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Coordinating Office Position Paper

AP08-2014: Minimum Content of a Country Specific Variant Database ICO Comments

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Authorisation:

This Position Paper has been prepared by Timothy D. Smith and represents the official position of the Human Variome Project Coordinating Office only. It does not represent an official position of the Human Variome Project, its Consortium, Advisory Councils or International Scientific Advisory Committee.

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Richard Cotton

I. Context

The International Coordinating Office (ICO) received the attached Activity Proposal (AP) on the 25th of February, 2014, from Human Variome Project Consortium member Martina Witsch-Baumgartner. Pursuant to the Project's Standards Development Process (PD06-2011) it was forwarded to the International Scientific Advisory Committee (ISAC) for referral to a Sponsoring Council (SC). Further pursuant to the Standards Development Process we have prepared the following comments on the AP for the SC.

II. Comments

Background

This Activity Proposal is concerned with developing a set of minimal content requirements for country specific variant databases, or HVP Country Nodes. As the submitter rightly points out, under the Project's Global Collection Architecture—a proposed international data sharing ecosystem that operates in a harmonised, federated fashion—HVP Country Nodes are sources of data for international gene/disease specific databases. To assist the sharing of data between knowledge sharing infrastructure operating within the Global Collection Architecture, it would be beneficial for HVP Country Nodes to collect a standardised set of core data elements. Obviously, HVP Country Nodes would be free to collect and share additional data elements beyond this minimum core set, as long as such collection and sharing was done in compliance with the legal, regulatory and ethical frameworks that the Node is operating under.

The Human Variome Project is already in the process of defining a core set of minimum content for international Gene/Disease Specific Databases via the activities of [WG03-2012: Minimum content for gene variant databases \(LSDBs\)](#). It would make sense to complement this work with the activity proposed by the submitter.

Scope of Work

The ICO believes that the scope of work outlined in the Activity Proposal is necessary and sufficient to achieve the stated objectives of the proposed activity. However, we do note that the question of defining minimum content requirements to assess the quality of data in a country specific database may be premature without first undertaking a work package that researches and defines the quality parameters that this class of database should be aiming to achieve. A similar activity is being undertaken for Gene/Disease Specific Databases by [WG05-2013: Variant Database Quality Assessment](#).

The submitter proposes that the final outcome of this activity be an HVP Standard: Minimal requirements for country/regional specific variant databases. The ICO concurs with this proposal, noting that it is consistent with approaches taken by similar Working Groups.

Possible Working Group Members

A number of members of the International Confederation of Countries Advisory Council (ICCAC) are responsible for the development and maintenance of existing HVP country Nodes. These members include the representatives from the following countries:

- Australia
- Malaysia
- Mexico; and
- the United Kingdom.

The ICO recommends that representatives from these countries make up the initial membership of any Working Group to undertake this activity.

Available Assistance from the ICO

The ICO is able to assist any chartered Working Group in the manner described by the submitter:

- Web meeting facilities;
- Room and materials for face to face meetings during meetings that Working Group members are attending; and
- administrative support.

The ICO is unable to provide direct funding to support this activity, but may be able to assist in applying for funds. The ICO notes that there are several EC Horizon 2020 funding calls open (e.g. [H2020-INFRAIA-2014-2015: Integrating and opening research infrastructures of European Interest](#)) in which this proposed activity might have a place within a broader program of work.

III. Summary of Recommendations

We recommend that:

- The proposed activity be taken up by the Human Variome Project
- The ICCAC act as Sponsoring Council for this Activity
- The ICCAC charter a Working Group to undertake the activity as proposed
- The resultant work be published as an HVP Standard

ATTACHMENT A

Activity proposal

AP-08 Minimum content of a country specific database (CN-01-2014) (2nd draft version)

Need being addressed

A country specific database is meant to be the base for data which are later on introduced to international databases, the interest there will be probably to get information on country- and ethnic specific variants with corresponding frequencies. Hence the national databases should contain minimum information which makes it possible at the end in the international databases to characterize/define variants regarding their pathogenicity.

At the moment there is no recommendation regarding minimal content of a country/regional specific database.

Scope of proposed activity

Generation of a recommendation list defining minimum set up of a database (regarding the variant, the patient and the phenotype)

Recommendations about definition of a minimum of description regarding the database and its webpage, for example regarding the collection of data and curation, describing laws and ethics underlying the choice of included data.

Plan of action

Assign a working group of 3-5 persons from ICCAC who should define standards/suggestions regarding a minimum set up of a database regarding (draft version), which will be reviewed by other HVP members and submitted as a publication.

Required resources

Webmeeting possibilities (calliflower, Skype or other), face to face meeting during upcoming meeting, administrative help by ICO staff?, Time

Expected deliverables

Recommendation publication: "Minimal requirements for country/regional specific variant databases ". Preferably the document will be published in a peer-reviewed scientific journal.

Recommended publication class

Publication should be finally a document describing a (minimal) standard for country/regional specific variant databases.