

A SOCIO-TECHNICAL PERSPECTIVE ON ONTOLOGY DEVELOPMENT IN HEALTHGRIDS

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Abstract

The paper focuses on the alignment of technical and human infrastructures in the semantic web, taking the development and alignment of ontologies in eHealth as a vehicle for exploring the barriers to achieving this. The authors report on the outcomes of a recent eScience workshop looking at data integration and ontology alignment across Grid projects in the same disease domain. It is premised on a view of Grids as socio-technical systems aligning technical and human networks in ways which can create useful synergies, but may also work at cross purposes, generating unpredicted costs and risks and barriers.

1. Socio-technical Alignment

The eHealth vision of large-scale, seamless data-sharing for research and clinical trials faces real barriers in the development of shared infrastructures for collecting, coding, cleaning and representing data across multiple distributed communities of practice. Ontology development is a significant strand in that vision, premised on semantic alignment, and the collective definition and representation of terms and frames of reference across communities. We look here at some of the more persistent semantic and systemic alignment issues in the context of neuro-psychiatric disease, as experienced by Grid projects working in this domain in the UK, EU and US, where the opportunities and the challenges of integration have been particularly evident.

The paper takes a perspective on Grids and other complex, distributed networks as co-evolving socio-technical systems [Joslyn, 1999; Kling 2003]. Their design can be seen as an opportunity to align technical and human networks to create useful synergies, as for example in the case of many Web 2.0 applications, or as a challenge, where technical and distributed human infrastructures may work at cross purposes to generate unpredicted costs and risk [Luna-Reyes 2005].

The authors draw on the outcomes of a UK eScience funded workshop bringing together multiple Grid projects in the UK, EU and US to roadmap the issues in data integration and ontology alignment in HealthGrids working in the same disease domain [Ure et al 2007].

The presentations and a shared wiki are available on the UK e-Science site¹.

The event was co-hosted by the National e-Science Centre, the Generation Scotland national population genomics project² and the NeuroGrid³ project developing a platform for sharing clinical and brain imaging data relating to schizophrenia, dementia and stroke. This also built on the ongoing road-mapping process to support collaboration in data sharing in particular disease domains initiated in the HealthGrid Share project⁴. Conclusions are also derived from a recent eSocial Science workshop on ontology development in the Humanities and the Social Sciences Grids [Lin Y. and Procter R. 2007], as well as other published work on data integration and semantic alignment in eHealthGrids [Breton 2006; Wilson and Lessens 2006] and in the semantic web [Goble 2006; de Roure 2006] more generally.

1.2 Issues in Ontology Harmonisation

Gruber [1993] uses the term ontology to mean a specification of a conceptualization, and as 'a description of the concepts and relationships that can exist for an agent or a community of agents.' This formal specification is dependent on the agreement of common terms, referents and relationships, however

¹ https://wikis.nesc.ac.uk/mod/Main_Page

² www.generationscotland.org

³ www.neurogrid.ac.uk

⁴ <http://www.eu-share.org/>

there is also a need for harmonization of the protocols, tools and other processes through which that data is derived.

These are increasingly issues of concern given the vision of international, multi-centre clinical trials, translational medicine, and interoperability in the European Health Area. Several Grid projects now operate in the same disease domains, and in taking one such disease domain (schizophrenia) as a test case it was possible to engage clinicians, ontologists, imaging scientists, e-scientists, and geneticists on real-world issues in the integration of heterogeneous datasets from diverse sites, populations and working contexts.

2. Issues in Alignment

The data ‘supply chain’ underpinning eHealth and translational medicine is a gradual conversion process where many types of error or bias can arise at different stages from sampling, collection, coding, aggregation, analysis or use, sometimes referred to as the ‘social life of information’ [Duguid, 2000].

