	EUROPEAN COMMISSION RESEARCH AND INNOVATION DG	Periodic Report

Project No: 305207

Project Acronym: SUPPORT-IRDiRC

Project Full Name: Support for international rare disease research to serve the IRDiRC objectives

Periodic Report

Period covered: from 01/10/2012 **to** 31/03/2014

Start date of project: 01/10/2012

Project coordinator name:
Dr. Ségolène AYME

Version: 1

Date of preparation: 26/05/2014

Date of submission (SESAM): 27/05/2014

Project coordinator organisation name:
INSTITUT NATIONAL DE LA SANTE ET DE LA RECHERCHE MEDICALE (INSERM)

Periodic Report

PROJECT PERIODIC REPORT

Grant Agreement number:	305207
Project acronym:	SUPPORT-IRDiRC
Project title:	Support for international rare disease research to serve the IRDiRC objectives
Funding Scheme:	FP7-CSA-SA
Date of latest version of Annex I against which the assessment will be made:	05/10/2012
Period number:	1st
Period covered - start date:	01/10/2012
Period covered - end date:	31/03/2014
Name of the scientific representative of the project's coordinator and organisation:	Dr. Ségolène AYME INSTITUT NATIONAL DE LA SANTE ET DE LA RECHERCHE MEDICALE (INSERM)
Tel:	+33 1 56 53 81 37
Fax:	+33 1 56 53 81 38
E-mail:	segolene.ayme@inserm.fr
Project website address:	http://www.support-irdirc.eu/

Declaration by the scientific representative of the project coordinator (1)

I, Dr. Ségolène AYME INSTITUT NATIONAL DE LA SANTE ET DE LA RECHERCHE MEDICALE (INSERM) , as scientific representative of the coordinator of the project SUPPORT-IRDiRC and in line with the obligations as stated in Article II.2.3 of the Grant Agreement declare that:

The project has achieved most of its objectives and technical goals for the period with relatively minor deviations.

The attached periodic report represents an accurate description of the work carried out in this project for this reporting period.

The public website is up to date.

To my best knowledge, the financial statements which are being submitted as part of this report are in line with the actual work carried out and are consistent with the report on the resources used for the project (section 6) and if applicable with the certificate on financial statement.

All beneficiaries, in particular non-profit public bodies, secondary and higher education establishments, research organisations and SMEs, have declared to have verified their legal status. Any changes have been reported under section 5 (Project Management) in accordance with Article II.3.f of the Grant Agreement.

Name	Dr. Ségolène AYME INSTITUT NATIONAL DE LA SANTE ET DE LA RECHERCHE MEDICALE (INSERM)
Date	27/05/2014

This declaration was visaed electronically bySegolene AYME(ECAS user name naymease) on 27/05/2014

1. Publishable summary

Summary description of project context and objectives

Context
Research into rare diseases is badly needed as many patients still lack proper diagnosis and most of them are left without effective treatments. It is also an area where experts are rare as well. In addition, research in this area is very relevant from a scientific point of view as rare diseases are model diseases for more common disorders and are strong drivers of innovation. However, research in this field faces some specific constraints stemming from the low prevalence that represents the defining characteristic of rare diseases, the sheer number of rare diseases (an estimated 6,000 to 8,000 depending on the granularity of the disease definition) and their high phenotypic heterogeneity. Increasing the number of diagnostic and therapeutic options for patients suffering from rare diseases requires an acceleration of the identification of yet unknown critical genes, an improvement of gene defects identification, an increase in the knowledge of pathophysiology and natural history of rare diseases, an identification of potential therapeutic targets, discovery of new biomarkers and definition of appropriate surrogate end-points to adequately evaluate treatments and therapies.

It is now recognised that a global coordination of efforts is necessary to raise substantial investment. This awareness has led to the launch of the International Rare Diseases Research Consortium (IRDiRC), whose mission is to coordinate and foster international collaborative research on rare diseases, with the ambitious objectives of developing 200 new therapies and producing diagnostic tools for a majority of rare diseases by 2020. The idea behind the creation of IRDiRC is to provide a framework for the international effort with a development of necessary policies that will foster international collaboration. Policies in priority areas of research will allow the coordination of rare disease research worldwide in order to avoid duplication, fragmentation, redundancy and research gaps, and therefore ensure rapid translation of results from invested research into diagnostics and treatments that are beneficial to patients.

Objectives
The overall objective of SUPPORT-IRDiRC is to provide organisational and communication support to IRDiRC and its various members, and thereby contribute to the development of policies and guidelines aimed at accelerating research on rare diseases, and reinforcing international research cooperation. To this aim, SUPPORT-IRDiRC has gathered a group of motivated and highly experienced investigators who will work together during six years to help IRDiRC members remain on the right track in order to reach its ambitious objectives.

