Reviewer’s report

Title: The EPIRARE Proposal of a Set of Indicators and Common Data Elements for the European Platform of Rare Disease Registries.

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Reviewer: S. Faisal Ahmed

Reviewer’s report:

It is useful to see the outcome of the EPIRARE project as a summary in this report. The authors have done a lot of work at agreeing on a come on data set which should be collected by a rare disease registry. In this report they have categorised them into three domains. The first one relates to identification details of the case, second one relates to the process of care and the willingness of the case to participate in studies and the third one relates to outcome. Whilst this is a laudable objective, registries are resource intensive and the it is highly likely that there is an inverse relationship between quality of the data in a registry and the number of fields. In addition, a number of fields that the authors have described will already be captured in local health service records especially in countries that have these infrastructures. It is highly likely that firewalls between these local health informatic systems and a common rare disease registry will prevent automatic uploading of data and the user may have to rely on manual entry of data. The point I would like to make is that it would be useful for EPIRARE to define a much smaller set of data elements that it thinks should be mandatory from the four domains. In future directions, it should try to to field test in a model to see how feasible would it be to do this.

In addition, I understand EPIRARE has already performed a survey of what rare disease registries already collect. The EPIRARE report would be enhanced if it can provide the data from this survey as I am presuming the common data elements have been developed from this survey as there would have been a common denominator amongst all these registries.

Level of interest: An article of importance in its field

Quality of written English: Acceptable

Statistical review: No, the manuscript does not need to be seen by a statistician.

Declaration of competing interests:

I declare that I have no competing interests