

3.2 Core of the report for the period: Project objectives, work progress and achievements, project management

3.2.1 Project objectives for the period

The overall purpose of this project is to provide organisational and communication support to the International Rare Diseases research Consortium (IRDiRC) and its various members, thus to contribute to the development of policies, guidelines, and tools aiming at accelerating research on rare diseases and at reinforcing international research cooperation.

The specific objectives of the project are:

Objective 1: support the work of IRDiRC bodies – an Executive Committee, three Scientific Committees, and 12 Working Groups - by organising meetings and teleconferences, assuring secretarial work for them and keeping all members updated, assuring a smooth organisation of the consortium’s work. This objective is linked to the activities performed in the WP1.

Objective 2: support advances in research on rare diseases by collecting and diffusing pertinent information and results (projects funded by IRDiRC members, policy and guidelines relevant to the field, outcomes of research, etc.) to IRDiRC members, and to facilitate cooperation with other stakeholders and other projects of the HEALTH programme in rare diseases. This objective is linked to the activities performed in the WP2.

Objective 3: assure the dissemination of IRDiRC activities and research initiatives, and of progress towards IRDiRC goals with different means of communication such as logo, website, flyer, newsletters, annual “State-of-Art” report, etc. This objective is linked to the activities performed in the WP3.

Objective 4: Manage the SUPPORT-IRDiRC project itself to ensure a smooth running of the project between the two partners and ensure all actions are in compliance with the FP7 rules. This objective is linked to the activities performed in the WP4.

3.2.2 Work progress and achievements during the period

3.2.2.1 WP1 “Support to the implementation of IRDiRC”

- Work progress and achievement during the period

The main aim of this WP is to provide secretarial assistance to the IRDiRC bodies (Executive Committee, Scientific Committees, and Working Groups) by organising meetings and teleconferences, providing preparatory documents and writing the reports. A second aim is to ensure a liaison between the different bodies to optimise the complementarity and coherence of their specific activities contributing to the common goals.

The secretarial support was established for the Executive Committee, three Scientific Committees and 12 Working Groups, which constitute the consortium and are now all running.

Task 1.1 – Organisational support to the IRDiRC Executive Committee

The secretariat of the IRDiRC Executive Committee was moved to the SUPPORT-IRDiRC project on 15 April 2013 at the request of the European Commission, when the chairmanship of the Executive Committee was transferred from Ruxandra Draghia-Akli (European Commission) to Paul Lasko (University McGill, Canada).

Between April 2013 and March 2014, one meeting of the Executive Committee (23-24 September 2013, Miami, USA) and two teleconferences (25 November 2013 and 3 February 2014) were organised. For each of these meeting/teleconferences, agenda and preparatory documents were prepared in collaboration with the Chair, and reports were drafted and circulated to all members of the Executive Committee for approval (Deliverable D1.1).

Other tasks include the maintenance of an up-to-date list of members on the IRDiRC website, finalisation of the policy document (layout and final corrections), update of the governance documents, which was approved in September 2013, and invitation of new members to the Scientific Committees whose nominations were approved by the Executive Committee.

Procedure of work with the Chair of the Executive Committee and the Executive Committee were established during a visit of the SUPPORT- IRDiRC project coordinator and project manager to the Chair of the Executive Committee in Montreal, Canada, in February 2013, and a visit of the Chair to the IRDiRC secretariat in Paris, France, in June 2013 (Milestone MS1). Teleconferences with the Chair and the Scientific Secretariat (SUPPORT-IRDiRC project) are organised when necessary (e.g., before each meeting or teleconference of the Executive Committee for finalisation of the agendas, etc.). The Secretariat provided the Chair with a letterhead and business card.

Task 1.2 – Scientific Committees and Working Groups secretariat

Secretariat activities for both Scientific Committees and Working Groups include setting up meeting/teleconference dates by Doodle based on communication with the corresponding chairs, disseminating the agenda and relevant documents, preparing and circulating lists of research projects (at the specific request of Working Groups), and writing reports of these meetings/teleconferences (Deliverable D1.1).

