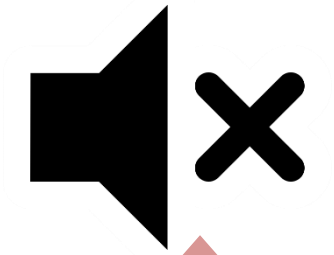
@swglh

Welcome to the 'Bristol open event for Scientist Training Programmes in Genomics'



Please Mute your Microphones either on headset/phone or Webex 'bubble' option on bottom of screen

Please follow guidance on use of hand raising and prompt to open microphone.

We will start promptly at 2pm

We will be video and audio recording the session – if you don't want to speak out use the chat function from bubble at bottom of screen

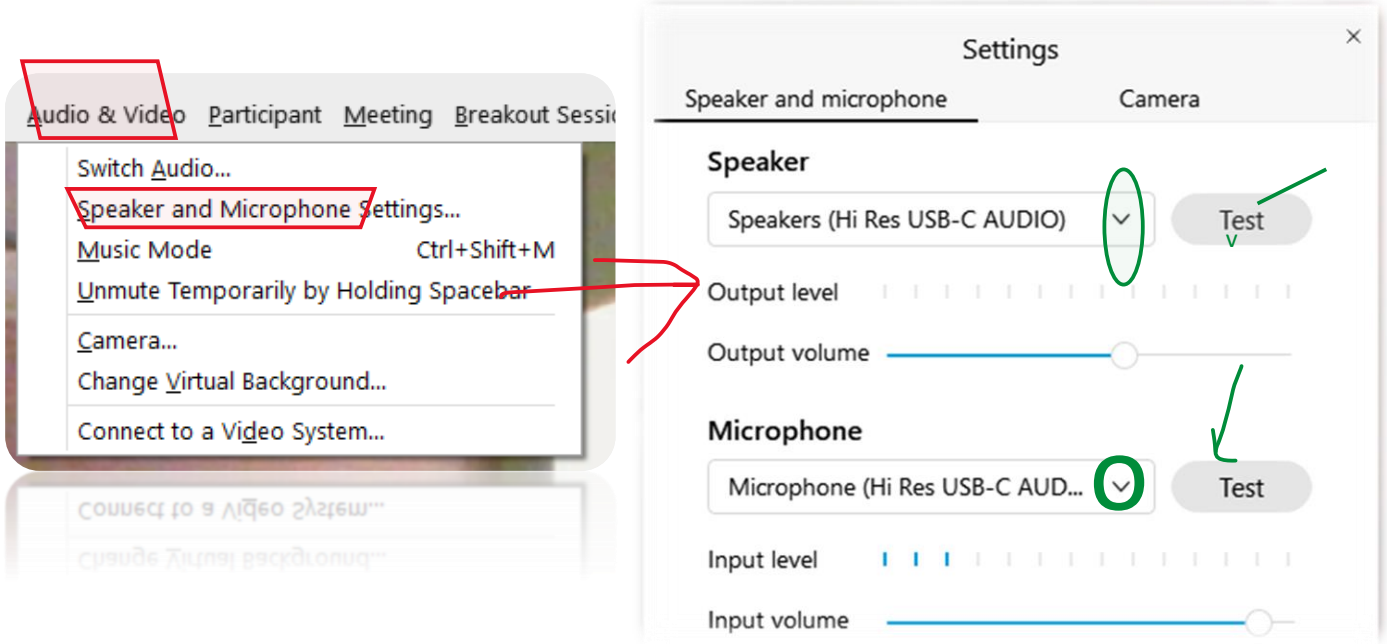
Rules of Engagement

Management of interaction – a lot of you on line

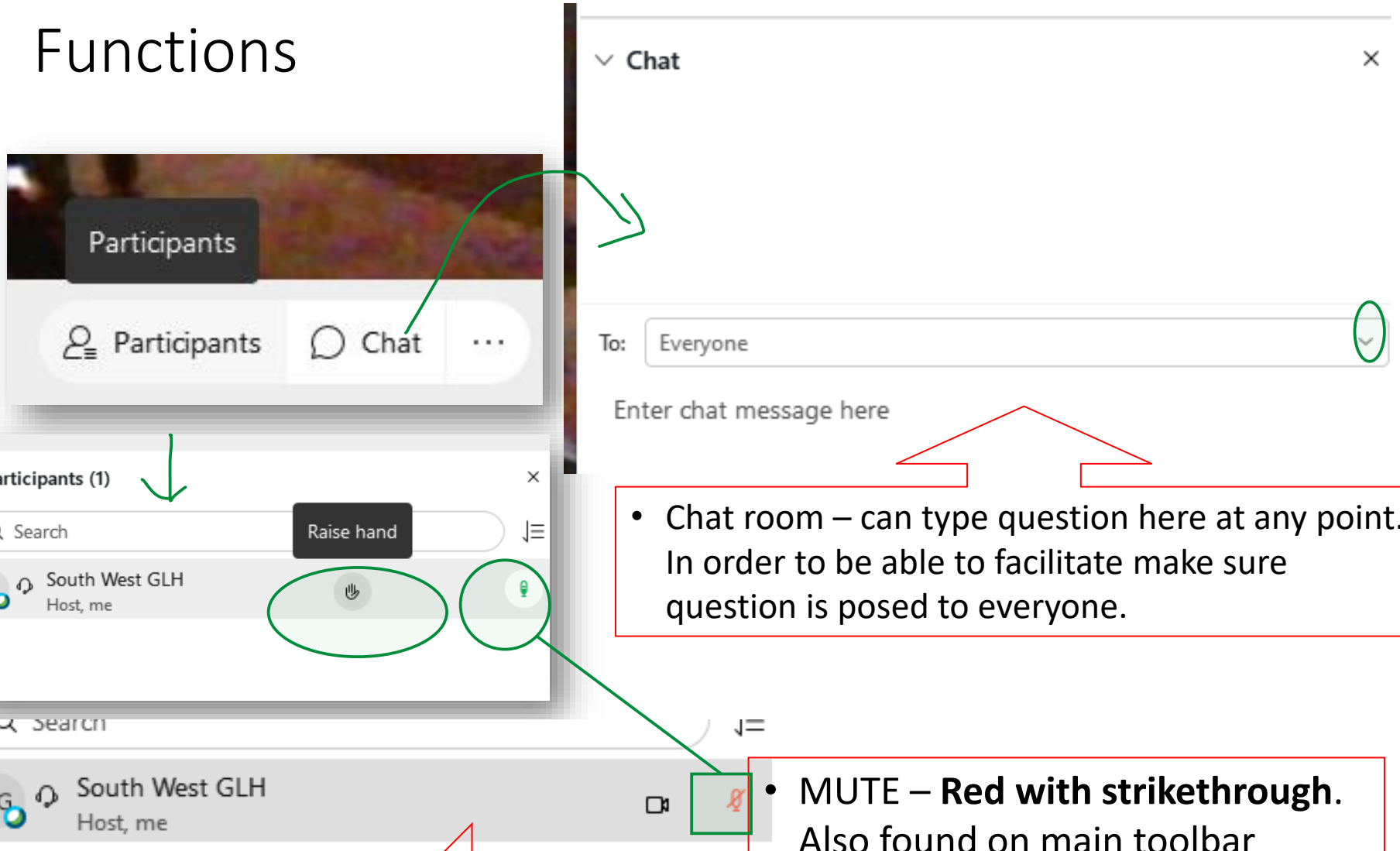
- **Touch points for questions**
  - End of Catherine Delmege's session
  - Break out sessions
- **Keep muted**
  - Invitation to speak during breakouts
- **Use chat room function**
- ***No question is a silly question!***
  - ***Be respectful – to presenters and peers***

Trouble shooting

- Audio and technical issues
  - Self management – check audio and microphone set up
- If all else fails – this is recorded so can be viewed after



Functions



- Chat room – can type question here at any point. In order to be able to facilitate make sure question is posed to everyone.

- MUTE – **Red with strikethrough**. Also found on main toolbar
- **Raise hand** – used when asked for questions – when asked can then unmute

The Programme

Time	Topic	Facilitator
13:50	**Room opens**	*Opportunity to familiarise yourselves with functions of WebEx.*
14:00	**Overview of session** • Rules of engagement • Breakout sessions • Post event resources **Team Introductions**	Mel Watson SWGLH Education Lead Bristol team
Part 1 - Overview of Genomic STP programmes in Bristol and programme training officers' perspectives		
14:10	**Overview of programmes in Bristol & Genomics programme Q&A**	Catherine Delmege SWGLH- BGL training officer
14:40	**Genomic Counselling programme**	Sally Monks Genomic Counsellor Bristol Clinical Genetics Dept.
15:00	**Cancer Genomics programme**	Kirsty Russell Clinical Scientist and Higher Specialist Scientific Trainee (HSST)
15:10	**Clinical Bioinformatics in Genomics programme**	Matthew Wherlock Clinical Scientist and Higher Specialist Scientific Trainee (HSST)

Part Two: The Trainee Perspectives		
15:20	**Genomics & Genomic Counselling**	Catherine Fielden, Emily Arbuthnot, and Emma Charlton
15:40	**Cancer Genomics**	Elle Mortensson
15:50	**Bioinformatics**	Clare Kennedy and Andrew Smith
16:00	**Breakout sessions**	*lead by trainees*
	Breakout 1: Genomics Q&A	Catherine, Emily, and Rosie
	Breakout 2: Cancer Genomics Q&A	Elle, and Jordi
	Breakout 3: Bioinformatics Q&A	Matt, Clare & Tom
	Breakout 4: Genomic Counselling Q&A	Poppy, Emma, and Jake
16:30	**Closing remarks & feedback**	*Regroup in main room*

Introductions

Genomics
- Catherine Delmege
- Catherine Fielden
- Emily Arbuthnot
- Rosie Woodruff

Cancer Genomics
- Kirsty Russell
- Eleanor Mortensson
- Jordi Baitup
- (Katie Jones)

Clinical Bioinformatics
- Matt Wherlock
- Clare Kennedy
- Andrew Smith
- Tom Scott- Adams

Genomic Counselling
- Sally Monks
- Emma Charlton
- Poppy Emmett
- Jake Povah

Bristol open event for Scientist Training Programmes in Genomics

Catherine Delmege
Bristol Genetics Laboratory
Training Officer

Aims for today

At the end of this event we hope you will understand more about

- STP Genomics training in Bristol
  - NBT - Bristol Genetics Laboratory
    - Genomics
    - Clinical Bioinformatics – Genomics
    - Cancer Genomics
  - UHBristol – Genomic Counselling
- Job Roles of Clinical Scientists in genomic specialties
- Recruitment process

NHS Genomic Medicine Service (GMS)

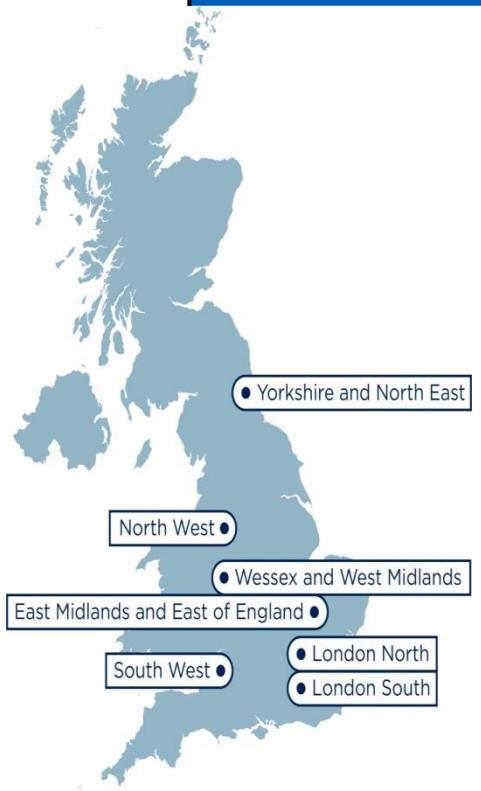
Launched in 2020:

Large expansion in Genetic/Genomic testing

Service providing consistent & equitable care for the country's 55 million population

- Delivering to a single national testing directory defining
  - **Available tests**
  - **Technology**
  - **Eligibility criteria**
  - **Tests will be centrally funded**

02/10/18 Genomic Laboratory Hubs



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Home › Genomics › Genomic Laboratory Hubs

Genomic Laboratory Hubs

From October 2018, Genomic testing in the NHS is being provided through a single national testing network, consolidating and enhancing the existing laboratory provision. This will create a world class resource for the NHS and underpin the future Genomic Medicine Service. It will also support the delivery of the Government's Life Sciences Strategy and the broader research and innovation agenda, building upon the NHS contribution to the 100,000 Genomes Project.

This new network will be delivered through a network of seven Genomic Laboratory Hubs (GLHs), each responsible for coordinating services for a particular part of the country.

