Job title	Senior Postdoctoral Genomics Scientist
Division	Medical Sciences
Department	Nuffield Department of Medicine
Location	Centre for Human Genetics, Building for Genomic Medicine, Roosevelt Drive, Old Road Campus, Oxford, OX3 7BN
Grade and salary	Grade 8: £45,585 - £54,395 with a discretionary range to £59,421 pa
Hours	Full time
Contract type	Fixed-term contract until 30 November 2027 Funding is provided by the NIHR
Reporting to	Prof Jenny Taylor, Professor of Translational Genomics
Vacancy reference	173349

Hybrid working arrangements	**The successful person will need to work on site for a minimum of 3 days per week**
Additional information	This role meets the eligibility requirements for a Skilled Worker Certificate of Sponsorship under UK Visas and Immigration legislation. Therefore, the Nuffield Department of Medicine welcomes applications from international applicants who require a visa.
About us	• University of Oxford - www.ox.ac.uk/about/organisation • Nuffield Department of Medicine (NDM) - https://www.ndm.ox.ac.uk • Unit - www.chg.ox.ac.uk
What we offer	https://hr.admin.ox.ac.uk/staff-benefits • An excellent contributory pension scheme • 38 days annual leave • A comprehensive range of childcare services • Family leave schemes • Cycle loan scheme • Discounted bus travel and Season Ticket travel loans • Membership to a variety of social and sports clubs • A welcoming and diverse community

Research topic	Human genetics and genomic medicine
Principal Investigator / supervisor	Prof Jenny Taylor

Project team	BRC / Taylor group
Project web site	www.ox.ac.uk/ https://www.chg.ox.ac.uk/people/jenny-taylor; https://oxfordbrc.nihr.ac.uk/research-themes/genomic-medicine/
Funding partner	The funds supporting this research project are provided by NIHR Oxford Biomedical Research Centre
Recent publications	The impact of inversions across 33,924 families with rare disease from a national genome sequencing project. Pagnamenta et al 2024. *American Journal Human Genetics*. Am J Hum Genet. 2024 Jun 6;111(6):1140-1164. doi: 10.1016/j.ajhg.2024.04.018. Epub 2024 May 21. Structural and non-coding variants increase the diagnostic yield of clinical whole genome sequencing for rare diseases. Pagnamenta et al. Genome Med. 2023 Nov 9;15(1):94. doi: 10.1186/s13073-023-01240-0. PMID: 37946251;

The role

Applications are invited for a Postdoctoral Scientist in Genomics to join the Oxford Biomedical Research Centre (BRC) Genomic Medicine Theme, based at the University of Oxford's Centre for Human Genetics (WHG), within the group of Professor Jenny Taylor. The Centre for Human Genetics has an outstanding history of genetics research and gene discovery in rare and common diseases whilst the Oxford BRC's Genomic Medicine Theme has an impressive track record of translating novel genomics tests and technologies into the clinic to inform diagnosis and treatment of patients. Increasingly our vision is to use genetic information in the design and development of individualised therapeutics and we have close links with the Oxford Harrington Centre to achieve this.

You will be familiar with a variety of methods for generating and analysing human genome sequence data, including exome and whole genome sequencing. A requirement of this role is to be able to identify potential pathogenic variants from genome sequencing data for rare disease patients. Familiarity with a range of genomics resources and publicly accessible databases to assess deleteriousness of genetic variants is required. The position offers access to substantial genome datasets both in-house and from our involvement with national programmes such as the Genomics England (GEL) 100,000 Genomes Project. You will be expected to take a pro-active role in contributing to the analysis of these national datasets where Oxford has research and clinical interests. Understanding the mechanisms of rare disease and the links with common disease is also important and familiarity with common disease datasets including UK Biobank will therefore be an asset.

You will also have excellent laboratory skills relevant to genomics including DNA sequencing (short and long read), RNASeq, qPCR and RT-PCR. Expertise in a range of other cellular or molecular biology techniques, including cell culture, would also be an advantage. Demonstrable ability to manage laboratory work, including experimental design and execution, and development of protocols and methods is required.

This is an exciting opportunity for a highly motivated and ambitious individual to apply and develop their genetics expertise in a cutting-edge field of translational genomics. The internationally renowned research of the University of Oxford's Centre for Human Genetics, combined with the translational

genomics undertaken by the Oxford BRC, provides an active and rich research environment in genomics in which to pursue these goals.

