Rationale for a job planning document for Consultants in Clinical/ Medical Genetics

The reason for writing this document is to describe the effect of the changing landscape of genomic medicine on the evolving role of the Clinical Geneticist and integration with mainstream medicine. With recent advances in genomic medicine such as 100,000 Genomes Project, the imminent implementation of NHS whole genome sequencing (WGS) and access to such technology by mainstream clinicians, our role has become more integrated with mainstream clinical practice than ever in the form of more inter-disciplinary team working, multidisciplinary team meetings and interpretation of complex genomic results.

Clinical Geneticists work in teams that cover a large geographical area (now defined by the boundaries of the Genomic Medicine Service Alliances) and generally sub-specialise in areas of practice such as Cancer Genetics, Adult Care, Paediatric Genetics, and Prenatal Medicine. This job planning document is an iterative document taking into account: the changing role of Clinical Geneticists and need for sub-specialisation as more genetic disorders are identified; increasing knowledge of evolving phenotypes; the need to ensure that we are up-to-date with the fast-moving pace of technological advances and keeping abreast with latest published literature. Our role in genomic variant interpretation and correlating genotype with clinical phenotype in order to provide best possible, evidence-based care for our patients and their families is crucial.

Main Principles Underlining Job Planning for Consultants in Clinical Genetics

The job plan is similar for most consultants in all Genetic Centres in the UK, although there is a degree of variability due to local geography, local mainstream expertise, specialisation and on-call clinical commitments.

Job content

1. The job plan is based on a weekly model bearing in mind most Consultants work to a monthly schedule and an annualised job plan.
2. All consultants have a commitment to clinics in their tertiary centre and peripheral hospitals which may change over time according to service needs. Personal circumstances may also be taken into account.
3. The number of clinics per week cycle is based on a formula which is equivalent for all consultants. The content of specialist clinical work may be taken into account as some clinics require more input. Other aspects of the job plan may also affect this.

4. Consultants may have a specialty interest which will influence the location and timing of their clinics.
5. In certain departments, most LTFT consultants attend departmental meetings on an equal basis to FTE. This is important for input into clinical case discussions, laboratory meetings and CPD. However, in certain departments with larger Consultant numbers, LTFT may attend such meetings proportionate to their job plan.
6. On call weeks rotate and may coincide with any week in the monthly cycle. LTFT consultants who participate in the rota on a full time basis may take time *in lieu*, which does not coincide with fixed clinical commitments and is recorded alongside annual and study leave.
7. Urgent prenatal and acute ward work in Clinical Genetics may vary across departments based on size and links with their local feto-maternal unit, intensive and neonatal care units.
8. The commitment is to provide a fixed number of clinics per 42 week year (annualised job plan).
9. Post-COVID-19, the clinic content and number of face-to-face appointments is likely to change and there is an acknowledgement that video and telephone consultations may replace some of the in-person clinic appointments (see below).
10. Additional roles that influence job content:
    - Clinical Lead — 1.0PA (trust-dependant)
    - TPD/Educational Lead — 0.5-1PA (trainee & deanery-dependant)
    - Governance Lead — variable (trust-dependant)
    - Research funded or recognised by Trust — variable
    - Other areas by specific agreement — variable
11. Additional internal roles are included in SPA and may not be specifically recognised (such as audit lead, teaching and organisation of regional and national meetings).

Example Formula for job content

Days	Five day	Four day	Three day
Basic PAs*	10	8	6
OP clinics/week [F2F/ telephone/ video appointments]	2	1.5	1.0
Ratio clinics: support + clinical	1:1	1:1	1:1
Clinic support sessions (Pre-clinic case preparation and immediate post-clinic work)	2	1.5	1.0
Clinical (urgent/ ad hoc/ telephone contact/ ward in-patient) sessions	1.5-2	1-1.5	1.0-1.25
Specialty MDTs, variant interpretation and additional genomic work	1.5-2	1-1.5	0.5-1.0
GC/ Trainee Clinical Supervision	0.5	0.5	0.25
Main departmental clinical meeting	0.5	0.5	0.25- 0.5
SPAs$^{\pi}$	1.5-2.5	1.5-2.0	1-1.5
Other renumerated roles	Locally agreed	Locally agreed	Locally agreed
Total PAs	10.0	8.0	6.0

Notes

*On appointment the number of basic PAs is likely to be 10, 8 or 6. The PA allocation will then be adjusted according to additional commitments, travel commitments to be consistent with the existing consultant team. However, Consultant appointments with 7 or 9 PAs have been made.