The seven GLHs are:

- Wessex and West Midlands GLH led by Birmingham Women's and Children's NHS Foundation Trust
- East Midlands and East of England GLH led by Cambridge University Hospitals NHS Foundation Trust
- North West GLH led by Manchester University NHS Foundation Trust
- London North GLH led by Great Ormond Street Hospital for Children NHS Foundation Trust
- London South GLH led by Guy's and St Thomas' NHS Foundation Trust
- South West GLH led by North Bristol NHS Trust
- Yorkshire and North East GLH led by The Newcastle upon Tyne Hospitals NHS Foundation Trust

Expanding service

Alternative route to Clinical Scientist registration:–

- Alternative to STP training

We recruit Healthcare scientists at band 6 (non-registered).
Applicants require either:

- Comprehensive knowledge and experience of a wide range of laboratory genetics techniques and their clinical application and interpretation
- Experience of genetic/genomic research and development

Genetic Technologists – alternative career path

Graduate roles start at band 5

At BGL we have Genetic Technologists working in bands 6,7 and 8

Bristol Genetics Laboratory

BGL

- One direct entry STP post starting Sept 2021 in each of
  - Genomics
  - Bioinformatics Genomics
  - Cancer Genomics

NHS West of England NHS Genomic Medicine Centre

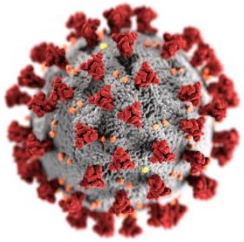
The 100,000 Genomes Project

BGL is an experienced training laboratory

- STPs since 2009
- 10 STPs at BGL currently

- Accredited by the National School of Healthcare Science (NSHCS) as a training laboratory

Training during the pandemic



- BGL Staff, including our STPs, work from home where possible with some attendance days at BGL
- Workplace training resources have been adapted to be used remotely
- FutureNHS collaboration platform – used for document sharing
- MSTeams – training tutorials, Q and A sessions with trainers

Modernising Scientist Careers Training Programmes

Scientist (STP)

- work towards Masters degree
- register with (HCPC) as a Clinical Scientist
- roles as a Healthcare Scientist (Afc band 6) or Clinical Scientist (band 7 onwards)

Higher Specialist (HSST)

- for a smaller number of experienced Clinical Scientists
- eligibility to apply for posts as a Consultant Clinical Scientist
- fellowship of the Royal College of Pathologists
- Taught doctorate

National School of Healthcare Science

Oversight, coordination and delivery of training

- Responsible for centralised Recruitment
- Produce the Curricula/learning guides
- Monitor the progress of trainees
- Maintain consistency and standardisation
- Manage the final exit examination

MANCHESTER 1824
The University of Manchester

Academic provider
MSc in Clinical Science

Scientist Training Programme

Has the same format for all specialties

Three year training programme

- Trainees are employed on a three-year temporary contract on the Agenda for Change pay scale at band 6

Agenda for Change (Afc) is the pay system which covers all NHS staff except doctors, dentists and very senior managers. Each of the nine pay bands has a number of overlapping pay points. Staff will normally progress to the next pay point annually until they reach the top of the pay band

- **STP Salary – band 6:**
  - £31,365 year 1 and 2, £33,176 for year 3 (Afc 2020/21)

Scientist Training Programme

- Over three years, trainees will complete:
  - ✓ an online portfolio of evidence of completion of work-based learning and assessments (OneFile)
  - ✓ MSc in Clinical Science – Manchester University
  - ✓ Final Assessment

- Some study time for the MSc is provided in the workplace (usually 1.0 day per week)
- MSc –supports workplace training; funded; travel and accommodation expenses provided; includes research project
- Additional study time outside the normal working week is required
- STPs are supernumerary members of staff with opportunities to work as part of a team and to gain experience in service delivery

Work-place training

First year: rotational year

- Trainees complete **four rotations** in their relevant field of healthcare science, one of which will be in own specialism
- Genomics trainees complete two rotations in-house (Genomics & Bioinformatics)
- Two external rotations – Genomic counselling; Reproductive Medicine

Second and third year: specialist years

- All trainees have specialist modules and a research project

Genomics modules:
- prenatal genomics
- paediatric genomics
- adult genomics
- cancer genomics

Bioinformatics modules:
- programming
- research methods
- advanced clinical bioinformatics
- whole systems molecular medicine
- IT for advanced BI applications
- applied NGS

Genetic Counselling modules:
- counselling & communication skills
- applied genomics in clinical care
- advanced counselling and ethical practice
- applied genomics and BI in advanced clinical care

Cancer Genomics modules:
- solid tumours (1&2)
- Haematological malignancies (1&2)

Broad training experience

In addition to work-place training in specialist modules, trainees complete:

- **Clinical Experiential Learning:** complements specialist knowledge
  - Patient facing experiences
  - Attendance at multi-disciplinary team meetings
- **Professional Practice**
  - Conferences, national or regional trainee events
  - Development of communication and interpersonal skills
- **Elective:** designed to facilitate a wider experience of healthcare and/or the practise of healthcare science in a cultural and/or clinical setting that is different from the usual training environment
  - Could be in a different area of the health service
  - Up to 4 weeks
  - Trainee plans and arranges elective with support from laboratory staff

Support for Trainees

Workplace training:

- Named trainer for each module
- Tutorials with trainers
- Training reviews with Training Officer
- Trainee networks – local/regional/national
- Focus on 'self-directed learning'
- Be responsible for your own training progression

The recruitment process

- National process managed by the NSHCS
- See NSHCS website for information
- Application includes a “situational judgement test” – replaces previous numerical and logical test

- NSHC Q&A webinar on **Thursday 28th January** at 2:00pm

Status	STP Direct entry	STP In-service
Applications open	Monday 25th January 2021 at 11:00am	Monday 25th January 2021 at 11:00am
Applications close	Monday 22nd February 2021 at 4:00pm	Friday 26th February 2021 at 4:00pm

What are we looking for?

Applicants must have an honours degree (1st or 2.1) in a pure or applied science relevant to the specialism for which they are applying. Applicants with a relevant 2.2 degree will also be considered if they have an MSc or PhD in the specialism for which they are applying.

Good personal organisational skills

For all candidates evidence of research experience, e.g. in the form of a higher degree or equivalent evidence of scientific and academic capability, is considered desirable.

Good attendance record

Effective team worker, willing to adopt a role working in collaboration with others.

A passion for (committed, in depth interest in and enjoyment of) scientific practice and its application to direct clinical care of patients in a clinical environment.

Ability to work under pressure (emotional resilience and ability to prioritise and plan work)

Websites

- Bristol Genetics Laboratory website
- https://www.nbt.nhs.uk/south-west-genomic-laboratory-hub

- National School of Healthcare Science: includes information on recruitment, available posts, curricula/ learning guides
  - www.nshcs.hee.nhs.uk

Thank you for listening

Any questions?

Clinical GENETICS Service

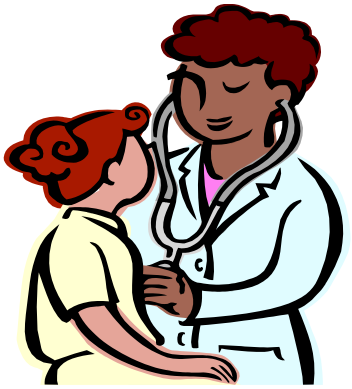
University Hospitals Bristol NHS
NHS Foundation Trust

STP Genomic Counselling Programme

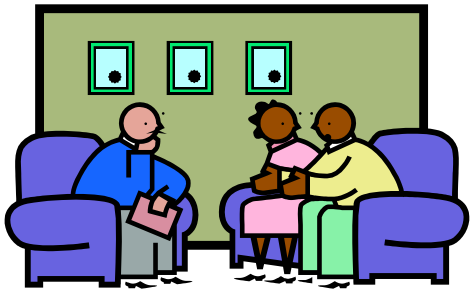
Clinical GENETICS Service

University Hospitals Bristol NHS
NHS Foundation Trust

Regional Genetics Service
A Multidisciplinary Team



Consultant Geneticists and Speciality registrars in Clinical Genetics



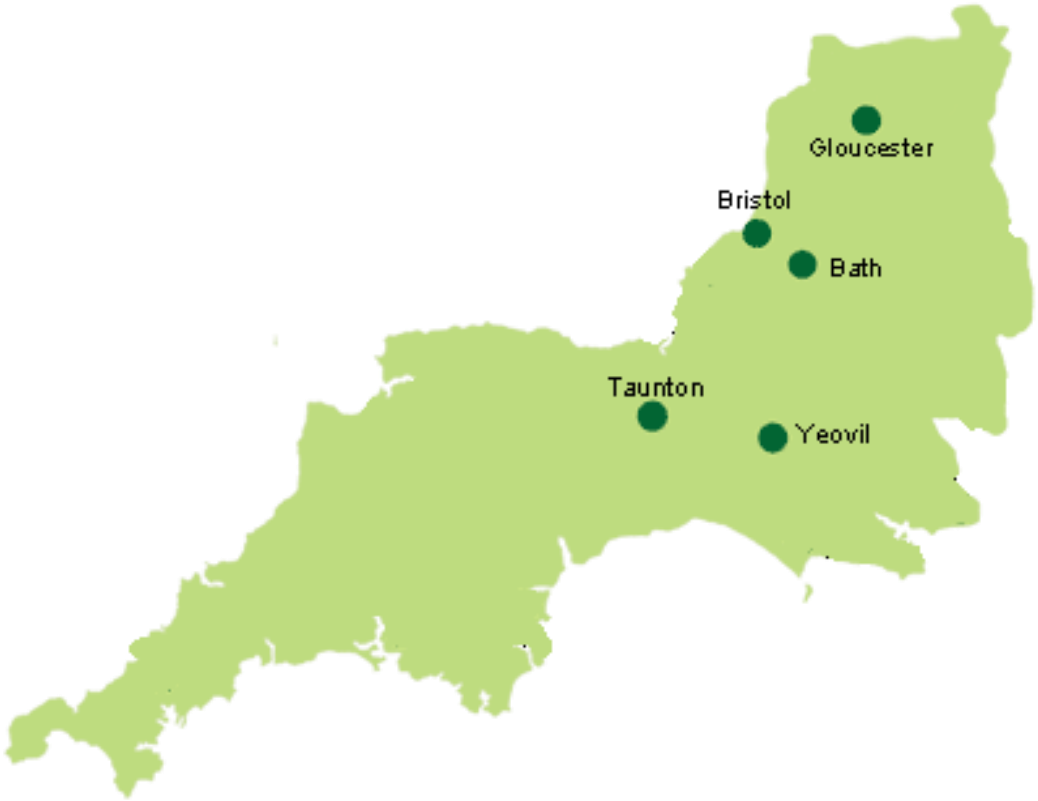
Genetic Counsellors

Molecular and Cytogenetic lab staff

Clinical GENETICS Service

University Hospitals Bristol NHS
NHS Foundation Trust

Regional Genetics Service



Bristol Regional Genetics Service
St Michael's Hospital
Bristol

Peripheral departments:

Royal United Hospital
Bath

Gloucestershire Royal Hospital
Gloucester

Musgrove Park Hospital
Taunton (incl. a Yeovil clinic)

Bristol Genetics Laboratory
Southmead Hospital

REASONS FOR REFERRAL

- Patient with known or suspected genetic condition
- Family history of genetic condition or pattern of disease suspicious of genetic disorder e.g. cancer, cardiac
- Child with developmental delay
- Abnormalities identified during pregnancy
- Prenatal genetic testing or advice
- After reproductive loss

Clinical GENETICS Service

University Hospitals Bristol NHS
NHS Foundation Trust

What is Genetic Counselling?

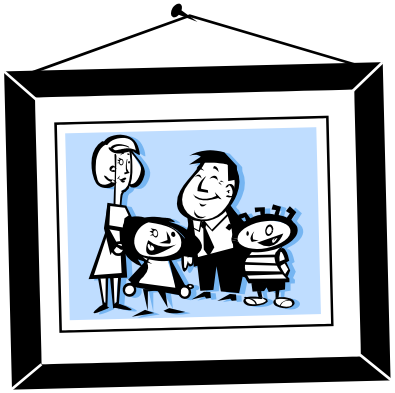
The communication process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease

GENETIC COUNSELLING INCLUDES:

- DIAGNOSIS or confirmation of diagnosis
- ASSESMENT of risk and gathering family and medical history
- EXPLANATION of the genetic facts and recurrence risks
- DISCUSSION of options including genetic testing and prenatal diagnosis
- SUPPORT towards making the best possible adjustment/decision

Genetic Counsellors can also help with:

Advice on disclosing risk to relatives



Informational support

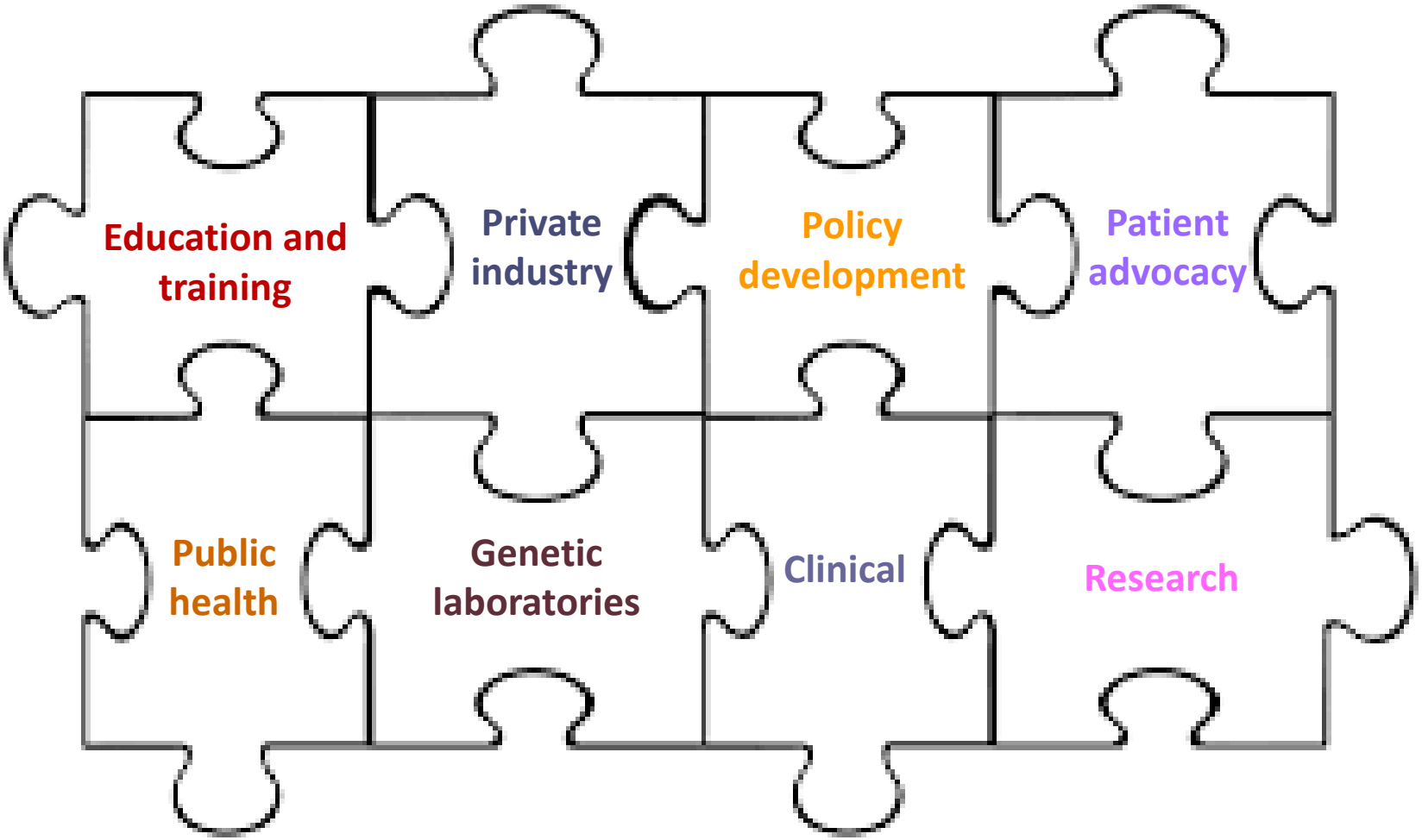
Emotional support

GENETIC COUNSELLOR'S WORK:

- Autonomous and with medical staff
- Face-to-Face, virtual or phone clinic appointments
- Pre-clinic preparation (clinic planning, confirmation of diagnosis, reviewing literature)
- Post-clinic follow-up (letters, arranging testing/screening/referrals, further support)
- Giving and explaining genetic testing results
- MDT working
- Training colleagues (Genetics and non-genetics)
- Continuing professional development
- Audit/research

.....and More

The varied roles of genetic counsellors



Becoming Genetic Counsellor

- Entry routes:
  - STP Genomic Counselling Programme (England)
  - MSc Genetic and Genomic Counselling (Cardiff and Glasgow)
  - Via nursing or midwifery (set B criteria)
  - International courses

What we are looking for in a Genomic counselling application

- Why do you want to be a GC? What experience do you have that will be relevant? Have you attended an open event, met with or shadowed a GC?
- What caring experience do you have and to what level? Reflect on how this will benefit you working as a GC
- What human genetics experience do you have?
- What counselling skills have you used in your experience? Can you reflect on the use of these? Do you have any formal counselling training?
- Don't make statements about yourself without reflection and/or examples from your experience

Further information

- Association of Genetic Nurses and Counsellors
  - www.agnc.org.uk
- Genetic Counselling Registration Board
  - www.gcrb.org.uk
- British Society of Genetic Medicine
  - www.bsgm.org.uk
- Gemomics Education
  - www.genomicseducation.hee.nhs.uk
- Genetic Alliance
  - www.geneticalliance.org.uk

STP in Cancer Genomics

Kirsty Russell

Clinical Scientist and Higher Specialist Scientific Trainee (HSST)

Oncology Genomics

Bristol Genetics Laboratory

Introduction

Bristol Genetics Laboratory

- The STP in Cancer Genomics is a relatively new training scheme, which was added to the STP training programme in 2018.
- Bristol Genetics Laboratory (BGL) currently has three STPs (Katie, Elle and Jordi) in this discipline.
- The role of a Clinical Scientist in Cancer Genomics is to deliver diagnostic services for patients who have a possible or confirmed diagnosis of cancer.
- Training will be mainly based at BGL but regular interaction with other healthcare professionals such as cellular pathologists and haematologists will be an important part of the role to provide integrated services.

Introduction Cont.

Bristol Genetics Laboratory

- This will be provided by:
  - two three month rotations (one in cellular pathology and one in haematology) in year one
  - attendance at Specialist Integrated Haematological Malignancy Diagnostic Service (SIHMDS) MDT meetings in years 2 and 3
  - attendance at the paediatric MDT meetings at the Bristol Children's Hospital in years 2 and 3
- Also need to complete a project and an elective within the 3 year training programme.

STP in Cancer Genomics – Role Summary

Bristol Genetics Laboratory

Throughout the training period the STP will:

- Process diverse clinical samples referred via a range of cancer diagnostic pathways covering both haematological and solid malignancy
- Provide analysis and full interpretation of genomic results that will inform the integrated clinical report
- Communicate clinical information to a range of service users
- Provide active participation in the multi-disciplinary environment

Role Summary Continued…

Bristol Genetics Laboratory

- Take responsibility for the work and development of themselves and others
- Take responsibility for aspects of the day to day running of the service
- Take responsibility for the identification and implementation of new ways of working that will promote the use of latest scientific advances into clinical practice for the benefit of the patient
- Contribute to research and development in the area of practice

(source: the National School of Healthcare Science)

Module Plan

Bristol Genetics Laboratory

	Module Title	Module Aim
Year 1	Introduction to Healthcare Science, Professional Practice and Clinical Leadership	To provide broad knowledge and understanding of science and scientific knowledge, contextualised to the practice of healthcare science and the services.
	Introduction to Cancer Genomics	*TBC*
	Genetics, Genomics and Molecular Science	To provide an introduction to human genetics, genomics and molecular science
	Introduction to Clinical Bioinformatics and Genetics	To provide knowledge of genetics and knowledge and understanding of bioinformatics tools and infrastructure.
Year 2	*Solid tumours (1)* • *Colorectal* • *Melanoma*	*To provide in-depth knowledge of the molecular mechanisms of colorectal cancer and melanoma, and the associated genomic testing and clinical skills.*
	Haemato-oncology Tumours (1) • *CML* • *Burkitts Lymphoma*	*To provide in-depth knowledge of the role of molecular pathology in the diagnosis of lymphoma, and of the analysis of haemo-oncological disease.*
	Research Methods	
	Research Project Part 1	
Year 3	*Solid tumours (2)* • *Ovarian* • *Lung* • *Breast*	*To provide specialist understanding of the biological process and molecular mechanisms of cancer and the organisation and delivery of the associated diagnostic genotyping service.*
	Haemato-oncology Tumours (2) Myeloid Conditions	*To provide specialist knowledge of the role of molecular pathology in the diagnosis, prognosis and molecular monitoring of patients with a haemato-oncolgical malignancy.*
	Research Project Part 2	

Existing modules/*New Cancer Genomics specialty modules*

Future Career

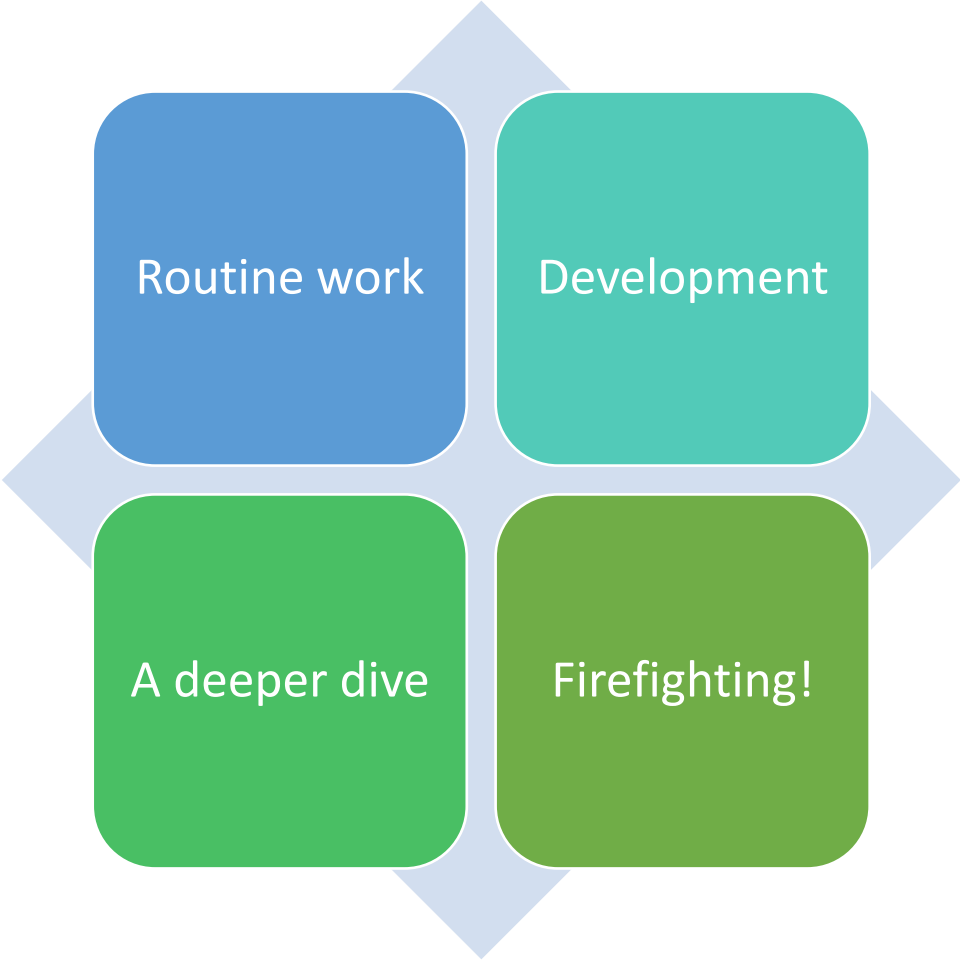
Bristol Genetics Laboratory

- Health and Care Professionals Council (HCPC) Registered Clinical Scientist in the Oncology Genomics team within a Genetics Laboratory – in the future this may be more of a moving role between pathology departments?
- Opportunity for further study to consultant clinical scientist level by participation in Higher Specialist Scientific training (HSST) in Molecular Pathology of Acquired disease.
- Stand alone RCPath in Molecular Oncology of Acquired disease.

Clinical Bioinformatics (Genomics) STP

Matt Wherlock

The role



Work-based training

- Mostly hosted here
- Cover competencies and assessments
- Training mostly delivered within the team
- Projects cover most competencies
- Individual professional development

Year 1

- Introduction to Clinical Bioinformatics and Genetics
- Computing for Clinical Scientists
- Information and Communications Technology in the Clinical Environment
- Introduction to Health Informatics Science

Years 2 & 3

- Advanced Clinical Bioinformatics
- Programming
- Applied Next Generation Sequencing
- IT for Advanced Bioinformatics Applications
- Whole System Molecular Medicine

Academic

University of Manchester

Mostly course work and group work

Some health informatics and physical sciences in the first year

More specialised in the second and third years

Research project in department but assessed by Manchester

HSST

SPECIALIST TRAINING

CONSULTANT LEVEL

5 YEARS

PROFESSIONAL DOCTORATE

NHS
South West Genomic Laboratory Hub

Clinical GENETICS Service

The trainee perspective – Genomics and Genomic Counselling

Cath Fielden (Genomics)

Emily Arbuthnot (Genomics)

Emma Charlton (Genomic Counselling)

Clinical GENETICS Service

University Hospitals Bristol NHS
NHS Foundation Trust

Genomic Counselling STP: a day in the life

A typical day for me might include:

- Attending a clinic
- Writing notes and letters
- Gathering information for patients and their relatives for clinic
- Attending a meeting
- Work towards competencies
- Work towards my research project

Module 1: Principles and practice of genomic counselling

1	Critically reflect on the role of the genetic counsellor within genetic services and patient pathways through observing consultations involving an adult for the common types of referrals to a clinical genetics service.	• The role of the genetic counsellor within genetic services and patient pathways. • The range of patient pathways for an adult patient (e.g. prenatal, cancer genetics, cardio-genetic, neuromuscular, neurological, connective tissue disorders). • How the clinical genetic service fits within the patient pathway for an adult patient.
3	Critically reflect on the roles of the professional groups involved in delivering an NHS clinical genetics service.	• The role of laboratory staff in an NHS clinical genetics service. • The role of clinical geneticists in an NHS clinical genetics service. • The role of genetic counsellors in an NHS clinical genetics service. • The role of administrative staff in an NHS clinical genetics service.
4	Appraise how clinical genetics and other health professionals work together in multidisciplinary teams.	• Composition, role and working-practice of multidisciplinary teams. • Inputs and outputs from these multidisciplinary teams.

Module 1: Principles and practice of genomic counselling

5	Identify the individual's agenda in five observed genetic counselling sessions.	• Role and process of active listening. • Importance for establishing agenda.
6	Identify the individual's psychosocial concerns in five observed genetic counselling sessions.	• Theories of psychosocial adjustment. • Responses to loss (bereavement, loss of imagined future). • Responses to uncertainty. • Family life cycle. • Impact of illness/disability on the family at different stages of the family life cycle • Impact on the family when one or more family members have complex needs
15	Use a model of reflective practice to describe what happened and what could have been done differently to achieve a better outcome for the patient in three observed genetic counselling sessions.	• Different models of reflective practice. • Strengths and weaknesses of each model. • Counselling theories. • Genetic counselling practice.

Module 1: Principles and practice of genomic counselling

14	Accurately communicate risk under direct supervision.	• Strategies to convey risk and other genetic information relevant to the client's agenda.
11	Gather a comprehensive 3-generation family history relevant to the clinical question and construct a clear 3-generation family tree under direct supervision.	• Questions to ask when obtaining a family history. • Pedigree symbols. • Drawing a 3-generation family history.
8	Actively listen whilst establishing a relationship with the patient in a genetic counselling context in order to establish the patient agenda under direct supervision.	• Principles of patient centred counselling. • Role and process of active listening. • Importance for establishing agenda. • Role in ascertaining medical and family history.

NHS South West Genomic Laboratory Hub

North Bristol NHS Trust

Genomic STP: a day in the life

The training programme is a balance between service work vs gathering evidence for competencies vs studying. **It's not all analysing and reporting cases.**

Day to day I'm:

- Observing technical processes performed by GTs
- Writing up notes/essays for my online portfolio as evidence of knowledge and understanding or preparing for assessments
- Reading journals, textbooks, best practice guidelines, SOPs
- Analysing and reporting both training cases and live service work (under supervision)
- Supporting scientists (e.g. performing audits/export work/answering phones)
- Attending clinics, MDTs, conferences, workshops
- Participating in public engagement
- Planning my own workload and how I am going to provide evidence for competencies

Module 1: Genetics, Genomics and Molecular Science

1	Apply infection control risks in accordance with departmental protocols.	• Protocols and requirements for hygiene and infection control related to the relevant range of investigations, including preparation, conduct and completion of investigation. • Protocol for hand washing and how effective hand washing contributes to control of infection.
3	Critically reflect on referral patterns for genetic investigation following standard laboratory practices including sample receipt.	• Minimum data set required for identification of samples and the importance of ensuring that this is complete, correct and appropriate. • Factors affecting sample integrity and appropriate corrective action. • Procedures for handling samples which may contain category 2, 3, and 4 pathogens. • Use of laboratory and hospital information systems to identify and record patient demographics, clinical details and relevant laboratory results. • The importance of maintaining correct and unique labelling, including transfer of labels throughout the preparation.

Module 1: Genetics, Genomics and Molecular Science

5	Perform a basic chromosome analysis on a minimum of three cases that demonstrate different chromosomal syndromes or anomalies.	• Basic chromosome identification. • Karyotype construction. • G-banding. • Numerical and structural anomalies and normal variation. • Relationship of basic chromosomal anomalies to clinical features in patients. • Correct ISCN nomenclature.
8	Perform a basic molecular analysis on three cases demonstrating common genetic condition.	• Analysis of results following standard laboratory procedures. • The clinical background and molecular pathology of the disorder being investigated. • The range of tests available for the individual or the family. • Significance of previous results in relation to the current sample. • Relevant professional guidelines and correct interpretation.
11	Support the preparation of reports and the reporting process for patients being investigated for genetic disorders.	• Range of reporting formats and options. • Relevant professional guidelines for reporting. • Policy for authorisation and disclosure of results and the need for confidentiality and information governance. • Factors involved in evaluation of clinical risk to the patient and their family. • Procedures for issuing written results, verbal results or for faxing. • Patterns of inheritance (Mendelian and non-Mendelian), including imprinting.

