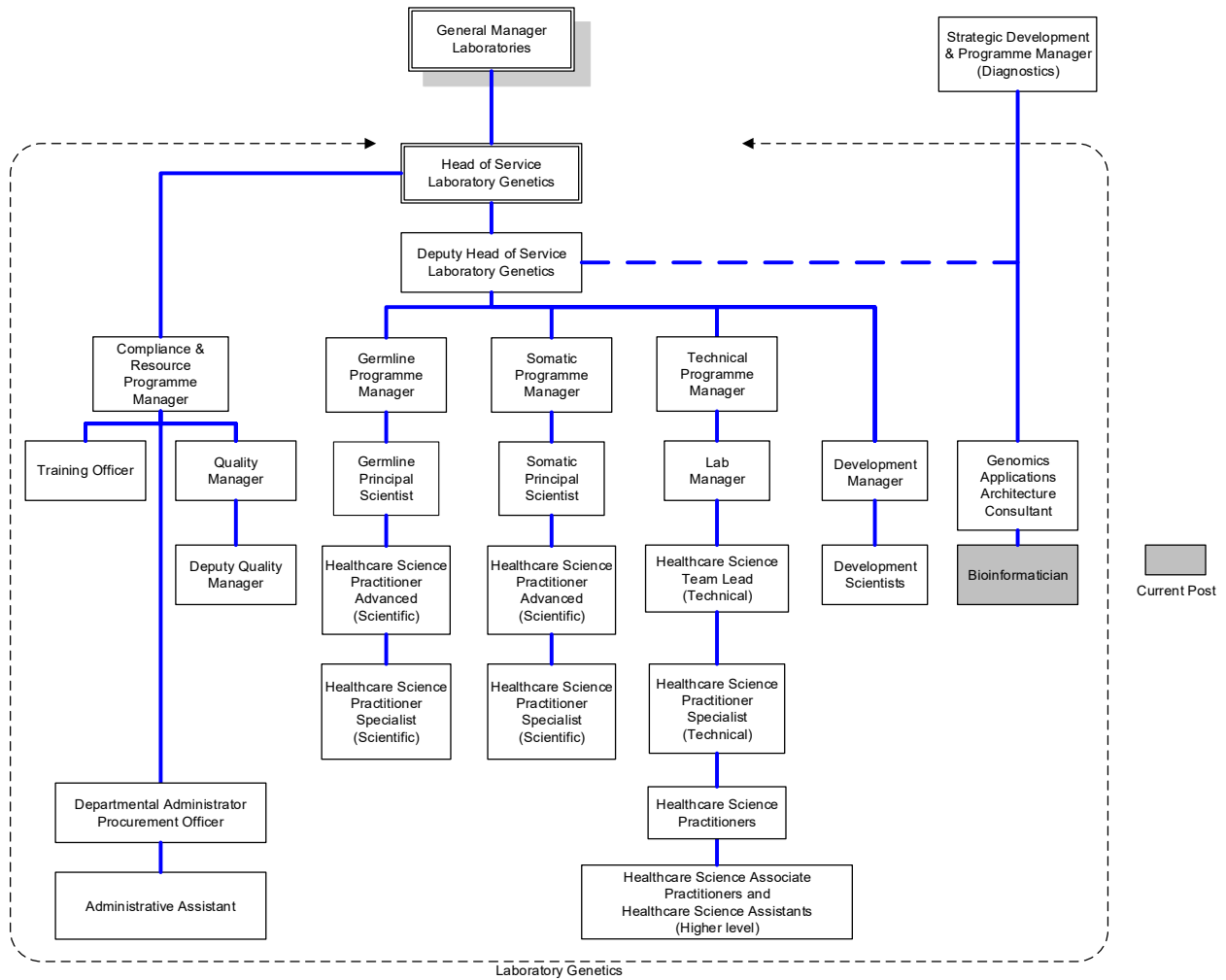


**NHS GREATER GLASGOW & CLYDE
JOB DESCRIPTION**

1. JOB IDENTIFICATION	
Job Title:	Bioinformatician, Band 7
Responsible to:	Genomics Applications Architecture Consultant
Department:	Laboratory Genetics
Directorate:	Acute Diagnostics
2. JOB PURPOSE	
<ul style="list-style-type: none"> • Responsible for the development and maintenance of bioinformatic pipelines in the Laboratory Genetics department. • To maintain, enhance and develop reliable and efficient infrastructure for bioinformatic tools and processes to ensure the highest quality diagnostic genetics service. • Responsible for ensuring the implementation of standard operating procedures (SOPs) and systems documentation. • To ensure that Laboratory Genetics complies with legal and professional requirements for safe transmission and storage of large amounts of genetic data. • Provide highly specialised advice and training for bioinformatics to colleagues within the West of Scotland Centre for Genomic Medicine. 	
3. ROLE OF DEPARTMENT	
<p>The Laboratory Genetics department, which forms part of the West of Scotland Centre for Genomic Medicine provides a comprehensive diagnostic genetic service for the patients of the West of Scotland (population >2.7 million) and as part of the Scottish Genetics Laboratories Consortium and the Scottish Molecular Pathology Consortium, and specialised testing for particular disorders to the whole of Scotland, the UK and overseas. Based at the state of the art Laboratory Medicine building at the Queen Elizabeth University Hospital in Glasgow, the Laboratory Genetics department is responsible for the specialist diagnosis and/ or monitoring of patients with constitutional (prenatal and postnatal) and acquired (malignancy) genetic abnormalities in hereditary genetic disease, solid tumours as well as adult and childhood leukaemia. The service is funded by National Services Division, NHS Scotland.</p> <p>The Laboratory Genetics department is the largest of their type in Scotland and they process in excess of 35,000 specimens a year. They collaborate closely with other laboratories in the Laboratory Medicine building including pathology, and with various research groups at the University of Glasgow. The genetic laboratories provide a specialist education and training programme for our healthcare scientists and other healthcare professionals, including continuous professional development, ensuring our workforce is appropriately trained and developed to deliver a high quality diagnostic genetics service. In addition, the genetic laboratories deliver a component of the MSc in Medical Genetics in collaboration with the University of Glasgow.</p>	

4. ORGANISATIONAL POSITION



5. SCOPE and RANGE