For the Scientific Committees, six face-to-face meetings and five teleconferences were organised. For the Working Groups, three face-to-face meetings and 28 teleconferences were organised.

In addition, the Scientific Secretariat sent invitation emails to all members of the 12 Working Groups at the request of the Scientific Committees, between February 2013 and October 2013.

Procedure of work with the chairs of the Scientific Committees were determined either during a visit to the Chair (Diagnostic Scientific Committee, February 2013, Ottawa, Canada), a visit from the Chair (Therapies Scientific Committee, July 2013, Paris, France) or by phone (Interdisciplinary Scientific Committee, February 2013) (Milestone MS1). Since then, communication with the chairs is conducted essentially by email or phone calls, with occasional face-to-face discussion at the Executive Committee meetings.

Task 1.3 – Management of logistics related to the implementation of IRDiRC

Support for the organisation of meeting depends on the meetings:

- Executive Committee meetings are hosted by a member of the Committee and the work of the Scientific Secretariat essentially consisted of circulating relevant information (announcement of the meeting, location of the event, hotel information, etc.).
- Regarding the meetings of the Scientific Committees, two out of six were organised with the help of the member hosting the meeting (Montreal, Canada, October 2013 and Prague, Czech Republic, December 2013). The work of the Scientific Secretariat essentially consisted of circulating relevant information and providing reimbursement for travel expenses.
- Four Scientific Committee meetings were completely organised by the Scientific Secretariat as the meetings were held in a location without presence of local members (three meetings in Dublin, April 2013, right before the 1st IRDiRC conference) or in Paris (March 2014). In these situations, there was additional work including: finding a meeting place, accommodation, catering, etc.

A member of the Scientific Secretariat was present at each meeting to provide logistic help if necessary, and to take minutes of the meeting.

- Deviation

The task “Establish contacts with potential new members so as to introduce them to the IRDiRC project” is not carried out by the Scientific Secretariat as the Chair of the Executive Committee decided to take on this responsibility.

- Statement on the use of resources

Help in-kind was provided by Partner 2 (Fondation maladies rares) or employees of Inserm US 14 (Orphanet) in drafting some reports from the Scientific Committees and Working Groups meetings/teleconferences. Partner 2 drafted 20% of the reports (8/39) and employees of Inserm US 14 18% (7/39) of these reports. The reminder were carried out by the SUPPORT-IRDiRC staff (24/39 reports).

Time allocation of the staff for the WP:

Assistant: 60%

Communication manager: 10%

Project manager: 40%

Research assistant: 10%

3.2.2.2 WP2 “Facilitation of cooperation between relevant RD stakeholders”

- Work progress and achievement during the period

WP2 is dedicated to the communication between stakeholders in the field of rare disease through the collection and dissemination of the projects funded by IRDiRC members, collection and dissemination of relevant guidelines and policies, and publication on relevant projects, through the IRDiRC website for all stakeholders, as well as a private website and monthly newsletter to members of the Executive Committee, Scientific Committees and Working Groups.

Task 2.1 – Liaison between the research consortia funded under HEALTH 2012.2.1.1-1

SUPPORT-IRDiRC follows the activities of the EUrenOmics, NeuOmics and RD-Connect through the participation to their joint annual meetings (25-27 January 2013, Sitges, Spain; and 23-26 February 2014, Heidelberg, Germany so far) and by publicising their outcomes on the IRDiRC website (three articles in the “Research Highlights” section). In addition the project coordinator is member of their Scientific Advisory Board.

Task 2.2 – Liaison with other IRDiRC research projects and rare disease research initiatives

The main tasks consist in compiling research projects funded by IRDiRC members, making it known to IRDiRC participants, entering the information in the Orphanet database, and publishing it on the IRDiRC public website for further dissemination.