NHS South West Genomic Laboratory Hub

North Bristol NHS NHS Trust

Clinical Scientists

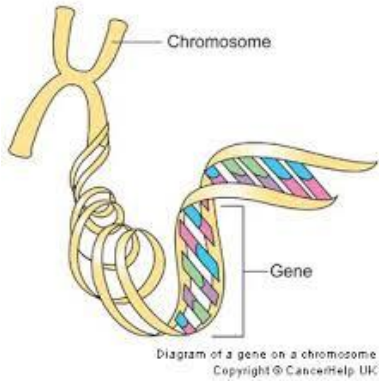
They don't have a lab coat* and work in an office at a computer!

Responsibilities:

- Analyse and interpret scientific data (clinical context also important)
- Report findings in a clear and unambiguous manner.
- Service management
- Service development
- Problem solving, troubleshooting assays and technical direction
- Report authorisation
- Export samples through the UKGTN
- Audit data
- Write Standard Operating Procedures (SOP) for laboratory processes
- Complete External Quality Control schemes
- Continued professional development
- Present data at external/internal meetings
- Training

NHS
South West Genomic Laboratory Hub

Clinical GENETICS Service



What types of test do we perform at BGL?

Cytogenetics: a method of visualising changes to the whole genome

e.g. chromosome analysis.

Molecular genetics: methods of visualising changes to the sequence of DNA

e.g. testing for cystic fibrosis (CF)

Case study

- 26 year old man with unexplained infertility

South West Genomic Laboratory Hub

Case Study: Referral

North Bristol NHS Trust

Genetics Request Form

MANDATORY FIELDS are indicated in blue type
See reverse for additional information on sample requirements
Please fill out as completely as possible

North Bristol NHS Trust

BGL is a UKAS accredited medical laboratory No.9307

Tubes/volumes

BGL FM296 V8
Active 09/04/18

For BGL use only: DO NOT affix labels here

Please affix patient label here

SURNAME: STUDY
FIRST NAME: Case
ADDRESS
POST CODE

DOB | SEX: M | CG number | CONSULTANT

HOSPITAL NUMBER
NHS NUMBER
REFERRING HOSPITAL

Address for report (full address please if GP surgery)
Purchase Order no:

BILLING ADDRESS (if different to report address)

Date/time taken and by whom

Mother's name/DOB (IMPORTANT for all infant/fetal samples)

Date/time received

Previous genetic investigations in family: Yes/ No
If yes, please give brief details below (include laboratory numbers)
Mother (name & DOB).....
Father (name & DOB).....
Sibling/s (name/s & DOB).....

For LEUKAEMIC samples please indicate:
- Diagnostic ☐
- Follow-up (Remission) ☐
- Follow-up (Relapse) ☐
- Bone marrow transplant, specify sex of donor ☐
- MRD ☐

IMPORTANT for all fetal samples
Gestation..... Parity.....
Gravida..... LMP.....
EDD.....
Multiple pregnancy ? NT≥3.5mm ☐

Date and time of next appointment

Sample type
- Private ☐
- NHS ☐

Priority
- Urgent ☐
- Routine ☐

Specific molecular genetic tests — Consent required for DNA extraction and storage — **EDTA TUBE**

Specific cytogenetic tests — **LITHIUM HEPARIN TUBE**

DANGER OF INFECTION? YES / NO
(NBT The lab works to containment level 2+)
- Risk of blood borne pathogen? ☐
- Recent blood transfusion? ☐
- Recent cytotoxic drugs? ☐

Please give additional details:

CLINICAL SUMMARY/ADDITIONAL INFORMATION/SPECIAL REQUESTS (if several tests are required please indicate order of testing required)

Unexplained infertility

BGL acceptance of a testing request acts as an agreement with the requestor

- DNA extraction and storage ☐
- DNA Export (Please attach letter) ☐
- DNA testing (please specify test required) ☐
- Array CGH testing ☐
- Karyotyping ☐
- QF-PCR: aneuploidy ☐
- QF-PCR: gender ☐
- Mosaicism ☐
- FISH (please specify) ☐
- Breakage studies: Fanconi anaemia/ Ataxia Telangiectasia/Other ☐
- Fixed cell storage for 2 years (blood only) (stored routinely for 4 months) ☐
- Fixed cell storage (oncology only) ☐
- Cell freezing (solid tissues/prenatal samples) ☐

BGL use only

Lab N°(s)

CONSENT STATEMENT (please see overleaf): It is the referring clinician's responsibility to ensure that the patient/carer knows the purpose of the test and that the sample may be stored for future diagnostic testing. In signing this form the clinician has obtained consent for testing, storage and for the use of this sample and the information gathered from it to be shared with members of the donor's family through their health professionals (if appropriate). The patient should be advised that the sample may be used anonymously for quality assurance and training purposes. If the patient does not wish information to be shared please write this clearly in the clinical summary box. Certain disorders with particular counselling issues e.g. HD may require a specific consent form (see website for further details).

NAME: SIGNATURE: BLEEP No:

BRISTOL GENETICS LABORATORY, SOUTHMEAD HOSPITAL, BRISTOL, BS10 5NB Tel: 0117 414 6168/6167/6174 Fax: 0117 414 6464 General enquiries email (not results) : nbn-tr.geneticsenquiries@nhs.net

Case Study: Test Directory

Referral: Unexplained infertility

R297 Possible structural chromosomal rearrangement - karyotype

Testing Criteria

Possible structural chromosomal rearrangement requiring karyotype including:

1. Possible Robertsonian translocation, reciprocal translocation, ring chromosome or other microscopically visible structural rearrangement indicated by findings from microarray, WGS or other laboratory technique, OR
2. Recurrent miscarriage (defined as three or more consecutive miscarriages) in whom testing of products of conception has not been possible. Note: this should not be performed routinely but can be used in exceptional circumstances where testing of products of conception has not been possible, for example because no testable material has been stored or retained, OR
3. A family history suggestive of familial balanced translocation, OR
4. Unexplained infertility who are going to undergo infertility treatment, OR
5. Patient with ambiguous genitalia potentially caused by a sex chromosome rearrangement not detectable via other tests

