

EMPOWERING PEOPLE

GENETIC COUNSELLING IN FOCUS

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This exhibition was developed in partnership between the Public Engagement and Society and Ethics Research teams at the Wellcome Genome Campus. These teams are part of Connecting Science; enabling everyone to explore genomic science and its impact on research, health and society.

The exhibition would not have been possible without the support and involvement of the following people:

Anna Middleton, Christine Patch, Lauren Robarts, Jonathan Roberts, Gemma Chandratillake, Helen Jolley, Georgina Hall, Sara Levene, Amy Goldman, Laura Olivares Boldú, Mark Danson, Kenneth Skeldon, Becky Gilmore, Emily Boldy, Chrystal Ding, Paul Fenn Films, Association of Genetic Nurses and Counsellors, Stylographics, Venue AV.

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www.wgc.org.uk/engage

PREFACE

The Wellcome Genome Campus is home to cutting-edge scientific research but also to social science researchers leading the way in examining the ethical, legal and social implications of genomics. With a subject as broad and fast-moving as genomics, why choose to stage an exhibition about genetic counselling? The increase of genetic testing means that all of us are closer to potentially discovering things about ourselves - whether we want to know them or not. Genetic counselling is vital in empowering people to make decisions about genetic information that are right for them. Everyone in society should have the opportunity to consider what these tests could mean, and be able to contribute their voice to the discussion. This, in addition to the growing evidence base of genetic counselling research at the Wellcome Genome Campus, meant that an exhibition wasn't just timely but necessary.

The exhibition uses work from the Society and Ethics Research team and contains the voices of eight genetic counsellors sharing what it is that they do and how this supports people to make choices that are right for them and their unique circumstances. To explore the work of these counsellors in further detail we commissioned documentary photographer Chrystal Ding to capture them in their working environment. Most importantly we invite you to consider what this subject means to you. What are your views?

Becky Gilmore

Exhibitions & Interpretation Coordinator
Wellcome Genome Campus

INTRODUCTION

What is genetic counselling?

Genetic counsellors work with people to understand their family history in relation to a particular genetic condition, a bit like carrying out a genetic risk assessment. The tool that they use is a family tree, also known as a 'pedigree'.

Our genome is our entire set of genetic instructions, encoded in DNA. We have a copy of our genome in most cells in our body. The DNA in our genome is packaged up into pairs of chromosomes, which we inherit from our biological parents. Because this genetic information is passed on through generations of a family, a pedigree is a useful family history tool that enables a genetic counsellor to understand patterns of inheritance and risk factors.

ANNA MIDDLETON

Walking in someone else's shoes

Professor Anna Middleton is Head of Society and Ethics Research at the Wellcome Genome Campus. This research group are social science academics who explore the ethical, legal and social issues raised by genomics. Anna is a Registered Genetic Counsellor and Chair of the Association of Genetic Nurses and Counsellors (AGNC), the professional body that represents those working in the discipline of genetic counselling and services in the UK. She is also an affiliated staff member at the Faculty of Education, University of Cambridge.

In this short film she explores the importance of genetic counselling and how research is providing an evidence base for improving counselling approaches and informing service provision.

Link to the film: bit.ly/empoweringpeople-anna

Portrait and environment

Chrystal Ding, 2019

Here Anna is depicted in her office at the Wellcome Genome Campus. As an active researcher, and head of a research team, Anna's work often finds her at her desk analysing information, writing up findings and communicating across a wide network of health professionals, policy makers and academics.





SOULS 2019

Richard Seminar
- ON AI
- OR DEC

Media Launch
YDYS - Feb

Music of Life
- Recruitment
- Analysis
- write up

ABC vs St George's
- Survey
- Recruitment
- write up

Socialising the Genome
- August
- Blog, Seminars/paper

YDYS
Finish reviews - collaborators all meet

Film for World Congress
- ask participants to comment on GGI ideas e.g. exploring use of CRISPRs, need to measure outcomes

"Bench to Bedside + Beyond"

↓
Singer ↔ see ↔ DE

1. Post DOC fte
2. Team Manager (Laver) fte
3. Full time Admin support

Steve Palmer
Send meta-analysis to for hrs excel
Select routes outcome

Jojo FutureLean Cause Grant
x 2 papers - from April onwards

Post Legx Cause
- adjustment
- GGI claim
- singing ball

UK paper → Jon Jerome + data support

REFERRAL

Who sees a genetic counsellor, and how are they referred?

