be accomplished by various 55 methods, depending on what that thing is. For example, -56

<sup>\*</sup> Fax: +1 212 305 3302.

E-mail address: jjc7@columbia.edu.

<sup>&</sup>lt;sup>1</sup> Consider, for example, the nodes in a hidden layer of a neural network. Each node represents coordination of values from input nodes and conveys some information to the output nodes, but we do not characterize these nodes as corresponding to any real or conceptual intermediate entity.

7 December 2005 Disk Used

**ARTICLE IN PRESS** 

114

150

2

J.J. Cimino | Journal of Biomedical Informatics xxx (2005) xxx-xxx

57 if the thing is a particular object in reality (say, a particular 58 person or automobile), its meaning might be conveyed by a 59 proper noun or some unique identifier (e.g., the person's name or the automobile's registration number). If the thing 60 61 is a generalization of some particular set of things in reality (that is, a universal that has some extension, or set of 62 63 instances in reality), its meaning might be conveyed by a general term for the things or by some set of characteristics 64 that all these things have in common (e.g., "person" or 65 "passenger vehicle, self-propelled, four wheels ... "-of 66 course, this raises the need to define these additional 67 terms). Finally, if the thing is an idea, we might refer to 68 69 it with some description of the idea or by describing the 70 particular instances on which the idea is based.

71 A particular term is not restricted to a single method for 72 the conveyance of its meanings. Depending on how the 73 thing to which the term corresponds are viewed, it might 74 have multiple aspects that could be conveyed in multiple 75 of ways. For example, a controlled terminology for speak-76 ing about celestial bodies might contain a term with the name "solar planet," the meaning of which might be 77 78 expressed by referring to a list of known planets ("any of 79 Mercury, Venus, Earth ... ") or by a intentional definition 80 ("a large body that orbits around the Sun").<sup>2</sup>

81 The names (terms) used for these things usually serve the 82 purpose of conveying the meanings, when the reader has 83 appropriate background knowledge. The Desiderata 84 attempted to stress the fact that something more formal than the language-specific linguistic shorthand with which 85 86 humans are comfortable was required to convey meanings 87 in useful ways. Specifically, some formal representation to 88 convey the intended meanings of the data and knowledge 89 elements was requested. Formal representations could, 90 themselves, be represented symbolically (that is, using a 91 controlled terminology) and would be independent of the 92 fashions of language. In the "solar planet" example above, 93 this might be accomplished by including in the terminology 94 elements corresponding to each of the planets, by including 95 elements that correspond to the Sun and the notion of 96 orbit, or by both.

97 While the formal representation of terms constitutes a 98 type of knowledge, the Desiderata stopped short of using the word "ontology" to refer to this knowledge. This omis-99 sion was due, in part, to the absence of the term in the 100 101 reviewed informatics literature, although speakers at the 102 time sometimes referred to it [3], usually as "the O Word" because of the perceived overuse of the term by the com-103 104 puter science community [4,5]. Instead, the Desiderata 105 attempted to describe this focus on meanings to be "con-106 cept oriented." The use of "concept" was chosen as a way to refer to the meanings of the symbols, as understood 107 by humans. But, the notion of "concept"-an abstract idea 108

generalized from specific instances [2]—is only one aspect 109 of the data and knowledge items that we might wish to represent. As a result, while the Desiderata may be interpreted 111 narrowly, in fact, the work from which they were synthesized related to broader aspects of symbolic representation. 113

### 2. A conceptual terminologies

In "From Concepts to Clinical Reality: An Essay on the 115 Benchmarking of Biomedical Terminologies" [6], Smith 116 holds that terminologies should not rely on the use of con-117 cepts. He is troubled by the fact that concepts can be 118 viewed linguistically, psychologically, epistemologically, 119 and ontologically. He sees concept-oriented terminologies, 120 on the one hand, as collections of elements that may or 121 may not correspond to things in reality and, on the other 122 hand, as little more than groupings of the synonymous 123 terms by which humans express ideas. 124

Smith provides many examples of ways in which termi-125 nologies composed of expressions of ideas lead to difficul-126 ties: in one, taken from Campbell et alia's description of 127 the National Library of Medicine's (NLM) Unified Medi-128 cal Language System (UMLS), "aspirin" and "Aspergum" 129 refer to the same meaningful element in the UMLS's Meta-130 thesaurus [7]. He holds this paradox to be an artifact of the 131 "conceptualist" view. 132

He prefers to focus, instead, on terminologies composed 133 of references to universals only, and to their corresponding 134 instances in reality-what he calls the "realist" view. In this 135 view-which assumes that there is only one, universal 136 objective reality-only things in reality would be consid-137 ered, such as pieces of Aspergum and portions of aspirin. 138 Each of these objects would be instances of one of two, 139 mutually exclusive universals (a universal of gum products 140 made from aspirin, perhaps called "Aspergum," and uni-141 versal of portions of aspirin, perhaps called "aspirin"). 142 More general universals would be considered (such as "as-143 pirin-containing drugs" and "organic acids") which would 144 also extend to real world objects, including (respectively) 145 pieces of Aspergum and portions of aspirin. All of this is 146 147 done without the messiness of concepts and without any confusion of what is the chewable thing and what belongs 148 on a chemist's shelf. 149

