

## YHNE GLH

### Job Description

#### 1 JOB DETAILS

**Job title:** Rare Disease Clinical Lead

**Pay band** Medical Consultant

**Job Tenure:** Fixed Term Contract to 31<sup>st</sup> March 2020

**Job plan allocation:** Up to 3 PAs

**Base:** Current location of employment but will be required to travel to all parts of the YHNE GLH region

#### Essential Requirements:

- Medical Practitioner with GMC licence to practice & entry on specialist register
- CCT or equivalent in speciality relevant to genomic medicine
- Membership or Fellowship of relevant Royal College
- Medical Consultant in rare disease genomics
- Experienced clinician with expertise in and experience of clinically interpreting and advising on the clinical actionability of complex genomic data up to the level of whole genome sequencing
- A leading clinician, with:
  - Proven experience of managing teams and leadership roles at Directorate or Divisional Level or equivalent, across professional and specialty boundaries
  - A recognised national /international expert in own field as evidenced by publication record and in contributing to national and international initiatives in genomic science/medicine
- In depth knowledge of the NHS and its recent, ongoing reforms, and likely future strategic direction. Ability to develop and maintain consensus and collaboration within and between teams
- Strong persuasion and influencing skills with ability to present proposals effectively to Executive level across organisations

#### Desirable Requirements:

- Higher Degree or equivalent

#### 2 JOB PURPOSE

- Acts as the point of contact for clinicians seeking advice on appropriateness of referrals for genomic testing, whom to test within the family, use of HPO terms, understanding the Genomic Test report and actionability of findings and downstream management of the patient and their relatives and when to refer to a clinical genetic clinical or counselling service
- Liaises with and advises the staff in the GLH on referrals, testing strategy, phenotypes, actionability etc.

- Plays a key role in the Genomics MDT and the production of the final clinical interpretation in the context of the presenting phenotype  
Plays a key and substantial role in education and training across the clinical specialisms within the host Trust but also across the wider Geographical Areas

### **3 DIMENSIONS**

- The reconfiguration of Genetic Laboratory service provision in England is supporting the creation of a world class resource in the use of genomics and genetic technologies within the NHS. It is based around the consolidation of services in Genomic Centralised Laboratory Hubs that will work together in networks with Genomic Local Laboratory Hubs to create a genomic laboratory service infrastructure for the NHS in England.
- YHNE GLH is bidding to become a Genomic Laboratory Hub (GLH) covering the North and East of England. It comprises a partnership arrangement between several organisations (Leeds Teaching Hospitals Trust, Sheffield Children's Hospital FT, Newcastle Upon Tyne Hospitals FT, Sheffield Teaching Hospitals FT and NewGene Ltd.).
- Key responsibilities of YHNE GLH:
  - Consolidate services to deliver significant efficiencies
  - Operate within a national testing network
  - Operate a coherent informatics infrastructure
  - Sample collection, processing and logistics
  - Technical and analytical platforms
  - Bioinformatics, annotation and validation
  - Clinical interpretation, reporting and returning of results
  - Quality management and assurance of both processes and data
  - Plan & develop workforce for now and the future
  - Organisational governance and partnership arrangements
  - Promotion of research and innovation
- YHNE GLH will:
  - Work together with other Genomic Laboratory Hubs (GLHs) to form part of a national network of genomic laboratories to provide a comprehensive and coordinated national genomic laboratory service that is an integral part of the planned NHS Genomic Medicine Service.
  - Deliver a prescribed list of genomic tests, for a defined geography, as outlined in the National Genomic Test Directory.
  - If appointed as a provider of a national specialist testing service, YHNE GLH will be responsible for providing the genomic testing service for prescribed GLHs within the national network and will work with clinicians and genomic MDTs within the prescribed geographies.
  - Participate in regular meetings convened by NHS England's Genomic Unit as part of the national network.
  - Establish local networks of any subcontracted laboratories and designated providers for Rare Disease genomic testing to provide a coordinated and comprehensive service across the defined geography to deliver standardised outcomes, ongoing consolidation and adoption of new technologies.
  - Consolidate the existing specialist genetic laboratories into state of the art, high throughput, high quality and standardised (ISO Accredited) laboratories through the processing of the majority of genomic tests for its geography, as prescribed in the National Genomic Test Directory, through a single laboratory based in Leeds. Operate enhanced working practices to ensure genomic testing services are

available to support the delivery of seven-day services across the geography and ensure that genomic technology platforms are fully optimised.

