Pain Assessment Terminology in the NCBO BioPortal: Evaluation and Recommendations

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***Abstract*—The International Association for the Study of Pain (IASP) publishes since 1986 a relatively frequently updated list of pain terms currently known as the 'IASP Taxonomy'. It was examined how nine terms defined in this taxonomy and used by pain specialists to describe findings of somatosensory testing and pain assessment are classified in the representational artifacts accepted in the NCBO BioPortal. It was found that the majority of the BioPortal resources cover the terms poorly and that the quality of the hierarchies and the mappings are below acceptable quality standards. It is concluded that without the BioPortal studies of this nature are hard to perform, but also that for the BioPortal to become an instrument which is useful for other purposes than determining that its content is of poor quality, the internal quality assurance principles used for its development and maintenance need to be improved and documented.**

Keywords—pain terminology, NCBO BioPortal, quality assurance

I. Introduction

Findings based on the various kinds of responses that patients may report when subjected to stimuli to test their somatosensory status, are typically described using terms such as '*allodynia'*, '*hyperesthesia'*, and so forth. Standard definitions for these terms were first proposed in 1979 [1] and are since then regularly updated by the International Association for the Study of Pain (IASP), in print for the last time in 1994 [2], with more regular electronic updates on the IASP webpage [3], the last one May 2012 (subset in Table 1).

These definitions, together with the IASP definition for '*pain'* as '*an unpleasant sensory and emotional experience associated with actual or potential tissue damage, or described in terms of such damage*', suggest the hierarchy displayed in Fig. 1 in which terms displayed in **Small caps** are the immediate superordinate terms found in the definitions and the arrows stand for the classical subsumption relation. Although the individual definitions follow the Aristotelian style '*an A is a B which C*', the defined terms do not lead all together to a complete directed graph with an overarching top, not even if all 29 IASP terms would be included. Furthermore, the terms '*allodynia'* and '*hyperalgesia'* have superordinate terms which under their standard meanings should represent disjoined classes: although sensation and sensitivity are certainly related, nothing which ***is*** a kind of one can also be a kind of the other.

TABLE I. Pain terms analyzed

Allodynia: pain due to a stimulus that does not normally provoke pain. Note: The stimulus leads to an unexpectedly painful response.
Analgesia: absence of pain in response to stimulation which would normally be painful.
Dysesthesia: an unpleasant abnormal sensation, whether spontaneous or evoked. Note: Special cases of dysesthesia include hyperalgesia and allodynia.
Hyperalgesia: increased pain from a stimulus that normally provokes pain.
Hyperesthesia: increased sensitivity to stimulation, excluding the special senses. Note: Hyperesthesia includes both allodynia and hyperalgesia, but the more specific terms should be used wherever they are applicable.
Hyperpathia: a painful syndrome characterized by an abnormally painful reaction to a stimulus.
Hypoalgesia: diminished pain in response to a normally painful stimulus.
Hypoesthesia: decreased sensitivity to stimulation, excluding the special senses.
Paresthesia: an abnormal sensation, whether spontaneous or evoked. Note: it has been agreed to recommend that paresthesia be used to describe an abnormal sensation that is not unpleasant while dysesthesia be used preferentially for an abnormal sensation that is considered to be unpleasant. There is a sense in which, since paresthesia refers to abnormal sensations in general, it might include dysesthesia,

It is therefore not possible to use these definitions in the Ontology for Pain-Related Mental Health and Quality of Life (OPMQoL) which is being developed as part of the NIDCR-funded project R01DE021917 with the goal to integrate five datasets gathered in four different countries from patients suffering from one or other form of orofacial pain [4, 5].

