

Solve-RD Publication Policy

Version V3.1

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1 Solve-RD Publication Board

Solve-RD established a Publication Board. The Publication Board is chaired by Holm Graessner and engages all WP leads and is open for interested DITF and WG leads to join. The Publication Board shall strategically plan publications and shall ensure transparent communication about planned Solve-RD publications. The Publication Board shall also provide advice on authorship discussions.

2 Solve-RD publication tiers

The Publication Board defined **three tiers of Solve-RD publications** in terms of scope and coordination requirements. The three-tier model has been approved by the Solve-RD Steering Committee on 16 Feb 2021.

Tier 1 "flagship" publications

- Require input from the entire Solve-RD consortium and central coordination
- ERN/DITF and/or WP-level (or even cross WP) coordination/lead required
- Example: massive re-analysis of all existing WES/WGS data

Tier 2 "central" Solve-RD publications

- Requires coordination within ERN/DITF or DATF/WG
- Example: individual manuscript of re-analysis effort of a WG; or individual ERN yield/new phenotype/new gene(s)

Tier 3 "local" Solve-RD publications

- Individual solved cases/disease
- Case reports

3 Solve-RD fairness principles to determine authorship

The Publication Board defined the following principles to determine authorship:

- (1) Authorship reflects (scientific) contribution
 - Infrastructure and support roles (such as clinicians contributing cases or infrastructure enabling joint analysis) will be taken into consideration, too.
 - (Governance) role in the project should not define authorship (position).
- (2) Fair and justified designation of authors
 - Designation of authors will be based on submitted analysis projects and in line with the Solve-RD Publication Policy.
 - Prominent authors shall take responsibility for the manuscript.
 - Both "fairness towards the consortium" and "fairness towards individuals" will be taken into account, however, individual contribution has to be appreciated foremost.



Proposed process to determine authorship (for tier 1 publications):

- Manuscript leads propose authorship positions based on contribution taking into account the Solve-RDs fairness principles.
- All authors and contributing Solve-RD sub-structures (DATF-WGs, DITFs, etc.) are being informed and explicit agreement is requested.
- Proposal of authorship positions is being run by the Publication Board. Only in case of serious issues the SC is being involved. Sub-structures (e.g. DITF) propose and harmonise respective authorship rules.

4 'Solve-RD consortium' as an author and additional Solve-RD corporate authorship lists

An affiliation list has been created and is regularly being updated containing all members of the Solve-RD consortium and associated partners (see Annex I). This list determines the Solve-RD author ('Solve-RD consortium'). Additional authorship lists of Solve-RD subgroups (i.e. for ERN-specific Data Interpretation Task Forces (DITF)) have been created and contain all Solve-RD partners contributing to the respective subgroup.

The 'Solve-RD consortium' author list and all additional Solve-RD corporate authorship lists are administered and regularly updated by the project management team (lead: Holm Graessner).

'Solve-RD consortium' as an author shall be used for all publications that require input from the entire Solve-RD consortium (Tier 1).

Other Solve-RD corporate authorship lists will be used for publications that cover the work or contributions from the respective groups.

For all publications including 'Solve-RD consortium' or Solve-RD corporate authorship lists as an author, the responsible authors shall contact the project management office. The management office provides the most recent version of the respective list, provides advice for the use and support for appropriate formatting of the authorship lists considering the requirements of the respective journal and publisher. The management office is responsible to inform all list authors about the planned publication that makes use of the corporate author list.

5 Notification and authorship policy with regard to shared data

All Solve-RD publications are acknowledged to be based on the fundamental principles of open scientific collaboration, reciprocity, attribution and benefit sharing. For any publication resulting from work carried out using data shared or generated through Solve-RD (e.g. for identifying a novel gene), including where data has been accessed through the RD-Connect Genome-Phenome Analysis Platform, the authors should in all cases acknowledge and give appropriate authorship positions to all relevant parties in line with best practice for acknowledgement of scientific contribution including submission of the primary data.

Examples and further principles are described below.

- 1. A publication arising from research in which the party leading the publication ("the PI team") is primarily analysing their own submitted data (example: novel gene discovery by a submitter analysing their own patient cohorts in the RD-Connect GPAP):
 - i. Where a publication only includes data and hypotheses from the PI's own research group, key authorship positions may be held by this group, but the software, tools and resources made use of for the research should be duly acknowledged and referenced in line with the policies for those resources (e.g. see RD-Connect GPAP policy below). Where justified,