### 3. What is on the side of the patient

Smith considers clinical terminologies, in particular, as 151 domains where concept-orientation should be eschewed 152 in deference to realism. It is natural to consider that a 153 patient in reality has real attributes that can be described 154 155 and used to support activities such as diagnostic and therapeutic decision-making. Each of these (the patient 156 and the attributes) would be represented in a biomedical 157 terminology as a universal. These universals might or 158 might not have names associated with them as linguistic 159 dressing. The important thing is that their meanings are 160 determined solely by their extensions in reality, not by 161

<sup>&</sup>lt;sup>2</sup> With the recent discovery of a new large body orbiting the Sun outside the orbit of Pluto, the extremtional meaning will need to be updated with some reference to the new body, which will be awkward until it is named; meanwhile, the intentional meaning remains valid.

7 December 2005

**ARTICLE IN PRESS** 

162 concepts spawned in human minds as ways of thinking163 about reality.

**Disk Used** 

164 Strong arguments can be made for the ontological puri-165 ty of this approach and for the clarity by which it can sup-166 port reasoning. After all, the reality is what it is, and is not 167 perverted by the fashionable views of society. A patient 168 who behaves in the same particular manner from year to 169 year might be labeled as having a psychological disease 170 one year, as having a personality disorder the next year, 171 and to be a normal variant the year after that, without 172 any change in the actual patient. Recording the manner-173 isms themselves, rather than some momentary conceptual 174 perspective, allows us to reconsider past information in 175 the past (and future) contexts, as our understanding of 176 the causes of the mannerisms becomes better understood. 177 In fact, one of the novel ideas in Ledley and Lusted's 178 1950 landmark paper was a call for the use of such primary 179 patient attributes for automated medical diagnosis [8].

180 This approach is strengthened by including unique iden-181 tifiers not just in the terminology of universals but "on the side of the patient"—that is, each attribute of the patient is, 182 183 itself, a unique entity in reality and is assigned its own iden-184 tifier. When a patient's temperature is measured, the mea-185 surement is an instantaneous entity (a *perdurant*), while 186 the polyps seen on colonoscopy are persisting entities 187 (endurants); each is given an identifier by which they can 188 be referenced, so that multiple temperature measurements 189 can be related to each other and individual polyps can be followed over time. 190

## 191 4. Terminologies of universals

192 The notion of terminologies that are limited to well-be-193 haved universals, each one clearly understood because of 194 its extension in reality, is appealing and, if possible, would make the lives of clinical system developers much simpler. 195 196 In fact, many patient records today do, to a limited extent, 197 implicitly represent patient information as extensions in 198 reality, as discussed below, through their data models. 199 While a clinical record may appear to capture that, as 200 Smith charges, a physician fits together a patient and a con-201 cept (e.g., a disease), the clinical record is usually capturing 202 (with some unique identifier composed of time, patient, 203 physician, and concept) that at some point in time, the phy-204 sician is observing attributes of the patient that leads the 205 physician to believe that the signs the patient is manifesting 206 are evidence that the patient has some disease. Thus, while 207 the concept may appear to be separate from the patient, 208 there is a deeper connection that is implied by the design 209 of the patient record [9].

The idea of unique identifiers for aspects "on the side of the patient" that are instances of universals in the terminology is not entirely new. Clinical laboratory systems have long included controlled terminologies for specimens. The terms in such terminologies arguably correspond to universals, since they have extensions in reality, in the form of actual instances of specimens collected from actual patients. When an actual specimen is collected from an 217 actual patient, it is assigned a unique identifier (at New 218 York Presbyterian Hospital, these are called "accession 219 220 numbers"). Using such identifiers, it is possible to perform multiple measurements on the same specimen (to check for 221 consistency), to perform different measurements on the 222 same specimen (to correlate findings), and to report multi-223 224 ple results on the same specimen (such as preliminary and 225 final results). It is even possible to invalidate a previous measurement, if some problem with an analysis is 226 discovered. 227

The idea of uniquely enumerating each of the patient's 228 problems can be traced back to Weed's landmark paper 229 230 on problem-oriented medical records [10], later implemented in his PROMIS system. A more elaborate example of 231 unique identifiers for patient attributes can be found in a 232 paper by Barrows and Johnson [11] that describes the 233 assignment of persistent identifiers to patient problems. 234 As a particular problem (or the understanding of the prob-235 lem) evolves over time, new interpretations can be assigned 236 without losing the reference to the original problem. Smith 237 promotes the extension of this approach (as proposed with 238 Ceusters and Smith [12]) to permeate the patient record. 239 Everything about the patient that is medically salient 240 241 receives a unique identifier instance that relates it back to a universal in the terminology. 242