πSupporting professional activities (SPAs) for Clinical Genetics according to Royal College of Physicians for approval of job plan is 2.5 SPAs for a 10PA job plan; however, some Consultants are on 1.5-2 SPAs (1-1.5 core SPA and 0.5 common SPA). The proposed example allows flexibility for those Consultants on 2.5 SPAs in their job plan currently.

Travel times are flexible and depends on distance travelled to peripheral clinics which may vary based on geographical spread and clinic allocation, hence this will be local trust-dependant.

Definition of work undertaken in Clinical and SPA sessions

Outpatient clinics

Patient numbers per clinic are determined based on the complexity of patient caseload, new versus follow-up appointments and travel time required. In certain weeks, OP clinics may be partially replaced by ward referral/ ward round rota and written or telephone advice and guidance to referrers and patients, as per local practice.

Post COVID-19 experience has demonstrated that it is apparent that some patients can be managed with telephone / video consultations without the need to see face-to-face, but it is likely that pre- and post-clinic work required for such consultations will be the same or possibly greater than a face-to-face clinic appointment. It is important that there is an acknowledgment of this recent adaptation to clinical practice and funding allocated accordingly. Further information on the efficiency and efficacy of virtual working in Clinical Genetics will be available from the Clinical Genetics Society audit of working practices and patient surveys during the COVID19 pandemic.

It is expected that a full time Consultant will deliver ~2 clinics/ week for 42 weeks in an annualised job plan. See variable above.

Some clinics may be done as Multi-disciplinary clinics (MDC) with involvement of more than one (or in some situations, several specialities) to enable a one-stop clinical visit for patients and their families.

With evolving roles of Clinical Geneticists and a move towards a more advisory role, face-to-face clinics will be partially replaced by genomic variant interpretation, genomic MDT discussions, liaison with the laboratory in interpretation of complex genomic results and mainstream clinical MDTs forming part of DCC delivery. It is anticipated that this may reduce direct clinic referrals, but acknowledges that this is a reflection of how we deliver the service in our changing role more efficiently with increased mainstream access to genomic testing.

Clinic support sessions (one per clinic) include activities such as:

1. **Pre-clinic preparation**

Appraise past medical notes and family history, research condition, consult latest literature, identify likely investigations, identify key investigations in Genomic Test Directory, contact Genomic Laboratory Hub (GLH) and determine turnaround time for investigations.

For Consultants involved in Cancer Genetics work, this would also involve reviewing patient family histories, questionnaire assessment and advising on screening and management of moderate cancer risk referrals by letter rather an appointment.

2. **Post-clinic patient management**

Dictate clinic letters, organise DNA sample export and testing with laboratories, update review, preparation for clinical meetings. Identify family members at risk, arrange on-going referrals and investigations as required. Facilitate and administer opportunities for patients to participate in clinically relevant research including clinical trials. Accessing and providing patient support group information.

Other Clinical sessions (one per clinic) include activities such as:

1. Clinical follow-up & management of genetic results: Interpret genetic test reports, utilise databases, liaise with laboratories, undertake MDT case discussion, telephone consultations and write to professionals and patients about results, plan required follow-up or discharge.
2. Seeing urgent ward referrals-covering all specialities, neonatal and intensive care unit referrals; undertaking ward consults over the phone, advise on diagnosis, genetic investigations and follow up.
3. Provision of clinical service to pregnant mothers with fetal abnormality, increased risk of genetic disorder and facilitate prenatal testing. Involvement in Fetal medicine / neonatal MDTs and perinatal mortality meetings.
4. Provision of telephone advice to primary and secondary care clinicians and other health professionals on all aspects of genetics including complex ethical situations. Triage of referrals to ensure appropriate care pathways.
5. Provision of care to existing families with genetic disorders when a new event arises.
6. Telephone, video and email consultations with patients - clinical advice and discussion of results.
7. Clinical and organisational work relating to specialist services (Consultant-specific).

