Summaries NSS Applications round 1 2020

Application nr.	Title	Summary	Comments - ethics
40-46300-98-1002	**European Aniridia Conference 2020**	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 or part 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.	The only reccomendation for the organisers is related to the involvement of patients and their related personal data in the event. Patients should be contacted to take part in this event only by their caregivers/people having access to their medical data and **being aware of their medical condition** or by their patients' association or if they realeased their consent to be contacted for events related to their diseases. However, they may also directly contact the organiser following the advertements of the event. Where applicable, the organiser should also guarantee that the patients sign an informed consent form allowing the use of data/photos/videos collected during the event for dissemination or other purposes. If some patients do not allow the use of data/share of photos/videos for any purpose, the organiser has to ensure that data/photos/videos for these patients will not be used for dissemintation activities.The applicants may also refer to the indications provided in section **5.2.3.Ethical evaluation** of the call.
40-46300-98-1003	**European Neurofibromatosis meeting 2020, transporting knowledge between NF patients, scientists and clinicians**	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.	The only reccomendation for the organisers is related to the involvement of patients and their related personal data in the event. Patients should be contacted to take part in this event only by their caregivers/people having access to their medical data and **being aware of their medical condition** or by their patients' association or if they realeased their consent to be contacted for events related to their diseases. However, they may also directly contact the organiser following the advertements of the event. Where applicable, the organiser should also guarantee that the patients sign an informed consent form allowing the use of data/photos/videos collected during the event for dissemination or other purposes. If some patients do not allow the use of data/share of photos/videos for any purpose, the organiser has to ensure that data/photos/videos for these patients will not be used for dissemintation activities.The applicants may also refer to the indications provided in section **5.2.3.Ethical evaluation** of the call.
40-46300-98-1004	**Responsible use of genome editing tools in rare disease research : a multi-stakeholder interactive workshop**	What if we could cure patients with genetic diseases by editing the responsible genes ? Our genomes are made of a material called DNA which encodes information on how to make proteins. The information to make each protein is called a gene. When a gene is changed (mutated), duplicated, or deleted, part of the DNA may be changed in such a way that the protein it encodes no longer functions correctly. Genome editing tools are revolutionizing sciences and medicine. The new generation led by CRISPR-Cas enables scientists to make precise modifications in targeted DNA sequences – in other words, they allow for the insertion, removal or replacement of a given sequence. Among their many potential applications, such technologies raise significant hopes for the treatment of rare diseases, 70% of which have a genetic cause. But what is the current state of medical research? What are the ethical and societal concerns at stake? How can we empower and engage patients on these topics? This interactive workshop will bring together a broad range of stakeholders - including scientists, ethicists, policymakers, representatives from patient organisations, industry and regulatory agencies in Europe – to explore issues relating to the use of genome editing tools for RD research. The original format of this 1,5 day workshop, involving presentations on the latest scientific and medical advances and thematic break-out sessions, aims at facilitating communication between all stakeholders and developing a set of key recommendations to ensure responsible use of these novel technologies in RD research.	No patients involved in the event, therefore no sensitive/health data are planned to be processed. The applicants may refer to the indications provided in section **5.2.3.Ethical evaluation** of the call.
40-46300-98-1005	**Alex TLC Community Weekend Conference 2020 (scientific presentations and round tables)**	Leukodystrophies are genetic disorders primarily affecting the white matter of the central nervous system (i.e. the brain or spinal cord). The majority are degenerative, causing symptoms such as impaired mobility, vision, speech and hearing, incontinence, inability to swallow and loss of cognitive skills. In some cases, they may cause death. There are over 90 leukodystrophies, with some so rare they are yet to be named. The Community Weekend Conference is a unique opportunity for patients, families, doctors, researchers and scientists to discuss how the various leukodystrophies affect individuals and families.Held in a central UK hotel, the event gives patients and family members the opportunity to talk openly about their condition and gives medical professionals the opportunity to hear directly from those affected, as well as learn from each other. From a clinical viewpoint, the weekend agenda includes round table discussions and presentations and presents a rare opportunity for major stakeholders to progress ideas which would otherwise take months, or even years, to develop; ideas such as 'What does best practice for leukodystrophy patients look like?' and 'Which symptoms are the most important for researchers to address?'. A key focus is to strengthen the relationships between patients, medical professionals and research bodies. Attending professionals are encouraged to mix with each other, patients and their families through evening schedules of entertaining and inclusive activities, with opportunities for stakeholder discussions and questions throughout the event.	The only reccomendation for the organisers is related to the involvement of patients and their related personal data in the event. Patients should be contacted to take part in this event only by their caregivers/people having access to their medical data and **being aware of their medical condition** or by their patients' association or if they realeased their consent to be contacted for events related to their diseases. However, they may also directly contact the organiser following the advertements of the event. Where applicable, the organiser should also guarantee that the patients sign an informed consent form allowing the use of data/photos/videos collected during the event for dissemination or other purposes. If some patients do not allow the use of data/share of photos/videos for any purpose, the organiser has to ensure that data/photos/videos for these patients will not be used for dissemintation activities.The applicants may also refer to the indications provided in section **5.2.3.Ethical evaluation** of the call.