Set up cell culture

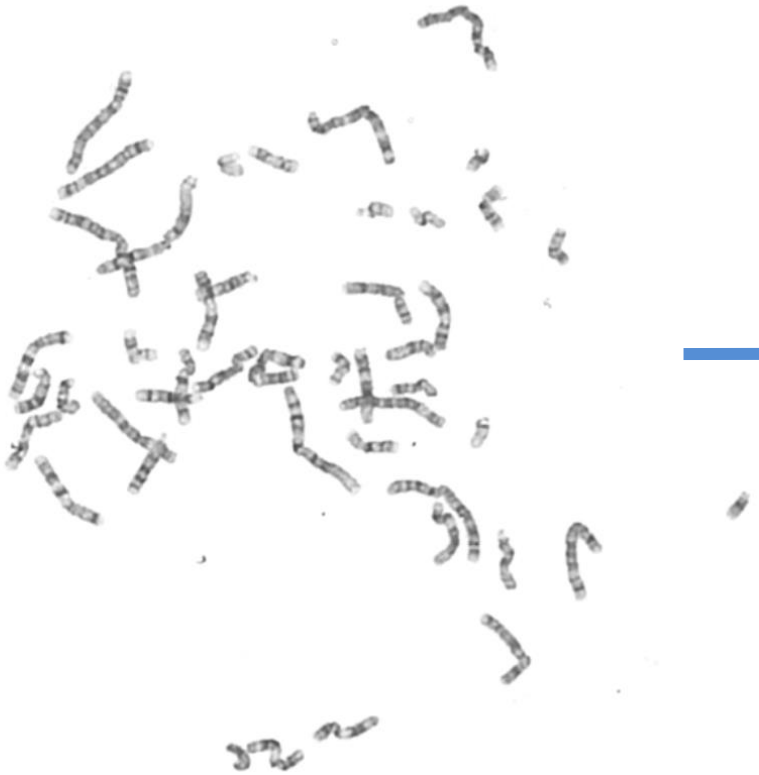
Harvest

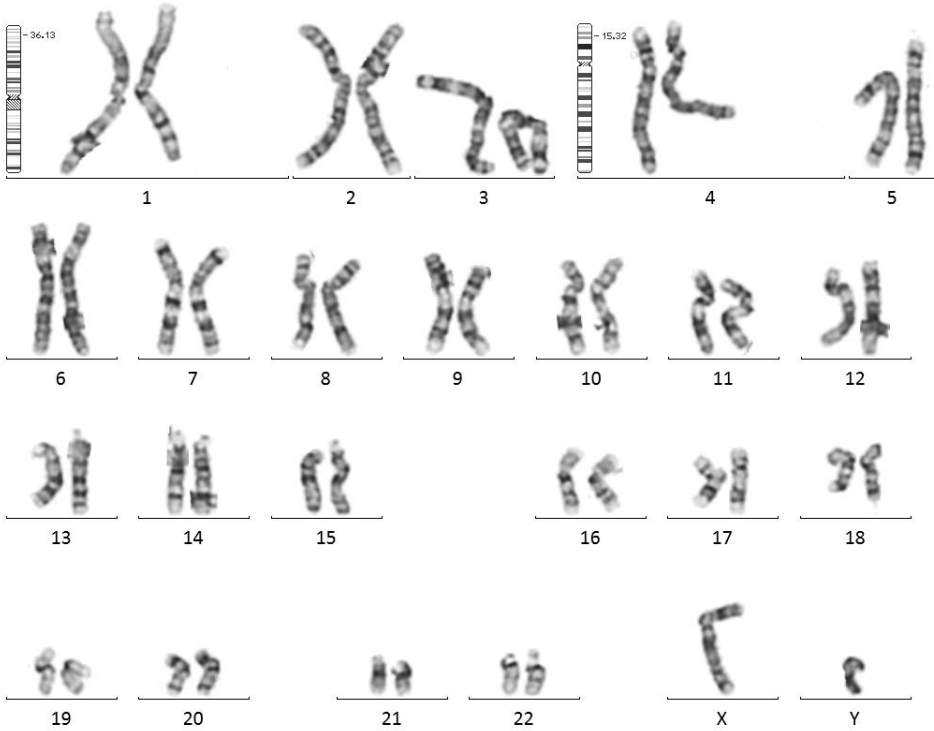
Slide preparation

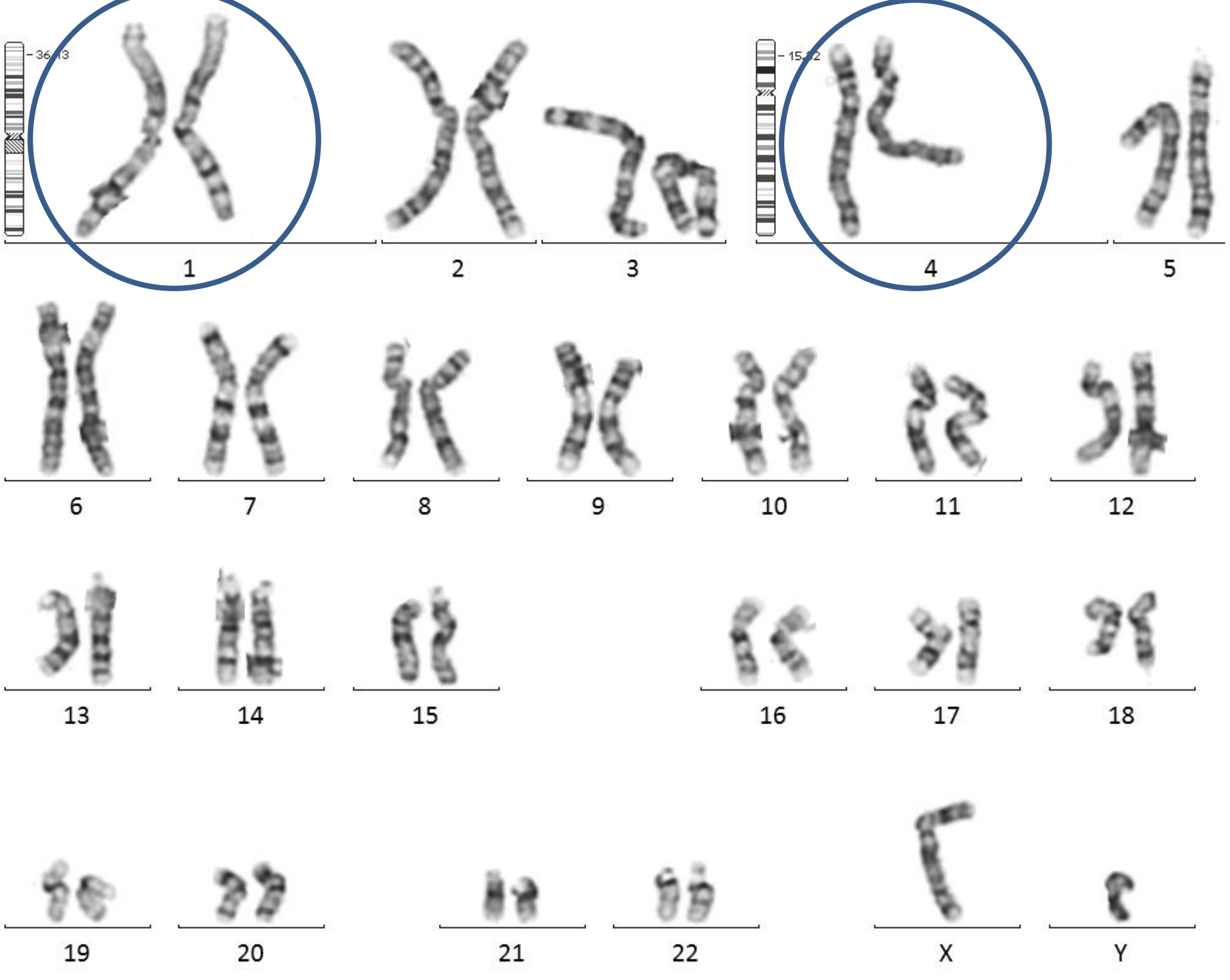
G-Banding

Analysis

Karyotyping



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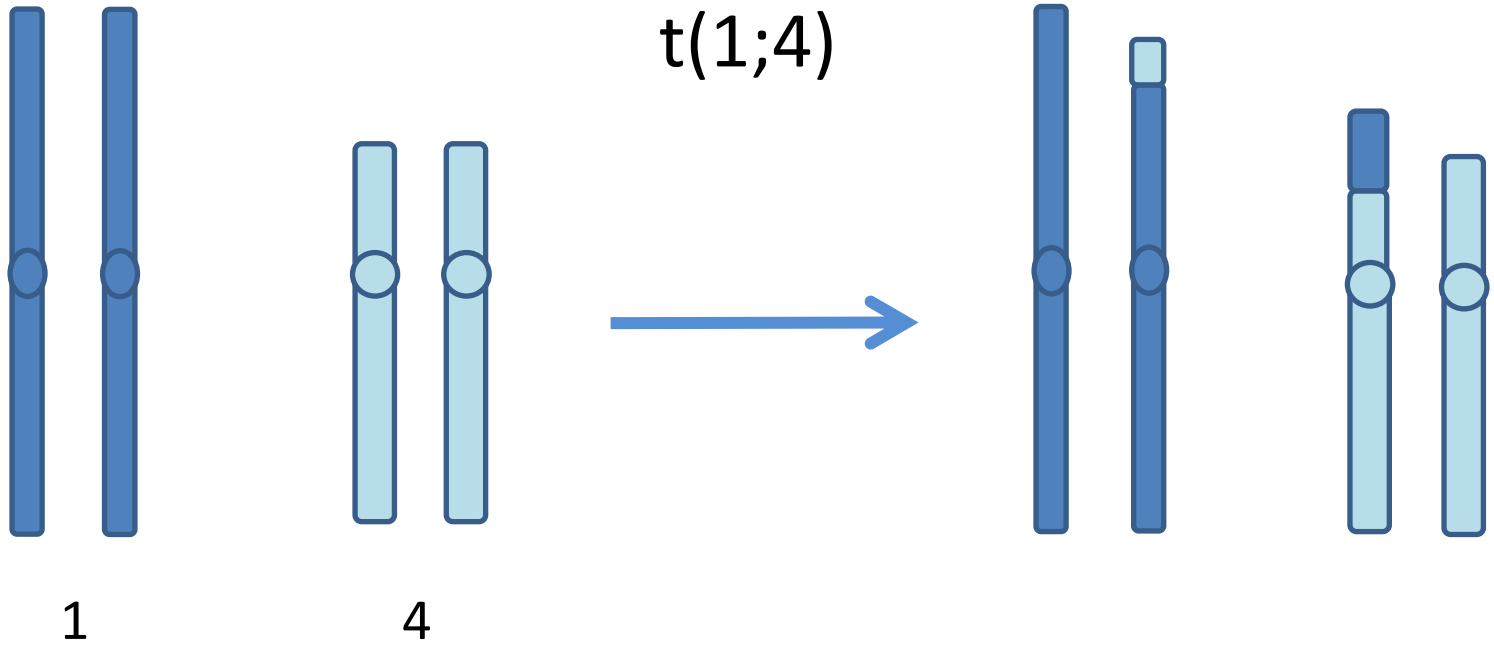
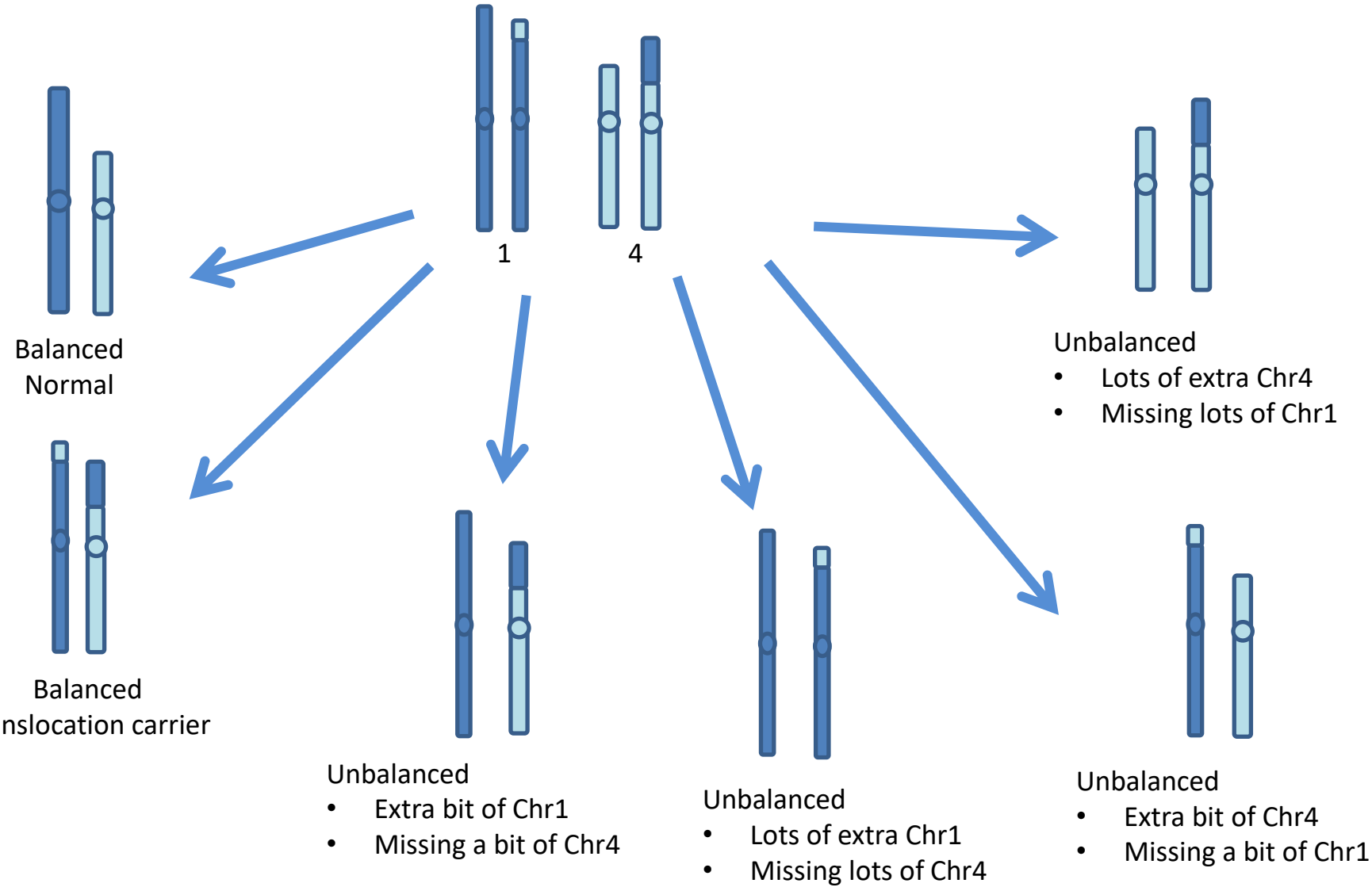
Translocation



Reproductive Implications



NHS South West Genomic Laboratory Hub

Case Study: Report

North Bristol NHS Trust

Genetics Report

Full Name:	**Case Study**			Sample Type:	**Whole Blood**
Date of Birth:	**Aged 26**	Sample ID:	**Case Study**	Collected:	
Sex:	**Male**	CG No.:		Received:	
NHS No.:		External ID:		Activated:	
Postcode:		Hospital No.:		Authorised:	

Clinical summary: Fertility investigations

Results & Report

Case Study has a balanced reciprocal translocation between chromosomes 1 and 4. This confers a risk of liveborn offspring with intellectual disability and dysmorphism. Prenatal diagnosis should be considered in any future pregnancy. Wider family studies are advised.

Karyotype: 46,XY,t(1;4)(p36.13;p15.32)

Analysis showed a male karyotype including a balanced reciprocal translocation between the short arm of chromosome 1 and the short arm of chromosome 4, with breakpoints at 1p36.13 and 4p15.32. Balanced translocations are usually carried without phenotypic effect.

This translocation is consistent with recurrent miscarriages in CS's partner, and confers a risk of future conceptions with an unbalanced chromosome complement which may result in the live birth of an infant with substantial physical and intellectual disabilities therefore, CS and his partner should be advised to consider prenatal diagnosis in any future pregnancies. Please refer to table below with possible viable outcomes (*i*).
NB: a normal pregnancy with a normal or balanced karyotype may also be achieved, although some translocation carriers may experience recurrent miscarriage.

In order to establish whether there are any other relatives to whom a similar reproductive risk applies a family study is advised, beginning with chromosome analysis of CS's parents. See accompanying patient information leaflet and karyotype.

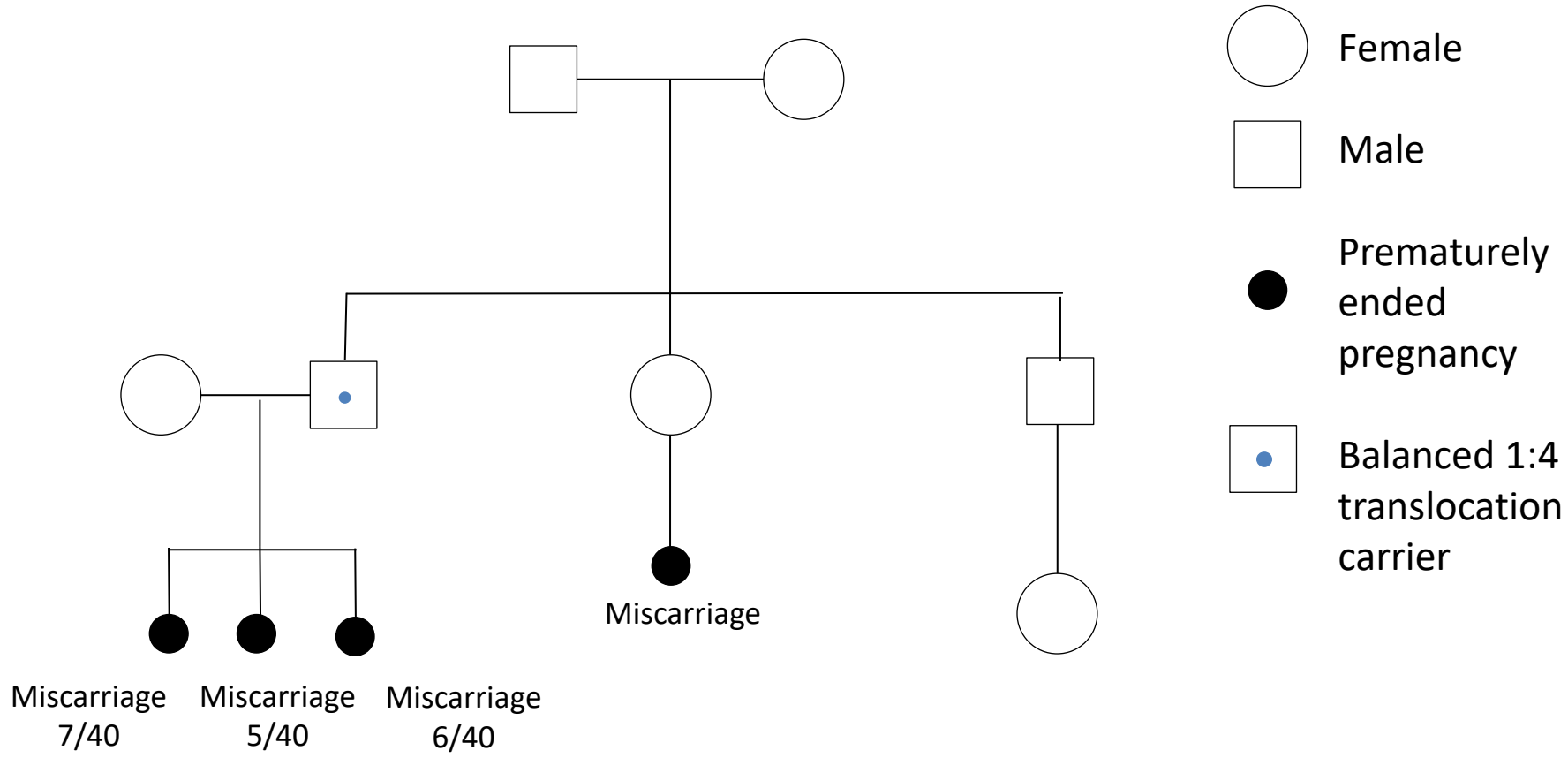
Unbalanced segregants which may result in live birth

	Mode of segregation	Karyotype	Trisomic segments	Monosomic segment
(*i*)	**Adjacent I**	**46,XN,der(1)t(1;4)(p36.13;p15.32)**	**4pter – 4p15.32**	**1pter – 1p36.13**

Other unbalanced segregants, not listed may occur, resulting in pregnancy loss.