The post holder is a bioinformatician who has day to day responsibility for the development and maintenance of bioinformatic pipelines within the Laboratory Genetics department and will report to the Genomics Applications Architecture Consultant. There are >100 members of staff in the Laboratory Genetics department section including Consultant Clinical Scientists (Head of Laboratory and Deputy Head), Principal Clinical Scientists, Principal Healthcare Scientists, Clinical Scientists/ Healthcare Scientists Advanced, Healthcare Scientists, Biomedical Scientists, Genetic Technologists, Healthcare Scientist Practitioners, Healthcare Scientist Assistants, Healthcare Science Support Workers and admin and clerical staff.

The Laboratory Genetics department receives specimens from hospitals, health centres and general practitioners from the West of Scotland and offers a comprehensive genetic service to these users. It works cooperatively with the genetics, molecular pathology and molecular haematology laboratories in Aberdeen, Dundee and Edinburgh as part of the Scottish Genetics Laboratories Consortium and the Scottish Molecular Pathology Consortium. The post holder works under the direction of the laboratory's scientists to help deliver this service.

6. MAIN TASKS, DUTIES AND RESPONSIBILITIES

The post holder will be mainly responsible for developing and maintaining pipelines for the primary and secondary analysis of next-generation sequencing (NGS) data for hereditary and somatic genetic disease. They will work with the scientific staff to transform large volumes of statistical and genetic data into annotated results for clinical reporting. The duties of the post holder are:

Clinical, scientific and technical

- To adhere strictly to the departmental policies and Standard Operating Procedures.
- To follow Health and Safety regulations, as outlined in the laboratories protocols and policies.
- To perform, organise and prioritise their own work in the laboratory.
- To be responsible for the time management of multiple tasks and to respond to the changing requirements of the laboratory by taking on additional tasks and responsibilities as required, in relation to bioinformatics.
- To demonstrate and apply a thorough understanding of the scientific principles involved in the delivery of a genetic diagnostic service.
- To write and maintain Standard Operating Procedures relating to procedures involving bioinformatics, ensuring that they comply with standards for accreditation.
- To deliver bioinformatic pipelines in the diagnostic Laboratory Genetics department.
- To check analytical data extracted in relation to results and reports, which include statistical analysis.
- To provide specialised advice to colleagues within the West of Scotland Centre for Genomic Medicine, in matters relating to bioinformatics.
- To participate in internal and external quality control procedures and assessments, as directed by the quality manager.
- To monitor the quality of the data produced by next generation sequencing, microarrays and other technologies to ensure that it meets the standards required for diagnostic service, according to laboratory protocols and national professional standards.
- To help determine and establish internal quality control parameters and quality indicators for bioinformatic analysis and pipelines.
- To perform other duties as deemed appropriate by the head of laboratory.

