School of Health and Medical Sciences

City St George's, University of London

Cardiovascular and Genomics Research Institute

Bioinformatician/Statistical Geneticist

Ref: 119-25

JOB DESCRIPTION

Post Title	Bioinformatician/Statistical Geneticist
Grade	Grade 7
Contract type	Fixed term until September 2027
Responsible to	Professor of Cardiovascular Medicine
Accountable to	Professor of Cardiovascular Medicine, Lecturer in Cardiovascular Science
Responsible for	Nominated junior researcher(s)
Liaises with	Clinical and scientific research staff in the Cardiology Section, members of the Bioinformatics Centre, Genetics Centre, University as directed.

Overall purpose of job

This post is funded by an exciting project grant recently awarded by The British Heart Foundation (BHF), 'The rare arrhythmia syndrome evaluation (RASE) 100K Genomes Project: Enhanced phenotyping for greater insights'.

The successful applicant will be joining the Cardiovascular and Genomics Research Institute at the School of Health and Medical Sciences at City St George's, working for the Inherited Cardiac Conditions (ICC) team, part of the Cardiology Clinical Academic Group (CAG), in liaison with the Genetics Research Centre. The post holder will primarily be expected to engage in processing, annotation, association and statistical analyses of whole genome sequencing data from participants with rare cardiac arrhythmias, who have been sequenced in the 100,000 Genomes Project, Genomics England. Data from control subjects from 100,000 Genomes Project and UK Biobank will be available for analysis. The successful candidate will

perform in-depth genetic analyses to uncover inheritable risk factors for arrhythmia syndromes. Data for replication analyses will be available from international collaborators. We expect to identify new and improved ways to prevent sudden death in patients and their relatives.

Main Duties and Responsibilities

Data acquisition and analysis

1. Undertake variant association studies and statistical modelling utilising the aforementioned cohorts and control datasets.
2. Provide support for genetics and genomics in the ICC Research Group including developing and applying bioinformatics pipelines for analysis of human genome sequencing data.
3. Conduct bioinformatics and computational analysis of sequencing data from patient samples leading to identification of disease-causing variants, to be validated in the wet lab.
4. Maintenance of the pipeline and data management ensuring quality and version control and integrity and security of genome sequencing datasets.
5. Incorporate protein informatics and pathway analyses to support a comprehensive assessment of variants and integrate data from other relevant public available 'omics data sets as required.
6. Collaborate and meet with project team.

Publications & Presentations

7. Prepare talks for presentation of research work at group research meetings and scientific conferences.
8. Assist in the preparation of publications of associated research projects.

Teaching

9. To supervise and train where necessary new member of the research team and assist in the project supervisions of students.

Administration

10. Help with development, organisation and maintenance of the project database and ensure data provenance.
11. Ensure requirements of patient confidentiality and Information Governance are met.
12. Provide written reports on activity for internal use and for preparation of project reports to funding agencies.
13. Assist in the preparation of grant applications of associated research projects, taking the initiative to identify sources for funding of research projects.

General

14. Communicate frequently and effectively with the ICC Research Group, including scientists and clinicians, to discuss the bioinformatics requirements of the project, report and review results.

15. To attend and participate in the Cardiology CAG's academic activities e.g. research group meetings and seminars.
16. Keep abreast of methodology developments relevant to the field and integrate with existing data analysis pipelines.

It is expected that staff working with the School of Health and Medical Sciences, will be involved in our mentoring and tutoring activities, as appropriate, as well as supporting admissions, student recruitment and access and widening participation activities (MMI interviews, Open Days, school visits, clearing etc) where applicable. All academic staff are expected to act as a personal tutor.

You are also expected to undertake other activities appropriate to your grade as directed by your manager. This job description reflects the present requirements of the post. As duties and responsibilities change, the job description will be reviewed and amended in consultation with the post holder from time to time. City St George's, University of London aims to provide opportunities for all its employees to develop the skills required to be successful in their role and to further develop their careers.

St George's School of Health & Medical Sciences, University of London, is committed to the San Francisco Declaration on Research Assessment (DORA) principles.

