ICR The Institute of Cancer Research

in partnership with CANCER RESEARCH UK

JOB DESCRIPTION

JOB TITLE:	**Clinical Scientist for Cancer Genetics**
DIVISION:	Division of Genetics and Epidemiology
GRADE:	Scientific Professional 5 (Analytical Scientist 2)
RESPONSIBLE TO:	Professor Clare Turnbull

OBJECTIVE OF THE POST:
To manage a package of work focused on clinical variant interpretation of cancer susceptibility genes. This workpackage is part of CanGene-CanVar, which is a complex 5-year £4.3 million CRUK-funded programme, which is aimed at delivering data resources and tools to support implementation of cancer susceptibility genomics within NHS services. The main elements of the workpackage upon which the postholder will concentrate are: (i) data extraction, assembly and novel analyses to gain insight into variant pathogenicty. (ii) coordination of development of a datasystem for clinical variant interpretation for cancer susceptibility genes (CanVar-UK) (iii) management of a national working group involving >100 clinicians and scientists from NHS Molecular diagnostics laboratories (CanVIG-UK-Cancer Variant Interpretation Group UK) (iv) evolving of UK contribution to international variant interpretation collaborations. This post will be well suited to an individual with (i) a background in diagnostic molecular laboratory science who wishes to combine clinically-directed and academically-directed endeavours AND (ii) who wishes to advance their analytical and bioinformatics skills AND (iii) who wishes to build on pre-existing strong organisational and project management skills.

PROJECT DESCRIPTION:
A **Clinical Scientist in Cancer Genetics** is required to work with us on delivery of our recently awarded £4.3 million CRUK Catalyst Award: “CanGene CanVar: Data Resources, Clinical and Educational Tools to leverage Cancer Susceptibility Genetics for Early Detection and Prevention of Cancer”, running 2019-2024 ((http://cangene-canvaruk.org/). This large programme has been designed to deliver research and resources that will improve how we deliver national NHS testing for cancer susceptibility genes such as BRCA1, BRCA2 and Lynch Syndrome genes. It will involve working with a large team of scientists, clinicians and patients highly passionate about achieving this goal.

This complex programme of work involves 16 PIs from six Higher Education Institutes (ICR, Cambridge, Southampton, Manchester, Oxford and St George’s University London) as well as Public Health England (Health Data Insight). The programme is structured into six inter-related Work-Packages of which the postholder will be focused on WorkPackage 2.

1: Collection, linkage and analyses of nationally-collected genomic and cancer data
2:Data Clinical Laboratory Resources for Genomic Cancer Variant Interpretation
3:Dynamic Evidence-based Guidance for Clinical Geneticists, Oncologists and GPs
4:Patient-facing tools for decision-making and understanding risk
5:Medical Education
6:Ethics, Governance and Policy

Professor Dame Sue Hill and Ellen Graham from NHS England are named collaborators on the award on behalf of the NHS Genomic Medicine Service and we shall be working closely to coordinate national working groups. Delivery of the educational component will be in collaboration with Health Education England; Dr Anneke Sellars from the Genomics Education Programme is likewise a named collaborator. There are also over 40 additional collaborators on the Programme, involving international researchers, NHS clinical geneticists, genetic counsellors, clinical scientists, oncologists and GPs, commercial partners and a PPI group comprising representation from various relevant patient groups.

Key to WorkPackage 2 is CanVIG-UK (Cancer Variant Interpretation Group UK), established in 2017 by Turnbull as the cancer susceptibility specialist subgroup of UK Association of Clinical Genomic Science (UK-ACGS) activity in adoption of the ACMG framework for variant interpretation. CanVIG-UK currently comprises >100 members including clinical and laboratory representation from each of the 25 Molecular Diagnostic Laboratories and Clinical Genetics Services of the UK (NHS) and ROI. CanVIG-UK meets monthly and undertakes multidisciplinary review and consensus variant classification of problematic clinical cases. CanVIG-UK has also generated a consensus specification of the ACMG and UK-ACGS variant interpretation frameworks, as applicable to cancer susceptibility genes.

We are seeking an individual with an exceptional track-record in molecular laboratory science and diagnostics, likely coming from a background of working as an accredited clinical scientist or in an equivalent role. It is anticipated that the postholder will have (or seek to evolve) strong skills in data analysis and thus will take advantage of relevant in-house and external opportunities to advance programming/analytical skills. It is anticipated that the post-holder will have intuitive organisational skills: a formal qualification in project management is not required but there may be opportunity within the role for pursuing this. The postholder will work alongside a highly proficient bioinformatician-software engineer and able clinical fellow, supported by a programme administrator. The postholder will both coordinate the activities of workpackage 2 and also be involved hands-on with component projects. Key activities:

(i) Coordination of the CanVIG-UK (Cancer Variant Interpretation Group UK). Management of this group offers great opportunity for collaborative co-working with colleagues in molecular diagnostics/clinical genetics, including:
- **a.** consultative generation of national consensus guidance
- **b.** management and documentation for monthly meetings
- **c.** management of variant review ('Variant(s) of the month')
- **d.** evolution of systems for community communication

(ii) **Coordination of the Expert Clinical Laboratory Scientist Group (ECLSG).** The ECLSG comprises seven senior Clinical Scientists who are contributing to planning of outputs and activities for the workpackage. Again, management of this group offers great opportunity for collaborative co-working with senior colleagues in molecular diagnostics/clinical genetics

(iii) Development of a datasystem supporting cancer susceptibility gene variant interpretation (CanVar-UK (http://www.canvaruk.org/)). This will involve identification of input data, curation of content, design and presentation, working closely with our bioinformatics-software engineer.