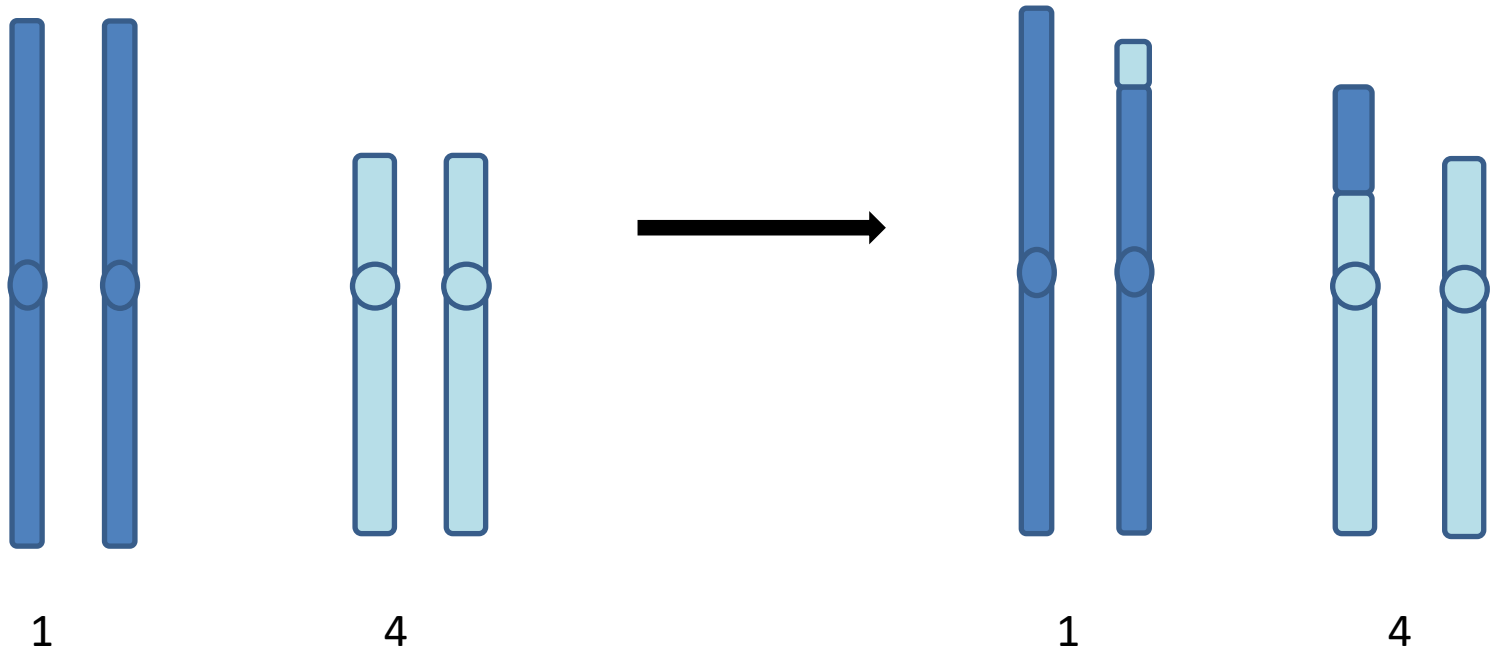
In Clinic

- Take a brief medical history and family history



In Clinic

Explanation of results: balanced reciprocal translocation



In Clinic

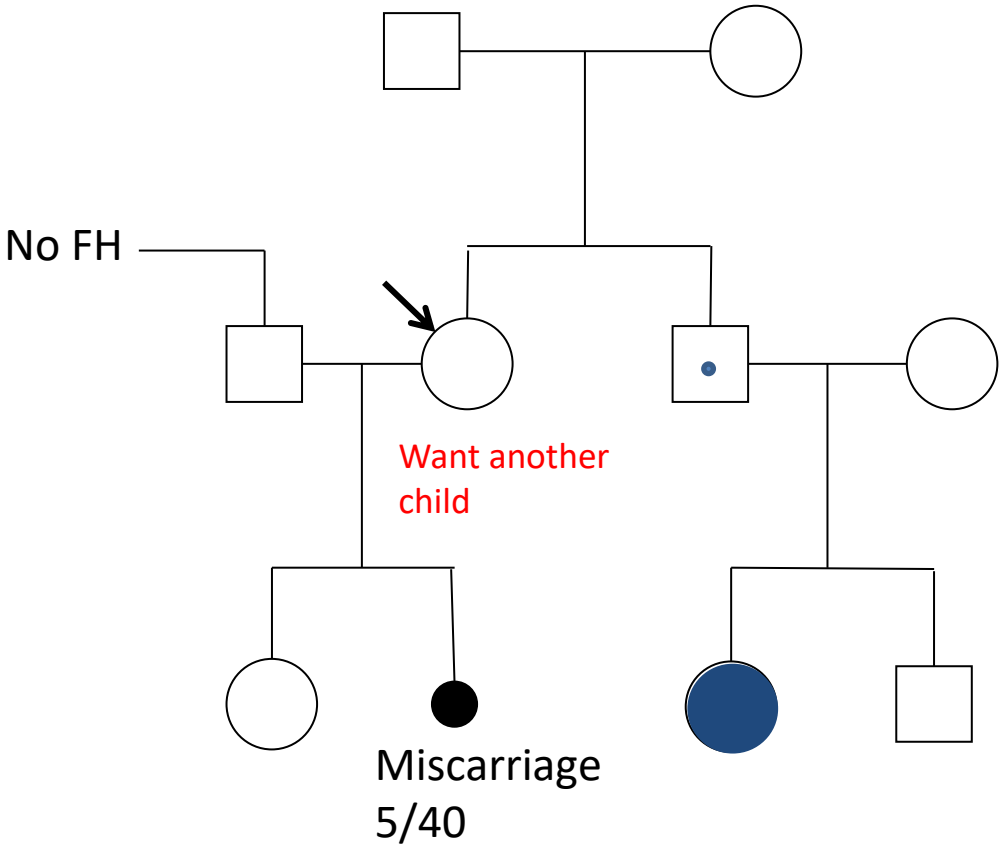
- Implications- reproductive options
  - Testing before (PGD), during (CVS or Amniocentesis) or after pregnancy
  - Using donor sperm or adopting

- Implications for wider family
  - The patient's brother and sister may also be carriers of the balanced translocation- may have testing too.

Case study

- 33 year old woman coming in to discuss carrier testing for Cystic Fibrosis (CF)
- Niece was diagnosed with CF shortly after she was born.
- Brother found to be a carrier.

In Clinic



Female

Male

Prematurely ended pregnancy

Carrier of CF

Affected with CF

In Clinic

Explain what CF:

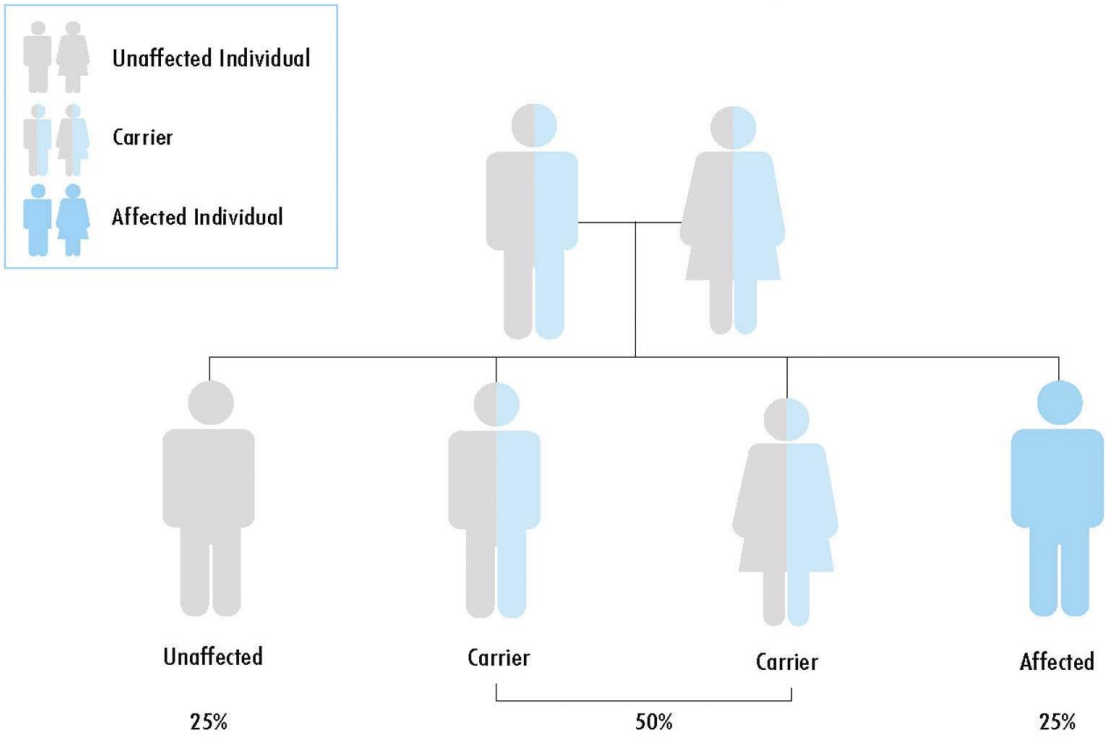
- Causes thick, sticky mucus which can affect the lungs, reproductive tract and digestive system.
- Currently, no cure but it can be well treated and managed.

Explain the genetics of CF:

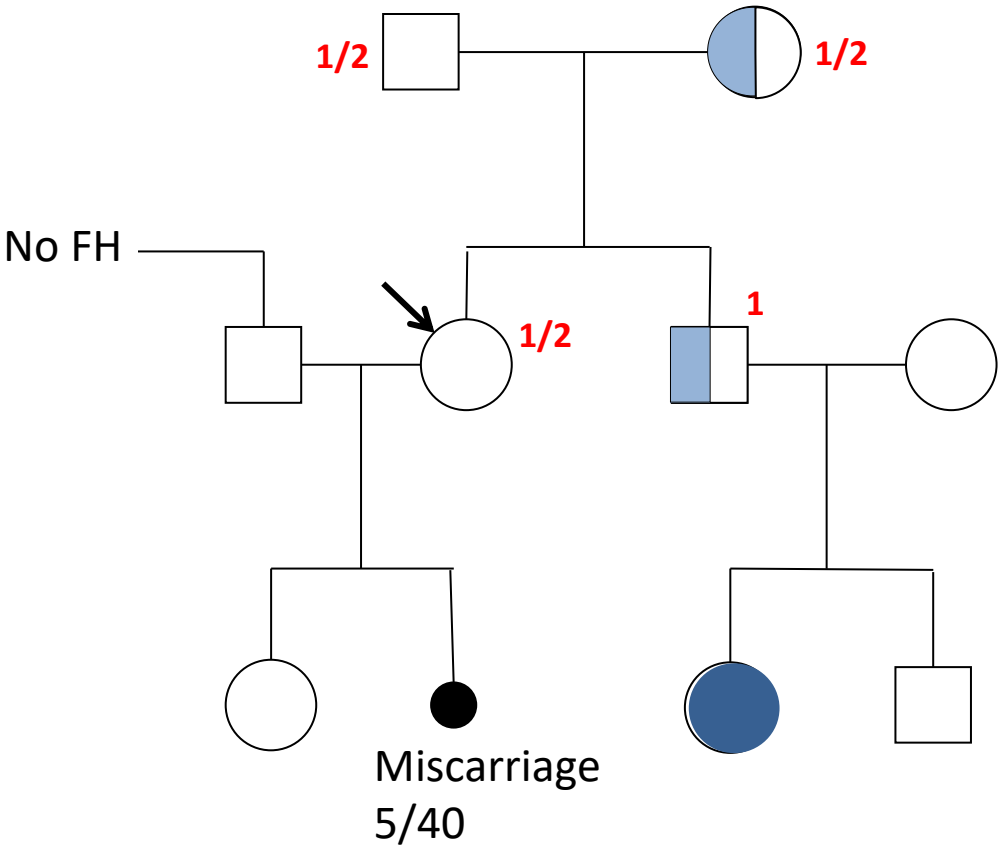
- Cells → DNA → Genes
- CF is caused by small spelling mistakes/ alterations in the gene called CFTR

In clinic

Explain how CF is inherited:



In Clinic



Female

Male

Prematurely ended pregnancy

Carrier of CF

Affected with CF

Clinical GENETICS Service

University Hospitals Bristol NHS
NHS Foundation Trust

In Clinic

If patient wants testing:

- Explain what results they might get
- Explain what testing involves: Blood test
- Take consent and bloods
- Arrange result appointment

NHS South West Genomic Laboratory Hub

Case Study: Referral

North Bristol NHS Trust

NHS North Bristol NHS Trust

Genetics Request Form

MANDATORY FIELDS are indicated in blue type
See reverse for additional information on sample requirements
Please fill out as completely as possible

BGL is a UKAS accredited medical laboratory No.9307

Tubes/volumes

BGL FM296 V8
Active 09/04/18

For BGL use only: DO NOT affix labels here

Please affix patient label here

SURNAME: STUDY

FIRST NAME: Case

ADDRESS

POST CODE

DOB

SEX: F

HOSPITAL NUMBER

NHS NUMBER

REFERRING HOSPITAL

CG number

CONSULTANT

Address for report (full address please if GP surgery): Clinical Genetics

Purchase Order no:

BILLING ADDRESS (if different to report address)

Date/time taken and by whom

Mother's name/DOB (IMPORTANT for all infant/fetal samples)

Date/time received

Previous genetic investigations in family: Yes/ No
If yes, please give brief details below (include laboratory numbers)

Mother (name & DOB).....

Father (name & DOB).....

Sibling/s (name/s & DOB).....

For LEUKAEMIC samples please indicate:

- Diagnostic ☐
- Follow-up (Remission) ☐
- Follow-up (Relapse) ☐
- Bone marrow transplant, specify sex of donor ☐
- MRD ☐

IMPORTANT for all fetal samples

Gestation..... Parity.....

Gravida..... LMP.....

EDD.....

Multiple pregnancy ? NT≥3.5mm ☐

Date and time of next appointment

Sample type

Private ☐

NHS ☐

Priority

Urgent ☐

Routine ☐

Specific molecular genetic tests
Consent required for DNA extraction and storage
EDTA TUBE

Specific cytogenetic tests
LITHIUM HEPARIN TUBE

DANGER OF INFECTION? YES / NO
(NBT The lab works to containment level 2+)

Risk of blood borne pathogen? ☐

Recent blood transfusion? ☐

Recent cytotoxic drugs? ☐

Please give additional details:

CLINICAL SUMMARY/ADDITIONAL INFORMATION/SPECIAL REQUESTS (if several tests are required please indicate order of testing required)

Brother reported to be a carrier of c.1521_1523del p.(Phe508del) *CFTR* mutation; for *CFTR* mutation analysis

BGL acceptance of a testing request acts as an agreement with the requestor

- DNA extraction and storage ☐
- DNA Export (Please attach letter) ☐
- DNA testing (please specify test required) ☐
- Array CGH testing ☐
- Karyotyping ☐
- QF-PCR: aneuploidy ☐
- QF-PCR: gender ☐
- Mosaicism ☐
- FISH (please specify) ☐
- Breakage studies: Fanconi anaemia/ Ataxia Telangiectasia/Other ☐
- Fixed cell storage for 2 years (blood only) (stored routinely for 4 months) ☐
- Fixed cell storage (oncology only) ☐
- Cell freezing (solid tissues/prenatal samples) ☐

BGL use only

Lab N°(s)

CONSENT STATEMENT (please see overleaf): It is the referring clinician's responsibility to ensure that the patient/carer knows the purpose of the test and that the sample may be stored for future diagnostic testing. In signing this form the clinician has obtained consent for testing, storage and for the use of this sample and the information gathered from it to be shared with members of the donor's family through their health professionals (if appropriate). The patient should be advised that the sample may be used anonymously for quality assurance and training purposes. If the patient does not wish information to be shared please write this clearly in the clinical summary box. Certain disorders with particular counselling issues e.g. HD may require a specific consent form (see website for further details).

NAME: SIGNATURE: BLEEP No:

BRISTOL GENETICS LABORATORY, SOUTHMEAD HOSPITAL, BRISTOL, BS10 5NB Tel: 0117 414 6168/6167/6174 Fax: 0117 414 6464 General enquiries email (not results) : nbn-tr.geneticsenquiries@nhs.net

Case Study: Referral

Referral: Brother reported to be a carrier of c.1521_1523del p.(Phe508del) *CFTR* mutation; for *CFTR* mutation analysis.