This required an evolution of the Orphanet database to allow storing of information of research projects funded by IRDiRC bodies. This was performed at month 6.

The strategy for data collection depends on the presence of a national Orphanet team in the countries of interest:

- If a national team is present in the country, they collect the data, enter the data in the Orphanet database, and provide the Scientific Secretariat with the information to be displayed on the IRDiRC public website. Countries concerned are Australia, Canada, Germany, Finland, Italy, Spain, UK, and the Netherlands.
- These national teams were trained to collect the appropriate data set on month 6.
- If there is no Orphanet team in the country, the Scientific Secretariat collect the data and enter them in both Orphanet database and the IRDiRC website. Countries concerned are China, Korea and USA.

State of data collection and entering is summarised in the table below.

Member	Published in IRDiRC website	Entered in Orphanet database
Academy of Finland	Yes	Ongoing
Agence National de la Recherche (ANR)	Yes	Ongoing
BGI	a)	a)
Canadian Institutes for Health Research (CIHR)	Yes	Ongoing
Chinese Rare Disease Research Consortium	a)	a)
Children's New Hospitals Management Group	a)	a)
E-Rare	Yes	Yes
European Commission	Yes	Ongoing
Federal Ministry of Education and Research (BMBF)	Yes	Yes
Orphan Products Grants Program from Food and Drug Administration (FDA)	Yes	Yes
French Foundation for Rare Diseases	a)	a)
French Muscular Dystrophy Association (AFM)	Yes, but not completely	No
Genome Canada	Yes, but not completely	No
Istituto Superiore de Sanita	a)	a)
Korea National Institute of Health	Yes, but not completely	Yes

National Institute of Health Carlos III	Yes	Yes
National Institute for Health Research	Yes	Yes
Eunice Kennedy Shriver National Institute of Child Health and Human Development (NIH)	Yes	No
National Cancer Institute (NIH)	Yes	No
National Center for Advancing Translational Sciences (NIH)	Yes	No
National Eye Institute (NIH)	Yes	No
National Institute of Arthritis and Musculoskeletal and Skin Diseases (NIH)	Yes	No
National Institute of Neurological Disorders and Stroke (NIH)	Yes	No
National Human Genome Research Institute (NIH)	Yes	No
NKT Therapeutics	Yes	Yes
Sanford Research	Yes	Yes
Telethon Foundation	Yes	Yes
The Netherlands Organisation for Health Research and Development	Yes	Yes
Western Australian Department of Health	a)	a)

a) List of projects not received

For privacy concern, research projects funded by Industry members were not collected. Several funding agencies have not yet provided us with the list of research projects that they funded since they became IRDiRC members.

It is not yet possible to enter projects funded by NIH members in the Orphanet database as there are over three thousands of such projects funded since 2010, and there is no Orphanet team in USA or workforce available in SUPPORT-IRDiRC for such a task. There is an ongoing discussion with NIH members to determine if there is a possibility of sending us the information in a format that would allow direct injection into the Orphanet database.

For information purpose, projects funded by IRDiRC members were regularly promoted on the IRDiRC website. Ten articles were published in the 1st reporting period.

The Scientific Secretariat also participated in the preparation of two workshops, by inviting participants, circulating information or drafting the report, depending on the workshop:

- “The 2013 international workshop on rare disease system for pathogenicity inference” (18-19 April 2013, Dublin, Ireland)
- The “Rare genetic diseases: diagnosis and discovery workshop” (3 December 2013, Prague, Czech Republic).

Task 2.3 – Support to IRDiRC Working Group

An online platform, called private website (<http://www.irdirc.org/priv/restricted/wp-login.php>), was launched in August 2013. It is a password-protected site accessible to members of the IRDiRC Executive Committee, Scientific Committees, and Working Groups (Deliverable D2.2).

The purpose of this website is to share all documents related to IRDiRC committees (report of meetings and teleconferences, PowerPoint presentations of the meetings, documents created by the committees, etc.) and also documents of interest such as policies and guidelines in the field of rare diseases, etc.

