**BSGCT PPIE Award – APPLICATION FORM**

Nominations must be submitted by the advertised deadline to secretary@bsgct.org

**SECTION 1**

**APPLICANT’S NAME:**

James Arwyn-Jones

**APPLICANT’S AFFILIATION AND CONTACT DETAILS (including email address):**

UCL Great Ormond Street Institute of Child Health

**CONFIRMATION THAT APPLICANT IS A CURRENT BSGCT MEMBER:** Yes

**CONFIRMATION THAT APPLICANT WOULD BE AVAILABLE TO RECEIVE TROPHY AT THE NEXT BSGCT ANNUAL CONFERENCE:** Yes

**SECTION 2**

 **BRIEF STATEMENT FROM THE APPLICANT ADDRESSING THE JUDGING CRITERIA** (up to 1 page)**:**

***The Norrie Disease Conference 2023***

Norrie disease is an ultra-rare X-linked recessive condition, resulting in congenital blindness and early-onset progressive hearing loss. Our group led by Prof. Jane Sowden at the UCL Great Ormond Street Institute of Child Health are pursuing the development of a gene therapy to ameliorate hearing loss in Norrie disease.

***Motivation for the interaction***

In December 2023, in collaboration with the Norrie Disease Foundation, our group hosted a one-day event for anyone interested in learning about the latest in Norrie disease. This included clinicians and researchers, but was largely targeted towards patients and families living with the condition. The motivation for this event came from a need to bridge the gap between families, scientists and clinicians, to enable the sharing of information and encourage collaboration for this ultra-rare disease. Given its rarity, our group recognises that to make meaningful conclusions in clinical research for Norrie disease, it is vital to engage with the patient community to gain their insights and involve as many of them as possible in research design and as participants where feasible. Our programme included talks by two teenagers living with Norrie disease, as well as a parent of a child with the condition; all of whom shared their experiences of Norrie disease and highlighted important areas of focus for the research community – largely around the progressive hearing loss in Norrie disease. Talks and discussions were also delivered by experts in the field, with presentations on neurodevelopmental and retinal aspects of Norrie disease, hearing loss, underlying biological mechanisms of the disease, and therapeutic approaches for Norrie disease with lessons from other rare conditions.

***The nature of our PPIE activity***

A specific goal of the conference was to highlight the importance of PPIE and collaboration within our research, and PPIE was intertwined throughout the event. As well as an interactive sensory and music environment for younger patients, we designed a tactile interactive learning area for those who wanted to learn more about the science behind sight and sound, and ran a mock PPIE session with attendees at the end of the day’s schedule. The PPIE session was designed in conjunction with the UCLH BRC Hearing Health Theme PPIE Panel and their PPIE lead, and included salient points taken from NIHR’s PPIE briefing notes for researchers and learning modules from the BRC’s PPIE Panel training. It had the educational intent to highlight the importance of PPIE in translational research and encourage patients and families to get involved. First, an overview of PPIE was provided, with an explanation of what PPIE is, why it is important, and how it is practically done, including various examples in research. The session then progressed to interactive mock examples to engage the audience in discussion. Members of the audience were separated into groups online and in person, and given handouts (text and/or Braille) of mock Plain English Summaries for two fictional studies. These sparked lots of interaction between groups and the main discussion points were drawn back again to the core elements of successful PPIE in research.

***The impact of the interaction***

The impact of the PPIE session, and whole event in general, was extremely positive. Our research group found interactions with the patient community both rewarding and inspiring, and brought the experiences of families into sharp focus. As a result of hosting this event, we have developed research collaborations and met interested clinicians, but most importantly strengthened our relationship with a very engaged patient group. As part of our project in developing a gene therapy for hearing loss in Norrie disease, we are undertaking a natural history study of the disease to better understand its natural course and genotype-phenotype correlations, as well as gaining clinical insights necessary to inform the potential clinical trials of the future. Our event was instrumental in increasing our reach for recruitment of patients and families to the natural history study, and highlighting its existence to relevant clinicians who care for these patients. Through contacts made and our relationship with the Norrie Disease Foundation, we have increased our patient cohort more than three-fold and this now extends outside the UK across the world, with participants in Australia, Hong Kong, Iran, and beyond. We now have regular meetings with the Norrie Disease Foundation and continue to involve the community closely in our work to pursue therapeutic options.

