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Resource Title: Guidelines for the informed consent process in international collaborative rare disease research
Website: http://www.nature.com/ejhg/journal/vaop/ncurrent/abs/ejhg20162a.html
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Name of Project Leader (please bold the last name): Deborah **Mascalzoni**
E-mail: deborah.mascalzoni@crb.uu.se
Institution Name: University of Uppsala, Sweden
Address: Husargatan 3, BMC, entrance A11. Box 564, SE-751 22 Uppsala

Summary description of the resource (250 words max):

The Guidelines are meant to provide guidance for an effective informed consent process focusing on RD patients involved in international research. The Guidelines were developed with the input of patients collected during a workshop in Rome and the paper itself, including the very last version, was submitted and commented by the patients representatives involved in RD-Connect. For RD research it is especially relevant to ensure the best use of available resources but at the same time protect patients' right to integrity.
The paper proposes solutions to some of the challenges faced by international RD research requiring the sharing of clinical and genomics individual level data implicating some level of coordination between research centers worldwide. What are the minimal requirements and a good standard for meaningful informed consent taking into account opportunities, risks and transparency in the management of genomic data? The informed consent core elements identified are meant to effectively serve patients and research in order to achieve a good ethical standard and take into account possible changes for 1. future research and 2. Legacy data and collections. The principle is to plan in advance the best possible consent procedure to avoid ethical and legal hurdles that could hamper research in the future and make the most of existing resources without overriding patients rights in the process.

Resource's contribution to IRDiRC's focus and mission (250 words max):

To achieve IRDiRC main goals, to deliver 200 new therapies for rare diseases and means to diagnose most rare diseases by the year 2020, there is a clear need to promote international collaboration among RD researchers and maximum use of existing clinical and genetic data, while ensuring patient

integrity and involvement in research. To achieve this it is essential to streamline ethical and regulatory procedures for prospective and retrospective research. The current guidelines provide core elements that should be included in informed consent templates international collaborative RD research and procedures for the re-contact and re-consent of patients.

Resource's terms-of-use, licence policy (250 words max):

The Guidelines are published in a paper licensed under a Creative Commons Attribution-NonCommercial-ShareAlike 4.0 International License. The images or other third party material in the paper are included in the article's Creative Commons license, unless indicated otherwise in the credit line;

Process in place for quality control, life cycle management and scientific peer-review:

The guidelines were developed following a workshop involving bioethicists, researchers and patient representatives, and they were published in a peer reviewed journal.

Core impact indicators (e.g. numbers of users per year, number of visits to site, number of downloads, number of partnerships):

1 citation and 930 page views as recorded by the EJHG on June 9, 2016;

Size and constitution of the maintenance team, where applicable:

NA

Funding scheme (budget and sources):

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Resource's uniqueness:

Although a body of literature exists to describe informed consent in general, this is not the case for the rare disease field. Also, to our knowledge there is no clear guidance on the issue of re-consent and re-contact of patients for the use of their collected data and samples in international collaborative research and genomics research. The guidelines include new core elements that should be addressed in such research: Hosting of the data in open access databases; Use of interoperable identifiers for the de-identification of participants; Access by industry if foreseen and prospects for third-party commercialization and intellectual property; Possible linkage to different data (registries, medical records, etc.).

Publication(s):

Gainotti S, Turner C, Woods S, et al. Improving the informed consent process in international collaborative rare disease research: effective consent for effective research. European Journal of Human Genetics 2016; doi: 10.1038/ejhg.2016.2