Bioinformatics and HI&T Resources

- To be responsible for the development and maintenance of bioinformatic pipelines within the Laboratory Genetics department.
- Experience working with various operating systems (e.g. Windows Server, LINUX); knowledge of relational database management systems (e.g. SQL Server) and SQL; knowledge of web technologies (e.g. IIS, [ASP.Net](#), [VB.Net](#)); knowledge of communications infrastructure (e.g. LANs, WANs and associated security hardware); expertise in programming, scripting and markup languages (e.g. Visual Basic, BASH, PERL, C++, Java, Python, JSON, XML).
- To plan, develop, validate and implement major bioinformatics tools, pipelines and processes for efficient analysis, annotation and interpretation of large amounts data generated from next generation sequencing, microarray and other technologies as required within the laboratory.

- To review, assess and validate bioinformatics tools and resources available to annotate and aid in the interpretation of novel gene sequence and structural variants.
- To validate and implement robust bioinformatic workflows for data processing, analysis, annotation, interpretation, storage and backup, including the application of an independent checking system.
- To create intuitive macros and other software tools to allow user-friendly manipulation and annotation of data by other laboratory staff.
- To implement and maintain robust version control for each aspect of the bioinformatics pipeline, in line with existing departmental quality management system.
- Working with laboratory management and HI&T colleagues to monitor and evaluate new developments in bioinformatics to ensure that the laboratory uses the most appropriate tools and methods available to deliver a high quality diagnostic service.
- To perform routine maintenance and backup on high-specification NHS GG&C computers, working with HI&T colleagues.
- To support the development and maintenance of effective databases for storage of scientific and clinical information and to help establish links with the LIMS, working with HI&T colleagues.
- Working with HI&T colleagues to develop and maintain effective methods for robust storage and transmission of large amounts data generated by different processes, ensuring that the appropriate regulatory and legal standards are met.

Managerial

- Participate in the relevant laboratory meeting, lectures, seminars and courses to facilitate personal development.
- Prepare and update documentation in work involving bioinformatics as directed by the quality manager to ensure that the department maintains UKAS accreditation status.
- To participate in the laboratory internal audit programme as directed by the quality manager.
- To present the results of audit work to colleagues at internal and national meetings.
- To communicate non-compliances to the Genomics Applications Architecture Consultant, head of laboratory and/or quality manager.
- To be aware of and follow the current regional and national policies and legislature, along with UK best practice guidelines for laboratory genetics, and promote these to others.
- To assist in any other aspects of the laboratory management, including administration, and policy and procedure updating, as directed by the Genomics Applications Architecture Consultant, head of laboratory and/or quality manager.
- Ensures that the laboratory complies with requirements of data protection, liaising with the NHS GG&C information governance and IT security managers as required.
- Working closely with laboratory management, the Genomics Applications Architecture Consultant, and the Service Delivery Programme Manager for Diagnostics to advise in the evaluation and procurement of software and IT hardware in relation to bioinformatics.
- Be responsible for maintaining, implementing and notifying all relevant laboratory staff of software and pipeline updates, ensuring that all changes and procedures are documented properly.

- Assist with general IT issues within Laboratory Genetics, liaising with appropriate laboratory and HI&T colleagues as required.

Research and development

- Work in collaboration with laboratory management, the Genomics Applications Architecture Consultant, and the Service Delivery Programme Manager for Diagnostics to develop and validate bioinformatics initiatives designed to improve the efficiency of existing services.
- To undertake and collaborate in relevant research and development on the application of bioinformatics for new diagnostic techniques.
- To present the results of bioinformatics service development to colleagues at internal meetings.
- Working with laboratory management, the Genomics Applications Architecture Consultant, and the Service Delivery Programme Manager for Diagnostics to incorporate new applications of bioinformatics robustly and safely into routine testing.
- To participate in the evaluation and validation of changes to standard operating procedures which involve bioinformatics.
- Maintain an awareness of the relevant scientific literature.

Teaching and training

- In consultation with laboratory management, train and supervise relevant laboratory staff on bioinformatics applications.
- To report any training issues to the head of laboratory or the training officer.
- To take part in Continuing Professional Development activities to acquire new knowledge and skills for service and personal development.
- To represent the laboratory at local, national and international meetings, as deemed appropriate by the head of laboratory, and to disseminate information gathered at these meeting back to laboratory colleagues.