Task	Responsibility
Authorisation of testing (review of referral, assign test) and batching of tests	Scientist Band 6/7
Phone calls, where required, regarding incomplete referral information	Scientist Band 6/7
Proceed cases on STARLIMS onto relevant worklist, according to Scientist's batching	GT 5/6 (Fragment PCR team)
Printing out worksheets, undertaking PCR/ABI analysis and processing of the results on GeneMarker	GT 5/6 (Fragment PCR team)
Results check 1	GT 5/6 (Fragment PCR team)
Results check 2 and results recording	Registered Scientist Band 6/7
Report writing	Scientist Band 6/7
Report authorisation	Senior Registered Scientist (Band 8)

NHS South West Genomic Laboratory Hub

North Bristol NHS NHS Trust

Case Study: Test Directory

Referral: Brother reported to be a carrier of c.1521_1523del p.(Phe508del) *CFTR* mutation; for *CFTR* mutation analysis.

R244 Carrier testing for known familial mutation(s)

Testing Criteria

Patient requiring carrier testing for specific disorder where the familial mutation(s) have already been identified in a relative

The range of specialties who will request this test will depend on the disorder in question

Referrals for testing will be triaged by the Genomic Laboratory; testing should be targeted at those where a genetic or genomic diagnosis will guide management for the proband or family.

Where in Pathway

As dictated by clinical situation

Requesting Specialties

- Clinical Genetics

Specialist Service Group

- Core

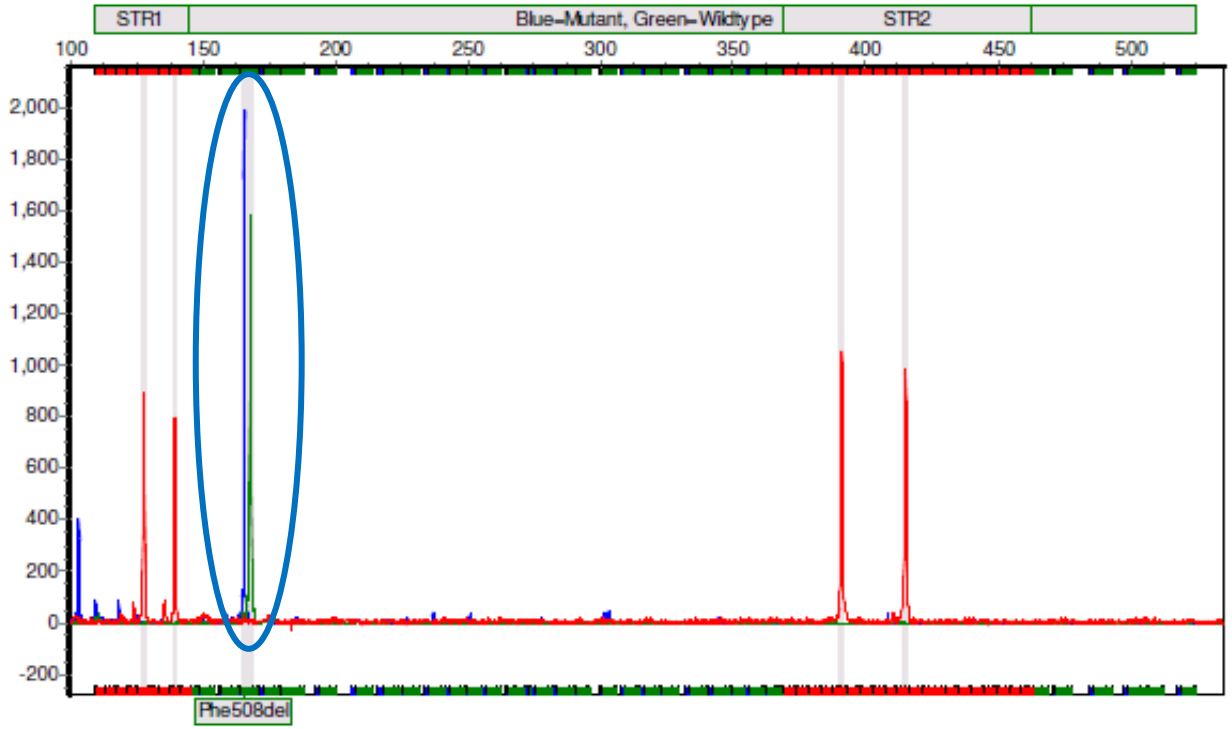
Associated Tests

Code	Name	Optional Family Structure	Scope(s)	Target Type	Target Name	Method
R244.1	Specific target Targeted mutation testing	Singleton	Small variants	Single interval	Specific Target	Targeted mutation testing

South West Genomic Laboratory Hub

North Bristol NHS Trust

Case Study: Analysis



Conclusion	F508del/Normal	
Comments:		
	Date	Initial
Authorization 1		
Authorization 2		

ARMS result: 2 peaks at the Phe508del locus – a variant and wild-type

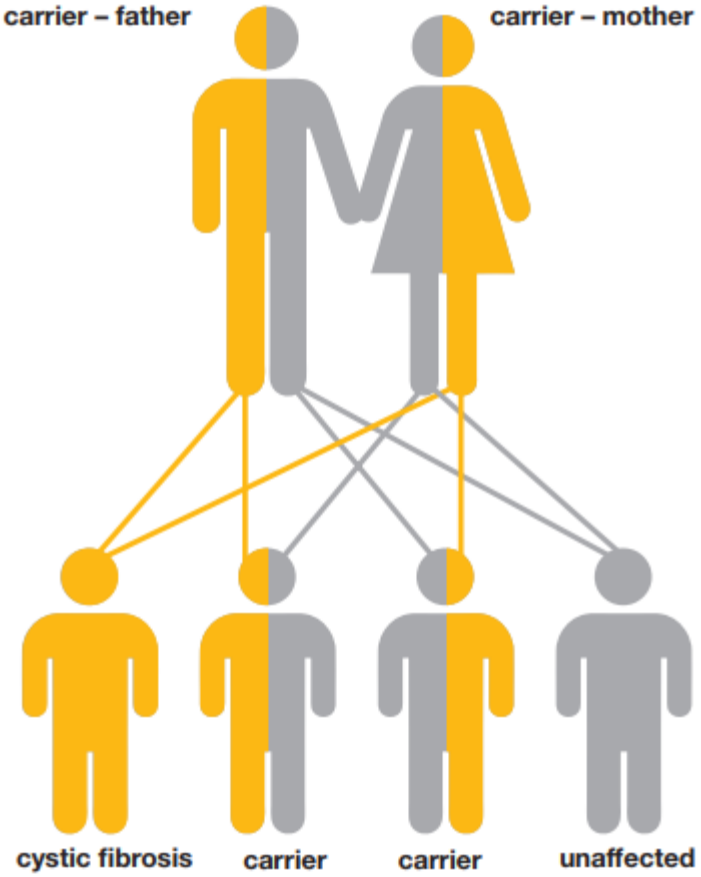
Analysis - Is the patient a carrier?

Interpretation - What does this mean for the patient? Can we offer testing to anyone else?

[Preliminary analysis can be performed by a GT. It is the scientist's responsibility to thoroughly check the analysis and write a report to reflect the correct interpretation.]

Case Study: Interpretation

- This patient is heterozygous for the p.(Phe508del) variant found in her brother
- She does not carry any of the remaining 49 variants in the Elucigene kit
- As this patient is of a reproductive age we can offer carrier testing to her partner if she is considering a pregnancy
- Any other adult relatives can be offered testing



NHS South West Genomic Laboratory Hub

Case Study: Report

North Bristol NHS Trust

Genetics Report

Full Name	**Case study**				
Date of Birth	**? Age 33**	Sample ID		Sample Type	**Whole blood**
Sex	**Female**	CG No		Received	
Postcode		External ID		Activated	
NHS Number		Hospital No		Authorised	

Clinical Summary: Brother reported to be a carrier of c.1521_1523del p.(Phe508del) *CFTR* mutation; for *CFTR* mutation analysis.

Results and Report

Test Performed	Result: Familial *CFTR* pathogenic variant PRESENT	
	cDNA	Protein
Elucigene CF-EU2v1 ARMS *CFTR* pathogenic variant screen	c.[1521_1523del)];[=]	p.[(Phe508del];[=]

This patient is a carrier of (is heterozygous for) the c.1521_1523del p.(Phe508del) familial *CFTR* pathogenic variant. This patient does not carry any of the remaining 49 *CFTR* pathogenic variants listed below.

Any offspring of this patient would be at 50% risk at least of being a carrier of a *CFTR* pathogenic variant.

If appropriate, this patient's partner can be offered *CFTR* pathogenic variant analysis, following genetic counselling, to ascertain their carrier risk (general population 1/24) and hence the risk of this couple having a child affected with CF in each pregnancy.

Close adult relatives may now be tested for the above familial *CFTR* pathogenic variant, following genetic counselling.

1. Bombieri C, *et al.* Recommendations for the classification of diseases as *CFTR*-related disorders. J of Cystic Fibrosis 2011; (10) : Suppl 2: S86-S102.
2. Yu, J *et al. CFTR* mutations in men with CBAVD: a systemic review and meta-analysis. Human Reproduction, Vol27, No 1 pp.25-35, 2012.

This patient was tested using the Elucigene CF-EU2v1 ARMS variant screen which tests for the presence of the following *CFTR* pathogenic variants: p.Phe508del, p.Gly551Asp, p.Gly542*, c.489+1G>T, p.Arg553*, c.1585-1G>A, p.Trp1282*, p.Asn1303Lys, c.3717+10kbC>T, p.Arg117His, p.Arg334Trp, p.Ala455Glu, c.3528delC, c.948delT, p.Ile507del, p.Arg347Pro, p.Ser1251Asn, p.Glu60*, c.2988+1G>A, c.1766+1G>A, c.579+1G>T, p.Gly85Glu, c.2052delA, p.Arg560Thr, p.Asp1152His, p.Pro67Leu, c.54-5940_273+1025del21kb, c.1679+1.6kbA>G, c.262_263delTT, c.313delA, p.Leu671*, p.Arg117Cys, c.2215delG, p.Tyr122*, p.Trp846*, c.2657+5G>A, p.Gln890*, p.Leu206Trp, c. 3140-26A>G, p.Arg1066Cys, p.Tyr1092*, p.Arg347His, p.Met1101Lys, p.Arg1158*, p.Arg1162*, p.Tyr515*, p.Val520Phe, c.3773dupT, p.Ser549Arg, p.Ser549Asn. intron 8 polyT and TG tracts
Notes: Any remaining DNA is currently retained in long-term storage and may be used anonymously for quality control of this assay. *CFTR* variant Nomenclature according to HGVS guidelines (www.hgvs.org). *CFTR* reference sequence Gen Bank Ref NM_000492.3 Nucleotide 1 corresponds to the first nucleotide of the first codon ATG. The CF-EU2v1 ARMS determines zygosity for all above variants except p.Ser549Arg.

Results

- Usually done via telephone call or face-to-face (pre-COVID)
- Discuss result and what it means for patient

1 x 1/25 x ¼ = 1/100
Patients risk x Partners risk x chance will pass it on

- Psychosocial aspects
- Implications for family members
- What next?
- Sometimes refer on to other services and/or continue follow-up.

STP in Cancer Genomics

Elle Mortensson (Yr 2)
STP Cancer Genomics
Bristol Genetics Laboratory

Cancer Genomics Manchester University teaching

Teaching takes the form of online learning, conventional lectures and group work.

University modules:

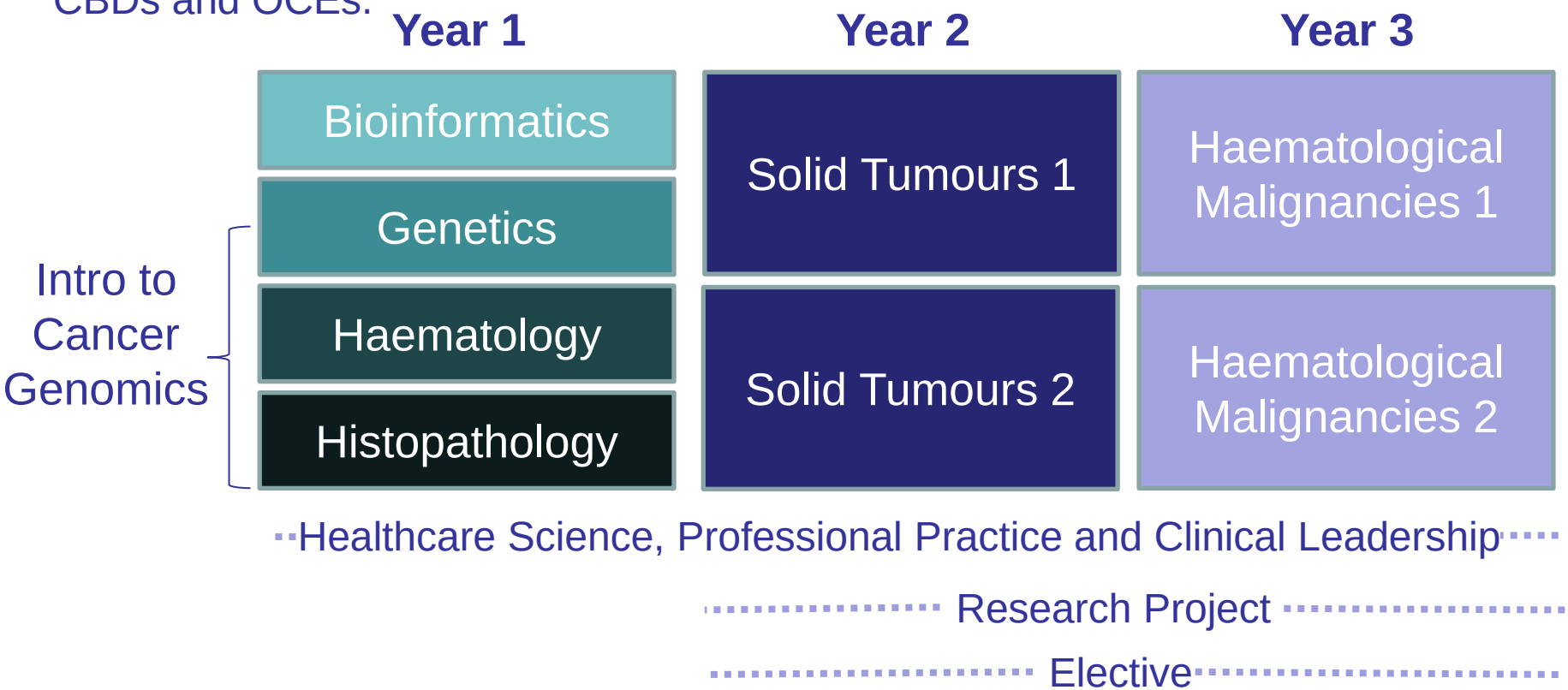
Year 1	Year 2	Year 3
Professional Practice & Intro to Healthcare Science and Clinical Leadership (Sept/Oct)	Haematological Malignancies 1 (Feb)	Haematological Malignancies 2 (Feb)
Intro to Bioinformatics (Sept/Oct)		
Intro to Human Genetics (Sept/Oct)	Solid Tumours 1 (May)	Solid Tumours 2 (May)
Intro to Cancer Genomics (March)		

Assessment can be group presentation, essays or exams.