Responsibilities

You will:

- Conduct analysis of whole genome sequencing data from rare disease patient samples leading to comprehensive identification of candidate disease-causing variants, including single nucleotide, structural, splicing and non-coding variants.
- Use a range of algorithms, resources and publicly accessible datasets for assessing deleteriousness of genetic variants from in-house and national cohorts and combine this with knowledge of genetic inheritance and segregation data to refine candidate list.
- Undertake analysis of rare disease cases in the Genomics England (GEL) national 100,000 Genomes Project, liaising with bioinformaticians to assess pathogenicity of candidate variants and with referring clinicians to gain additional clinical data and samples.
- Collaborate with members of GEL research networks and other collaborative programmes, for instance at monthly MDT meetings.
- Explore potential of rare disease variants to impact common disease pathogenesis using cohorts such as UK Biobank.
- Apply novel bioinformatics pipelines, algorithms and tools including publicly available or commercial software packages for genome sequence analysis. Liaise closely with the bioinformaticians in the group to develop and apply such tools.
- Identify genetic variants potentially amenable to therapeutic intervention eg with nucleic acid therapeutics using anti-sense oligonucleotides, CRISPR-editing and gene therapy.
- Utilise multi-omics datasets, including transcriptomics, proteomics and metabolomics, to support or refute candidate genes emerging from genome sequencing data.
- Design and run experiments to provide functional validation of genetic variants including DNA (Sanger, short and long read) and RNA sequencing, qPCR and RT-PCR, mini-gene analysis.
- Apply additional molecular and cell biology techniques as appropriate to validate functional effect of variants which may include Western blotting, immunofluorescence, cell culture, generation and differentiation of iPSCs and organoids.
- Keep up to date with advances and to implement novel laboratory methodologies.
- Liaise effectively with core facilities and collaborators for such purposes.
- Assist with translation of sequencing technology / pipelines into clinical use. Contribute ideas and communicate effectively with other bioinformaticians, geneticists and clinicians.
- Be meticulous about record keeping and maintaining lab notebooks for both analytical and experimental work, to ensure patient cases are tracked and followed up. Provide written reports on activity for publication or internal use and for funding agencies.
- Communicate frequently and effectively with the BRC project teams, including laboratory scientists, clinicians and PIs, to discuss the genomics requirements of projects, report data, review results and ensure that the tasks are completed satisfactorily. Present at, and participate in, lab group meetings and contribute to discussions at these.
- Establish effective working relationships with various individuals and groups as defined in ‘Relationships’ below.
- Supervision of students and support to the academic programmes run at the Centre for Human Genetics

General Responsibilities:

- Report to and meet regularly with Professor Jenny Taylor, PI at CHG and Co-Theme Leader of the Genomic Medicine Theme.
- Meet with the other members of the BRC Genomic Medicine Theme, including genomics scientists, bioinformaticians and clinicians to design studies and review results.
- Continue to develop research interest's genome sequencing data analysis and genomics laboratory techniques in keeping with the aims of the Taylor Group and the Centre.
- Liaise with other bioinformaticians in CHG and be aware of relevant complementary projects in other Oxford centres including the Big Data Institute, Weatherall Institute for Molecular Medicine and Centre for Medicines Discovery to support variant annotation and interpretation.
- Be accountable for your professional conduct within the BRC and the Centre.
- Ensure such that all conduct is with due regard to the University Equal Opportunities and Data Protection policies.
- Participate in and support the public engagement and widening access activities of the Department and the University. This is anticipated to be not more than 2 days per year.
- Undertake mandatory training as required by the University, Division and Department. The specific list of training courses may change from time-to-time, in response to both legal and internal University requirements.

Selection criteria

Essential

- Hold a PhD in Genetics or related subject.
- Strong postdoctoral experience in analysis and interpretation of genome sequencing data to include single nucleotide and structural variants, copy number abnormalities as well as assessment of variants in regulatory or non-coding regions.
- Knowledge of genetic variant databases such as Decipher, ClinVar, Clingen, gnomAD and disease-specific databases to inform variant annotation.
- Experience of identifying pathogenic variants underpinning rare disease pathogenesis from genome sequencing data.
- Expertise in analysis of multi-omics datasets.
- Demonstrable commitment to contribute to collaborative programmes, including Genomics England and commercial partnerships, to analyse data within the environments and according to the regulations of these programmes.
- Motivation to assess relevant new pipelines and commercial software packages relevant to genomics and apply them efficiently, to improve data analysis.
- Expertise in laboratory genetics techniques including Sanger sequencing, Illumina next generation sequencing, PCR, Western blots and core cellular biology techniques.
- Demonstrable experience in conducting independent research delivering high quality scientific outputs and with proven ability to write up scientific findings in peer-reviewed journals.
- A high degree of self-motivation and initiative and ability to work independently and collaborate within teams to meet project deadlines.
- High level of attention to detail, self-organisation and timeliness.
- Excellent communication skills (both written and oral) with the ability to communicate effectively at all levels (of technical knowledge).
- Experience working in a fast moving and motivated scientific environment.