It was hypothesized that alternatives could be found in the BioPortal of the National Center for Biomedical Ontology

Fig. 1. IASP pain assessment terminology hierarchy

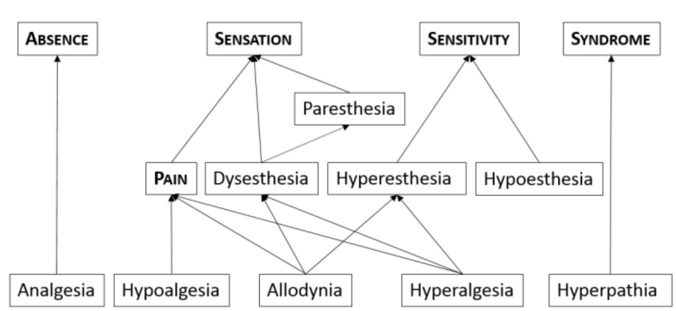

TABLE II. SUMMARY ASSESSMENT OF TERMINOLOGICAL AND ONTOLOGICAL QUALITY OF THE SEARCH TERM RELATED BIOPORTAL CLASSES RETRIEVED

Representational Artifact → / **Assessment Parameter**	**Norm**	**BDO**	**COSTART**	**CRISP**	**CSSO**	**CTCAE**	**DOID**	**GALEN**	**HP**	**ICD10CM**	**ICNP**	**ICPC2P**	**MEDDRA**	**MESH**	**MP**	**NCIT**	**NDFRT**	**NIFSTD**	**OMIM**	**PDQ**	**PHARE**	**RCD**	**RH-MESH**	**SNMI**	**SNOMEDCT**	**SOPHARM**	**SYMP**	**WHO-ART**	**Totals**
AP1 IASP search terms covered	9	1	6	2	1	2	1	0	5	1	2	2	8	3	4	4	4	1	1	0	1	5	3	7	9	4	6	2	
AP2 Number of direct class matches	>8	1	6	2	1	2	1	1	5	1	2	8	9	7	4	5	5	1	1	1	1	5	11	7	14	4	6	2	113
AP3 Direct classes with wrong IASP synonymy	0								1					2										1	3			2	9
AP4 Direct classes with definitions	=AP2			2	1	2	1		4					7	4	5	3	1									6		36
AP5 Number of direct classes with inappropriate homonymy	0											6											6		5				17
AP6 Number of additional direct classes through spelling variants	0	1											3			1						2	3						13
AP7 Number of class matches	>AP2	7	24	12	2	8	7	9	39	5	16	16	15	72	31	35	40	3	5	3	6	56	60	38	164	39	24	26	762
AP8 Foreign classes in hierarchy	0											8		7			5		1	1		5		7	6				40
AP9 Number of hierarchy classes with disjointness violations	0		7	4		2	4	1				6		15			11	1	3		4	5	2		49			24	60
Evaluation																													
Maximum number of norm violations		6	8	8	6	8	6	1	8	6	8	8	8	8	8	8	8	6	6	1	6	8	8	8	8	8	8	8	
Number of norm violations (except P8)		4	4	3	2	3	3	1	4	3	3	6	3	5	2	3	5	3	5	1	4	6	5	5	5	3	2	5	

(NCBO) [6] which contains to date 370 representational artifacts with over 5.6 million classes. The objectives of the work reported on here were to assess (1) whether these resources offer a more adequate view on pain assessment terminology, and (2) to what extent the BioPortal is a useful instrument in determining whether (1) is indeed the case.

II. METHODOLOGY

The nine terms – henceforth called '*search terms*' – from Table 1 were submitted to the on-line version of the BioPortal Annotator [7] thereby using the following annotator options: (1) 'longest match only' unselected, (2) manual mappings included, and (3) inclusion of all ancestors. With these options thus set, the annotator returned for each search term ***ST*** in this step one or more records, each such record containing (1) the unique identifier of a class ***CL*** in relation to which ***ST*** was found (2) the name of the representational artifact ***RA*** to which ***CL*** belongs, (3) whether ***CL*** was retrieved on the basis of what the annotator qualifies as a '*direct match*' between ***ST*** on the one hand and a preferred term, synonym or identifier of ***CL*** on the other hand, or on the basis of being – mostly within ***RA***, but occasionally also within a representational artifact other than ***RA*** – an *ancestor* of a class which matches directly, and (4) the preferred term ***PT*** of ***CL*** [8].

In a second step, all detailed terminological information available for each ***CL*** matching directly was retrieved, including a visualization of the subsumption graph and all the mappings – if any at all – of ***CL*** to classes in other representational artifacts within the BioPortal. The raw data and analysis file is available as [8]. Mappings between classes from different representational artifacts are further qualified by the BioPortal as being the result of enjoying shared Concept Unique Identifiers (CUIs) from the Unified Medical Language System (UMLS), and/or being automatically generated using the Lexical OWL Ontology Matcher (LOOM), which generates mappings based on lexical similarity of the preferred name and synonyms between pairs of ontologies [9].