Nature and Scope of Job

The Inherited Cardiac Conditions (ICC) service at St. George's University Hospitals NHS Foundation Trust is provided by Professor Elijah Behr, Professor Sanjay Sharma, Professor Maite Tome-Esteban, Professor Michael Papadakis, Dr Mark Specterman and Dr Gherardo Finocchiaro who have a world-renowned reputation in the investigation of SUDY, sports cardiology, cardiomyopathies and ion channelopathies. The ICC team undertakes translational research with Professor Mary Sheppard and Dr Joseph Westaby (cardiac pathology), Dr Angeliki Asimaki and City St George's genetics researchers (Dr Roddy Walsh, Dr Alan Pittman and Prof Pia Ostergaard). Our research utilises large genomics datasets including the 100,000 Genomes Project and UK Biobank, for numerous cardiovascular diseases including Familial Hypercholesterolaemia, led by Dr Marta Futema. The academic and clinical staff are members of the Genomics England Research Networks and Professor Behr leads the Predisposition and Screening domain. This project has the potential to transform the future of healthcare by improving the prediction and prevention of disease, enabling new and more accurate diagnostic tests and allowing the personalisation of drugs and treatments to specific gene variants.

The candidate will work as a member of the ICC team in the Cardiovascular CAG and play an integral role in our research around long QT syndrome, Brugada syndrome, and catecholaminergic polymorphic ventricular tachycardia (CPVT). The position offers access to substantial genome datasets in-house. These include WGS, GWAS and RVAS data form sudden cardiac arrest and death cohorts. The post holder will be expected to take a proactive role in contributing to the analysis of these datasets, provide informatics expertise and where required, lead the informatics and computational components of our on-going research activities. The post holder will seek to set up custom methods/algorithms for 'omics data analyses and will be expected to work together with other bioinformaticians in the institution to strengthen our informatics and computational expertise.

In this role, the candidate will collaborate with The Rare Arrhythmia Syndrome Evaluation (RASE) consortium, currently co-chaired by Professor Behr, which is a UK-wide network of collaborators from 20 Trusts working collaboratively by aggregating cohorts of arrhythmia syndrome patients to enhance power to conduct more effective research.

The post holder will work closely with scientists and clinicians and push for project completion and publication to the highest standard and also support grant applications from the group.

This is an exciting opportunity for a highly motivated and ambitious individual to apply and develop their bioinformatics expertise in a cutting-edge field of cardiac genetics.

The role is offered as a full-time post but applicants who wish to work less than 1.0 FTE on a pro-rata basis or take part in a job-share will also be considered. The post holder should ideally have post-doc experience of genomics data analysis. Detailed knowledge of cardiology will not be essential as there will be every opportunity to learn once in post. The post holder will be familiar with a variety of methods for generating and analyzing human genome sequence data, such as WGS, WES, PRS, SNP data and mtDNA sequencing analysis. Experience using a range of genomic bioinformatics pipelines to provide a comprehensive annotation and interpretation of variants, including single nucleotide, copy number and structural variants, is required. Familiarity with UNIX/LINUX environments and modern programming languages, e.g. Python or R, is essential; experience with High Performance Computing Clusters and modern research data management requirements is also desired. Knowledge of publicly accessible databases and resources is expected. In addition, familiarity with use of other 'omics data sets to provide a detailed catalogue of information supporting candidate disease variants would be desirable enabling the ICC Research Group to carry out more integrated omics research.

Special Factors

The role is fixed term for 24 months with the possibility of extension and is offered as a full-time post. Applicants who wish to work flexibly will be considered. Proposed starting date is Oct 1st 2025.

Person Specification

Criteria	Description	Essential/ Desirable	How it is to be tested
Qualifications	A PhD with a human genomics data analysis focus	E	AF
Experience	Experience in statistical analysis, genetic statistics and interpretation of large-scale data especially NGS.	E	**SS1**, INT
	Experience with protein informatics to inform assessment of impact of variants on proteins' secondary and tertiary structures, protein-protein interactions and protein expression.	D	INT
	Practical experience of working with RNAseq data, miRNA and large-scale omics data	D	INT
	Experience in genome-wide and exome-wide common and rare variant based association studies	E	**SS2**, INT
	Experience of improving and maintaining bioinformatics pipelines and demonstrable commitment to quality and version control and secure data management.	E	**SS3**, INT
Knowledge/ Skills	Proven track record in a relevant subject (e.g. Bioinformatics/Computational biology/Computer Sciences/Human Genomics)	E	INT
	Excellent understanding of Unix, and a scripting language such as R, Perl or Python.	E	**SS4**, INT
	Experience of additional programming languages such as Java, SQL or C/C++	D	INT
	Ability to develop a database of multiple data types	E	**SS5**, INT
	Knowledge of genetic variant databases such as Decipher, ClinVar, Clingen and disease-specific databases to inform variant annotation.	D	INT
	Ability to assess relevant new bioinformatics developments relevant to the field, and apply them efficiently, as appropriate to improve data analysis.	E	INT

Key:
PQ=Prerequisite Qualification, AF=Application Form, CV=Curriculum Vitae,
SS=Selection/Supporting Statements, ST=Selection Test/Presentation, INT=Interview

Note: Elements marked SS (Supporting Statements) in the Person Specification will be highlighted in Step 6 (Supporting Statements) on the online application form. Applicants' answers to Step 6 are an essential part of the selection process. Applicants should write individual supporting statements to demonstrate how their qualifications, experience, skills and training fit each of the elements highlighted in this section.