The tokenization of the entire patient record in this way 243 is a bold proposal and, if technically feasible, would open 244 up entire new pathways to patient care, epidemiology, 245 quality assurance, and various other ways to use and reuse 246 patient data. There is no question that such recording 247 should be done more often than it is now, and that coded 248 electronic record systems as they exist today actually do 249 obliterate some of the instance-recording that occurs in 250 251 patient care. Most notably, effort spent on encoding billable diagnoses and procedures divert valuable resources 252 away from recording the "what it is on the side of the 253 patient" in favor of data that are less-resusable [13]. 254

I set aside here questions of whether the complete exten-255 sion of this approach can actually be accomplished without 256 paralyzing care providers with attribute-identification tasks 257 (will endoscopists tattoo each polyp so that they can iden-258 259 tify them later?) and how such an approach could be studied to gather evidence of its cost-effectiveness and safety. 260 261 More immediately, we must consider whether a terminology that supports this approach can truly be composed of 262 only universals and not include what have been traditional-263 ly understood as conceptual entities. 264

Laboratory specimen terminologies, as they exist today, 265 typically do not include definitional information about the 266 various specimens (collection methods, equipment used, 267 body parts collected, etc.) beyond their name (e.g., "blue 268 top blood specimen"). However, the meanings of these 269 terms are generally understood, at least to the laboratory 270 technicians, and these meanings are based on general attri-271 butes (e.g., "5 ml test tube with heparin and venous blood 272 from the patient") rather than a set of extensions in reality 273

4

7 December 2005 Disk Used

### J.J. Cimino | Journal of Biomedical Informatics xxx (2005) xxx-xxx

274 (e.g., "Specimens 22122, 37812, 45092, ..."). In fact, the 275 terminology could include terms for specimens that don't 276 actually exist in reality ("blue top saliva specimen"). Such 277 terms would not be included for reasons of pure fantasy 278 (such as support for a pathologist-turned-fiction writer), 279 but rather in anticipation of the eventual occurrence of 280 such a specimen [14]. While there is no need to precoordi-281 nate every possible permutation of body part and collec-282 tion method, there is a need to create terms in a 283 terminology for specimens that will soon be needed, but for which no actual instance yet exists. Thus, if we assume 284 285 that recording what is on the side of the patient will involve instances of laboratory specimens collected from the 286 287 patient, and that the laboratory information system would 288 benefit from having some controlled set of well-defined 289 terms for universals to process the actual specimens, we 290 cannot escape the practical need to include terms for things 291 that do not yet actually exist—that is, terms that are, at 292 least temporarily, purely conceptual. The alternative, to 293 add a new specimen term to the laboratory system's termi-294 nology when the first instance of specimen is created, is 295 simply not practical: laboratory systems dictate to the clini-296 cians which specimens are allowed, not the other way 297 around.

298 Other attribute domains may prove equally messy when 299 trying to limit a terminology to a set of terms for univer-300 sals. For example, the clinical collection of patient temper-301 ature is a routine procedure that will continue regardless of 302 how we represent the resulting data. Recording a number 303 of degrees in the patient record, along with a unique iden-304 tifier for that recording, is not sufficient for the information 305 to be used in care of the patient; somehow, the notion that 306 it is a body temperature must be conveyed. To really know 307 the patient's body temperature, we would need to know the 308 kinetic energy of every one of the patient's molecules. Any 309 measurement process that attempted to detect this would 310 (according to Heisenberg) render the result moot by 311 destroying the patient. Instead, we use a device that detects, 312 imperfectly, the average kinetic energy of a small subset of the patient's molecules. From this, we make a guess at the 313 patient's actual temperature, taking into account that the 314 reliability of our estimate is further influenced by the orifice 315 316 containing the aforementioned molecules. I submit, then, 317 that when we record a patient's temperature, we can *only* 318 do so through reference to concepts; we do not have the 319 luxury of "real" universals. We might allow that since this 320 is the best approximation we can make, that patient body 321 temperature is, for all intents and purposes, a universal, 322 or we can acknowledge that we use the instances of mea-323 surements to help us create a conceptual representation 324 from which we can reason. The result—what we think we 325 are dealing with and what we do about it—is the same. 326 The advantages of an imperfect universal over an imperfect 327 concept are not self-evident.