40-46300-98-1006	**Cilia2020 - Interconnect**	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 only reccomendation for the organisers is related to the involvement of patients and their related personal data in the event. Patients should be contacted to take part in this event only by their caregivers/people having access to their medical data and **being aware of their medical condition** or by their patients' association or if they realeased their consent to be contacted for events related to their diseases. However, they may also directly contact the organiser following the advertements of the event. Where applicable, the organiser should also guarantee that the patients sign an informed consent form allowing the use of data/photos/videos collected during the event for dissemination or other purposes. If some patients do not allow the use of data/share of photos/videos for any purpose, the organiser has to ensure that data/photos/videos for these patients will not be used for dissemintation activities.The applicants may also refer to the indications provided in section **5.2.3.Ethical evaluation** of the call.

40-46300-98-1007	**The AKU Scientific Conference**	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 severe 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 Piestany, 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.	No patients involved in the event, therefore no sensitive/health data are planned to be processed. The applicants may refer to the indications provided in section **5.2.3.Ethical evaluation** of the call.
40-46300-98-1008	**WAS2020**	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.	No patients involved in the event, therefore no sensitive/health data are planned to be processed. The applicants may refer to the indications provided in section **5.2.3.Ethical evaluation** of the call.
40-46300-98-1009	**Newborn screening in Inherited Metabolic Diseases; multi-stakeholder meeting moving towards equity and a common approach**	There are big differences between European countries when it comes to newborn screening (NBS). Taking into consideration the overall needs and priorities regarding health conditions and system resources, there is room for a feasible improvement on NBS programmes in EU by means of a shared consensus document (roadmap). We consider it crucial to initiate a broad discussion involving the whole spectrum of stakeholders participating in debates about 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 to deepen collaboration between stakeholders: a brainstorming session in April organized by MetabERN and a meeting under the EJP-NSS, which will integrate wider stakeholder perspectives to advance the topic of NBS. This meeting will be organised back-to-back with the 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 and reignite discussions on areas for potential European-level collaboration. The final result will be a consensus paper with steps toward identifying potential barriers and finding common grounds for NBS, involving pre-post born management, follow up of the affected child, family assistance and discuss technical discussion regarding criteria for the expansion of NBS . The outcome of this process will be a roadmap for policy-makers, the scientific community and advocacy organisations.	No patients involved in the event, therefore no sensitive/health data are planned to be processed. The applicants may refer to the indications provided in section **5.2.3.Ethical evaluation** of the call.
40-46300-98-1010	**European Inherited Optic Neuropathy (ION) Research Network**	In the light of the expansion of ERN-EYE to more than double in size with several new countries being represented for the first time, this event is proposed to relaunch Working Group 2 (Neuro-ophthalmology) and to focus on a specific group of diseases in order to encourage one or more transnational collaborative research projects under the auspices of ERN-EYE. Inherited Optic Neuropathies (IONs) are an under-researched disease area with high unmet need and the selection of this group of conditions allows for a broad multi-disciplinary approach while remaining specific enough to generate realistic research objectives from the meeting.	No patients involved in the event, therefore no sensitive/health data are planned to be processed. The applicants may refer to the indications provided in section **5.2.3.Ethical evaluation** of the call.