(iv) Coordination of interactions/data-sharing etc with interaction endeavours including ENIGMA, InSIGHT, ClinGen Expert Groups

(v) Delivery of novel analyses to improve our understanding of variants in cancer susceptibility genes, guided by and working with ECLSG members, our bioinformatics-software engineer and clinical fellow. This will include project coordination, data analysis, data presentation and drafting of manuscripts.

(vi) **Development of a definitive atlas of pathogenic and non-pathogenic BRCA1/BRCA2 variants.** This will include close working with Dr Mike Hubank (NHS Centre for Molecular Pathology, Royal Marsden Hospital) with opportunity for close working with CMP staff and joining of CMP lab meetings. The variant atlas, once validated, will be used for development of a wholly automated pipeline for germline BRCA diagnostics (which will underpin the BRCA-DIRECT BRCA-testing project). The outputs also will be ratified by the CanVIG-UK group and will be used to populate the CanVar-UK web-resource.

The post holder will report to Turnbull (at least) weekly. The post will be located at ICR Sutton, where several of the other programme clinical and scientific appointees/students will be located. Occasional UK travel may be required. For the right candidate, we are very open to flexible daily/weekly work-patterns including part-time working.

The post will be situated within the Division of Genetic and Epidemiology in the laboratory of Professors Clare Turnbull and Richard Houlston FRS, which comprises 20-25 post-doctoral scientists, bioinformaticians and students involved in a range of projects relating to Cancer Genomics. A well as leading on delivery of this CRUK Catalyst Programme, Professor Turnbull also undertakes a range of clinically-focused research work on susceptibility, somatic and functional cancer genomics. The BRCA-DIRECT project has been funded by CRUK 2020-2022 and involves building a digital platform to deliver “Clinically-Integrated, Clinician-Independent, Rapid, Digital, Genetic Susceptibility Biomarker Testing”.

Informal enquiries are welcome and can be made to Professor Clare Turnbull (Clare.Turnbull@icr.ac.uk). Please note – this address is for enquiries only and applications must be submitted on-line via the e-recruitment system.

KEY DUTIES & RESPONSIBILITIES

- Manage (closely supported by Turnbull) the national clinical laboratory scientist/clinical genetics group (CanVIG-UK: Cancer Variant Interpretation Group-UK, >100 members), including management of agenda of monthly meetings, communications and coordination of activities/outputs.
- Manage (closely supported by Turnbull) the Expert Clinical Laboratory Scientist Group, including communications for and between monthly meetings and coordination of activities/outputs.
- Co-develop in-house with our bioinformatics-software engineer CanVar-UK (http://www.canvaruk.org/), a datasystem supporting cancer susceptibility gene variant interpretation. Activities will include identification of input data, curation of content, interface design and graphic/graphical data presentation.
- Perform ‘research’ analyses of variant data to address novel research questions.
- Undertake written presentation for publication of research analyses
- Undertake verbal presentation of research analyses at internal and external meetings
- All staff must ensure that they familiarise themselves with and adhere to any ICR policies that are relevant to their work and that all personal and sensitive personal data is treated with the utmost confidentiality and in line with the General Data Protection Regulations.

APPOINTMENT DETAILS

Appointment will be on a Fixed Term Contract for 2 years in the first instance, extendible to 4 years pending review of requirements of the role. The full salary scale is in the range from £32,000 to £44,400 p.a. inclusive. Appointments are normally made at the start of the range which represents the market rate for the role but consideration will be given to experience and skills. Future progression is based on annual performance review.

Annual leave entitlement is 28 days per annum. There is an additional entitlement to 8 bank/public holidays and 3 ICR-set privilege days.

Benefits include a contribution-defined benefit pension scheme

This job description is a reflection of present project portfolio and is subject to review and alteration in detail and emphasis in the light of scientific developments.

PERSON SPECIFICATION

CRITERIA	Essential or Desirable?
Education & Knowledge	
MSc/PhD degree in a relevant subject OR professional healthcare qualification (eg NHS clinical scientist, clinical bioinformatician)	E
Experience	
Experience in management of genomic data	E
Experience in ACMG classification of pathogenicity for germline variants	E
Experience in ACMG-AMP evaluation of somatic variants	D
Experience of evaluation of variant data for cancer susceptibility genes	D
Experience in interactions with senior clinicians/scientists/researchers	E
Experience in development of software/bioinformatics tools	D
Experience in communications/management of social media	D
Experience in committee/working group management	D
Skills	
Application of ACMG Genomic Variant Interpretation System	E
Application of HGVS nomenclature	E
Management of NGS data	E
Ability to undertake basic analysis and presentation of data eg Excel	E
Sourcing of genomics information using online resources	E
Programming/data management using a package such as R, STATA	D
Image generation using software packages	D
General	
Excellent organizational skills and attention to detail	E
Ability to work independently and problem solve	E
Excellent written communication skills	E
Excellent verbal communication skills	E
Interest in cancer research and genomics	E
Interest in clinical transformation	E
Ability to work professionally and collaboratively within a team	E