328 Patient temperatures are sometimes needed as primary 329 parameters for patient management, such as hypo- and 330 hyperthermia. Most of the time, however, the temperature is used as a proxy for detecting and monitoring underlying 331 conditions. We speak of the term of "fever" and its more 332 specific, yet less-well-defined term "low-grade fever." If 333 these terms correspond to the ways we think about various 334 states of patient temperature, then they are concepts. Can 335 we at least do away with these concepts and reason from 336 first principles about our patients? This approach is appeal-337 ing, until we consider that we don't have much of a clue 338 about the first principles that relate disease processes to 339 particular values of body temperature. Is a patient with a 340 temperature of 39 °C twice as sick as a patient with a tem-341 perature of 38 °C, or only 2.6% sicker? Of course, neither is 342 the case, but we have no algorithm nor body of experience 343 with which to characterize patient states based on temper-344 ature. Instead, we mentally convert temperature measure-345 ments into conceptual representations of the patient's 346 true body temperature (as above) and then further use con-347 cepts like "low-grade fever" to help us match patients tac-348 itly to conceptual patterns that correspond to various 349 disease states (such as mild upper respiratory tract infec-350 tion—a cold). 351

The medical literature that is part of the foundation of 352 clinical education, and the original studies on which that 353 literature is based, have been derived in part from patient 354 data that were recorded conceptually, rather than realisti-355 cally. Perhaps past behavior is no excuse: if we begin today 356 to record patient data through the exclusive use of observa-357 tions on the side of the patient, we might eventually reach 358 some point at which we can compare a patient before us 359 with our experience by matching patient attributes, rather 360 than concepts. Homer Warner and others have argued that 361 such data should be the basis for logical diagnostic reason-362 ing, rather than reliance on abstractions that are the prod-363 uct of human experts, no matter how experienced [15]. 364

### 5. What is on the side of the clinician

Even if we are to discard the past hundred years or so of 366 clinical literature, we are still faced with the fact that 367 human beings reason based, necessarily, on concepts; the 368 best clinical reasoners rely on tacit knowledge that not only 369 is conceptual in nature but is, by definition, inexpressible 370 371 [16,17]. Our eventual liberation from the vagaries of human expert reasoners may not relieve us of this reliance on con-372 ceptual representations. When Warner attempted to inte-373 grate his diagnostic expert system with his clinical 374 information system, he found the mapping of patient data 375 to clinical concepts to be a significant challenge [18]. 376

365

Consider, for example, "severe acute respiratory syn-377 drome" (SARS). When the condition first arose, we might 378 379 have chosen to define this term based on a set of actual cases in reality that shared a set of particular attributes 380 381 (i.e., certain clinical manifestations with particular geographic and chronological characteristics). While such 382 characteristics were certainly true for each individual 383 patient, we must also consider how clinicians dealt with 384 this condition. Did they hold in their minds the unique 385 7 December 2005

386 identifiers of the individual cases or did they use some abstract representation, based on their understanding of 387 the disease at the time? It is certainly the latter, for without 388 the conceptual representation, they would have no way to 389 390 consider cases that fit some of the pattern of the disease 391 without having all of the characteristics of the initial 392 cases-for example, they would have to treat cases arising 393 in Canada as a different set of instances of a different uni-394 versal, since they do not match the previously identified 395 geographic characteristics. Smith does not say how we 396 would know to relax those constraints to recognize these 397 cases as being instances of the SARS universal. Humans, 398 however, achieve such reclassification readily, even 399 subconsciously.

**Disk Used** 

Instead, clinicians made use of a SARS concept, which 400 included conjectures, such as "probably viral." This 401 402 allowed them to consider aspects that would not be evoked 403 solely by the then-known characteristics of the individual 404 cases-for example, the recognition of cases that fit the pat-405 tern but not the definition based on known cases. Canadian 406 SARS cases can be classified as such according to the con-407 ceptual representation, allowing us to relax the geographic 408 constraints retroactively. It is fitting, then, that a controlled 409 terminology contain terms corresponding to such concepts, to record, in the patient's record, what diagnosis the clini-410 411 cian is considering at some point in time. If new informa-412 tion arises, the clinician might discard the previous 413 diagnosis and consider a new one. The terminology can 414 be employed to record both considerations.

# 6. If universals are insufficient, can we live with concepts?

415

416 We appear to be in a quandary: Smith would like us to 417 work with patient information at the instance level, and 418 reason with universals, thus avoiding the muddiness inher-419 ent in concept-level representation in reasoning. However, 420 we are trapped into using concepts, as long as we deal with 421 human reasoners (and their computer systems) and cannot 422 be able to escape them when dealing with human patients 423 (as per the fever example, above). Perhaps, though, things

424 may not be as bleak as they seem. 425 An analysis of the great aspirin-Aspergum controver-426 sy leads to the conclusion that it is a problem not of 427 semantics but of language. It is possible that someone, 428 somewhere, refers to this chewable product of the Insight 429 Pharmaceutical Corporation (Plymouth Meeting, PA) as 430 "aspirin." This is an example of the rhetorical device 431 known as synecdoche, in which a general word or phrase 432 is used to stand in for a more specific one (or vice versa). 433 In this case, there is no conceptual dilemma-when the speaker says "aspirin," he is actually attempting to com-434 435 municate a meaning that is synonymous with the mean-436 ing generally associated with "Aspergum." That he risks 437 being misunderstood is merely the effect of the ambigui-438 ties that beset human language. Had he, instead, used 439 some agreed upon identifier (whether a code or an agreed-upon name, recognized by speaker and listener 440

as being a unique preferred term in a controlled termi-441 442 nology), there would be no conceptual dissonance.