In the UK genetic counselling is a service provided within the NHS for people who have, or have a potentially increased risk of, an inherited genetic condition. These could be, for example, cancers or conditions with known genetic causes such as cystic fibrosis or Huntington's disease. A patient is referred to a genetic counselling service by other healthcare professionals, such as a GP.

People may see a genetic counsellor for many reasons: to explore their risk of having or developing a condition, to make sense of the results of a genetic test, or to discuss the possibility of taking a test, and to understand the possibility of passing on a genetic alteration to their children.

AMY GOLDMAN

The subtleties and nuances

Amy Goldman is a Registered Genetic Counsellor working at Guy's and St Thomas's NHS Trust in London. During her time as a trainee she was elected to the committee of the AGNC.

In this film Amy discusses the skills needed in genetic counselling and how a counsellor utilises these skills as they move between different types of cases.

Link to the film: bit.ly/empoweringpeople-amy

Portrait and environment

Chrystal Ding, 2019

Amy is photographed in the Genetics Clinic at Guy's Hospital in London. Hospital consultation rooms are used for both genetic counselling appointments and for examination. Amy prepares the room for her appointments by arranging the chairs to create a space for conversation. If someone is attending with a partner, she finds that the armrests meeting is a subtle cue for them to hold hands. A box of tissues sits on the nearby desk; out of prominent view but in arm's reach if needed.





SARA LEVENE

Working through the pros and cons of difficult decisions

Sara Levene is Consultant Genetic Counsellor at The Centre for Reproductive & Genetic Health in London and is a committee member of the AGNC. Sara specialises in pre-implantation genetic diagnosis (PGD), which she discusses in this short film.

PGD is a method to check embryos for serious inherited genetic conditions. Only embryos that are free of the condition are implanted into the woman's womb. PGD can only be used in the UK to test for conditions that have been approved by the Human Fertilisation & Embryology Authority.

Link to the film: bit.ly/empoweringpeople-sara

Portrait and environment

Chrystal Ding, 2019

Chrystal captured this portrait of Sara in her clinic at The Centre for Reproductive & Genetic Health in London. An accompanying photograph shows Sara drawing a pedigree (a shorthand chart, also known as a family tree). These charts are an important tool for understanding inheritance in a family.





JONATHAN ROBERTS

Fostering resilience

Dr Jonathan Roberts is a Pre-Registration Genetic Counsellor at Addenbrooke's Hospital in Cambridge. He is also a Postdoctoral Researcher in the Society and Ethics Research team at the Wellcome Genome Campus. Jonathan's research explores how genomics can be made more accessible, focusing on family storytelling and genetics in popular culture.

He discusses the psychological dimensions of genetic counselling, the importance of empathy and the experience from both the counsellor and the patient perspective. In preparing for a new case, Jonathan sees the importance in taking the time to understand someone else's story and their motivations.

Link to the film: bit.ly/empoweringpeople-jonathan

Portrait

Chrystal Ding, 2019

Jonathan's portrait was taken at the Wellcome Genome Campus. His experience of working in the genetic counselling clinic has helped shape the research that he does. He investigates ways to make genomics more understandable and accessible to all. By exploring genetics in popular culture and the stories we tell ourselves as families and societies, Jonathan finds evidence for playful, creative tools and language choices that are sensitive to people's experiences but can be used to improve access.



Alastair Smith's case

Alastair has been referred for genetic counselling by a consultant neurologist. This is a senior medical doctor who specialises in the nervous system. He has been referred because his father has been diagnosed with Huntington's disease and Alastair wishes to consider whether to undergo a predictive genetic test to find out if he is also affected. Huntington's disease is an inherited condition that causes degeneration in the brain's nerve cells. This has wide-ranging and debilitating effects on a person's movement and cognitive abilities. Symptoms usually appear in a person's 30s or 40s, and there is currently no cure. Because the symptoms usually appear later in life, many people discover that they are affected after having their children, meaning that there is a 50% chance that each child will have inherited the condition.