The website was launched at month 11 instead of month 6 as initially planned due to the lack of a communication manager for 3 months.

In addition to the organisation of teleconferences and drafting of reports, support to the Working Groups mostly consists in collecting and diffusing guidelines and policies in the area of rare diseases relevant to the Working Groups (Deliverable D2.1), and providing list of research projects and clinical trials in their area of expertise at their request.

For the first reporting period, 13 guidelines and policies were collected and disseminated on both public and private IRDiRC websites. A strategy has yet to be developed for systematic collection of policies and guidelines as the process is currently not optimal.

- Deviation

Data portal for IRDiRC-affiliated investigators to encourage the sharing of preliminary results (Deliverable D2.3) was not established as members of the Executive Committee and Scientific Committees did not consider it necessary.

Report to the Executive Committee and Scientific Committees was not conducted at month 11 as initially planned (Milestone MS3) but provided through PowerPoint presentations at each Executive Committee face-to-face meetings where the chairs of the three Scientific Committees were also present (months 4, 7 and 12). The PowerPoint presentations are available on the private website for consultation.

- Statement on the use of resources

Most of the work was conducted by Partner 1, with in-kind help from national Orphanet teams to enter research projects funded by IRDiRC members in the Orphanet database.

Time allocation of the staff for the WP:

Assistant: 10%

Communication manager: 30%

Project manager: 20%

Research assistant: 80%

3.2.2.3 WP3 “Engagement of the community and dissemination of outcomes”

- Work progress and achievement during the period

The objective of this WP is to achieve an effective dissemination of IRDiRC activities and research initiatives, and of progress towards the IRDiRC goals through IRDiRC website, newsletters, and other dissemination tools.

Task 3.1 – Develop an IRDiRC communication strategy and communication plan for approval by the IRDiRC Executive Committee

The communication strategy was presented to the Executive Committee for formal adoption during their meeting held in Evry, France, on 25 September 2012. During this meeting, the logo and the outline of IRDiRC communication supports was approved.

Since, progresses and proposed next actions were presented at each Executive Committee meetings.

Task 3.2 – Logo and graphical chart for IRDiRC

The IRDiRC logo and graphical chart (front, letterhead, PowerPoint, etc.) were created by a contractor (Deliverable D3.1).

Layout for IRDiRC documents (reports, agenda, etc.), and a cover for the Executive Committee report and official documents were created by the communication manager of Orphanet.

Task 3.3 – Website on IRDiRC research

The IRDiRC public website (<http://www.irdirc.org/>) was launched on January 2013 to inform the community at large on IRDiRC, its activities, projects funded by IRDiRC members, and also news of interest in the field of rare diseases, such as conferences, policy and guidelines documents.

Information available related to IRDiRC:

- Goals
- Background and history
- Governance and policies
- Members (all Committees and Working Groups)
- IRDiRC activities (full reports of meeting/teleconference of all IRDiRC bodies, conference, workshop, etc.)
- List of projects funded by IRDiRC members
- IRDiRC in the news
- IRDiRC-related calls

Other information:

- About rare diseases
- Research on rare diseases
- Current results of research in relation with IRDiRC goals
- Selected reports and policy documents
- Research highlights

The IRDiRC website is updated daily. More work is necessary on the page “Current results of research” to include indicators that the Scientific Committees wish to see included.

The IRDiRC website was accessed 14 905 times in 9 months.

Task 3.4 – Annual State of Art and monthly newsletter or RSS flux

An internal newsletter is published and circulated to all 225 members of IRDiRC committees and Working Groups, monthly since July 2013. Nine newsletters were published in the first reporting period. Content of the newsletter includes information on:

- New IRDiRC documents available on the website
- New selected reports and policy documents available on the website
- New list of research projects published on the website
- Highlight on research projects
- News published on the website
- IRDiRC related calls
- Website update

- Other news (new members, planning of IRDiRC teleconference, conference of interest, etc.)