To assess the extent to which the search terms are adequately covered in the individual BioPortal resources, and in the BioPortal as a whole, well-known quality assessment criteria and recommendations – see results and discussions for details – for terminologies [10, 11] and ontologies [12] were used. To assess the adequacy of the backbone hierarchy within individual resources 7 disjoint collections of in total 10 high level *groupings,* inspired by the various preferred terms that were retrieved, were constructed: [*Adverse event*], [*Body part*], [*Discipline*], [*Disease, Disorder or Finding*; *NON-pain disorder*; *Pain / sensation finding*], [*Pharm. Effect / Endpoint*], [*Function / Process*; *Technique / Therapy*] and [*Meta / Top*]. Each class (with disambiguation where required as for instance for 'analgesia') was classified into one of these groupings on the basis of its preferred term. Examples of classes labelled *Meta* are classes with preferred terms such as *Inactive Concept* and *Unclassified*, whereas the *Top* labelling include classes such as *Snomed CT Concept* and *Topical descriptor* [8].

The adequacy of the mappings between directly matched classes was assessed semi-automatically. Mapping records in which the semantics of at least one of the classes could not be determined, were excluded. Records where only one of the classes was marked as being *Meta*, were automatically tagged as obsolete. Records for which the preferred names of both classes were identical, except in the case of 'analgesia' given its homonymous semantics, were automatically assigned as being correct. All other cases were assessed manually.

III. RESULTS

Querying for the 9 search terms in the BioPortal Annotator exactly as displayed in Table 1 returned 762 annotation records of which 113 were about in total 104 candidate annotation classes labelled by the Annotator as 'direct' and which originated from 27 different sources [8] out of the 371 total artifacts at the time this work was performed. 17 annotation records revealed that in the ICPC2, RH-MeSH and SNOMED CT some of the search terms matched directly to more than one class (Table 3, AP5 in Table 2) – thus reflecting homonymy, while 9 records showed that some of the classes were mapped to by distinct search terms (AP3 in Table 2) – thus reflecting synonymy for the terms involved within the context of that source. Ignoring capitalization, the 104 direct annotation classes exhibited in total 25 distinct preferred terms. In Table 3 it is displayed how these preferred terms are related to the original search terms in each resource.

225 additional candidate annotation records [8] were retrieved by querying for three of the spelling variants suggested by some of the retrieved preferred terms obtained by querying for the original search terms (Table 3): 77 for *hyperaesthesia*, 76 for *hypesthesia*, and 72 for *hypoaesthesia*. These records reveal that these terms match directly with 14 classes that were not matched with the original search terms, thereby bringing ICD10 on board as extra representational artifact. These records are not included in any further analysis. 649 annotation records were labelled by the Annotator as containing hierarchical ancestors of the classes matched directly, totaling 206 distinct ancestor classes with together 169 distinct preferred terms [8]. One class, labelled '*UMLS:OrphanClass*' appeared in 40 records involving the 8 representational artifacts labeled ICPC2, MESH, NDFRT, OMIM, PDQ, RCD, SNMI, and SNOMEDCT. 1036 mapping records were retrieved for all 104 classes matched directly to the search terms, of which 71 duplicates, yielding 965 records further analyzed [8]. 399 of those records required manual assessment.

A. *Quality of BioPortal Resources Retrieved*

Table 2 provides – with the exception of assessment parameter AP8 – a summary assessment of the terminological and ontological quality of the classes (and by extension of the resources from which they originate) that were retrieved for the 9 search terms. Further details about certain aspects are available in Table 3 and Table 4. 9 APs are considered, and for each AP a norm is determined. Table 2 thus illustrates that:

- only SNOMED CT covers the 9 search terms in the lexical form provided by the IASP (AP1), while MeDDRA has complete coverage if lexical variants are taken into account (AP6), (it was not checked whether resources contained atomic terms that through post-coordination would allow to express the terms),
- 5 resources do not make the distinctions in terminology made by the IASP (AP3, details in Table 3),
- 11 resources provide textual definitions for at least some of the classes (AP2, AP4),
- 3 resources exhibit inappropriate homonymy for some of the search terms (AP5),
- more than half of the resources exhibit for at least some of the search terms a hierarchy which on the basis of the face value of the preferred terms is composed of disjoint classes (AP9, details in Table 4),
- none of the representational artifacts cover the domain delineated by the IASP search terms adequately when taking all assessment parameters into account.