After discussing his options with a genetic counsellor Alastair decided to take a predictive genetic test to find out if he has the altered Huntington's disease gene; if he does, this will confirm that he will definitely develop the condition. Unfortunately his test result was positive, meaning he has the altered gene. Alastair came back to the genetic counselling clinic to discuss the implications of his result, and alongside his partner explored options for starting a family that could avoid passing his inherited condition to future children. Whilst the genetic test result was not the outcome that he wanted, Alastair noted that the process enabled him to be empowered to make decisions for him and for his future family.

EXPLORING FAMILY HISTORY

Family tree

Genetic counsellors work with people to understand their family history in relation to a particular genetic condition, a bit like carrying out a genetic risk assessment. The tool that they use is a family tree, also known as a 'pedigree'.

Our genome is our entire set of genetic instructions, encoded in DNA. We have a copy of our genome in most cells in our body. The DNA in our genome is packaged up into pairs of chromosomes, which we inherit from our biological parents. Because this genetic information is passed on through generations of a family, a pedigree is a useful family history tool that enables a genetic counsellor to understand patterns of inheritance and risk factors.

How is a family tree drawn?

Although they share similar details, pedigrees are different to family trees that you might compile when tracing your family's genealogy. For your genealogical story you might be interested in relatives going back many generations, or identifying distant cousins, but a genetic counsellor usually takes a three-generation family tree and uses this to work out whether there are clear disease inheritance patterns showing in the family.

In the clinic the family members of interest are the ones you are most closely related to, so most family trees only go back as far as your grandparents and will capture details of your parents, your siblings, your children and your parents' siblings. This is because you are more genetically similar to these individuals, meaning that an alteration that is present in your DNA may be present in theirs, and vice versa.

	male	female	sex unknown	
Individual				Marriage/partnership
Affected individual (symbol coloured in)				Divorce/separation
Multiple individuals				Where the partners are blood relatives (consanguineous relationship)
Deceased				Children/siblings
Pregnancy				Identical twins (monozygotic)
Miscarriage				Non-identical twins (dizygotic)
Person providing pedigree information				

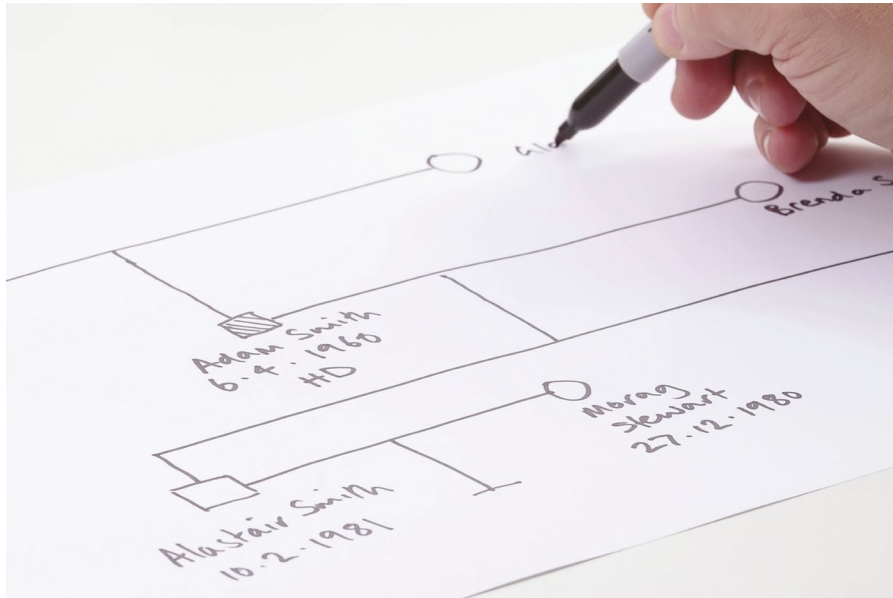
Standardised pedigree symbols and nomenclature

In the 1990s a standardised set of symbols was developed and agreed for use in drawing pedigrees. The purpose was to create a common language that would make the details of a diagram recognisable and intelligible to anyone accessing the information. Whether drawing by hand or using computer software, the same symbols are used. They show details such as biological sex, whether someone is affected by a condition (and their age of diagnosis), and if someone is alive or not. They can also include information about social relationships (e.g. divorce or adoption) as well as gender identification.

