HipSci

Human Induced Pluripotent Stem Cell Initiative

iPS cells for Rare disease cases: call for proposals

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. The project collaborators aim to generate over a thousand 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. Further information is available at www.hipsci.org.

We are seeking clinicians (or scientists with access to relevant patient cases) to provide samples from patients with inherited genetic diseases for the disease component of the HipSci project. We request that samples are provided in groups of between 10 and 50 with related phenotypes, and that all samples already have the genetic defect identified (the aim is to support gene analysis rather than gene discovery), with typically multiple cases with defects in the same gene. We encourage clinicians/scientists with only small numbers of samples to form larger groups with others who can contribute similar cases.

Sample providers will receive back the iPSC produced to allow them to work with their cell lines, but the lines will also be available via a stem cell bank under an MTA to others for research purposes, either in academia or industry. We are in the process of obtaining overarching ethics permission for the genetic disease component of the study, so that contributing clinicians/scientists will only need to provide Site Specific Information for their samples.

Proposals can be submitted online from 20 March 2014 until 23rd of May 2014, with decisions being made in June 2014. Proposals will be evaluated by a committee involving independent external experts working together with members of the consortium, chaired by Veronica van Heyningen.

Please send your proposal to **HipSci-proposals@ebi.ac.uk** .

Submission of Proposals

Proposers must be UK-based principal investigators or equivalent.

Applications should be in 12 point font in Word or PDF format with the following information:

1. Title and main applicant.
2. Provide clear scientific rationale for the iPSC work to be undertaken (500 words max)
    a. Describe the disease and what is known of its aetiology
    b. What will iPSC add to the study of this disease?
    c. How will future studies be carried out – differentiation and phenotyping methods?
3. Specify numbers for iPSC production and description of disease subjects to be studied, with affected gene, family structure and the extent of phenotype information available. (500 words max).
4. Give proposed time line for sample collection/submission. The expected cell type currently for iPSC generation is fibroblasts from skin biopsies. We would accept either existing fibroblasts[1] or 2-4mm fresh punch skin biopsies. It is possible that in special cases peripheral blood mononuclear cells (PBMC) may be accepted. (200 words max)
5. Demonstrate that the applicant is set up to study the iPSCs, once these are available, either in their own lab or by pre-arranged collaboration with another expert lab (200 words max)
6. Include a justification for the likely need for the lines and how many lines you and/or collaborators will utilise
7. Provide one page CV for main applicant(s), including current post and brief career history, 3 relevant key publications, short summary of current funding, and other relevant material up to a maximum of one page

Prioritisation of iPSC requests

Submissions will be prioritised by the panel, according to defined criteria:

i. Tissues to be studied are not readily available from diseased subjects
ii. The disease is not readily modelled in the laboratory
iii. Diseases to be studied will be monogenic. We will not accept polygenic disease cases
iv. Diseases studied will be ones in which no or limited previous iPSC studies are available
v. There should be extensive phenotyping information available to allow linking between clinical and iPSC studies

vi. The submitting group should demonstrate understanding of and viable arrangements for carrying out informative studies with the iPSC obtained

vii. The generation of the required cell type(s) from iPSC should be feasible or under development at the time of experiment start

viii. The willingness to share iPSC with other scientists is an absolute requirement for participants

[1] Should you consider providing fibroblast for reprogramming, the requirements are 2-3 vials (1-2 milion cells per vial) at passage 3.

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