TABLE III. MAPPING OF SEARCH TERMS TO PREFERRED TERMS IN THE REPRESENTATIONAL ARTIFACTS

Representational Artifact → / Search Term / Preferred Term	BDO	COSTART	CRISP	CSSO	CTCAE	DOID	GALEN	HP	ICD10CM	ICNP	ICPC2P	MEDDRA	MESH	MP	NCIT	NDFRT	NIFSTD	OMIM	PDQ	PHARE	RCD	RH-MESH	SNMI	SNOMEDCT	SOPHARM	SYMP	WHO-ART	Grand Total	Occurrence
Allodynia								1		1		1	1	1							1			1	1	1		9	9
Allodynia								1		1		1		1							1			1	1	1		8	8
Hyperalgesia													1															1	1
Analgesia		1	1			1	1					1	1	1	1	1			1	1	1	2	1	1	1			17	16
Analgesia			1				1					1	1	1						1		2	1		1			10	9
No sensitivity to pain																					1			1				2	2
pain agnosia						1																						1	1
Analgesia [PE]																1												1	1
Hypalgesia		1																										1	1
Pain Therapy															1				1									2	2
Dysesthesia					1			1				1	1		1								1	2				8	7
Dysesthesia					1			1				1			1								1	2				7	6
Paresthesia													1															1	1
Hyperalgesia		1	1							1		1	1	1	1	1	1				1	3	1	3	1	1	1	20	16
O/E - hyperesthesia present (& [hyperalgesia])																								2				2	1
Hyperalgesia [Disease/Finding]																1												1	1
Hyperalgesia		1	1							1		1	1	1	1		1				1	3	1	1	1	1		16	14
HYPERAESTHESIA																											1	1	1
Hyperesthesia		1						1	1		4	1	1			1						3	1	1		1		16	11
Hyperesthesia [Disease/Finding]																1												1	1
HYPERAESTHESIA											4																	4	1
Hyperesthesia		1						1	1			1	1									3	1	1		1		11	9
Hyperpathia												1									1		1	2				5	4
Hyperalgesia																							1	1				2	2
Hyperpathia												1									1			1				3	3
Hypoalgesia		1										1		1							1			1	1	1	1	8	8
HYPALGESIA		1																										1	1
HYPOAESTHESIA																											1	1	1
Hypoalgesia												1		1							1			1	1	1		6	6
Hypoesthesia		1						1				1	1		1	1							1	2		1		10	9
Hypesthesia [Disease/Finding]																1												1	1
Reduced sensation of skin																								1				1	1
Sensory impairment								1																				1	1
Hypesthesia		1											1											1				3	3
Hypoesthesia												1			1								1			1		4	4
Paresthesia	1	1		1	1			1			4	1	1		1	1		1				3	1	1		1		20	15
Paresthesia [Disease/Finding]																1												1	1
paraesthesia											4																	4	1
Paresthesia	1	1		1	1			1				1	1		1			1				3	1	1		1		15	13
Grand Total	1	6	2	1	2	1	1	5	1	2	8	9	7	4	5	5	1	1	1	1	5	11	7	14	4	6	2	113	
Occurrence	1	6	2	1	2	1	1	5	1	2	2	9	7	4	5	5	1	1	1	1	5	4	7	9	4	6	2		95

B. *Adequacy of the NCBO BioPortal*

Out of the 27 representational artifacts which have at least one class with a direct match to a search term, 22 have classes which by the BioPortal are mapped to at least one other class from another artifact. 618 of these mappings are within these 22 sources whereas 347 mappings are towards classes from 18 target representational artifacts outside these sources. Of these 18, MeDDRA and RH-MeSH are the only two that have classes directly matched with the search terms, thus reflecting the BioPortal documentation that mappings are not always bidirectional.