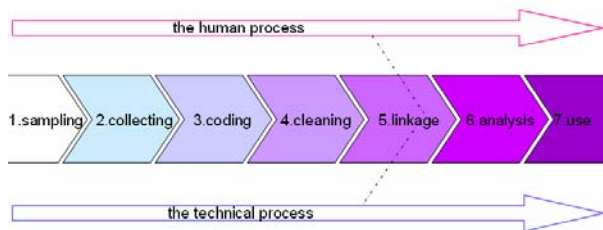


Fig. 1 Socio-technical Alignment Scenarios in eHealth

At the level of the data itself, there were many examples of real challenges in ensuring that this could be integrated without introducing confounding effects. These derived from differences in the make or calibration of equipment for example, differences in the interpretation of scan data or psychiatric tests for example, or differences in test populations. Many of the challenges in aligning or integrating distributed datasets, and their associated metadata derive from the very diverse contexts in which they are collected, shared and used. In the first instance we will outline some of these issues in aligning data, metadata and ontologies across multiple HealthGrids. The discussion section will then pull together some of the implications of this for ontology-building, drawing also on the experiences of the ‘Building Building Ontologies for the Humanities and Social Sciences’ Workshop at the National Centre for eSocial Science (NCeSS)⁵.

2.1 Across Communities

⁵ <http://www.ncess.ac.uk/events/ASW/ontologies/>

Within the same disease domain, concepts were different in scope, conceptualization, scale and semantics in the different projects, and an emerging consensus was to consider identifying core measures of symptom as a starting point for alignment, rather than attempting to align disease concepts in the first instance. In addition to this, within projects teams there were difficulties in achieving domain models that met the criteria of both clinicians and ontologists, with disengagement by clinicians from the process typically reported.

2.2 Across the Project Lifecycle

A number of projects reported problems associated with the way metadata (and requirements) evolved and changed significantly in the course of the project. Extending or changing metadata fields is hard [Pleiter 2006], and can generate a cycle of cost / delay and impacts negatively on system design from a technical perspective. It also generates a degree of hostility in some cases to users or intermediaries. In this it mirrors the problems associated with ‘requirements creep’ so often reported in the design of business systems.

2.3 Across Populations

Many of the datasets were drawn from regional or national studies from very different populations, raising the risk of confounding artifact with effect. In the case of neuroscience, for example, there are physiological differences in brain shape and size that reflect ethnic differences rather than disease effects. Establishing such differences is difficult in practice, and often impossible in retrospective studies. Statistical epidemiology provides some strategies for estimating and obviating some of these risks, however there were serious questions about the value of aggregated population data where effects are small or difficult to disentangle.

2.4 Across Sites: Collection, Coding and Collation

The practical experience of project managers and ontologies consistently highlighted the difficulty of ensuring the quality and consistency of collection, coding and collation, all of which were subject to significant rates of human error, misinterpretation or site-specific effects that were not directly.

Known error rates of 30% were reported as not uncommon when matching test data against patient data, raising serious questions about conclusions from multi-site studies involving small effects. Many of the problems related to the quality, sequence, correctness or completeness of the data originally filled in. For example, if the patient’s age is recorded differently on different files. Are these files of the same patient or not?

Protocols were often interpreted in different ways, and local events impacted on the administration of the test. In aggregated data, without local information, anomalies were hard to identify. Strategies for addressing this included wireless notepads or pens used at the data upload stage, so that data incorrectly loaded was automatically validated against the main database as it was stored. As indicated earlier, error trapping software was also used, together with metrics using probabilistic linkage.

It was also seen as important to keep links to raw data (or data owners) where possible. Metadata on provenance was seen as very important here [Goble 2006]. Cleaning and error trapping software can only capture particular types of error and many anomalies could only be recognized by those with local knowledge of the population, the context and the method of data collection on the ground. e.g. one study cited measures of heart rate captured in one site which were consistently higher in one site, and where this was consonant with known population differences. Data quality evaluation between sites in the study highlighted the fact that participants in one site had to climb six flights of stairs before the ('resting') heart rate test while the lift in the hospital was out of order. This would not have been evident from inspection of the data alone but required local input. Guidelines, checklists and toolkits were also used for enhancing data quality.

Interestingly, the same communities whose unique contexts generated these differences were seen by many as particularly well placed to evaluate, quality assure and enhance it. In this context, one study proposed to have a panel responsible for ethics, linkage and data quality issues mediating requests for data as well as submission of data, building on their ability to interpret and also act on local processes [Smith 2006; McGilchrist 2007].

