

The rare diseases GO FAIR implementation network

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Introduction

The key driver for the Rare Diseases Global Open FAIR implementation Network (RDs GO FAIR) is the need to substantially speed up the process towards earlier diagnosis and new treatments of rare diseases (RDs), in line with the vision and goals set by the international rare diseases research consortium (IRDiRC)¹. Clinicians, patients and researchers should be able to retrieve information and perform analyses across all potentially relevant data resources efficiently and correctly. This is much more likely when data that are collected are Findable, Accessible (under well-defined conditions), Interoperable, Reusable (FAIR), for analysis by computer. This is especially important, because rare disease data are collected everywhere around the world. A comprehensive ecosystem of FAIR RD data and services will increase the accuracy, efficiency, efficacy, and reliability for the use of RD data, and thereby support the IRDiRC goals, in particular towards earlier diagnosis and new treatments.

Implementing FAIR in the rare disease community requires generic and RD-specific standards and tools. Moreover, these need to be adopted and integrated into the tools and practices already commonly utilised by RD stakeholders. The RD community is motivated to make the accessibility and analysis of RD data as efficient as possible for all potential users, but is still far from having an established FAIR-minded culture in support of this goal. A GO FAIR implementation network for the RD community is required, because the current standards and tools, as well as the pathways to their adoption, are insufficiently mature for RD resources to become FAIR at source and by design to establish a functioning FAIR ecosystem. Adoption will require community-tailored guidelines, standards, and tools.

¹ <http://www.irdirc.org/irdirc-goals-2017-2027-new-rare-disease-research-goals-for-the-next-decade/>

Purpose of the Implementation Network

Within the context of supporting the IRDiRC goals, the main purpose of RDs GO FAIR is to establish a culture in the RD community where members help each other choose, adopt, and tailor guidelines, standards, and tools to implement FAIR principles. RDs GO FAIR will first aim towards reaching a tipping point for change, of culture, by supporting the RD community stakeholders to engage in the implementation of FAIR principles. This will be supported by materials for understanding the FAIR principles and how to apply existing global metrics of FAIRness. Finally, it is of critical importance to develop and apply RD-community-specific FAIR Metrics through building effective tools and technologies. Following successful progress, the network will scale up its ambitions to help expand the ecosystem of FAIR RD data and services.

Overarching Principle of Operation

“We commit to comply with the [Rules of Engagement](#) of GO FAIR Implementation Networks”

Targeted Objectives of the implementation network

1	Foster and strategically oversee the expansion of the adoption of FAIR data principles by the RD community towards collecting a critical mass of FAIR data, providing a home for stakeholders seeking advice on FAIR data stewardship and guiding the transition of the rare disease community towards understanding, embracing and refining the FAIR Principles and Goals.
2	Enable support for the RD community in FAIRifying the data and metadata that stakeholders collect and curate, defining strategies and sustainable service models for adoption.
3	Ensure that RD Patients and patient representatives are actively engaged in each phase of planning and implementation, so that the individual patient experience and the wider patient community perspectives are leveraged to facilitate implementation.
4	Ensure that FAIR sharing in the RD community is respectful and responsible towards RD patients.
5	Collect existing -and specify novel- metrics to formally and quantitatively assess FAIRness of rare disease data resources; commit to their regular application to monitor improvement and success.
6	Ensure that information standards relevant for implementing FAIR in the domain of rare diseases are identified, applied, aligned, improved, extended to fit to purpose, and shared following the FAIR Principles. New standards should only be developed when existing standards are not sufficient and cannot be extended or adapted..
7	Identify overlaps/duplications of effort towards implementation of FAIR in the RD community, and work with the identified groups to help foster collaborations and funding initiatives, thus minimizing duplication and maximizing harmonization.
8	Foster examples demonstrating how a critical mass of FAIR data facilitates information retrieval and federated integrative data analysis to answer key questions for rare and ultra-rare diseases beyond current capabilities, and support the dissemination of these examples.
9	Ensure that the analytics developed for rare and ultra-rare diseases are increasingly capable of exploiting FAIR data and guarantee their applicability to automated, distributed analysis and distributed learning (the Personal Health Train paradigm). Work towards a data and tool sharing infrastructure for distributed analysis.
10	Ensure that RD analytical tools (software, workflows, training materials, and other digital objects) are available through public FAIR repositories, and can adequately address the requirements for rare disease data.

Tasks

The following tasks are foreseen to achieve the RDs GO FAIR objectives

	Task	Subtasks	Obj.
1	Complete the execution plan & roadmap as part of the process of becoming a GO FAIR Implementation Network (within 3 months)		
2	Organise and maintain the network	1. Organise the network of RDs GO FAIR handlers and liaisons (domain representatives)	1
		2. Link to or organise, and support, networks of FAIR enablers for the RD field, including data stewards, patient representatives and advocates, software solution providers (for profit and not-for profit), infrastructure developers, cross-project FAIR data experts, project-specific teams, industry, other GO FAIR networks.	2, 7, 9
		3. Annually assess objectives, organisation, and tasks against progress in the field.	1
3	Foster the definition of requirements, criteria and metrics for FAIR data and FAIR standards, in compliance with the RDs GO FAIR objectives	1. Define expectations of respectful and responsible use of rare disease data with patient representatives, and incorporate these in the criteria associated with this task. 2. Define criteria to measure progress towards a critical mass of FAIR data and services in support of achieving IRDiRC goals. 3. Define metrics of success of information standards (within a FAIR ecosystem) and guidelines for a path to reach success in the RD community. 4. Collaborate with liaisons to	1, 4, 5, 6, 7, 10

		<p>compile, peer-review, and disseminate information standards, such as via FAIRsharing.org.</p> <p>5. Assess the RDs GO FAIR network's ability to identify overlaps and duplication of effort and provide recommendations towards mitigation.</p>	
4	Engage with software solution providers to foster production of FAIR data generating and FAIR data analysing software in line with RDs GO FAIR objectives	<ol style="list-style-type: none"> 1. Identify and stimulate development of reusable specifications of FAIR data-generating components for software providers. 2. Organise engagement with the GO FAIR Personal Health Train consortium to convey requirements for rare disease data. 3. Convey criteria set by the PHT paradigm to developers of FAIR and data analytics workflows in the rare disease field. 	2,7,10
5	Foster the definition and implementation of a service model that can be sustained by the rare disease community to make and keep all their valuable data sources FAIR.		2
6	Monitor and advocate engagement of patient representatives in FAIR implementation activities that RDs GO FAIR engages with.	<ol style="list-style-type: none"> 1. Annually survey involvement of patient representatives via the RDs GO FAIR liaison network and provide recommendations. 2. In close collaboration with EURORDIS, identify patient representatives to propose for participation in activities related to planning and implementation of FAIR data principles. 	3
7	Foster example applications of a critical mass of FAIR data and services, and support their reuse.	<ol style="list-style-type: none"> 1. Identify, advocate, and help disseminate efforts that showcase the application of FAIR data and services for analysis that answers key questions for rare and ultra-rare diseases. 2. Identify which platforms rare disease researchers use to find their tools and (i) advocate greater FAIR-compliance of 	8

		<p>these platforms (if needed), (ii) advocate the use of FAIR-compliant platforms for sharing FAIR-based applications.</p> <p>3. Share successful approaches and tools with other communities, such as GO FAIR communities beyond rare diseases</p>	
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Membership list

We consider this Manifesto to be one way by which the undersigned stakeholders can speak with one voice on a number of critical issues that are of generic importance to the objectives of FAIR, and on which we feel we have reached consensus.

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