



iPS cells for Rare disease cases: Expression of Interest

The Human Induced Pluripotent Stem Cell initiative (HipSci), funded by the Wellcome Trust and the MRC, aims to generate iPS cells from healthy volunteers and patients with genetic disease, and carry out genomic and cell biological studies on them. The project collaborators aim to generate up to 800 iPS cell lines, on which they will conduct extensive genetic analysis, and characterise how the cells respond to specific external stimuli.

The resulting cell collection and data-set will be the UK's most comprehensive resource for investigating how genetic variation impacts on cell behaviour and how diseases linked to a specific genetic defect can result in a broad spectrum of clinical abnormalities. The project is led by King's College London and the Wellcome Trust Sanger Institute. During the initial phase of the project our main focus was on the generation of iPS lines from healthy individuals from the Cambridge BioResource as well as a small number of rare disease patients. We have subsequently selected seven additional rare disease cohorts for iPS line generation in a second phase of genetic disease studies. The purpose of this call is to gauge the level and nature of interest in a third disease sample phase.

If you are a clinician (or scientist with access to relevant patient cases) working with patients with inherited genetic diseases, where a collection of disease causing mutations has already been identified, and would be interested in having iPS lines generated from some of your patients that you would use in further studies, we would like to hear from you.

Please note that at this stage this is only an Expression of Interest and not a final call for proposals. The results will allow us to plan the use of our remaining resources most efficiently.

Please contact us if you have a group of (between 10 and 50) patients with related phenotypes where you would be able to provide us with patient material (either fibroblasts or a skin biopsy).

Further information about the project and our cell lines is available at www.hipsci.org.

The Expression of Interest should be submitted to **HipSci-proposal@ebi.ac.uk** before 3rd July 2015.

Please include the following information:

- Name
- Affiliation [Proposers must be UK-based principal investigators or equivalent.]
- Disease
- Justification for making iPSC for this disease
- Number of samples you would be able to provide
- How would future studies be carried out – differentiation and phenotyping methods?
- Would you be able to perform these follow up studies in your own lab or would you collaborate with another group?
- Are there other studies already making stem cell lines in this disease area, and if so how would this project relate to/differ from those efforts?
- What added value would there be to making lines from patients compared to engineering corresponding mutations using Crispr/CAS9 technology?

Note:

Lines generated within the HipSci project will be available via a stem cell bank under an MTA to others for research purposes, either in academia or industry.

Funded by

