**BSGCT PPIE Award – APPLICATION FORM**

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

**SECTION 1**

**APPLICANT’S NAME:**

Dr Stephanie Jones

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

RDM-NDCLS, University of Oxford

**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)**:**

**Motivation**: We are developing a gene therapy for a rare condition leading to Surfactant Protein Deficiency caused by mutations in the *SFTPB* or *ABCA3* genes. These can be fatal in infancy - for the most profound cases where babies cannot breathe independently, there is no treatment, and these babies typically die within their first few months. While pre-clinical work proceeds in the lab, we are setting up a study to establish just how rare this condition is in the UK, as there is no dedicated registry. To carry out our research study, we need to request existing genetic information from intensive care units, to supplement clinical data from National Neonatal Research Database (NNRD). This is considered especially sensitive information under data protection regulations. Our ethical dilemma was whether we should seek consent from the parents of those babies whose genetic data we would need. Clinicians in this area warned us that the rate of obtaining consent for participation in such a study would be very low (around 30%). Considering that we are only expecting a maximum of 5-6 cases per year of these ultrarare surfactant protein deficiencies, this rate of consent would make our study almost worthless. To help us come to a decision about whether or not to seek consent, we advertised on a range of platforms to speak with parents whose baby had been treated in an intensive care unit soon after birth and/or had had genetic diagnostic tests in infancy. Compensation of £25 was offered for those who agreed to have an interview for up to an hour.

**Nature of PPIE:** We advertised on a disease-oriented website & Facebook group as well as more generic platforms and received responses from 19 people. Initial interactions via email, conversation on Teams or by phone, identified eight individuals with relevant experience and an interest in contributing to our ethical dilemma. Several others had PPI experience but did not give evidence of lived experience that was relevant to this project. Individual interviews were arranged online via Teams. A structured interview was used: introductions of the interviewer and interviewee; background of the research team; the project being established; and presentation of the ethical dilemma. The specific question was whether we should make every attempt to seek consent for data our study, followed by a free discussion of the pros and cons of this approach.

On balance, the outcome showed that while all interviewees would themselves consent to their child’s data being requested for a study such as ours, they did not think consent was essential. They agreed that the risk of not being able to trace the parents, or parents not responding (a low response rate), meant that the value of the study would be reduced, and it might even become unfeasible. We therefore concluded we should proceed without seeking consent from parents, subject to regulatory approval.

**Was this rewarding from my point of view?** Absolutely. It was humbling, inspiring and highly motivating to continue with our research.

**Was this rewarding from the patient(s) point of view?** I spoke to the one teenager who was a survivor of ABCA3 surfactant deficiency teenager, who was very excited about our research even though it would come too late for them to avoid needing a lung transplant. Parents also seemed genuinely grateful that we were working on such a rare condition, and all said they would be happy to help further if needed.

**Impact of the interaction:** We hope that with this evidence from relevant parents as part of our application, we will be able to proceed with our study in the most time-efficient manner, because we will not have to trace parents and wait for their responses to request the crucial confirmatory diagnosis information. This also means that we will have the most representative data to guide future clinical trials. We will also bear in mind feedback on design of any parent-facing documentation for any clinical trial (needing input from families, and clear, layered, uncluttered communication to get our message across to families facing a most distressing diagnosis).

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

Statements from email correspondence before the interview, or in response to receiving a summary of the interview by email, along with a form to apply for the compensation.

From C., mum of a child ("M") with Surfactant protein deficiency (survivor) who subsequently did an interview with me:

"*Hello, I just saw your post on a chILD group on Facebook. First, I want to thank you for caring enough about these kiddos to research and hopefully help save lives! (...) If I can help in anyway, please don’t hesitate to reach out. I remember begging the doctors to find something to help her. They even went as far as contacting the manufacturers of Surfactin, to see if they could separate the proteins and just give the SPB. Unfortunately, they weren’t willing to do this due to cost and lack of evidence that it would help. M received Surfactin (in full) within a few days of birth and it caused her lungs to bleed. Whatever you need, I’m happy to share. Thank you again, for all you’re doing. In the sixteen years of M's life, this is THE first time I’ve ever seen anything about a possible treatment. I’m so very hopeful for your work and the possibility of families never having to experience the devastation of our situation*. "

and after the interview:

"*Please reinvest my compensation back to the program. I will reach out to M's pulmonary doctor for the results of M's genetic testing. If there is anything I can do to further help, please don’t hesitate to reach out! Thank you for your desire to help these kiddos and families! We appreciate your efforts more than you know!*"

From E., mum of a child with a still un-identified syndrome, under investigation since he was born:

"*I saw your ad on Facebook advertised by the genetic alliance. From reading your email l think that is an interesting ethical dilemma and I can see arguments for options*." Later adding:

"*Thank you Stephanie. I have read over what you have written up - looks great*."

From S., mum of a child with a different type of rare genetic condition:

"*It was lovely to talk to last week and hear about the project. Everything in the notes looks good. Thank you*.”

From R & E, parents of a baby boy who died in infancy of ABCA3 deficiency:

" …*we were glad to be able to contribute to your research. Your notes are correct. (...)*

*Again, please do get in touch if there’s anything else you’d like to ask*. "

From G., mum of a little boy who passed away in infancy from ABCA3 mutations:

"*Hi Stephanie, Hopefully you get the permission you need to proceed without any hiccups*."