Enabling the employer to meet statutory requirements

- Comply at all times with the departmental and NHS GG&C Health and Safety policies, security policies, departmental operating procedures and disciplinary codes.
- Report/ensure that any defect or occurrence which may affect safety at work is brought to the attention of the Safety Officer.
- Maintain an awareness of the NHS GG&C Information Governance and IT security policies and the Data Protection Act, preserving confidential patient information.

7a. EQUIPMENT AND MACHINERY

The post holder will:

- Use various computers and other appropriate expensive, specialist, high specification IT equipment.

7b. SYSTEMS

The post holder will use various computers to:

- To record and extract patient information, to generate patient data and reports using the bioinformatics pipelines.
- To update and manage disorder specific laboratory databases, for a subset of genetic disorders.
- To access the laboratory's document control system (INVU).
- To access the laboratory's quality management system (Q-Pulse).
- To analyse results using specialised, highly specialised or custom-written software packages and bioinformatics pipelines.
- To participate in departmental audits.
- To search for patient test information and simple audit to produce standard and non-standard reports as required by the head of laboratory.
- To produce electronic data e.g. Word, Access, Excel, PowerPoint.
- To access the intranet and internet including the e-library, for pertinent scientific literature, particularly important for the reporting and interpretation of genetic test results.
- For Datix incident reporting.

The post holder will use:

- Photocopier for duplicating documentation.
- Scanners for document archiving.
- Email
- Telephone for communication both internally and externally.

8. DECISIONS AND JUDGEMENTS

The post holder will report to the Service Delivery Programme Manager for Diagnostics, and operationally report to the Genomics Applications Architecture Consultant and must take responsibility for their work, prioritising workload when necessary. Decisions often need to be made which require an understanding of the laboratory's policies, procedures and methodologies. These include:

- The post-holder is responsible for delivery of agreed objectives within guidelines provided, but he/she will frequently be required to determine how this is best achieved
- The post holder is required to manage their own day-to-day workload but may have to reprioritise this depending on the requirement for urgent bioinformatics analysis such as data analysis for a pregnancy or an urgent cancer test result.
- The post holder will work closely with the Genomics Applications Architecture Consultant, the deputy head of laboratory and lead clinical scientists when designing bespoke bioinformatics pipelines. A portion of the work undertaken by the post-holder will be project-based i.e. designing bespoke bioinformatic pipelines.
- To help determine and establish internal quality control parameters and quality indicators for bioinformatic analysis and pipelines.

- Assessing whether data is of sufficient quality to progress analysis for a patient report, particularly to ensure adequate coverage and depth to avoid false negative results and to drive repeat testing when required.
- Working with the scientific team to decide whether supplementary or alternative analyses are required for those cases which produce equivocal or unexpected results, in consultation with the head of laboratory when necessary.
- Making decisions regarding complicated and/or problematic analysis, troubleshooting and offering advice and guidance to other staff where appropriate, in consultation with head of laboratory when necessary.
- The post holder will be required to juggle a large number of activities with varying priorities, ensuring that the needs of the service are met.
- Supporting the scientific teams in identifying whether variants are real biological changes or artefacts of the methodology to avoid false positive results.
- Develop processes for assessing run to run variation of each methodology/instrument to ensure that consistent high quality is maintained and any issues are identified early.
- Assessing data quality and coverage when procedures are improved or changed to ensure that validation is carried out appropriately before they are implemented into routine service.
- Compiling analytical clinical data reports to ensure that the information is clear, concise and unambiguous.

9. COMMUNICATIONS AND RELATIONSHIPS

The post-holder will:

- Communicate as an effective team member alongside colleagues, to ensure optimal use of resources and the delivery of an efficient, high quality service.
- Interact effectively with both technologists and scientific staff involved in the overall next generation sequencing procedure to ensure high quality data is generated.
- Maintain close links with colleagues both in other areas of Laboratory Genetics and also other departments within the NHSGG&C diagnostics directorate (eg Clinical Genetics, Biochemical Genetics, Pathology, Haematology, Microbiology, Virology, Biochemistry) to provide an integrated high quality service.
- Communicate collaboratively with colleagues from other Scottish Genetics Consortium laboratories, University departments and commercial suppliers to maximise the speed of development and implementation of next generation sequencing technologies and associated bioinformatics solution.
- Communicate regularly with colleagues within the HI&T Department. Such discussions will generally be technological or procedural in nature and will be aimed at ensuring the laboratory are provided with coherent and integrated solutions from the HI&T Department.
- Attend relevant laboratory meetings and discuss laboratory issues and developments with colleagues.
- Present laboratory data and research findings at local, national and international scientific meetings and conferences.