*Applicants should address other elements of the Person Specification in Step 7 (Additional Information). Shortlisting will be based on applicants' responses to Step 6 **and** Step 7. Therefore applicants should complete both sections as fully as possible on the online application form.*

Updated July 2025

About City St George's, University of London

City St George's, University of London is the University of business, practice and the professions.

City St George's attracts around 27,000 students from more than 150 countries.

Our academic range is broadly-based with world-leading strengths in business; law; health and medical sciences; mathematics; computer science; engineering; social sciences; and the arts including journalism, dance and music.

In August 2024, City, University of London merged with St George's, University of London creating a powerful multi-faculty institution. The combined university is now one of the largest suppliers of the health workforce in the capital, as well as one of the largest higher education destinations for London students.

City St George's campuses are spread across London in Clerkenwell, Moorgate and Tooting, where we share a clinical environment with a major London teaching hospital.

Our students are at the heart of everything that we do, and we are committed to supporting them to go out and get good jobs.

Our research is impactful, engaged and at the frontier of practice. In the last REF (2021) 86 per cent of City research was rated as 'world-leading' 4* (40%) and 'internationally excellent' 3* (46%). St George's was ranked joint 8th in the country for research impact with 100% of impact cases judged as 'world-leading' or 'internationally excellent'. As City St George's we will seize the opportunity to carry out interdisciplinary research which will have positive impact on the world around us.

Over 175,000 former students in over 170 countries are members of the City St George's Alumni Network.

City St George's is led by Professor Sir Anthony Finkelstein.

School of Health and Medical Sciences currently offers a range of employee benefits:

Salary: £51,039 pa, (pro-rated for part-time staff).

Hours: 35 hours per week which can be done flexibly in various ways or part time/job share can also be considered. Staff are expected to work the hours necessary to meet the requirements of the role and this will be dependent on the service area.

Annual leave: 30 days per annum. Plus eight UK public holidays and four days when City St George's, University of London is closed (usually between Christmas and New Year). Part time staff receive a pro rata entitlement.

Pension: Membership of competitive pension schemes with generous employer contribution and a range of extra benefits.

Superannuation Arrangements of the University of London (SAUL)

London Pension Fund Authority (LPFA)

Universities Superannuation Scheme (USS)

National Health Services Pension Scheme (NHSPS) (existing members only)

Flexible working Flexible working, including part-time or reduced hours of work, opportunities to work from home for many posts, compressed hours and local flexibility in agreeing start and finish times of work.

Travel City St George's, University of London offers an interest free season ticket loan and participates in the Cycle to Work Scheme.

Gift Aid If you would like to make a tax-free donation to a charity of your choice, this can be arranged through our Payroll.

Sports and Leisure Facilities Rob Lowe Sports Centre, situated on the St George's Healthcare NHS Trust site offers exercise facilities that can be utilised by City St George's, University of London staff.

Within walking distance from St George's is Tooting Leisure Centre. Facilities include a swimming pool, gym and various exercise classes. The Centre offers staff an all-inclusive corporate membership. For more information please contact Tooting Leisure Centre.

Shops and facilities There are a number of shops and facilities situated on site including ATMs, student bar and shop, Pret a Manger, M&S Simply Food store, library and multi-faith room.

Informal enquiries

Informal enquiries may be made via email to: Professor Elijah Behr (ebehr@sgul.ac.uk) and Dr Marta Futema (mfutema@sgul.ac.uk)

Making an application

All applicants are encouraged to apply on line at http://jobs.sgul.ac.uk as our system is user friendly and the online application form is simple to complete. Please note that CVs only will not be accepted.

For any accessibility issues please contact hrhelp@sgul.ac.uk

Closing date: **15 August 2025**

Interview date will be on 03 September 2025. As shortlisted candidates will be notified by email, it is imperative that you provide an email address that is accessed frequently.

Please quote reference **119-25**

We are delighted that you are interested in working at the School of Health and Medical Sciences. You will be notified of the outcome of your application by email. We aim to respond to all candidates within 5 weeks of the closing date of the vacancy.