EJP RD

European Joint Programme on Rare Diseases

H2020-SC1-2018-Single-Stage-RTD
SC1-BHC-04-2018
Rare Disease European Joint Programme Cofund

Grant agreement number 825575

D7.3

List of funded networks in the Networking Support Scheme due at Month 21

Organisation name of lead beneficiary for this deliverable:

Partner 34-ZonMw

Due date of deliverable: month 21

Dissemination level:

Public

Description

The aim of the Networking Support Scheme (NSS) is to encourage sharing of knowledge on rare diseases and rare cancers of health care professionals, researchers and patients. The NSS also aims to enable or increase the participation of usually underrepresented countries in Europe in new and in existing research networks on rare disease(s) or rare cancer(s).

The scheme provides financial support to applicants for fostering organization of workshops or conferences for new research networks or existing/expanding research networks. The focus of these workshops or conferences should be exchange of (the implications of) research results and innovative solutions as well as to strengthening the collaborations between different stakeholders. The results of these networking events may lead to future collaborative and novel research efforts, e.g. by applying to research funding within the EJP RD (e.g. Joint Transnational Calls), to COST actions or Marie Skłodowska-Curie Actions (mobility and training calls). The applying consortium is allowed to invite other participants to join the networking event. There is no limit to the number of participants; however, the maximum budget that a consortium can request is € 30,000 for a networking event.

The first round opened Mid-December 2019 and the 9 submitted applications were collected on March 3, 2020 at 14:00 CET. All 9 submitted applications were eligible. These applications were evaluated in May 2020 by the Networking Evaluation Committee (NEC). In addition to experts in clinic and research in rare diseases, the panel also included representatives of patient advocacy organisations. An independent observer was present at the plenary evaluation meeting. 6 proposals were selected for funding for a total requested budget of 168,619€.

The abstract of projects selected for funding are available in the Annex I.

Due to the Covid-19 crisis, the collection date for the second round of application planned for June 2020 was skipped and postponed to the next round in September 2020.

Networking Support Scheme list of proposals selected for funding at Month 21

Project title	Principal applicant
ROUND 1	
European Aniridia Conference 2020	D.M. McKay (UK)
European Neurofibromatosis meeting 2020, transporting knowledge between NF patients, scientists and clinicians	R. Oostenbrink (NL)
Cilia2020 - Interconnect	B. Schermer (DE)
The AKU Scientific Conference	N. Sireau (UK)
Wiscott Aldrich Syndrome 2020	A. Kedar (IL)
Newborn screening in Inherited Metabolic Diseases; multi-stakeholder meeting moving towards equity and a common approach	M. Scarpa (IT)

ANNEX I: Abstract of projects selected for funding

ROUND 1

European Aniridia Conference 2020

Partner	Country
Dave McKay **Aniridia Network**	**United Kingdom**
van Heyningen, Veronica University College London	United Kingdom
Sánchez de Vega, Rosa Aniridia Europe (European Aniridia Federation)	Norway
Moosajee, Mariya UCL Institute of Ophthalmology, Moorfields Eye Hospital, Great Ormond Street Hospital for Children and the Francis Crick Institute, London	United Kingdom
Bylė Irma Association "AniridijaLT"	Lithuania
Lima Cunha, Dulce UCL Institute of Ophthalmology	United Kingdom
Damante, Giuseppe University of Udine	Italy
Hall, Hildegard Nikki MRC Human Genetics Unit, MRC Institute of Genetics and Molecular Medicine, University of Edinburgh and Princess Alexandra Eye Pavilion, Edinburgh, Scotland, UK	United Kingdom
Grupcheva, Christina Varna Medical University	Bulgaria
Tsoneva, Elena Association Aniridia Bulgaria	Bulgaria

Summary:

This event enables sharing of specialist knowledge about the rare genetic eye condition aniridia. Its goal is to develop better understanding to tackle sight loss and other effects of to aniridia.
Professionals such as: ophthalmologists, researchers, vision scientists, and geneticists will gather with people who have aniridia and their relatives, to upskill each other.
Aniridia is visual impairment present at birth. Most people with aniridia have all orpart of their irises (the coloured rings in the eyes) missing. Other parts of the eye are typically under-developed. Other conditions often lead to further sight loss.
Aniridia is usually caused by an abnormality in a gene called PAX6. It also controls brain and pancreas functions. So patients may also have disturbed sleep and predispositions to obesity or diabetes. It can occur as part of more significant condition known as WAGR/11p Deletion Syndrome.
Understanding aniridia is challenging, due to the scattered patient population, its highly variable impact and complications of linked conditions. This event innovatively

addresses this by bringing all the stakeholders together for 3 days Researchers and clinicians will discuss their latest work and experience of the disease, with contributions from patients. New approaches to clinical management, drug development, and stem cell therapy will be presented.
There will also be tours of the laboratories at UCL Institute of Ophthalmology. Patients and relative have the unique chance to get a free 30 minute consultation with the world's top aniridia experts at Moorfields Eye Hospital.