Specific objectives of this project include:
a) Support the work of the IRDiRC bodies – an Executive Committee, three Scientific Committees and 12 Working Groups - by organising meetings and teleconferences, assuring secretarial work for them, keeping all members updated, and assuring a smooth organisation of the consortium's work (WP1).
b) 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 (WP2).
c) 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. (WP3).
d) 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 (WP4).

Description of work performed and main results

During the first 18 months of the project, SUPPORT-IRDiRC achieved significant results and the project developed as planned:
- Work procedures were established for each of the IRDiRC bodies (Executive Committee, Scientific Committees, and Working Groups) in agreement with the chairs of the Committees. A regular communication with and between the chairs of the Executive Committee and Scientific

Committees was instituted to ensure smooth running of the consortium.

- Support was provided in the organisation of meetings and teleconferences of the IRDiRC bodies. The secretarial activities for the Executive Committee were relocated to SUPPORT-IRDiRC at month 7 of the project when the chairmanship of the Executive Committee was transferred from the European Commission to another member of IRDiRC. In the past 11 months, SUPPORT-IRDiRC organised one face-to-face meeting and two teleconferences for the Executive Committee. Since the beginning of the project, SUPPORT-IRDiRC organised a total of six meetings and five teleconferences for the three Scientific Committees and a total of three meetings and 28 teleconferences for the 12 Working Groups.

- Secretarial assistance to IRDiRC bodies mostly included: maintaining an up-to-date list of members, providing agenda and preparatory documents ahead of meetings/teleconferences in agreement with the Chair of the body, drafting reports of these meetings/teleconferences for approval by members of the body, and sending invitation to members newly nominated to a Scientific Committee or a Working Group.

- To facilitate the cooperation between the stakeholders in the field of rare diseases, SUPPORT-IRDiRC collected information on research projects funded by IRDiRC members for diffusion on the IRDiRC public website and the Orphanet database. Other data collection includes policies and guidelines relevant to the field, to be published on the IRDiRC website. The collected data provide relevant information to IRDiRC bodies in order to determine the gaps and opportunities in the field of rare disease, and to suggest actions that foster research in this field to help IRDiRC to reach its goals. A password-protected private website was launched to facilitate communication among IRDiRC members.

- To foster collaboration in the field of rare diseases, SUPPORT-IRDiRC participated in the organisation of two workshops, “The 2013 international workshop on rare disease system for pathogenicity inference” (18-19 April 2013, Dublin, Ireland) and the “Rare genetic diseases: diagnosis and discovery workshop” (3 December 2013, Prague, Czech Republic), that gathered experts in these fields.

- Communication tools were developed in the past 18 months, including a logo, graphical chart, flyer, poster, PowerPoint presentations, monthly newsletters and a public website to keep members of the consortium as well as the public informed. The public website (http://www.irdirc.org/), launched in January 2013 provide information on IRDiRC and its activities, and also information about rare diseases, rare diseases research, and news from the field of rare diseases.

- Management of the project itself between the two partners involved is facilitated by a close physical proximity of the two partners, good communication, and monthly reporting.

Deviation from the plan included a delay in the publication of the first annual report of the “State-of-Art” due to a delay in the gathering of research projects funded by IRDiRC members.

Expected final results and potential impacts

Increasing the number of diagnostic and therapeutic options for patients suffering from rare diseases requires an acceleration of the identification of yet unknown critical genes, an improvement of gene defects identification, an increase in the knowledge of pathophysiology and natural history of rare diseases, an identification of potential therapeutic targets, discovery of new biomarkers and definition of appropriate surrogate end-points to adequately evaluate treatments and therapies. This require international collaborative research in rare diseases to avoid duplication, fragmentation, redundancy and research gaps, and therefore ensure rapid translation of results from invested research into diagnostics and treatments that are beneficial to patients.

One expected final result is the reinforcement of international cooperation in research on rare diseases, through the development of policies and guidelines aimed at accelerating such research. This will be reached by reinforcing international cooperation between funding agencies, between academia, industry and patient organisations, as well as between and within research group/clinicians. In the past few years, collaboration between industry and patients association

started to develop, as well as collaboration between funding agencies. IRDiRC members include funding agencies, academia, industry and patient association, spread across 13 countries in four continents, creating a favourable eco-system to foster international collaborations.