Another possibility (and the one that is involved in this 443 444 particular example) is that there was an error in judgment during the construction of the UMLS that led to the merg-445 446 ing of two meanings, such that the same unique identifier was assigned to both meanings (and their corresponding 447 448 terms)-an example of spurious synonymy-what the Desiderata called ambiguity. I believe that Smith would 449 argue that this is precisely his point: attempting conceptual 450 orientation inevitably leads to such muddiness. This may 451 452 be as true of concept orientation as any other orientation, but in this case the error was a systematic one that concept-453 454 orientation itself actually helped to resolve. It seems that, at the time that terms for drug products were added to 455 456 the UMLS, they were simply treated as instances of chemicals and were therefore indistinguishable from the chemi-457 cals from which they were composed (Nelson, personal 458 communication).<sup>3</sup> 459

Eventually, the NLM determined that two meanings 460 were present-one that referred to an organic acid (a chem-461 ical) and one that referred to a drug (a manufactured 462 object). The NLM determined that the chemicals and man-463 ufactured objects were mutually exclusive semantic types; 464 therefore, the assignment of these two meanings to the 465 same identifier could automatically be determined to repre-466 sent ambiguity. Today, the UMLS contains two separate 467 unique identifiers to which these terms are assigned. 468

I believe that other apparent contradictions in concept 469 representation can be peaceably resolved when they are dis-470 covered, not by throwing them away and replacing them 471 with something that extends to collections of real-world 472 instances, but by doing the hard work of understanding 473 their intended meaning(s) and purpose(s) to resolve the 474 475 contradictions through improved representation. It is my experience that not only can concepts and universals coex-476 ist in the same controlled terminology, but that this is a 477 desirable situation. 478

By way of example, consider the Medical Entities Dic-479 tionary (MED), the controlled terminology used at New 480 York Presbyterian Hospital [19]. The MED needs identifi-481 ers for a wide variety of entities that are represented in 482 patient records, including laboratory tests, laboratory spec-483 imens, radiological and cardiologic procedures, and diag-484 485 noses. Some of these can be considered to be universals (such as laboratory tests, medications, and the aforemen-486

<sup>&</sup>lt;sup>3</sup> This error originated with the general assumption that, if a UMLS source terminology considers two terms to be synonymous, and no other source terminology treated them as distinct concepts, then the UMLS would perpetuate the synonymy. In the Medical Subject Headings (MeSH), "Aspergum" was listed as an Entry Term that pointed to the MeSH Heading "Aspirin" as the preferred term to use for indexing (i.e., "Aspergum': see 'Aspirin'"). Entry Terms may be synonyms of the MeSH Headings to which they refer, but they are not necessarily so, as in this case. But, because no other UMLS Source Terminology, at the time, contained the term "Aspergum" as an entity distinct form "Aspirin," they were automatically assumed to be synonymous.

7 December 2005 Disk Used

**ARTICLE IN PRESS** 

6

J.J. Cimino | Journal of Biomedical Informatics xxx (2005) xxx-xxx

487 tioned specimens), while others are conceptual (such as 488 diagnoses). There is certainly room for many more univer-489 sals, should some method be developed for recording more 490 instances on the side of the patient. Meanwhile, far too lit-491 tle is understood about the diseases represented by the 492 diagnosis terms for us to represent them as universals, yet 493 they are far too useful to discard.

494 The MED attempts to adhere to the Desiderata, 495 including the use of formal definitions. While these defi-496 nitions are present for a only subset of terms in the 497 MED, and while the MED has been rightfully denied 498 the characterization of "ontology" [20], it nevertheless 499 contains ontological information and it provides an example of how concepts and universals can safely inter-500 mingle in the same terminology. Fig. 1 shows a small 501 sample of entities from the MED, drawn from laborato-502 ry tests, procedures, medications, and clinical informa-503 tion system constructs (summary reports). Also shown 504 is some of the ontological information contained in the 505 MED, expressed as semantic relationships among the 506 507 entities. These entities do more than share a common set of identifiers; their inclusion together in the MED 508 supports automated reuse of patient data (for example, 509 to aggregate comparable data into summary reports) 510 [21], automated inferencing (for example, in decision sup-511 port) [22], and automated translation [19,23]. 512