Table 5 quantifies the appropriateness of the mappings on the basis of our methodology. The 'B' and 'T' following the resource names in Table 5 indicate whether the resource exhibits mappings bi-directionally resp. only incoming. B-mappings are only counted once in the totals. Mappings are qualified as being excluded ('Excl.') from the analysis because of either ambiguity or missing information on the side of the classes mapped to ('T?') or being in the realm of the 22 source classifications ('S?'). 'Correct' mappings result from (1) the automatic assignment of the adequacy assessment for pairs of source and target classes with identical non-ambiguous preferred terms ('SAME'), and the manual verification of (2) classes with synonymous preferred terms, i.e. lexical variants or descriptions ('VARIANT') and (3) classes with ambiguous preferred terms. Erroneous mappings ('ERROR') are brought about by (1) automatic determination of mapping to or from inactive classes ('OBSO') and manual verification of (2a) mapping to or from classes with ambiguous meaning ('HOMONYM'), and (2b) inappropriate mappings between classes with unambiguous meanings ('WRONG'). Table 6 provides insight in the accuracy of the methods applied in the BioPortal to create mappings, i.e. whether on the basis of the UMLS Concept Unique Identifiers ('cui'), the LOOM algorithm ('loom') or both.

IV. DISCUSSION

During 'The Consensus Workshop: Convergence on an Orofacial Pain Taxonomy', held March 30 – April 1, 2009, Miami, Florida, which was attended by representatives from all major pain institutions, it was concluded that an adequate treatment of the ontology of pain together with an appropriate terminology, is mandatory to advance the state of the art in diagnosis, treatment and prevention [13].

As a first step, it was proposed to study the terminology and ontology of pain as currently defined. The ontological aspects have since then been covered in [4], and the underlying principles thereof been applied, for instance, in the definition of new pain-related disease entities and classifications [14, 15].

TABLE IV. GROUPING OF THE SEARCH TERMS IN DISJOINT UPPER CLASSES IN THE HIERARCHY OF THE REPRESENTATIONAL ARTIFACTS

Representational Artifact → / Search Term, Grouping	BDO	COSTART	CRISP	CSSO	CTCAE	DOID	GALEN	HP	ICD10CM	ICNP	ICPC2P	MEDDRA	MESH	MP	NCIT	NDFRT	NIFSTD	OMIM	PDQ	PHARE	RCD	RH-MESH	SNMI	SNOMEDCT	SOPHARM	SYMP	WHO-ART	Grand Total
Allodynia								7		8		4	11	8							11			15	10	5		79
Disease or Finding								6		4		4	8	8							9			11	10	5		65
Function / Process										4																		4
Meta / Top								1					3								2			4				10
Analgesia		4	5			7	9					1	6	7	6	8			3	6	11	5	6	9	9			102
Body part		1																										1
Discipline																				3								3
Disease or Finding		3	2			3	1					1	1	7						2	9	3	3	9	9			53
NON-pain disorder						4																						4
Technique / Therapy			3										1		3				2									9
Function / Process							6								3								2					11
Meta / Top							2						3			2			1		2		1					11
Pharm. Eff./Endpoint													1			6				1		2						10
Dysesthesia					4			9				1	11		8								5	22				60
Adverse event					1																							1
Disease or Finding					2			8				1	8		8								2	13				42
Function / Process																							2					2
Meta / Top					1			1					3										1	9				15
Hyperalgesia		4	7							8		1	11	8	5	8	3				14	19	6	29	10	4	13	150
Body part		1																										1
Discipline			4																									4
Disease or Finding		3	3							4		1	8	8	5	6	1				12	19	3	15	10	4	1	103
NON-pain disorder																											12	12
Function / Process										4													2					6
Meta / Top													3			2	2				2		1	14				24
Hyperesthesia		4						8	5		8	1	11			8						18	5	14		4		86
Body part		1																										1
Disease or Finding		3						7	4		4	1	8			6						18	2	10		4		67
Function / Process																							2					2
Meta / Top								1	1		4		3			2							1	4				16
Hyperpathia												4									10		6	23				43
Disease or Finding												4									8		3	14				29
Function / Process																							2					2
Meta / Top																					2		1	9				12
Hypoalgesia		4										1		8							10			17	10	4	13	67
Body part		1																										1
Disease or Finding		3										1		8							8			13	10	4	1	48
NON-pain disorder																											12	12
Meta / Top																					2			4				6
Hypoesthesia		4						7				1	11		8	8							5	20		4		68
Body part		1																										1
Disease or Finding		3						6				1	8		8	6							2	11		4		49
Function / Process																							2					2
Meta / Top								1					3			2							1	9				16
Paresthesia	7	4		2	4			8			8	1	11		8	8		5				18	5	15		3		107
Adverse event					1																							1
Body part		2																2										4
Discipline																		1										1
Disease or Finding	6	2		2	2			7			4	1	8		8	6		1				18	2	11		3		81
Function / Process																							2					2
Meta / Top	1				1			1			4		3			2		1					1	4				18
Grand Total	7	24	12	2	8	7	9	39	5	16	16	15	72	31	35	40	3	5	3	6	56	60	38	164	39	24	26	762