Task 3.5 – Other dissemination material

The IRDiRC activities and achievement were presented by the coordinator of SUPPORT-IRDiRC at three international events:

- S. Aymé: “IRDiRC: International Rare Disease Research Initiative”. 2nd annual orphan drug congress 2012, Barcelona, Spain, 17 October 2012.
- S.Aymé: “Tools to boost research in the field of rare diseases in 2013”. Seminar of the centre of comparative genomics, Murdoch University, Perth, Australia, 13 May 2013.
- S. Aymé: “Supporting the International Rare Disease Research Consortium: achievements and challenges in the field of interpretation of variants”. Japan Society of Pediatric Genetics annual meeting, Nagoya, Japan, 10 April 2014.

The following dissemination materials were produced:

- Flyer
- Poster
- Three PowerPoint presentations (“IRDiRC objectives and governance”; “IRDiRC achievement so far”; “Current status of rare disease research”).

These materials are regularly updated and available on the IRDiRC private website for use by its members.

IRDiRC poster was presented at:

- The European Society for Human Genetic Conference 2013 (8-11 June 2013, Paris, France)
- The American Society for Human Genetic Conference 2013 (22-26 October 2013, Boston, USA).

In addition, communication on IRDiRC is also done through regular publication in OrphaNews. OrphaNews is an electronic newsletter with 15,000 registered readers which is published bi-monthly. During the reporting period, 12 articles on IRDiRC and its activities were published.

Task 3.6 – Organisation of an annual IRDiRC conference for dissemination of information on IRDiRC activities and funded projects

The IRDiRC Executive Committee decided that there will not be an annual conference but fewer, depending on opportunities to organise such conferences and availability of resources.

The 1st IRDiRC conference, held in Dublin in April 2013, was organised by the European Commission with its own budget. The participation of the Scientific Secretariat was limited to the layout of the conference report (Deliverables D3.4) and the publication of the report, programme and speakers’ PowerPoint presentations on the IRDiRC website.

Since January 2014, work is ongoing to organise the 2nd IRDiRC conference to be held in Shenzhen, China on 7-9 November 2014, in collaboration with BGI. Three committees were established to manage the conference (Local Committee, Scientific Committee and International Organising Committee). Four teleconferences were organised between January and March 2014 to discuss the general organisation and the scientific program. The website for the conference is being

set up by the Scientific Secretariat in partnership with the University of McGill, Montreal, Canada. The staff of the Scientific Secretariat are member of the Organising Committee of this conference and manages the activities of the Scientific Committee of the conference.

Task 3.7 – Organisation of a satisfaction survey of key stakeholders

No deliverable is due for this task on the first reporting period.

- Deviation

The first edition of the annual report on “State-of-Art research in the field of rare Diseases” to disseminate IRDiRC outcomes (Deliverable D3.3) was not published at months 18 as previously planned as the collection of research projects funded by IRDiRC members was delayed due to the difficulty in obtaining the information. The first edition will be published at month 36.

Presence of IRDiRC on social media (Milestone MS4) was not established as members of the Executive Committee decided it was not necessary.

The flyer, poster and PowerPoint presentations are updated when necessary (new members or information, participation to a meeting) and do not follow the schedule planned in Milestone MS5.

- Statement on the use of resources

Most of the work was conducted by Partner 1, with in kind help from employees of Orphanet.

Time allocation of the staff for the WP:

Assistant: 10%

Communication manager: 60%

Project manager: 25%

Research assistant: 10%

3.2.3 Project management during the period

The overall purpose and mission of the “Management” work package (WP4) is to ensure the oversight, coordination and monitoring of the project in order to deliver as planned. Two partners were implicated in this project. As both partners are located in the same building, communication and coordination was easy.