Another expected result is the establishment of standard policies and guidelines aiming at accelerating research on rare diseases and at reinforcing international research cooperation, especially in the field of clinical and biological databases, through support to the elaboration of common SOPs, of a harmonised ethical approach and of unified rules to access patient data and samples. Areas where such policies and guidelines are necessary include genotypic and phenotypic data, animal and cellular models, data and samples collection. In addition, the sharing of databases, ontologies, and clinical trial information will foster research and international collaborations.

The project should contribute to the IRDiRC goals, i.e., finding a diagnostic for most rare diseases and developing 200 new therapies. Reaching these goals would be strongly beneficial to rare disease patients who often struggle for several years to obtain a diagnostic for a disease that, for most of the time, does not have an available treatment.

Project public website address:	http://www.support-irdirc.eu/

2. Core of the report

Project objectives, Work progress and achievements, and project management during the period

The Project Summary Pdf document contains the core of the report.

3. Deliverables and milestones tables

Deliverables (excluding the periodic and final reports)

Del. no.	Deliverable name	Version	WP no.	Lead beneficiary	Nature	Dissemination level	Delivery date from Annex I (proj month)	Actual / Forecast delivery date	Status	Comments
1	Preparatory documents and Post-meeting repo rts	0.0	1	INSTITUT N ATIONAL DE LA SANTE ET DE LA RECHER CHE MEDICALE (INSERM)	Other	CO	72	30/09/2018	Not submitted	
1	Collection of guidelin es and standards of i nterest in the field o f R&D for rare diseas es	0.0	2	INSTITUT N ATIONAL DE LA SANTE ET DE LA RECHER CHE MEDICALE (INSERM)	Report	PU	72	30/09/2018	Not submitted	
2	Online collaboration p lat form	0.0	2	INSTITUT N ATIONAL DE LA SANTE ET DE LA RECHER CHE MEDICALE (INSERM)	Other	RE	6	31/03/2013	Not submitted	
3	Data portal for IRDiRC -affiliated investiga tors	0.0	2	INSTITUT N ATIONAL DE LA SANTE ET DE LA RECHER CHE MEDICALE (INSERM)	Other	RE	6	31/03/2013	Not submitted	Data portal not subm itted as not consider necessary by Commi ttees members
1	Logo, graphical charter a nd official website of IR DiRC	0.0	3	INSTITUT N ATIONAL DE LA SANTE ET DE LA RECHER CHE MEDICALE (INSERM)	Report	PU	3	31/12/2012	Not submitted	
2	IRDiRC Communication s trategy and communica tion plan	0.0	3	INSTITUT N ATIONAL DE LA SANTE ET DE LA RECHER CHE MEDICALE	Report	PU	3	31/12/2012	Not submitted	

				(INSERM)						
3	Annual report on “State-of-Art of Research in the field of Rare Diseases”	0.0	3	INSTITUT NATIONAL DE LA SANTE ET DE LA RECHERCHE MEDICALE (INSERM)	Report	PU	72	30/09/2018	Not submitted	Delayed due to the difficulties to collect the necessary information from IRDiRC members
4	IRDiRC annual conference related documents	0.0	3	INSTITUT NATIONAL DE LA SANTE ET DE LA RECHERCHE MEDICALE (INSERM)	Other	PU	72	30/09/2018	Not submitted	
1	Performance review report	0.0	4	INSTITUT NATIONAL DE LA SANTE ET DE LA RECHERCHE MEDICALE (INSERM)	Report	CO	27	31/12/2014	Not submitted	Not due for the first reporting period

Milestones

Milestone no.	Milestone name	Work package no	Lead beneficiary	Delivery date from Annex I	Achieved Yes/No	Actual / Forecast achievement date	Comments
1	Working processes between IRDiRC Committees established	1	1	31/12/2013	Yes	15/12/2013	
2	Registration in Orphanet of all IRDiRC projects	2	1	30/09/2013	Yes	30/09/2013	
3	Annual policy report to the Executive COmmittee and the three Scientific Committees	2	1	30/09/2013	Yes	30/09/2013	
4	Establish presence on social media	3	1	31/10/2012	No	15/05/2015	Not established as considered not necessary by Executive Committee members
5	Flyers, posters, powerpoint presentations	3	1	31/03/2013	Yes	30/03/2013	

6	Work plan and internal communication plan def inition	4	1	31/10/2012	Yes	31/10/2012	

4. Explanation of the use of the resources

The **explanation on the use of resources** was removed from the scientific periodic reports in SESAM. These deta ils now have to be entered in the cost statement forms in FORCE instea d.

Attachments	SUPPORT-IRDiRC_Periodic_Report1_Core_report.pdf
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Period covered - start date:	01/10/2012
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This declaration was visaed electronically by Segolene AYME (ECAS user name naymease) on 27/05/2014