TABLE V. CORRECTNES OF DIRECT CLASS MAPPINGS

Representational Artifacts		Error			Correct			Excl.			
		WRONG	OBSO	HOMONYM	SAME	VARIANT	DISAMBIG.	S?	T?	TOTAL	% WRONG
ACGT-MO	T	1								1	100
AI-RHEUM	T	1								1	100
BDO	B		3		20	5			3	31	10.7
COSTART	B	31	12		44	40			44	171	34.4
CRISP	B	7	4		17	2		19	21	70	36.7
CSSO	B	1	2		17	8			3	31	10.7
CTCAE	B		4		19	4			4	31	14.8
GALEN	B	5		9	1	7	6	1	3	32	50
HIMC-ICD09	T		2						9	11	100
HIMC-LOINC	T								3	3	0
HL7	T					4		2		6	0
HOM-CLINIC	T								3	3	0
HOMERUN-UHC	T								3	3	0
HP	B	7	2		19	2			4	34	30
ICD10	T		1		2	3				6	16.7
ICD10CM	B		2		7	6			4	19	13.3
ICPC2P	B	1	6		21	48		1	10	87	9.21
IFAR	T					2				2	0
LOINC	T					6		3		9	0
MEDDRA	T		15						122	137	100
MESH	B	2	8	5	44	13	4	1	16	93	19.7
MP	B	6	6	13	29	8	6	1	7	76	36.8
NCIT	B	9	9	4	43	23	3	1	23	115	24.2
NCIt-Activity	T	2			2					4	50
NDFRT	B	5	10	8		60	1	1	28	113	27.4
NDF-RT	T	2	4	7	24	8	1	1	2	49	37
NIFSTD	B	2	2		17	4			2	27	16
OMIM	B	3	3		18	9			2	35	18.2
PDQ	B	3		4	4	8	3	1	11	34	31.8
PHARE	B	2		11			8	1	3	25	61.9
PMA	T					2		1		3	0
RCD	B	9	10	5	36	24	4	1	11	100	27.3
RH-MESH	T		12						86	98	100
RPO	T	2				2				4	50
SNOMEDCT	B	6	150	3	2	15	4	1	4	185	88.3
SOPHARM	B	6	8	13	29	4	6	1	10	77	40.9
SYMP	B	7	14		55	17			18	111	22.6
SYN	T	2			2					4	50
TRAK	T					2		1		3	0
WHO-ART	B	34	7		10	18			17	86	59.4
Grand Total		**78**	**148**	**41**	**241**	**177**	**23**	**19**	**238**	**965**	**37.7**
% of mappings		**27.67**			**45.70**			**26.63**			

The analysis performed here is another response to the workshop's recommendations with the goal to obtain more insight in how pain assessment terminology is dealt with in representational artifacts such as widely used classification systems, terminologies, and ontologies. At the same time, it provided an opportunity to assess the usability of the NCBO BioPortal for a task of this nature, and the appropriateness of the principles and methods applied in the BioPortal to present a unified, highly standardized and ontology-like view on resources which are qua structure and underlying design principles very different.