**BRIEF STATEMENT FROM REPRESENTATIVES OF THE PATIENTS INVOLVED, ASSESSING THE EXPERIENCE FROM THEIR POINT OF VIEW** (up to 1 page):

***Statement from Wendy Horrobin; mother to a son with Norrie disease & founder of the Norrie Disease Foundation***

In 2016 when the Norrie Disease Foundation (NDF) was founded, the NDF asked the UK community (at that time around 20 families were known to the NDF) what was important to them and what the charity could do to support them. The community told the NDF that they wanted to find a treatment for the devastating hearing loss which commonly starts in childhood and robs children already born blind, of their most valuable sense, the hearing. They also told us they wanted to be connected to more families.  At this point there was no research in the world doing this, and there was little reliable information on the condition. Families felt isolated and alone. In December 2023 the first Norrie Disease Research conference took place in London at UCL Great Ormond Street Institute of Child Health (GOS ICH). The Norrie Disease Foundation and Professor Jane Sowden’s research group at UCL Great Ormond Street Institute of Child Health (GOS ICH) came together with patients, families and clinicians in a milestone hybrid collaborative and patient-involving conference. The purpose of the conference was to enable the Norrie disease community to feel connected to the ongoing research into finding a treatment for the hearing loss as well as hear an informative range of presentations on Norrie disease and connect with other families.

The presentations were delivered carefully and thoughtfully, and every effort was made to use language that patients and families could understand. For the majority of the families and patients this was the first time they had listened to research content let alone been invited to join in, ask questions and be part of a mock PPIE session. The drive behind this was to help the Norrie disease community in the UK and globally feel more confident and empowered to ask questions about the research and the condition in general. The Norrie community wanted to find out how/if they could be involved in the research and to make sure that they were doing all they could to benefit and support their children, relatives, partners and friends who are living with the condition.

The conference achieved all of the above and enabled the Norrie disease community to find out how important their part in supporting the research efforts is and to recognise that finding a treatment for the hearing loss is a collaborative journey with the Norrie disease community, medical professionals and stakeholders; and not one where the researchers work in silos in their laboratories. The speakers worked hard at delivering grounding presentations, trying to make sure that the Norrie disease community could get a clear and realistic overview of what the research entails, and the various risks and unknowns and timescales. The conference was a brilliant way of addressing the barriers and fear that families and patients can feel about researchers and research.

The medical professionals worked really hard to make sure patients and families felt at the centre of the event. There were sensory rich activities including a tactile touch tour of the cochlear with brailled presentations on what was happening with the hearing research accompanied by talks. There was an interactive music room bursting with sensory toys and sounds with two musically trained play specialists supporting the families to enjoy music accompanied with dancing and movement.   Many of the teenagers were able to meet for the first time and parents bonded through their experiences.

The conference offered a rare and valuable opportunity for families, scientists, stakeholders, and medical professionals to mix freely outside of the clinical settings and hospital appointments and for families and patients to share insight into the reality of living with Norrie disease for the medical professionals. The conference has really helped the Norrie disease community to feel listened to and at the centre of the research and to appreciate where their fund raising is going, it has given them hope for the future and understanding on what getting to a treatment entails and how important their participation and support is and hope for global collaboration too.

The conference has also paved the way for an exciting new chapter for the Norrie disease community and the NDF will be planning regular zoom meets for both the UK and global Norrie disease community to connect more regularly and receive updates on the life changing research that is taking place inspired by them and for them. We can't thank Prof. Jane Sowden and her team enough for their support.