RARE EYE DISEASE ONTOLOGY CREATION AND EDITING

ERN-EYE

January 2019

Activity	Development of Rare Eye Disease (RED) Ontology using Orphanet and HPO base
Area	Research and registries
Duration	4 days
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1. SUMMARY

ERN-EYE recognized at an early stage that in order to facilitate collaboration across geographic and professional boundaries there was a clear need for a common language for use not only in research and registries but in presentation of cases for virtual consultations. A workshop was convened to revise two of the main existing ontologies to ensure that they were fit for purpose. The initial thinking was that, as this is a highly technical exercise, the involvement of patients would be a distraction and that non-scientists would be unable to engage in such high level debates.

This was queried by the ePAG and two members of the ePAG were invited to join the meeting. Following the discussion at the workshop was indeed challenging for the patient advocates, but everyone agreed that the outputs were improved as a result of their participation and the experience helped cement the role of the ePAG within the Network.

An article describing the process and the results of this exercise has been published in the Orphanet Journal of Rare Diseases: "An ontological foundation for ocular phenotypes and rare eye diseases"
https://ojrd.biomedcentral.com/articles/10.1186/s13023-018-0980-6

2. INITIAL SITUATION

Collaboration between healthcare providers even in the same country is complicated by different terminology being used for the same conditions or symptoms and different methods of coding this information leads to more confusion and risk of error. This is compounded when data crosses international and linguistic borders. Several "dictionaries" or ontologies exist for this purpose and ERN-EYE determined that we should settle upon two of these for the purposes of all internal communications within our network, the Orphanet and HPO ontologies.

The two ontologies were selected because they approach the same problem from totally different angles, with

Orphanet being designed from the outset as a genetically robust system of categorizing rare diseases and HPO taking a phenotypical approach. In that sense they complement each other and allow for a more complete coverage for our foreseeable needs.

However, it was obvious to the expert clinicians in ERN-EYE that ophthalmologists had not defined the categories and definitions that existed within these two existing ontologies and even among experts in the same field there are differences of opinion. It became very obvious, therefore, that we needed to revise these ontologies in a manner that made them more suitable for our purposes.

3. CONTRIBUTION TO PATIENT ENGAGEMENT AND/OR IMPROVEMENT OF CARE

The debates taking place over each definition were in some cases long, occasionally heated and complex. It is easy to see why the initial supposition that patients were not well equipped to add to this debate might have been framed. Indeed, meaningful involvement of patients in such debates requires a great deal of preparation and concentration on the part of the patient representatives and not every patient would be able to make a positive contribution.

However, in several cases where the clinicians present were finding difficulty reaching a consensus, the patient representative was asked to adjudicate, or at least to express an opinion, and at all times were able simply by being present to act as a reminder what all of this was about – the definitions acting in the best interests of patients. There is no question that the outputs were made more robust as a consequence.

4. SUCCESS FACTORS

The first point is the insistence by ePAG members that they must be full participants in every aspect of ERN activity where possible and not just observers, no matter how difficult that participation might be. Initially the request was acceded to probably out of a sense of maintaining harmony but in the event the majority of participants recognized the positive contribution of patients.

The second factor contributing to the success is a recognition by the ePAG members involved that in order to be credible as full participants there was an obligation to commit to the process and to prepare for the workshop as well as possible, but most of all to participate fully in what were exhausting sessions for all involved (running into late evening each day). This also helped develop a camaraderie among all participants.

5. LESSONS LEARNED

- Patients can and must be full partners in ERN activities wherever possible. This is clearly not realistic in situations where individual patient confidentiality is involved, eg virtual consultations, but complexity is not a valid reason for excluding patients

- Participating in all ERN activity can help build relationships of trust and mutual respect. Many clinicians and researchers will have had bad experiences of patient engagement to overcome (as we have with some researchers and clinicians) and we need to be aware of this and to demonstrate that we will recognize all the relevant professional boundaries

- In order to be able to participate at this level patients will need to invest a lot of time and recognize that it will not always be easy, nor will every ePAG member necessarily be able to provide the commitment or even the intellectual skills to participate at this sort of level. Hopefully each ePAG will nonetheless be able to put forward sufficient numbers of patient representatives to fully participate in this way.