It was clear here also that semantic alignment of data, and metadata as the basis for ontologies was of limited use where the underlying measurement processes were subject to differences in protocols, or equipment such as scanners. There was then a need for harmonization between different types of scanner, differences in the processes of registration or normalization, differences in the settings (even after servicing) etc. Similarly, in genetic analyses, results can be obtained by a range of different methods, such as micro-array, in situ hybridization, and immunocytochemistry, thus raising the possibility that differences between datasets may be a function of the testing and analysis process itself. A range of strategies were adopted here, including ongoing harmonization /data quality testing between sites, use of shared 'phantoms' as controls and use of common tools. In a number of other projects, early prototyping also provided a vehicle for community engagement and a

number of harmonisation studies were underway within and between participating Grids.

2.5 Across Scales

Neuroscientists now have access to a vast array of large, heterogeneous and multi-dimensional data from multiple sources, and across multiple scales. Data at molecular level on synaptic proteins involved in human mental illness, such as schizophrenia, bipolar disorder and mental retardation [Martone 2004] is even more valuable when integrated with scanning data, and genetic data yet this requires coordination in a spatial/anatomical frame of reference, using a shared data model, and ideally a human and machine readable format.

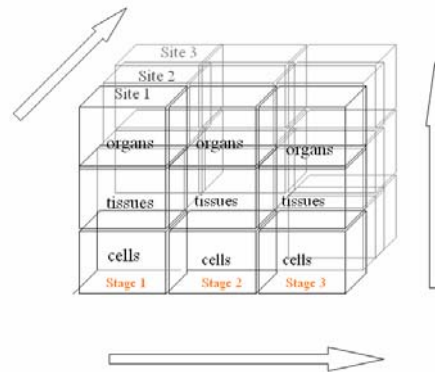


Fig. 2 Data integration across sites and scales

Much as existing pieces in a jigsaw can support new insights about the structure as a whole, and the missing parts of it, the aggregation of disparate information within a shared model can support both interoperability and understanding if there is sufficient opportunity and motivation for joint working. Integrating heterogeneous and distributed datasets is therefore a priority area for regional, national and international bodies supporting research in e-Health and e-Science. The challenge with heterogeneous and distributed data is to integrating data and information in a shared infrastructure that will allow human and machine users to make sense of it [De Roure 2003; Rector 2000] in relation to familiar frames of reference

3. Ontology Alignment Strategies – a socio-technical perspective

Ontology alignment studies, both technical and socio-technical, have been under-researched to date, both from the perspective of logical compatibility of Owl based ontologies [Cuenca et al 2006], and from the more socio-political perspective of forging agreement between disparate communities which is required to

underpin it. The issues are further confused by the difficulty of separating biomedical terms representing invariant concepts that can properly be treated as ontological classes, from other terms relating to local perception, measurement, understanding or use of these which are more epistemological, and therefore do not lend themselves to the kinds of logical classification and extension that are sought [Bodenreider, 2004]. We concentrate here on the latter, and draw analogies with strategies from other collaborative, distributed contexts such as the extended enterprise.

3.1 Open Source Collaboration

The approach adopted in the BIRN project, and co-opted by the collaborating Grids at the workshops was to facilitate collective development of the underlying semantics, and conceptual relationships using open source resources such as the BirnLex⁶ and Firework concept browser tools available through the BIRN Bioinformatics, Research Network project.

The strategic alliancing of disparate resources to common ends - has been increasingly seen as a means of adding value, cutting costs, benchmarking approaches and sharing risk towards in the development of many other socio-technical systems [Kling 2003] including Web2.0 applications. Leveraging wiki technology and distributed community knowledge provides a rationale for collaboration that works on several levels, providing a process for achieving critical mass, for de facto implementation of a standard and for leveraging distributed knowledge in synergistic ways.

3.2 Mediation

Bridging solutions included ontology mediators (such as the BIRN Mediator) mapping between existing ontologies, although many reported significant problems in practice. The use of shared tools and shared lexicons as free resources is increasingly a strategy, and obviated practical problems with sharing where members of consortia were using licensed or incompatible software. The BIRN project was the best example of this. The shared lexicon (BIRNLex) generated by BIRN collaborators, for example, provided standardized terms to describe subject information, neuroanatomy, molecular species, behavioral and cognitive processes, experimental practice and design, and associated elements of primary data provenance. The project also made available a software tool for browsing, extending, or assembling a set of existing ontologies/terminologies together with the latest version of the UMLS and NeuroNames terminologies.