- Consortium management tasks and achievements;

Task 4.1 – Contractual, financial and legal management

The Institut National de la Santé et de la Recherche Médicale (Inserm) is the coordinator and leader for each WP. Inserm has the structure necessary to provide contractual, financial and legal help.

Task 4.2 – Work plan definition

The work plan was established in the first monthly meeting of the project that was held in January 2013, once the staff was recruited. Since then, weekly meetings of the staff, with the project

coordinator, allow the review of the action plan of each staff and solve practical problems. In addition, the work plan is also discussed at the IRDiRC Executive Committee meetings as the Scientific Secretariat is intended to support IRDiRC activities.

Task 4.3 – Periodic reporting

A formal management meeting with all project members (Partner 1 and 2) and key people from Orphanet, is held monthly to report on the activities and monitor progresses towards goals (see dates below in the section “List of project meetings, dates and venues”). Reports of these meetings are available on the IRDiRC private website.

- Statement on the use of resources

Time allocation of the staff for the WP:

Assistant: 20%

Communication manager: 0%

Project manager: 15%

Research assistant: 0%

- Problems which have occurred and how they were solved or envisaged solutions;

The project manager, hired at the beginning of the project, quit after a few months in January 2013, to accept a permanent position in her country of origin (Portugal). She was immediately replaced by a new project manager.

The communication manager hired at the beginning of the project also suddenly quit for personal reasons in February 2013 but was only replaced in June 2013, leaving the project without a communication manager for 3 months. During this period, the tasks of the communication manager were divided between other employees. Although it delayed some of the work (setting up the private website, preparation of PowerPoint presentation, etc.), essential communication tasks (update of the public website, flyers and poster preparation, etc.) were carried out.

- Changes in the consortium, if any;

Partner 2 (Fondation maladies rares) requested to withdraw at the end of the first reporting period as this partner became an official IRDiRC member. All its tasks will be performed by the remaining unique partner, Inserm but without the in-kind contribution of Partner 2.

- List of project meetings, dates and venues;

Monthly meetings of the project were held in Paris, France, at the Rare Disease platform where both partner are located.

Date of the meetings:

- 13 January 2013
- 22 February 2013
- 22 March 2013
- 26 April 2013
- 23 May 2013
- 21 June 2013
- 12 July 2013
- 17 September 2013

- 8 October 2013
- 12 November 2013
- 10 December 2013
- 4 February 2014
- 3 March 2014

- Project planning and status;

In the next few months, the activities within the WPs will continue, with a particular emphasis on tasks that need improvement (collection of policy and guidelines, assessment of current results of research) or require a significant amount of time (entering IRDiRC funded project in Orphanet database) to enable to publication of a “State-of-Art” report in month 36. In addition, a performance review will be conducted.

A new staff will be hired to help organising the conference in Shenzhen and developing the “State of Art” report.

- Impact of possible deviations from the planned milestones and deliverables, if any;

Mapping of all IRDiRC projects at month 11 (Milestone MS2) and publication of the first Annual report of the “State-of-Art” at month 18 were delayed due to initial difficulty in the collation of research projects funded by IRDiRC members. The purpose of these milestone and deliverable is to be able to draw a picture of the research in the field of rare diseases, thus help the different IRDiRC bodies to define, among others, priorities and gaps in this field. Nonetheless, as members of the Working Groups are specialists in their topic, they were able to define the first priorities to tackle for IRDiRC for the next 3 years (2013-2016), in coordination with the Scientific Committees. The need for a “State-of-Art” report was judged low priority by the Executive Committee. The scheduled publication of the “State-of-Art” at month 36 will be more instrumental in improving the communication than in influencing funding priorities, which are currently well-identified by the Scientific Committees.

- Development of the Project website, if applicable;

The project website describing the objective of the SUPPORT-IRDiRC, its members and activities was created (<http://www.support-irdirc.eu/>). It gives access to the main IRDiRC website which contains all the documents produced by the IRDiRC thanks to the support of “SUPPORT-IRDiRC”.