European Neurofibromatosis meeting 2020, transporting knowledge between NF patients, scientists and clinicians

Partner	Country
Oostenbrink, Rianne ErasmusMC	**The Netherlands**
Topilko, Piotr Institut Mondor de Recherche Biomedicale	France
Varan, Ali Hacettepe University, Institute of Oncology	Turkey
Melegh, Bela University of Pecs	Hungary
Rohl, Claas NF Patients United – Global Network of NF Support Groups NFPU	Austria
Legius, Eric University of Leuven	Belgium
Korycinska, Dorota Stowarzyszenie Neurofibromatozy Polska Alba Julia	Poland
Karwacki, Marek Child health Institute, Medical University Warsaw	Poland
Koczkowska, Magdalena Medical University of Gdansk	Poland
Piotrowski, Arkadiusz Medical University of Gdansk	Poland

Summary:

Encompassing three conditions, NF1, NF2 and Schwannomatosis, neurofibromatosis is a rare disease which has highly variable presentation of symptoms and complications affecting approximately 2.5 million patients globally. For the NF patients, the main issues are: 1. Access to diagnostic tools; 2. Access to optimal care; and 3. Limited availability of therapeutic options. In 2020 the European NF meeting will take place in Rotterdam, The Netherlands, with the focus on sharing knowledge among NF patients, scientists and clinicians. Bringing together all different disciplines involved in NF research and care, the European NF meeting offers a forum for advancing basic, translational, and clinical research in NF and related fields, with one purpose: to

improve the quality of life of NF patients. A unique character of the European NF meeting is its parallel 1-day session for NF patients and their representatives organized by the local patient advocacy organization. The networking event aims to share clinical expertise of NF and to build scientific research consortia with NF health professionals and NF patients on a broad European level. We in particular aim to outreach patients and health professionals from underrepresented European countries. This specific strategy is a novelty compared to previous meetings, lacking health care professional and patient representative visitors of underrepresented European countries thus far. The Networking Support Scheme grant will be used to support participants from European underrepresented countries visiting the European NF meeting 2020.

Cilia2020 – Interconnect

Partner	Country
Schermer, Bernhard **University Hospital of Cologne**	**Germany**
Firat-Karalar, Elif Nur Koç University	Turkey
Čajánek, Lukáš Masaryk University	Czech Republic
Wloga, Dorota Nencki Institute of Experimental Biology, Polish Academy of Sciences	Poland
Harris, Tess Ciliopathy Alliance	United Kingdom
Mill, Pleasantine University of Edinburgh	United Kingdom
Blacque, Oliver University College Dublin	Ireland
Jurisch Yaksi, Nathalie Norwegian University ofScience and Technology	Norway
Meunier, Alice CNRS	France

Summary:

Cilia are tiny, antennae-like cell organelles that can be found on almost all cells of the human body. Research has revealed, that a growing number of genetic diseases result from defects in cilia collectively termed the 'ciliopathies'. Some cilia are motile and involved in moving either liquids or cells within the body, whilst other cilia are immotile and transmit key signals from outside to the cell's interior. Since cilia occur within virtually all tissues, the spectrum of cilia-associated diseases is very broad, affecting the kidneys, lungs, brain, eyes and many more. The ciliopathies represent a spectrum of ~40 overlapping syndromes caused by mutations in nearly 200 genes.

Whilst often very rare individually, collectively the ciliopathies are thought to affect 1:1000 births. In October, basic researchers and clinicians from all over the world will meet in Cologne at "Cilia2020". As the largest cilia conference, it uniquely integrates patients and their representatives in the program with the aim of promoting bidirectional interactions between scientists and those impacted by ciliopathies. In 2020, we aim to expand this meeting to countries typically underrepresented in any international research networks and to enlarge the community. Our "Cilia2020 – Interconnect" satellite events aim to sustainably enhance diversity of the cilia/ciliopathy community within and around Europe. This will accelerate gene discovery and improve understanding of disease mechanisms, as well as facilitating development of much needed therapies for ciliopathies.

The AKU Scientific Conference

Partner	Country
Sireau, Nicolas **The AKU Society**	**United Kingdom**
Santucci, Annalisa Università degli Studi di Siena	Italy
Lakshminarayan, Ranganath Royal Liverpool Hospital, Liverpool University Hospitals	United Kingdom
Zatkova, Andrea Institute for Clinical and Translational Research	Slovak Republic
De Kock, Joery Vrije Universiteit Brussel (VUB), Faculty of Medicine and Pharmacy	Belgium
Imrich, Richard Institute of Clinical and Translational Research, Biomedical Research Center, Slovak Academy of Sciences	Slovakia
Kujawa, Mariusz Institution Medical University of Gdansk	Poland
Gallagher, James University of Liverpool	United Kingdom

Summary:

Alkaptonuria, also known as AKU or Black Bone Disease, is an extremely rare genetic condition, which can cause significant damage to the bones, cartilage and tissues of those affected. AKU normally only affects one in every 250,000 people worldwide. It causes a build-up of a substance called homogentisic acid (HGA), which binds to cartilage and bone and turns tissues black, in a process called ochronosis. This causes severe early onset osteoarthritis. The nature of the disease leads to major disability and long-term pain.