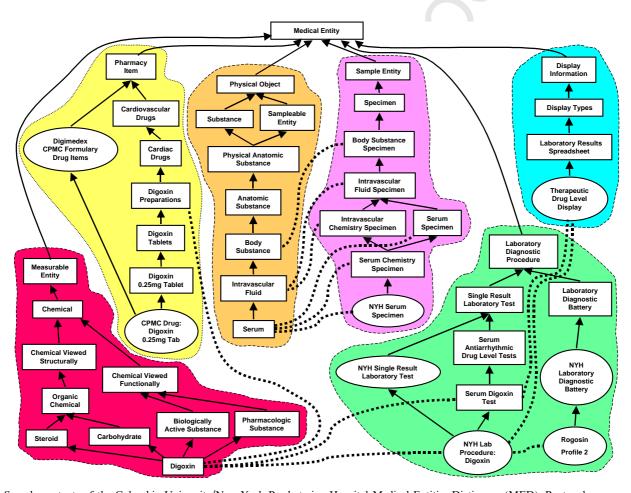


Fig. 1. Sample contents of the Columbia University/New York Presbyterian Hospital Medical Entities Dictionary (MED). Rectangles correspond to terms that represent universals. The extensions of the universals are actual tests, procedures and medications that are related to patients in the electronic health record. Dashed lines delineate different areas in the MED, such as (from left to right) measurable entities (including chemicals), specimens, sampleable entities (including body substances), diagnostic procedures (including laboratory tests), and the data dictionary, (including display information, which is used by the clinical information system for organizing patient data) The solid arrows represent is-a links in the MED, while the dotted lines represent nonhierarchical semantic relations, such as "measures substance" (between tests and chemicals), "has specimen" (between tests and specimens), "samples" (between specimens and anatomic substances), "has pharmaceutic component" (between drugs and chemicals), "has display parameter" (between laboratory results displays and laboratory tests) and "has test part" (between laboratory results displays, such as "Therapeutic Drug Level Display" refer to concepts used by the clinical information system for summary result reporting. The Therapeutic Lab Display shows results for digoxin tests, as well as other tests not shown here, such as "NYH Lab Procedure: Magnesium." For clarity, some high-level intermediate hierarchical terms between "Medical Entity" and "Laboratory Diagnostic Procedure" and between "Medical Entity" and "Pharmacy Concept" have been omitted, as has the is-a link between Chemical" and "Substance." "NYH" stands for "New York Hospital", a part of New York Presbyterian Hospital. More information about the MED, including a browser, is available at http://onto.cpmc.columbia.edu/medsite/med1.htm.

# **ARTICLE IN PRESS**

J.J. Cimino | Journal of Biomedical Informatics xxx (2005) xxx-xxx

### 513 7. Whither the Desiderata?

514 Although the Desiderata might be construed to be some kind of commandments ("no false gods"-concept orienta-515 516 tion, "thou shalt not kill"-concept permanence, "honor 517 thy father and thy mother"-multiple hierarchies, and so 518 on), they were only a synthesis of contemporary thought. 519 At the time they were presented (at the 1997 IMIA Work-520 ing Group 6 conference, in Jacksonville, Florida), the only 521 objection raised was to the rejection of "not elsewhere classified" (NEC) terms [24]. Eight years later, at a subsequent 522 523 meeting in Rome of the same working group, the NEC 524 issue was not even raised, most likely because the general 525 consensus is now that such terms are antithetical to 526 ontologies.4

527 It is a given that our knowledge expands and evolves. 528 Our knowledge about knowledge is subject to this same 529 evolutionary force. But rather than discard what we have 530 learned to replace it with this alternative world view, I 531 believe that we can expand our understanding of how con-532 trolled biomedical terminologies might be further devel-533 oped to embrace both perspectives. Smith chooses a path 534 he calls "realism." One cynical definition of reality is 535 "The dream of a mad philosopher" [25]. A more balanced 536 definition is "the totality of real things and events; some-537 thing that is neither derivative nor dependent but exists 538 necessarily" [2]. I suggest a path that acknowledges the 539 importance of representing reality, as best we can know 540 it, but accepts the need for concepts to help us, among 541 other things, reason under uncertainty. I consider this the 542 realistic path.

543 In the realistic approach, terminologies contain terms 544 that refer to universals and to concepts, along with various 545 names and unique identifiers for these. Sometimes, a single 546 term will refer to an entity that has both universal and con-547 ceptual characteristics. Terminologies also contain, to the 548 fullest extent possible, ontological information to include what we know about the meanings of the terms and the 549 550 entities that they represent. That ontological information 551 for terms referring to concepts is, as Smith argues, prob-552 lematic; I argue that it is no more problematic than onto-553 logical information for terms referring to universals. In 554 any case, being problematic does not render such informa-555 tion valueless and should not dissuade us from including it 556 where we can.

557 We therefore consider some desirable characteristics of 558 controlled biomedical terminologies that address not the 559 structural and content issues of the original Desiderata, 560 but their purpose:

561 (1) Terminologies should support capturing what is known562 about the patient. This is at the level of what is actually

observed, not just how we interpret our observations or 563 what we infer. For example, when recording the medication given to a patient, we should record the specific 565 product—"Aspergum," for example, when it is known. 566 However, when such details are not know, we will need 567 the terminology to provide us with a more general 568 term—"aspirin preparation," for example. 569