- individuals supporting the bioinformatics analysis or platforms may be approached for coauthorship based on individual scientific contribution.
- ii. Where a publication has involved the use or analysis of data from additional submitters, these submitters should be contacted as soon as possible ahead of publication and invited to provide input as co-authors. The PI team is strongly encouraged to share key authorship positions with other teams that have brought in similar intellectual input and/or fundamental data (e.g. "a second family"). Acknowledgement of bioinformatics support should also be considered as in (a) above.
- 2. A publication arising from the analysis of data where the party generating the hypothesis and carrying out the analysis is not themselves the data submitter (example: reanalysis of data by a Solve-RD bioinformatics group that did not submit the data or see the patients):
 - i. Submitters of the data used for the analysis should be contacted as soon as possible ahead of publication and invited to provide input as co-authors. The PI team is strongly encouraged to share key authorship positions with the submitting teams based on the value and amount of data contributed to the publication. If the primary data is the key to discovery, a key authorship position should be discussed with the owner of the primary data.
 - ii. Where a publication makes use of data from a large number of submitters or transversal analysis of the Solve-RD cohort, a group authorship for Solve-RD should be considered in order to acknowledge the role of all data submitters equally.

All data access through the RD-Connect genome-phenome analysis platform is monitored automatically by the system and all other data access for other Solve-RD activities is only to named individuals within the Solve-RD consortium, therefore any breach of the publication policy will be monitored and flagged up to the Solve-RD Steering Committee.

6 Confirmation of paper by Solve-RD Consortium

During the project and for a period of one (1) year after the project, every paper that is published with affiliation of Solve-RD or includes data produced or collated within Solve-RD has to be confirmed by the Solve-RD Consortium. The procedure is defined in Article 29.1 of the Solve-RD Grant Agreement and Article 8.4.2 of the Solve-RD Consortium Agreement. All Parties and associated partners (including associated ERNs) are obliged to follow this procedure:

Prior notice of any planned publication shall be given to the other Parties at least 45 calendar days before the intended date of publication. Any objection to the planned publication following the above notification shall be made in accordance with the Grant Agreement in writing to the Coordinator and to the Party or Parties proposing the dissemination within thirty (30) calendar days after receipt of the notice. If no objection is made within the time limit stated above, the publication is permitted.

An objection is justified if (a) the protection of the objecting Party's Results or Background would be adversely affected and/or (b) the objecting Party's legitimate academic or commercial interests in relation to the Results or Background would be significantly harmed.

The objection has to include a precise and reasonable request for necessary modifications, it being specified that any such modifications shall not harm the scientific content of the proposed publication or communication.

If an objection has been raised the involved Parties shall discuss how to overcome the justified grounds for the objection on a timely basis (for example by amendment to the planned publication and/or by protecting information before publication) and the objecting Party shall not unreasonably continue the opposition if appropriate measures are taken following the discussion. The objecting Party can request a publication delay of not more than 90 calendar days from date of submission to the other



Parties. After 90 calendar days the publication is permitted provided that Confidential Information of the objecting Party has been removed from the Publication and all reasonable modifications of the objecting Party have been implemented within the Publication as indicated by the objecting Party.

A decision has to be made and communicated within four weeks after submission of the publication draft (author list and abstract) to the Steering Committee.

7 Parallel Analysis of submitted data at centres

The Publication Board has to be informed if data sets that were submitted to Solve-RD for central analysis lead to publications based on separate (in-house) analysis.

8 Acknowledgements

Solve-RD funding acknowledgement

Any publications arising from Solve-RD project funding must be published Open Access and acknowledge EU funding in the following way:

"This project has received funding from the European Union's Horizon 2020 research and innovation programme under grant agreement No. 779257 (Solve-RD)."

RD-Connect GPAP acknowledgement

Authors should acknowledge the RD-Connect GPAP if tools, services or data provided by the platform have enabled or have contributed to research targeted for scientific publication.

RD-Connect GPAP should be acknowledged using the following wording:

"This study makes use of data and tools shared/provided through the RD-Connect GPAP, which received funding originally from the European Union Seventh Framework Programme (FP7/2007-2013) under grant agreement No. 305444."

In addition, the following paper should be cited:

Laurie S., Piscia D., Matalonga L., Corvo A., Garcia C., et al. The RD-Connect Genome-Phenome Analysis Platform: Accelerating diagnosis, research, and gene discovery for rare diseases. Human Mutation 2022 febr. 17. doi: https://doi.org/10.1002/humu.24353

Where appropriate, the RD-Connect GPAP should also be explicitly mentioned in the "Materials and methods" and/or "Results" section.

ERN acknowledgement

Any publications with contributions from ERNs should acknowledge involved ERNs in the following way:

"This study was supported by the European Reference Network(s) [add ERN names] (https://ec.europa.eu/health/ern/networks_en)."



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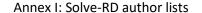
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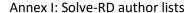
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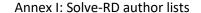




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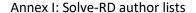
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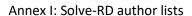




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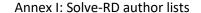




'Solve-RD DITF-EpiCARE'

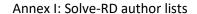
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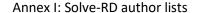




'Solve-RD DITF-RITA'

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