- Explain procedures and data output formats accurately and concisely to other staff within the laboratory.
- Liaise with the training officer when training other members of staff.
- Liaise with the quality manager for issues relating to UKAS accreditation.
- Liaise with senior management, including the head of laboratory, on other issues relating to laboratory management.
- To abide by the NHSGG&C policy on patient confidentiality.
- Attend an annual staff review.

10. PHYSICAL, MENTAL, EMOTIONAL AND ENVIROMENTAL DEMANDS OF THE JOB

Physical skills

- Keyboard skills are required.

Physical demands

- A combination of sitting, standing and walking is required.
- Frequent requirement for sitting in a restricted position for extended periods whilst using a computer workstation and specialist software to analyse data and results, or to write and authorise patient reports with little opportunity to exercise during this time.
- May require multi tasking and prioritisation of work often under stressful conditions.
- Some manual handling may be undertaken

Mental demands

- There is a frequent requirement for prolonged, intense concentration when analysing and interpreting genetic diagnostic data and results. Often processing very large and data sets.
- Work pattern can be unpredictable due to demands of the service, especially when an urgent specimen from a high risk pregnancy or new born baby must be processed immediately.
- Organisational skills, especially time management, and the ability to multi-task are very important.
- There is a requirement to prioritise workload to meet deadlines.
- This laboratory is a busy environment, which makes demands on the concentration.
- Cope with a wide variety of distressing or emotional circumstances eg diagnosing terminal conditions, abnormalities and genetic disease.

Emotional demands

- The handling of patient data and maintaining patient confidentiality whilst processing specimens which will undergo genetic testing.
- Processing urgent specimens for example from a baby on a ventilator, can be emotionally distressing.
- Ability to cope with unexpected problems and to take responsibility

Working Conditions

- Continuous use of display screen equipment (DSE) on a daily basis.

11. MOST CHALLENGING/DIFFICULT PARTS OF THE JOB

- Developing and implementing a bioinformatics pipeline for routine diagnostic use where the technology is highly developmental and with limited knowledge or support available from colleagues with a similar role, in collaboration with laboratory management.
- Undertaking research and development with a view to ensuring that NHSGGC adopts those emerging technologies which represent real service benefits.
- Working to very demanding Professional Standard Guidelines. These cover both the necessary quality of the work undertaken and also the acceptable turn-around times.
- Must have the ability to concentrate for long periods of time whilst analysing and interpreting very large genetics diagnostic data sets and results.
- Must be able to multi-task and deal with the unpredictable bioinformatics workloads.
- The acquisition and maintenance of knowledge with regards to laboratory procedures, and the interpretation and the reporting of results, which must be constantly refreshed as practice and guidelines change.
- Participation in continuous personal development where there are time constraints due to service commitments.

12. KNOWLEDGE, TRAINING AND EXPERIENCE REQUIRED TO DO THE JOB

- MSc or equivalent postgraduate qualification in bioinformatics with experience of human genetics OR MSc or equivalent postgraduate qualification in a human genetics based discipline with a significant bioinformatics component.
- Significant experience of developing and implementing bioinformatics tools and resources.
- Experience of programming and scripting language (e.g. C++, Java, Python).
- Bioinformatic analysis of next generation sequencing data.
- Thorough knowledge and understanding of next-generation sequencing approaches and statistical methodology used for the analysis of the large data sets generated.
- Thorough knowledge of bioinformatics tools available for the analysis and interpretation of genetic and genomic data.
- Knowledge of bioinformatics tool development guidelines and standards.
- Good knowledge of UNIX, programming and scripting language.
- Knowledge of information governance and data protection requirements.
- Capable of prolonged concentration and attention to detail.
- Ability to work as an effective team member in the delivery of a diagnostic service.
- Careful and meticulous, adhering to written laboratory policies and procedures.
- Enthusiastic and motivated.

- Good verbal and written communication skills.
- Ability to impart bioinformatics knowledge to less experienced colleagues.
- Demonstrate and participate in continuous professional development.
- Advanced IT skills including expertise in the manipulation of information/data and the ability to produce reports and statistical analysis.