(2) Terminologies should support retrieval. This has 570 implications for the how the terminology is used at the 571 time of data recording and at the time of querying. In 572 both settings, we should strive to make the meanings 573 of the terms universally understood; linguistic represen-574 tation should support, rather than obfuscate, this under-575 standing. For example, although "aspirin" is often a 576 shorthand form of the term "aspirin preparation," the 577 terminology should make the distinction between the 578 two terms clear to the person recording a patient's med-579 580 ications, such that someone later encountering the information in the patient's record will have to be able to 581 determine the meaning intended by the recorder. 582

(3) Terminologies should allow storage, retrieval, and 583 transfer of information with as little information loss as 584 possible. This has implications for how terminologies 585 evolve over time, while the data they are used to record 586 remain as frozen artifacts. Changes in terminologies 587 should not hamper our understanding of what was 588 stored on the side of the patient. For example, many 589 medical products that contained phenylpropanol-590 591 amine—a drug that has been prohibited by the US Food and Drug Administration-have continued to be manu-592 factured with a substitute ingredient (pseudoephedrine). 593 While the names of these medications have not changed, 594 595 the identifiers used to refer to them must change, so that we can know, from a patient's medical record, which 596 597 form of the medication the patient received.

(4) Terminologies should support aggregation of data. 598 While we want our terminologies to support those 599 who record data, we must recognize the legitimate needs 600 for abstraction of data, perhaps from multiple perspec-601 tives. For example, if we want to know which patients 602 are taking aspirin preparations, we will want to be able 603 to identify those patients whose records contain "Asper-604 gum" (or any of a large number of other specific prod-605 ucts), as well as those whose records merely show that 606 they are taking an "aspirin preparation." 607

(5) Terminologies should support reuse of data. Users of 608 609 data may wish to consider transformations other than simple aggregation, using what is known about the 610 terms by which the data are recorded. For example, if 611 we wish to know whether a patient is taking an anti-612 613 platelet agent, an antipyretic, an analgesic, or a nonste-614 roidal anti-inflammatory agent, we would want to be able to identify our Aspergum-taking patient as such. 615 (6) Terminologies should support inferencing. The knowl-616 edge underlying the terms used to record data should be 617 compatible with knowledge used for conceptual repre-618 sentations for reasoning (by humans and computers), 619

<sup>&</sup>lt;sup>4</sup> While the terms continue to exist, they are generally recognized to refer not to disease concepts or universals, but to instances of utterances made by clinicians when describing their beliefs and actions about a particular patient case.

8

7 December 2005 **Disk Used** 

676

Nathan (CE) / Karthikeyan (TE)

J.J. Cimino | Journal of Biomedical Informatics xxx (2005) xxx-xxx

620 such that the transformation from the former to the lat-621 ter can be accomplished. We need to be able to reach 622 across from what is on the side of the patient to use it 623 on the side of the clinician; terminologies can help. 624 For example, we would like to be able to use the knowl-625 edge that the (conceptual) condition "aspirin allergy" is 626 related to the chemical "aspirin" and, from that, infer 627 that we should be concerned about aspirin-allergic 628 patients (instances of a universal) who are given aspi-629 rin-containing products (instances of another universal).

630

631 If we can accept that the characteristics above are rea-632 sonable expectations for controlled biomedical terminologies, we can then proceed to determine how best to 633 634 realize them. We must recognize that, after all, everything 635 we say about the patient is, on some level, an abstraction 636 of reality and that how we record what we say-that is, 637 its context—is as important as what we say.

### 638 8. Conclusion

639 The original Desiderata paper discussed the entities repre-640 sented by controlled terminologies without reference to 641 ontologies, but it nevertheless reflected ontological princi-642 ples. While it referred to the terminologic entities as "concepts," it was describing desired characteristics of 643 universals as well. As long as we consider that the purpose 644 645 of terminologies is to support the recording and use of actual 646 data, rather than primarily as a pure knowledge base of what 647 is known in biomedicine, I believe that concepts and univer-648 sals can coexist and commingle in controlled terminologies, 649 to the advantage of those who seek to improve patient care 650 through symbolic representation of patient information.

### 651 Acknowledgments

652 I thank the IMIA Working Group 6 for the opportunity 653 to present this work in an intellectually stimulating setting. 654 In particular, I thank Dr. Barry Smith not only for his work on organizing the conference, but for his valuable 655 656 comments on earlier drafts of this paper. I also thank Mark 657 Musen and the anonymous JBI reviewers for their valuable 658 comments on the later draft of this paper.

### 659 References

- 660 [1] Cimino JJ. Desiderata for controlled medical vocabularies in the Twenty-First Century. Methods Inf Med 1998;37(4-5):394-403. 661
- 662 [2] Merriam-Webster, Incorporated. Merriam-Webster's Collegiate Dic-663 tionary, 11th ed. Springfield, MA: Merriam-Webster, Incorporated; 664 2005.
- 665 [3] Musen MA, Wieckert KE, Miller ET, Campbell KE, Fagan LM. 666 Development of a controlled medical terminology: knowledge 667 acquisition and knowledge representation. Methods Inf Med 668 1995;34(1-2):85-95.
- 669 [4] Schreiber G, Wielinga B, Jansweijer W. The KAKTUS View on the 670 'O' Word. Proceedings of IJCAI95 workshop on basic ontological 671 issues in knowledge sharing. Canada: Montreal; 1995.