3.3 Strategic alliancing: building around existing cognitive and social systems⁷

The success of Web2.0 applications underlines the effectiveness and intuitive usability of building on existing cognitive and social dynamics and frames of references. Socio-technical alignment can also generate useful synergies, leveraging the potential of diverse communities to adapt, innovate, validate, update and reconfigure around local requirements and using local knowledge.

Business models increasingly cite such approaches as a means of adding value, and enhancing usability [Sawhney and Parikh 2001]. Designers of systems dependent on speed, cost or quality (e.g. safety systems, P2P applications and those geared to transient e-business opportunities) have been early adopters of such strategies. One recurring strategy is the adoption of a shared core, aligned with multiple local domains.

3.4 Core: local strategies

A number of Grid projects have adopted a compromise approach. The Policy Grid project [Edwards 2006] provides an example of an eSocial Science Grid which deals with the tension between global and local factors by combining a fairly invariant core, with local variations based on the use of local folksonomies. Others have simply restricted the ontology to very well established and widely agreed concepts, and avoided attempts to encapsulate conceptual interpretations [Chapman 2007]. These run counter to the notion of a standard or static domain ontology and highlights concerns that ontology-driven interfaces might constrain social scientists within pre-specified frameworks. This has parallels with similar debates in requirements engineering. Shantenu Jha [2007] provides examples from Cactus Grid of the core: local approach, balancing the need for local purpose specific extensibility with a stable core. The design challenge in this context is then to find a means of harnessing the diversity of local resources as successfully as many social networking sites have done.

3.5 Agent /User Centred Strategies

Motta [2007] describes a shift from developing 'a centrally designed, monolithic ontology towards an automatic integration of ontology fragments, sourced from the semantic web....relevant to the current user need....and performing both ontology mapping and co-reference resolution on the fly', which has been termed faceted ontologies [Suominen 2007].

The concept of the single ontology can be seen as running counter to the concept of knowledge in research as a fluid, negotiated entity derived from

⁶ <http://www.nbirn.net/birnlex/index.htm>

ongoing challenge and renegotiation in distributed social contexts. Sacrificing local requirements in the interests of scalability, can result in a loss of currency, usability and relevance, making them increasingly unlikely to be adopted or maintained.

4. Ontology-Building Strategies

Discussions arising from the workshop series suggest that technical infrastructures can outstrip the human infrastructures required to agree and implement shared understandings, shared semantic representations, common collection or coding practices or ethical and legal governance of patient data across these different constituencies. This is a process which requires the explicit provision of formal and informal opportunities for collaboration to take place, and perceived rewards or benefits for so doing [Nonaka 2001, Wenger 1998]. Ontology-building, on the basis of shared definitions and representations of meaning, is necessarily dependent on this kind of collaboration process.

4.1 Ontology-building as a Socio-technical Enterprise

A key task recommended by the collaborating group from the multi-Grid workshop was to achieve agreement on core measures of symptom for psychoses. Not only are there different measures of symptoms, in relation to the data sets held, the same symptoms can be associated with different formal diagnoses and treatment recommendations. This provided another example of the need to go beyond the purely semantic to cultural and philosophical conceptualizations of disease. As in action research, the process of collaborative definition and discussion seemed itself to be an integral part of a process of change, accommodation and constituency –building.

Effective ontology building will depend on both opportunities for these new multi-local constituencies to work together, and incentives for doing so. The UK e-Science and e-Social Science Centres provide for collaboration and virtual collaboration infrastructure, [See Sattler 2006, Lin and Procter 2007; Ure 2007]. Currently, however, this is often additional effort, for which the rewards are not clearly defined. Clinicians for example, may be unwilling to invest time in developing approaches to share data with other groups with whom they compete for funding, for example, or where the rewards for devoting time to publishing research within their own constituency are much more tangible.