Speciality Clinical MDTs, Genomic Variant Interpretation & Additional Genomic Work:

MDT meetings to support Genomic Medicine Service Alliance and implementation of Genomic Test Directory by mainstream clinicians (terminology refers to the situation in England though similar need in devolved nations)

1. Ongoing work from 100,000 Genomes Project requiring interpretation of genomic results with review of clinical phenotype, with Clinical Scientists and mainstream specialities

2. Genomic variant interpretation is an important part of our workload and is likely to increase with increased access to whole genome sequencing. This involves researching several resources, literature search, database review and reverse phenotyping to assess significance of variant, subsequent facilitation of functional studies, contact with research groups, extended family studies (clinical assessment and genomic studies) to assess pathogenicity and feedback to patients. We will also need to input into databases to enhance knowledge of the specific variant but also allow further causal assessment depending on outcome of above.

3. Input into speciality MDTs ~ variable depending on service needs and speciality-specific. With MDT working, it is likely that there will be participation from different specialists and in certain MDTs, including Genomics MDTs, it is anticipated that there will be a wider participation of more than one Clinical Geneticist and with pre and post MDT preparation required. It is anticipated that where MDT discussions are chaired and led by Consultant Clinical Geneticist and/ or require substantial pre and post MDT contribution, this will require additional time and administrative support.

4. Implement new rapid exome service (R14) for children in NICU and PICU facilitating urgent exomes including assessment, referrals, interpretation of results and counselling parents within a short time scale.

5. Utilise and evaluate new Genomic Test Directory and provide feedback for annual update of panels.

6. Implement Patient Choice in the consenting process for Whole Genome Sequencing, and other genetic testing.

7. Provide additional advice for colleagues arising from the new Genomic Test Directory.

8. Contribute to the new Genomic Medicine Service Alliance to support NHSE deliverables.

9. Establish cross-boundary working in Genomic Medicine Service Alliance and contribute to appraisal and audit.

10. Enrol and facilitate patient participation in research projects and clinical trials relating to genomic medicine.

Other DCC sessions include:

1. Travel to clinics as above variable and trust-dependant.
2. Agreed additional flexible clinical working-such as specialist additional clinics either as part of MDC or individual based on specialist referrals and Consultant sub-specialty role.

Supporting professional activities include:

1. Management activities, including for sub-speciality roles
2. Clinical Governance
3. Appraisal and mentoring
4. Mandatory training
5. CPD
6. Educational supervision and training
7. Undergraduate and postgraduate teaching
8. Research and innovation
9. Research supervision of junior colleagues and GCs
10. Patient support group involvement activities
11. Professional liaison for 100 000 Genomes Project, Genomic Clinical Interpretation Partnership (GeCIP) activities & Genomic Medicine Service Alliance Liaison
12. Additional internal roles

Additional NHS responsibilities and external duties

1. Clinical Lead
2. Clinical director/ medical director roles
3. Remunerated Genomic Medicine Service Alliance roles
4. National and international council/ professional body membership and leadership activities
5. Royal College roles (RCP/ RCPCH)

References:

1. www.clingensoc.org/media/11296/theevolvingroleoftheclinicalgeneticist-290715.pdf

2. https://www.england.nhs.uk/genomics/nhs-genomic-med-service

Working Group:

Chair: Dr Meena Balasubramanian

Clinical Genetics Society: Dr F Elmslie, President CGS and Professor S Smithson, Vice-President CGS

Lead Clinicians: (with representation from every Genomic Medicine Service and the devolved nations):

Dr K Metcalfe (Chair), Dr T Dabir, Dr Nayana Lahiri, Dr E Kinning, Dr E Kivuva, Dr U Kini, Dr A Murray

Consulted and approved by the council of the Clinical Genetics Society, the council of the Cancer Genetics Group and all Clinical Genetics service lead clinicians