A. Are Resources in the BioPortal intrinsically flawed

As can be inferred from Table 2 and Table 3, all retrieved resources, with – at first sight – the exception of MeDDRA and SNOMED CT, seem to perform quite poorly in terms of coverage of the domain. Of course, some resources might have been designed with a specific purpose in mind and pain assessment terminology therefor being out of their scope. It is however hard to imagine for what sort of purpose a term such as *paresthesia* might be relevant and *dysesthesia* not: if one is present, all should be present. An exception is *analgesia* in the sense of a procedure rather than of a symptom: there would indeed be no place for any of the other terms in procedure terminologies. Although there are indeed a few resources retrieved for which *analgesia* is the only term matched, these resources are not restricted to procedures. Some resources turn out to exhibit a better coverage when spelling variants are used in the queries, but not to the extent that it can explain the overall lack of coverage.

Some resources, such as COSTART, MeSH and WHO-ART, suffer from the lack of discrimination between terms in pairs such as hypoalgesia/hypesthesia, hyperalgesia/hyper-esthesia, dysesthesia/paresthesia and analgesia/hypoalgesia. This was also found in SNOMED CT but only for classes that were labelled 'inactive' thus reflecting that these mistakes made in earlier versions were corrected afterwards.

15 resources exhibit through the eyes of the BioPortal a backbone structure which at least can be frowned upon (Table 4). How can *analgesia* be a kind of *nervous system* (COSTART), *communication disorder* (DOID - Human Disease Ontology), or *pharmacogenomics* (PHARE)? How can *paresthesia* be a kind of *peripheral nervous system* (OMIM), *hyperalgesia* a kind of *adrenal adenoma* (WHO-ART) or *neuroscience* (CRISP)? One can assume sloppy design on the side of the authors of these resources, or violation of the principle that preferred terms should have face validity [10]: thus in COSTART 'nervous system' might not mean *nervous system*, but rather *symptom related to the nervous system*. Or, and this leads to the next section, perhaps the BioPortal represents the structure of these resources erroneously?

B. *Is the BioPortal itself, or are some design or quality assurance principles behind it, intrinsically flawed?*

That something wasn't right with the representation of WHO-ART in the BioPortal was noted by Ruttenberg in 2011 and as such acknowledged by BioPortal staff who traced the issue down to be caused by the WHO-ART source codes, but nevertheless decided nothing to do about it at that time [16]. And apparently never since: the version of WHO-ART that showed up in the work reported about in this paper was version '2013AB' which was uploaded to the BioPortal, according to the summary page, February 18, 2014, indeed without any attention to the known issues. The data presented here demonstrate further that it is not just WHO-ART of which the representation in the BioPortal is problematic with respect to the semantics of the subclass relationship, but also 14 other resources that were retrieved on the basis of the search terms (Table 2, AP9).

Another indication that the BioPortal could benefit from some quality assurance introspection comes from the finding that for 8 of the 27 resources retrieved the Annotator returned 'UMLS:OrphanClass' as ancestor for 40 of the classes matched directly (Table 2, AP8).

Also the mapping results provide serious evidence in the direction that quality improvement is required.

TABLE VI. MAPPING SOURCES

Result	cui	cui, loom	loom	Grand Total
Error	29	20	218	**267**
WRONG	5	4	69	**78**
OBSO	24	16	108	**148**
HOMONYM			41	**41**
Correct	50	23	368	**441**
SAME	2	9	230	**241**
VARIANT	48	14	115	**177**
DISAMBIG.			23	**23**
Excluded	22	17	218	**257**
S?			31	**31**
T?	22	17	187	**226**
Grand Total	**101**	**60**	**804**	**965**
% Wrong	**36.71**	**46.51**	**37.20**	**37.71**

First there is the observation that through the mappings, 16 additional resources were discovered that contain classes which map directly to classes which were retrieved by means of the search terms. This can in part be explained by the absence of the search terms in the synonym set of these additional classes, but upon further inspection, it turns out that in case of in total 255 mappings for RH-MeSH and MeDDRA, as well as for (possible) resources which according to the syntax of the URIs of the classes mapped to might be named 'HOMERUN-UHC', 'HOM-CLINIC', 'HIMC-LOINC' and 'HIMC-ICD09', the URIs returned by the annotator do not resolve at all [8]. The former 4 resources are also not listed on the BioPortal webpage as being resources it contains, yet classes from them show up in the mapping results. In case of SNOMED CT, mappings are primarily involving classes which are marked as 'inactive'.