Cancer Genomics work based learning

NHS South West Genomic Laboratory Hub

Work based learning means: shadowing technologists and scientists, learning their work-flows and methodologies, experiencing how they manage quality assurance – then writing these up as competencies and completing DOPs, CBDs and OCEs.



Cancer Genomics typical working day

NHS South West Genomic Laboratory Hub

Friday 10th Jan 2020 (Yr1):

Recently started my new rotation in Histopathology

- Shadowed medical laboratory assistants (MLA) in specimen reception as they accepted different sample types, determined which required additional fixation and sent samples to the appropriate pathologists.
- Shadowed pathologists as they carried out macrodissection of specimens including a right hemicolectomy specimen (caecum, ascending colon and a portion of the transverse colon), hysterectomy specimen (uterus, cervix, fallopian tubes and ovaries) and prostatectomy (prostate).
- Wrote up what I had seen to contribute towards a competency in Introduction to Cancer Genomics work-based learning module.
- Spent some time revising for upcoming Professional Practice, Introduction to Healthcare and Clinical Leadership exam

Cancer Genomics typical working day

NHS South West Genomic Laboratory Hub

Friday 8th Jan 2021 (Yr 2):

I am undertaking work and training in the Solid tumour team and have recently started my new rotation in haematology

- Replied to STP applicant emails.
- Answered queries sent to the Solid Tumour email inbox regarding test results and expected incoming samples.
- Reviewed incoming samples and assigned these to the appropriate testing workflows.
- Analysed results from NGS DNA and RNA fusion panels.
- Wrote reports detailing the results of NGS DNA and RNA panel testing.

Bristol Genetics Laboratory

NHS North Bristol NHS Trust

National School of Healthcare Science

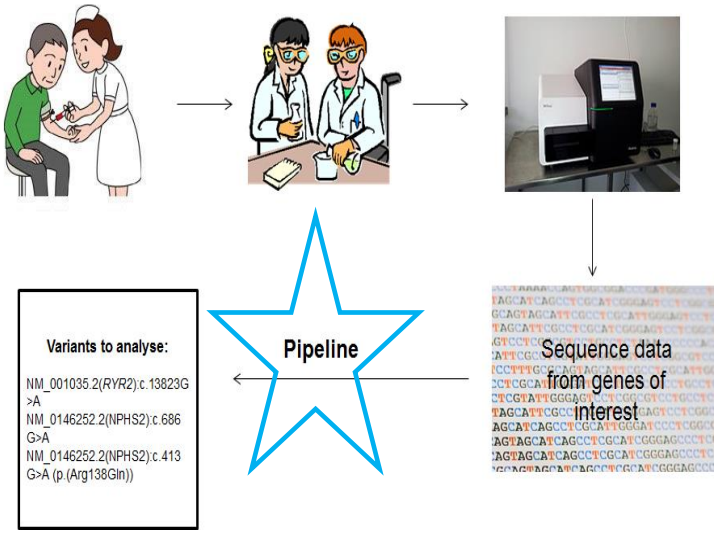
CLINICAL BIOINFORMATICS (GENOMICS)

NORTH BRISTOL NHS TRUST

STP OPEN DAY 2021

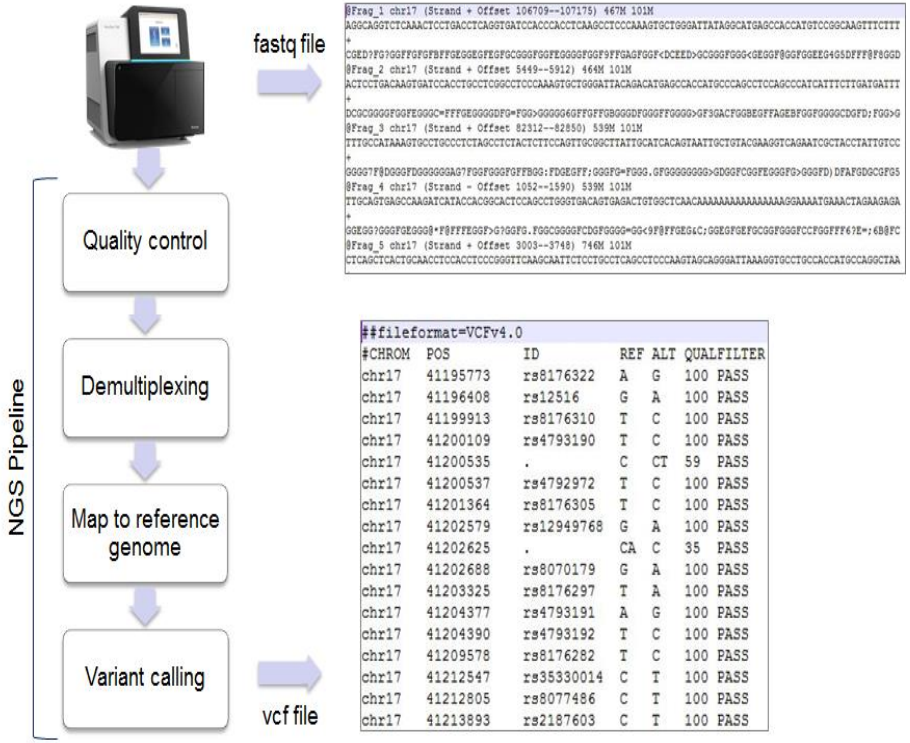
CLINICAL BIOINFORMATICS AND GENOMICS

- Area of healthcare science responsible for developing and improving methods for acquiring, storing, organising and analysing biological (genomic) data that supports the delivery of patient care.
  - Computing, biology and medicine.
  - Mathematics, statistics, coding and genomics
  - Interdisciplinary work and communication skills
- Develop NGS data analysis pipelines, fix bugs in code, create and manage databases, data storage, new advances, information governance.



BIOINFORMATICS TRAINING AT BGL

- Bioinformatics team: 3 qualified bioinformaticians and 3 trainees
- Two pipelines: constitutional and somatic.
- We are trained in running each pipeline by first observing and later running the pipeline under supervision several times
  - Competency records are kept for each pipeline we observe/run
  - Over time competency is built up
- Bioinformatics mailbox queries: assisting helps with our training.
  - Is this variant real?
    - Visualising NGS data, examining quality metrics.
  - Can you please make these files available to me?
    - Searching the drives for files using bash commands.
  - Can you extract the quality metrics from this subset of NGS runs?
    - Writing file parsers to extract the data you need from files.



BIOINFORMATICS TRAINING AT BGL

Portfolio

- Competency: generate a Binary Alignment Map (BAM) file.
- CBD: Implementation of a relational database for quality monitoring according to IG principles.
- OCE: Critically analyse data from a clinical audit, summarising key findings and indicating areas for future development.
- DOP: Create an SQL statement for extracting a defined subset of clinical data.

MSc Clinical Bioinform

MANCHESTER 1824
The University of Manchester

- Healthcare Science and Professional Practice
- Clinical Bioinformatics
- Health Informatics
- Applying ICT in the Clinical Environment
- Computing for Bio and Physical Science
- Programming
- Applied Next Generation Sequencing
- Research Methods
- Advanced Clinical Bioinformatics
- Whole Systems Molecular Medicine
- IT for Advanced Bioinformatic Applications

BACKGROUNDS OF BIOINFORMATICS STP'S

BSc Biomedical Science
- Haematology BMS

BSc Genetics & MSc in Genomics Medicine

BSc Medical genetics

BSc Biomedical Science
- Genomics Lab Tech

MNeuro Neuroscience

GEL Bioinformatician

BSc Biological Sciences

BSc Biological Science, MSc Bioinformatics & Systems Biology
- Healthcare Charity work
- Data Analysis Consulting

BSc Biology
- Lab technician
- Medical laboratory Assistant (NHS Blood & Transplant)

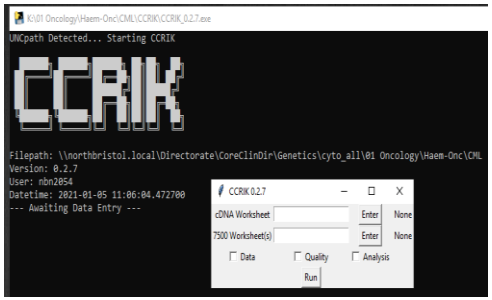
A DAY IN THE LIFE – TOM (1ST YEAR)

- As I came from a pure genetics background, most of time currently involves learning the essential programming skills required of a bioinformatician.
  - Most of this is taught in the MSc, but I've picked up a lot from my colleagues.
- Weekly team meetings where I hear about the work done by others in team.
- Observing runs of various binfx pipelines and soon will begin training to perform them myself
- Completing competencies for various STP modules.
- Completing assessments for the MSc

A DAY IN THE LIFE – CLARE (2ND YEAR)

- Biology background – BSc Biomedical science, PhD neuroepigenetics
  - Sequencing project: collaboration with bioinformaticians.
- Routine training
  - Service work: running pipelines and quality checks,
  - Bioinformatics mailbox queries
- Development work and projects
  - Quality monitoring database, somatic pipeline and CNV calling: DJANGO, SQL, python and R
- MSc work
  - Research project: developing a tool to detect microsatellite instability in NGS data from colorectal and endometrial cancer samples
- Quality improvement
  - Clinical and Internal Audits

A DAY IN THE LIFE – ANDY (3RD YEAR)



- Background
  - BSc Biomedical Science
  - 2 years as a Biomedical Scientist (Haematology)
- Service Work
  - Training and running pipelines
  - Helping with Bioinformatics Queries from Clinical Scientists
  - Undertaking audits for service improvement
  - Fixing bugs and working on updates for Bioinformatics tools
- STP Portfolio competencies and assessments
  - Primarily the specialist modules (Programming, Advanced Clin Bio, Applied NGS, Whole Systems Molecular Medicine)
    - Combination of projects and service work
      - CCRIK, aligner comparison (BWA-MEM/BWA-MEM2), MSc project
- MSc
  - Whole Systems Molecular Medicine, IT for Advanced Bioinformatics Applications
- Research project
  - Development of a tool to improve turnaround times for the clinical investigation and reporting of CNVs detected through microarray CGH analysis

USEFUL SKILLS

- DJANGO
  - https://tutorial.djangogirls.org/en/
- Python, R, SQL
  - Code academy, Data Quest, Coursera, Datacamp, Rosalind Bioinformatics,
- Linux
  - https://linuxjourney.com/
- Future learn: Clinical Bioinformatics: Unlocking Genomics in Healthcare

Breakout sessions

Bristol Genetics Laboratory

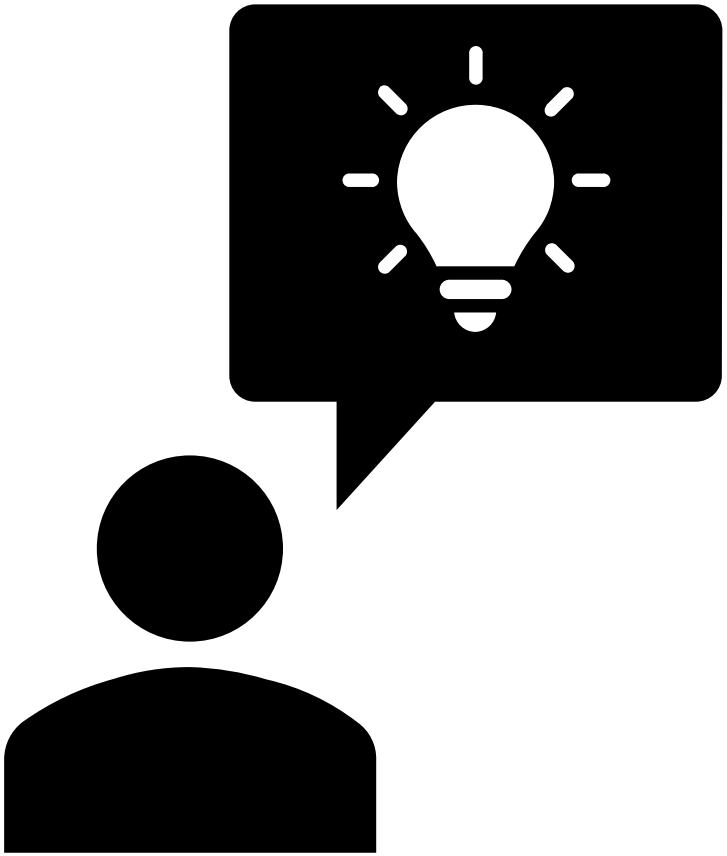
- 30 minutes
- 4 groups – allocated

Breakout 1: Genomics Q&A	Catherine, Emily, and Rosie
Breakout 2: Cancer Genomics Q&A	Elle, and Jordi
Breakout 3: Bioinformatics Q&A	Matt, Clare & Tom
Breakout 4: Genomic Counselling Q&A	Poppy, Emma, and Jake

- Regroup for sum – automatically
- Scribes in each group so FAQ tool will be made available to all post session

Points to feedback

Bristol Genetics Laboratory



Genomics Education Programme

NHS South West NHS Genomic Laboratory Hub

Genomic Education opportunities for you

Health Education England's Genomic Education Programme

https://www.genomicseducation.hee.nhs.uk

@genomicsedu

South West Genomic Laboratory Hub

Follow social media for information on the genomic medicine services and signposting to regional opportunities for education and training

Contact Mel Watson, E&T Lead at South West Genomic Laboratory Hub
Melanie.watson@nbt.nhs.uk
@mellyjwatson

https://www.nbt.nhs.uk/south-west-genomic-laboratory-hub

@swglh

Close

A huge **thank you** to the Bristol teams for their time and expertise

Thank you to all of you for engaging with us

Presentations and FAQ from breakouts will be available as soon as we can

Please can you complete **SurveyMonkey questionnaire** which will be emailed to you
https://www.surveymonkey.co.uk/r/Bristol_STP_OpenEvent_2021

We wish you all the very best in your endeavours and hope to see some of you as trainees later this year