- Be responsible for ensuring quality and the ongoing review of opportunities to further consolidate networked services, based on technological advances, the changing evidence base for genomic testing and the National Test Directory.
- Be accountable to NHS England's Genomics Unit within an agreed governance and operational framework that will oversee the test repertoire, co-ordinate the provision new national specialised testing services, monitor performance and finance.
- Have in place and maintain a partnership agreement with all sub-contracted partners and designated providers for Rare Disease genomic testing, as appropriate, setting out the working relationships and a sustainable operating infrastructure.
- Be responsible for establishing and maintaining a sustainable operational infrastructure and contracting arrangements for the geographical network inclusive of quality improvement.
- Fulfil the membership criteria for UKGTN (or any future successor arrangements) and any additional requirements as specified by the NHS England Genomics Unit.

#### **4 ORGANISATION**

Accountable to: Medical Director (Employing Organisation)

Responsible to: Scientific Director YHNE GLH

#### **5 KNOWLEDGE, TRAINING, EXPERIENCE**

As outlined under 'Essential Requirements'.

#### **6 SKILLS**

##### **Communication and Relationships**

- Builds extensive clinical engagement across tertiary, secondary and primary care
- Ensures strong linkages to Rare disease teams in partner organisations and relevant external partners
  - Advises and reports at a senior level on the GLH activity
- Communicates on service development.
- Communicates specialist information to conferences.

##### **Analytical and Judgmental Skills required for the post**

- Directs the medical input into the Rare disease testing pathway and the validation and other testing that is required to determine clinical validity and actionability
- Works with others to ensure that the referral pathways are established and feedback arrangements to requestors are clear and supported through Genomic MDTs and other mechanisms
- Acts as the senior medical advisor in YHNE GLH on the integrity and validity of results generated from genomic testing.

##### **Planning and Organisational Skills required for the post**

- Responsibility for the formulation and delivery of long-term, strategic plans as part of the YHNE GLH leadership team.

- Works collaboratively with Academic Health Science Networks (AHSNs) and is an active member of and helps to shape the Genomics Innovation Network.
- Works within the GLH National Network and with key stakeholders (NHSE, UK Genetic Testing Network (UKGTN), Genomics England and the sequencing provider) to ensure services are fit for purpose.

### **Physical Dexterity Skills required for the post**

- Standard keyboard skills.

## **7 KEY RESULT AREAS**

### **Patient/Customer Care**

- Provides senior consultant advice to clinicians on appropriateness of referrals for Rare disease genomic tests and for sign off for urgent requests particularly for WGS
- Plays a leading role in the establishment and ongoing development of Genomic MDTs especially associated with WGS and with any Specialist Test that the GLH delivers
- Oversees the monitoring of clinical outcomes from genomic testing to inform the national genomic testing strategy

### **Policy and Service Development**

- Set strategic direction in line with NHS England (NHSE) vision for the NHS Genomic Medicine Services and with any nationally, or internationally agreed projects and initiatives in genomics. along with others
- Maintains YHNE GLH services at the forefront of genomic science working with National Institute for Health Research (NIHR) and other bodies as appropriate.
- Contributes to evaluation of new technologies and approaches, including testing strategies and provides information on new tests to be included in the National Genomic Test Directory and composition of virtual gene panels.
- Lead on development and delivery of medical education and training events in YHNE GLH to promote and develop the genomic knowledge of the medical workforce

### **Financial and Physical Resources**

#### **Human Resources**

- Establish networks of other medically qualified personnel within the GLH who are contributing to the overall National Genomic Testing Service and the clinical interpretation of genomic results up to the level of WGS

#### **Information Resources**

- Advises and reports at a senior level on YHNE GLH Rare Disease genomics activity.

#### **Research and Development**

- Initiate, direct, coordinate and undertake research and innovation activity, playing a lead role in the development of YHNE GLH research capacity and capability building in Rare disease genomics.
- Work with Genomics England to support the national genomics research and industry agenda and is an active member of relevant Genomics England Clinical Interpretation Partnership (GeCIP) and discovery forum contributing to scientific research and industry collaboration.

## **8 FREEDOM TO ACT**

- Responsible for establishing the way in which general health and organisational policies are interpreted.
- Acts as senior medical adviser across the YHNE GLH patch.

## **9 EFFORT & ENVIRONMENT**

### **Physical**

- Sit in a restricted position for long periods on a regular basis.

### **Mental**

- Occasional requirement for intense concentration e.g. when analysing data, preparing detailed business plans or reviewing financial information.
- Workload unpredictable and interruptions often require immediate action.

### **Emotional**

- Occasional highly distressing or emotional circumstances e.g. handling difficult staffing issues.

### **Working Conditions**

- Occasional exposure to unpleasant conditions through interaction with laboratory services