A second observation is that – after excluding these 255 mappings as well as two others for which the meaning of the source class could not be disambiguated – still almost 38% of the mappings are inaccurate. There is no significant difference in accuracy between mappings produced using LOOM or UMLS CUIs alone. However, when both the LOOM and CUI-methods suggest a mapping, the error rate increases to over 46%, thus almost the equivalent of flipping a coin.

C. *Limitations*

The work reported on here bears certain limitations. Although the data demonstrate (1) that the domain of pain assessment terminology is poorly covered in the BioPortal resources, (2) that the way in which the BioPortal organizes the retrieved classes hierarchically using the subclass relation is debatable, and (3) that the techniques used to map these classes between resources are not quite adequate, no generalizations can be made to other domains. A further limitation is that the data were retrieved using the BioPortal website rather than the REST services. Perhaps these services offer better ways to filter inadequate data, but if that were the case, one could wonder why such filters are not used on the website.

Assessment of the correctness of the suggested hierarchy and the mappings was carried out with the quality criteria of the OBO Foundry and adherence to the principles of Ontological Realism in mind, neither of which are universally accepted [17] yet gaining considerable attraction [18]. Thus it is quite conceivable that reviewers outside the Foundry would report lower error rates, for instance by finding it perfectly acceptable that the 'concept' of analgesia as a pharmaceutical effect in some drug is considered equivalent to the 'concept' of analgesia as a procedure performed by an anesthesiologist or as a state of a patient brought about by such procedure. At the other hand, since the review here was based by first flagging results that for sure require manual evaluation (see methodology) it might very well be that certain mapping- or ancestor records were erroneously not flagged. In that sense, the error rates presented here could very well be – modulo mistakes made by sloppiness of the reviewer – the best case scenario. Another limitation is that this study does point out the kind of mistakes and how to find them semi-automatically, but is not conclusive on whether the root cause is in the source systems, the BioPortal, or a combination of both.

V. Conclusion and Recommendations

Without doubt, studies such as this one could not be carried out without a resource such as the BioPortal, or would require a lot more time and effort. Evenly without doubt, the BioPortal made it possible to reach the objectives of this study which were to find out (1) whether the sources in the BioPortal provide a more adequate view on pain assessment terminology – the answer being ***no***, and (2) to what extent the BioPortal itself is a useful instrument in determining whether (1) is indeed the case – the answer being ***yes***. As a side effect, this study raises serious questions about the quality assurance principles employed in the design and management of the BioPortal, more specifically (1) about the quality of the resources the BioPortal accepts for inclusion – it might seem unfair to criticize a lack of clear best practice policies in the investigated resources while not distinguishing their different semantic expressivity, the point being however that the BioPortal itself does not allow for such distinctions and 'promotes' all resources as ontologies, (2) the suitability of representing the hierarchy of these resources by means of the subclass relation, and (3) about certain house-keeping operations. Quality seems thus far not to have been much of a concern to the BioPortal scientific community, as witnessed by the presence of only one paper in Pubmed that addresses the topic [19]. Furthermore, although the BioPortal does indeed offer a mechanism to users to make notes on the quality of BioPortal content [6], it doesn't seem to be used much: the BioPortal homepage displays a list of the 5 last notes submitted, of which the last three were submitted 7 months prior to writing this paper, all three about a 'request' issued by user *rboden* – noted in the name of 'Jesus' as contact person – to add the following new term '*We need someone with qualifications*'. It is a bad sign that spam of this kind, whether unnoticed or noted but not acted upon, is accepted.

For the BioPortal to become an instrument which is useful for other purposes than determining that its content is of poor quality the following suggestions are in order: (1) do not accept resources that violate standard subsumption principles, (2) display for each resource quality metrics, rather than mere quantity metrics, for instance the extent to which they follow the principles of ontological realism or the OBO Foundry, and (3) provide better documentation about the methods and algorithms used to present hierarchies and mappings, and about the internal quality assurance principles.

Acknowledgment

The work described is funded in part by grant 1R01DE021917-01A1 from the National Institute of Dental and Craniofacial Research (NIDCR). The content of this paper is the responsibility of the author and does not necessarily represent the official views of the NIDCR or the NIH.

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