4.2 Ontology building as Knowledge Representation

Concepts of the nature of knowledge and knowledge building across distributed constituencies are relevant to such discussion, but are very often implicit rather than explicit in ontology development in eHealth. Both Dupre [2006] and Bodenreider [2004] points to some of

these difficulties in the bio-medical domain. Dupre points out the difficulty of separating facts and values, particularly in the nature of biomedical classification. Bodenreider highlights the difficulty of separating these in ontological contexts, outlining ‘the intrusion of the epistemological in the ontological’. Many of the difficulties reflect the way knowledge is generated, validated, reconstructed and implemented by and for local communities of practice [Wenger,1998].

4.3 Tensions and Trade-offs

It is arguable that the difficulties encountered both in the design and the usability of ontologies in practice have been instrumental in prompting a closer analysis of how the semantic web can most productively manage fluid concepts of bio-medical domains to facilitate data sharing, without constraining usability and flexibility. Generating a layer of semantic middleware that is human and machine readable is by definition a socio-technical enterprise, invoking tensions and trade-offs that are not always explicit. Moreover, like many other coupled socio-technical systems, the process of abstracting and creating them changes the process itself. Marc Berg [1997] argues that ‘disciplining a medical practice to a formalism tends to transform the use of medical criteria in different ways’ through the provision of tools based on simple, robust worlds, with simple, clear-cut variables, and low specificity. Tensions and trade-offs such as this are evident in the construction of shared representations of data, information and knowledge across disparate communities, as well as disparate social and technical systems.

The approach to this problem in many applications has been to separate out the core areas that can most easily/ usefully be standardized, and to allow a range of approaches to evolve ‘at the edge’ in an evolutionary manner, for example combining a core ontology with user based ‘folksonomies’ to better meet the needs of very diverse communities.

There was also a view expressed by some groups that distributed communities can themselves be used in enhance the quality of integrated resources in such distributed contexts, depending on the flexibility of the approach adopted. A growing number of researchers from different disciplines now view the diversity of communities as a potential resource for enhancing, currency, creativity and innovation rather than as merely a challenge to standardization and scalability [Berners-Lee 2007; Comfort 1999; Sawhney and Parikh 2001; McGilchrist 2007], especially where speed, data quality and rapid response to change are at a premium as they are in volatile environments such as safety, or ebusiness systems. Agent-based systems, at the furthest end of the spectrum, offer a particularly dynamic and adaptive approach to this. Motta [2007] for example, points to ‘a move from the concept of a centrally

designed, monolithic ontology towards an automatic integration of ontology fragments, sourced from the semantic web to meet current user needs.' Phil Edwards [2007] and Tim Berners-Lee [2007] both highlight the potential value of strategies adopted in social networking sites which leverage the community resource to actively create value from data. As value-added, usability and uptake become more significant drivers of funding for Grid applications, it may be that the approaches adopted in more competitive contexts are considered. One of the most significant paradigm changes in the design of complex distributed business systems in recent years has been the change in emphasis from the generation of value through scale and interoperability, to the generation of value from the leverage of local knowledge and innovation, and the localization of services [Sawhney and Parikh 2001]. As eHealth systems are rolled out, the combination of a stable core and dynamic local variants may emerge as a recurring pattern in the competition for quality and local usability in the design of such systems [Blair 2006].

5. Conclusion

While it is increasingly acknowledged that the technical aspects of ontology alignment need to be given more consideration, there is also an argument for more explicit consideration of how the socio-technical (and at times socio-political) aspects of alignment can best be fostered.

Ontologies provide a layer of semantic middleware multiple local and temporal conceptualizations of a domain, and between technical and human readable representations of that. There is therefore a tension between the technical ideal of a stable, interoperable information infrastructure and the view of knowledge as an evolving, socially/ locally constructed and often disputed entity [Garfinkel 1984]. The experience of ebusiness and social networking systems in particular highlight a range of strategies with potential applications in Grid contexts where the same tensions exist, and where quality, usability and cost-effectiveness are increasingly considerations.

It was clear from the experience of other Grid projects in the NeSC eHealth workshop that ontology development involves the building of an ontology development community. This is then dependent on the provision of effective support for facilitating (and rewarding) collaboration as an explicit part of system design. This was also an outcome of the NCeSS ontology building workshop which emphasized the need to support the development of stable socio-technical constituencies – an ensemble of technical components (hardware, software, etc.) and stakeholders (people, interest groups, visions, values). This was identified as a top down and a bottom up process that

did not fit easily with current support and funding structures [Procter R. 2007].

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