Desirable

- Experience analysing data within Genomics England 100,000 Genomes Project and UK Biobank datasets.
- Interest in or familiarity with selection of genetic variants for therapeutic intervention.
- Statistics knowledge.
- Experience of writing and securing grant applications.

Pre-employment screening

Standard checks

If you are offered the post, the offer will be subject to standard pre-employment checks. You will be asked to provide: proof of your right-to-work in the UK; proof of your identity; and (if we haven't done so already) we will contact the referees you have nominated. You will also be asked to complete a health declaration so that you can tell us about any health conditions or disabilities for which you may need us to make appropriate adjustments.

Please read the candidate notes on the University's pre-employment screening procedures at: https://www.jobs.ox.ac.uk/pre-employment-checks

How to apply

Applications are made through our e-recruitment system and you will find all the information you need about how to apply on our Jobs website https://www.jobs.ox.ac.uk/how-to-apply.

If you would like to apply, **click on the Apply Now button** on the 'Job Details' page and follow the on-screen instructions to register as a new user or log-in if you have applied previously.

As part of your application you will be asked to provide details of two referees and indicate whether we can contact them now. You will be asked to upload a CV and a supporting statement. The supporting statement must explain how you meet each of the selection criteria for the post using examples of your skills and experience. This may include experience gained in employment, education, or during career breaks (such as time out to care for dependants). Your application will be judged solely on the basis of how you demonstrate that you meet the selection criteria stated in the job description.

Please upload all documents **as PDF files** with your name and the document type in the filename. Please note using a long file name may prevent you from uploading your documents.

- http://www.ox.ac.uk/about_the_university/jobs/research/

All applications must be received by **midday** UK time on the closing date stated in the online advertisement

Information for priority candidates

A priority candidate is a University employee who is seeking redeployment because they have been advised that they are at risk of redundancy, or on grounds of ill-health/disability. Priority candidates are issued with a redeployment letter by their employing department(s).

If you are a priority candidate, please ensure that you attach your redeployment letter to your application (or email it to the contact address on the advert if the application form used for the vacancy does not allow attachments).

If you need help

Application FAQs, including technical troubleshooting advice is available at: https://staff.web.ox.ac.uk/recruitment-support-faqs. Non-technical questions about this job should be addressed to the recruiting department directly recruitment@ndm.ox.ac.uk

To return to the online application at any stage, please go to: www.recruit.ox.ac.uk.

Please note that you will receive an automated email from our online recruitment portal to confirm receipt of your application. **Please check your spam/junk mail** if you do not receive this email.
Important information for candidates

Data Privacy

Please note that any personal data submitted to the University as part of the job application process will be processed in accordance with the GDPR and related UK data protection legislation. For further information, please see the University's Privacy Notice for Job Applicants at: https://compliance.admin.ox.ac.uk/job-applicant-privacy-policy. The University's Policy on Data Protection is available at: https://compliance.admin.ox.ac.uk/data-protection-policy.

The University's policy on retirement

The University operates an Employer Justified Retirement Age (EJRA) for very senior research posts at **grade RSIV/D35 and clinical equivalents E62 and E82**, which with effect from 1 October 2023 will be 30 September before the 70th birthday. The justification for this is explained at: https://hr.admin.ox.ac.uk/the-ejra.

For **existing** employees on these grades, any employment beyond the retirement age is subject to approval through the procedures: https://hr.admin.ox.ac.uk/the-ejra.

There is no normal or fixed age at which staff in posts at other grades have to retire. Staff at these grades may elect to retire in accordance with the rules of the applicable pension scheme, as may be amended from time to time.

Equality of opportunity

Entry into employment with the University and progression within employment will be determined only by personal merit and the application of criteria which are related to the duties of each particular post and the relevant salary structure. In all cases, ability to perform the job will be the primary consideration. No applicant or member of staff shall be discriminated against because of age, disability, gender reassignment, marriage or civil partnership, pregnancy or maternity, race, religion or belief, sex, or sexual orientation.