The AKU Scientific Conference will facilitate the sharing of knowledge about the condition among AKU world experts and aspiring scientists wishing to excel in AKU

research in the future. It will focus on next steps in research following the recently ended DevelopAKUre clinical trials, a future gene therapy, an upcoming children's study and a co-therapy for patients to take alongside the vital drug nitisinone - a drug which improves symptoms and signs of AKU. This conference will be key for pushing forward the next exciting stages of AKU research as we now have a successful drug that is on its way to be licensed, and we are looking to advance research to cure AKU. This conference will be held at Smolenice Castle near Piešťany, Slovakia, the hub of European research into AKU. Our conference will be the first steps towards the final stage of the AKU mission, to find a cure for this debilitating disease.

The third international symposium for researchers and clinicians on Wiskott Aldrich Syndrome

Partner	Country
Kedar, Amir **The Israeli Wiskott Aldrich Syndrome Association (R.A)**	**Israel**
Thrasher, Adrian UCL Great Ormond Street Institute of Child Health	United Kingdom
Albert, Michael Dr. von Hauner University Children's Hospital, LMU	Germany
Villa, Anna IRCCS Ospedale San Raffaele	Italy

Summary:

The third international symposium for researchers and clinicians on Wiskott Aldrich Syndrome is being held in London UK and offers access to the latest research and analysis related to this rare disease. The participants will gain valuable insights into innovative perspectives in both basic and clinical research. The scientific programme will draw together experts from around the world to discuss breakthroughs in basic research, advances in clinical practice, novel therapeutic approaches and new insights into stem-cell and cellular therapies. This unique networking event is ideal forum to share knowledge, connect with colleagues and grow professional network. The goals of this meeting are:

- Expanding WAS/XLT and WASp research.
- Bridging the gap between basic and clinical research to speed up applications
- Foster collaboration among researchers.
- Attract young researchers to focus on WAS/XLT and WASp

Our three keynote lectures present the most recent knowledge in the research and clinical field of WAS. Prof. David Rawlings will talk about "Lessons learned regarding immune tolerance and progress towards new therapies for WAS", Prof. Anna Villa will shed light on "Platelets defects in Wiskott-Aldrich Syndrome" and Prof. Michael Albert will discuss about "HSCT for WAS – What have we learned in 50 years and what promises does the future hold?". As in our previous events, unpublished data regarding WAS and

WASp research will be presented and discuss among participants. The data presented and the different topics discussed in this event are rarely accessible elsewhere.

Newborn screening in Inherited Metabolic Diseases; multistakeholder meeting moving towards equity and a common approach

Partner	Country
Prof. M. Scarpa **Azienda Sanitaria Universitaria Friuli**	**Italy**
Dr. M. Jansen Netherlands National Institute for Public Health and the Environment	The Netherlands
Prof. dr. M. Cornel Amsterdam University Medical Center	The Netherlands
Dr. T. Tangeraas OUH Rikshospitalet	Norway
Prof. V. Kozich Charles University-1st Faculty of Medicine and General University Hospital	Czech Republic
Mr. J. Bonham Sheffield Childrens NHS FT	United Kingdom
M.Sc. V. Hedley Newcastle University Translational and Clinical Research	United Kingdom
Prof. dr. M. Baumgartner University Children's Hospital Zurich Eleonore Foundation	Switzerland

Summary:

There are big differences between European countries when it comes to newborn screening (NBS): for example in terms of different number of screened diseases with national programmes for screening range from 1-26 diseases, or Italy offering by law screening for at least 40 diseases.
Taking into consideration the overall needs and priorities regarding health conditions and health system resources, we think there is room for a feasible improvement on NBS programmes in EU by means of a shared consensus document (roadmap), especially if it can benefit from synergies deriving from a coordinated action of the EU. Thus, we consider it crucial to initiate a broad discussion involving the whole spectrum of stakeholders participating in debates about Newborn Screening (NBS) such as representatives of Scientific Organisations, Patient representatives, MetabERN and other stakeholders.
We therefore organize two meetings in 2020 to drive this topic forward and deepen collaboration between stakeholders. The first will be a brainstorming session (28 April 2020) organized by MetabERN and the second one (this application) falls under the EJP Networking Scheme, which will integrate this focused work with a wider audience of stakeholder perspectives, to advance the topic of newborn screening more

broadly. This meeting will be organised back-to-back with the annual conference of the International Society for Newborn Screening (ISNS).
It will build upon past discussions and first steps from Member States under the EU Committee of Experts on Rare Diseases (EUCERD), to reignite discussions on areas for potential European-level collaboration. The final result will be a paper in which we reach consensus on concrete steps toward identifying potential barriers and finding common grounds for the newborn screening, as a system involving pre-post born management, follow up of the affected child, family assistance together with some technical discussion regarding criteria for the expansion of the newborn screening offer. The outcome of this process will also be a roadmap for policy-makers, the scientific community and advocacy organisations.