- [5] Sim I. The "O" Word Revisited. <a href="http://smi-web.stanford.edu/">http://smi-web.stanford.edu/</a> 672 673 auslese/smi-web/archivedEventDetail.jsp?eventID=63>. 674
- Smith B. From Concepts to Clinical Reality: An Essay on the [6] Benchmarking of Biomedical Terminologies. J Biomed Inform; in 675 press.
- 677 [7] Campbell KE, Oliver DE, Spackman KA, Shortliffe EH. Represent-678 ing thoughts, words, and things in the UMLS. J Am Med Inform 679 Assoc 1998(5):421-31.
- 680 [8] Ledley RS, Lusted LB. Reasoning foundations of medical diagnosis; 681 symbolic logic, probability, and value theory aid our understanding of how physicians reason. Science 1959;130(3366):9-21. 682
- [9] Rector AL, Nowlan WA, Kay S. Foundations for an electronic 683 684 medical record. Methods Inf Med 1991;30(3):179-86.
- 685 [10] Weed LL. Medical records that guide and teach. N Engl J Med 686 1968;278(11):593-600.
- 687 [11] Barrows RC, Johnson SB. A data model that captures clinical 688 reasoning about patient problems. In: Gardner RM, editor. Proceed-689 ings of the nineteenth annual symposium on computer applications in 690 medical care. NewOrleans, LA: Hanley & Belfus, Philadelphia; 691 1995. p. 402-5.
- 692 [12] Ceusters W, Smith S. Strategies for referent tracking in electronic 693 health records. J Biomed Inform; in press.
- 694 [13] Jollis JG, Ancukiewicz M, DeLong ER, Pryor DB, Muhlbaier LH, 695 Mark DB. Discordance of databases designed for claims payment 696 versus clinical information systems. Implications for outcomes 697 research. Ann Intern Med 1993;119(8):844-50. 698
- [14] Wolff A, Begleiter A, Moskona D. A novel system of human J Dent 699 submandibular/sublingual saliva collection. Res 700 1997;76(11):1782-6.
- 701 [15] Lau LM, Warner HR. Performance of a diagnostic system (Iliad) as a 702 tool for quality assurance. Comput Biomed Res 1992;25(4):314-23.
- Patel VL, Arocha JF, Kaufman DR. Expertise and tacit knowledge 703 [16] 704 in medicine. In: Sternberg RJ, Horvath JA, editors. Tacit knowl-705 edge in professional practice: researcher and practitioner perspec-706 tives. Mahwah, NJ: Lawrence Erlbaum Associates, Publishers; 707 1999. p. 75-99. 708
- [17] Cimino JJ. Development of expertise in medical practice. In: Sternberg RJ, Horvath JA, editors. Tacit knowledge in professional 710 practice: researcher and practitioner perspectives. Mahwah, NJ: Lawrence Erlbaum Associates, Publishers; 1999. p. 101-20.
- [18] Li YC, Haug PJ, Warner HR. Automated transformation of 712 probabilistic knowledge for a medical diagnostic system. In: Ozbolt 714 JG, editor. Proceedings of the eighteenth annual symposium on 715 computer applications in medical care. Washington, DC Philadelphia: Hanley & Belfus; 1994. p. 765-9. 717
- [19] Cimino JJ. From data to knowledge through concept-oriented 718 terminologies: experience with the Medical Entities Dictionary. J Am Med Inform Assoc 2000;7(3):288–97.
- Pierre Zweigenbaum, Bruno Bachimont, Jacques Bouand, Jean 720 [20] Charlet, Jean-François Bosvieux. Le rôle du lexique sémantique et 721 722 de l'ontologie dans le traitement automatique de la langue médicale. 723 In: Le Beux P, Burgun A, editors. Actes du Colloque CRI-724 STAL'S. Saint-Malo; 1996. 725
- [21] Elhanan G, Cimino JJ. Controlled vocabulary and design of laboratory results displays. J Am Med Inform Assoc 1997;4(Suppl.):719-23.
- [22] Hripcsak G, Clayton PD, Jenders RA, Cimino JJ, Johnson SB. Design of a clinical event monitor. Comput Biomed Res 1996;29(3):194-221.
- 731 [23] Cimino JJ, Barnett GO. Automated translation between medical 732 terminologies using semantic definitions. MD Computing 733 1990;7(2):104-9. 734
- [24] Ingenerf J, Giere W. Concept-oriented standardization and statistics-735 oriented classification: continuing the classification versus nomenclature controversy. Methods Inf Med 1998;37(4-5):527-39.
- Bierce A. In: Schultz DE, Joshi ST, editors. The unabridged devil's [25] dictionary. Athens, GA: University of Georgia Press; 2000.

738 739

736

737

709

711

713

716

719

726

727

728

729