Full Application Due August 1, 2017

Genome Medicine Pilot Grants Genome England

I. Overview of the 100 000 genomes project and Rationale for Pilot Grants

The aim is to create a new genomic medicine service for the NHS – transforming the way people are cared for. Patients may be offered a diagnosis where there wasn't one before. In time, there is the potential of new and more effective treatments.

The project will also enable new medical research. Combining genomic sequence data with medical records is a ground-breaking resource. Researchers will study how best to use genomics in healthcare and how best to interpret the data to help patients. The causes, diagnosis and treatment of disease will also be investigated. We also aim to kick-start a UK genomics industry. This is currently the largest national sequencing project of its kind in the world.

If you have any questions, or if you would like assistance identifying appropriate resources to enrich your research proposal, please contact Dennis Wang at dennis.wang@sheffield.ac.uk or call 01142159102

II. Key Elements of the Pilot Grant Program

100 00 genome Pilot Grants are directed at utilising genome sequences to improve public health by scaling up the application of clinical interpretation of genomic variation.

The main impetus behind the grants is to support pilot studies that will lead to innovative projects that will impact clinical interpretation of genome data.

Pilot Grants will budget £50,000 per application for one year.

Pilot Grants should:

- Provide a compelling rationale for the choice of research into the clinically defined disease
- Address an area of direct interest to the applicant.
- Include a research plan that incorporates a description of the sequencing technology and provides a well reasoned analysis plan that clearly explains the choice of methods for analysis to include variant calling and filtering approach
- explain the interpretation of results to infer clinical outcome
- detail the use of any resources available through the Genome England 100 000 Genome project
- outline the potential for impact on the NHS and future work

Proposals should promote a clinically relevant approach to use of the human genome to better understand and treat a rare or complex disease

<u>Application and Review Processes</u> A three page complete application will be due <u>February 27, 2017</u>. Applications must be submitted electronically as a single pdf via the website. <u>Proposal structure.</u>

Project title

Abstract maximum of 250 words.

Research Proposal

- I. Scientific Proposal
 - A. Your scientific proposal is limited to three pages, not including references. Please use Arial font, size 11, single-spaced, with 0.7 inch margins. All figures should be included in the body of the application. Include the following sections (suggested lengths in parentheses):
 - I. Introduction: Describe the clinical background for your grant application. Critically evaluate existing challenges in understanding the genetic basis of the disease of interest; explain how your proposal addresses gaps in current knowledge; specifically discuss how your project has the potential to impact healthcare using the Genome England infrastructure and the current and future access to NHS facilities.
 - II. Project Details: describe the specific aim(s) that will be completed in the funding period. For each aim, provide the details of the approach, and provide anapproximate timeline for the activities related to that aim.
 - I. details should provide clear description and justify the choice of analytical approach. They should include:
 - I. How you will access the data generated and how you interact with it
 - II. What tools you will use for each step of its analysis and how you will ensure that the data is of high quality.
 - III. Any controls should be clearly explained in terms of measures of success.

III. Expected results

- I. Measures of success how will you judge if the project has been successful
- II. Preliminary data is not required, but a description of the nature the data that will be generated can be included if it speaks to feasibility.
- III. Impact: Describe the impact on Genome Medicine in the UK of your proposal if you are successful

References (limit to 1 additional page)

Appendix material is not allowed.

VIII. Grading

- 1. Does the proposal address an important problem and, if successful, will the results have an impact on have a sustainable impact on Genome Medicine in the UK? 10%
- 2. Is the choice of sequencing technology appropriate and well reasoned? 10%
- 3. Is the project focused and achievable? 10%
- 4. Are the methods and approach well reasoned and achievable? 50%
- 5. Are the measures of success appropriate? 10%
- 6. Does the project recognise and utilise Genomics England resources? 10%