Details unknown

Exploring and talking about families can raise many emotions and feelings for people, sometimes unexpectedly. For some people this is particularly challenging if there are circumstances that mean parts, or entire, family histories are unknown or if there has been emotional conflict or distress in a family.

In cases like these, genetic counsellors will work with an individual to understand and assess what is known and accessible and then discuss and work through their genetic testing and disease screening options based on their circumstances.



Sketching a family tree

There isn't a right or wrong type of family tree, there is just the one that someone has. Here a genetic counsellor sketches out the family trees of our case studies. Although the same style and symbols are used each time, the final tree is different for each person, reflecting our own unique stories and circumstances.

Link to the film: bit.ly/empoweringpeople-tree

HELEN JOLLEY

Treating entire families as the patient unit

Helen Jolley is a Registered Genetic Counsellor working at the Manchester Centre for Genomic Medicine.

Here, Helen discusses how in the genetic counselling clinic, a counsellor is not just considering the person in front of them but the possible impact on their wider family, and working with the patient to manage the implications of this.

Link to the film: bit.ly/empoweringpeople-helen



Portraits

Chrystal Ding, 2019

Helen's portrait was taken in the corridors of the Manchester Centre for Genomic Medicine. Genetic counsellors like Helen work with patients to discuss options for genetic testing but also to discuss results of tests; many of which may have complex factors to consider for the patient but also for communicating with their wider family. Helen uses her skills and training to work with her patients to find the most empowering path for them.



Hamid Ahmed and Yasmin Marshall's case

Yasmin and Hamid experienced the loss of several pregnancies and wanted to understand their circumstances and options using the expertise of a genetic counsellor to avoid repeating this experience. After their last pregnancy loss a chromosomal translocation was identified in Hamid. This means that there is an unusual arrangement of a person's chromosomes, the packages of DNA that we inherit from our parents. They were referred to their hospital's genetic counselling service by their GP.

Genetic counsellors work with parents and prospective parents who are concerned about passing on genetic conditions to existing or future children. Pre-implantation genetic diagnosis is a method to check embryos for serious inherited genetic conditions and Hamid and Yasmin wanted to have this test. Only embryos that are free of the condition are implanted into the woman's womb. Having this test would mean that the couple would need to conceive via IVF even though they are physically able to conceive naturally.

Hamid's chromosomal abnormality was a risk factor for future pregnancies and so the couple decided that, despite some of their misgivings about having IVF treatment, undergoing pre-implantation genetic diagnosis was the right option for them.

NEXT STEPS

What are my choices?

The right next steps at the end of an initial genetic counselling appointment will depend on the details of the particular case and the decision-making of the individual. This could be to take a genetic test, to make the next steps regarding a condition in another part of the healthcare system, or to talk about the outcomes with family members. A genetic counsellor will offer guidance about the process but the decision-making rests with the individual.

A key part of a genetic counsellor's skillset is to support a person through the complex language, steps and choices so that they feel equipped with knowledge and empowered to make a decision that is right for them at that time.

Should I tell my family?

Making a decision about a genetic diagnosis, whether actual or possible, raises many considerations for an individual. The process of working through a family history brings into focus our genetic connectedness; how something in our DNA that could impact us can also have implications for other people that we are related to.

Genetic counsellors provide guidance to enable people to make sense of these implications and find ways to approach having potentially difficult conversations with loved ones and relations. It can be a challenge to find the right language and to share advice, for example about the possibility of having testing of their own. Genetic counsellors can offer valuable support, which they are trained to deliver.



GEORGINA HALL

Ripple in the pond effect

Georgina Hall is a Consultant Genetic Counsellor at the Manchester Centre for Genomic Medicine and chairs the Joint Committee on Genetic Counsellor Regulation.

In this film Georgina examines how a genetic diagnosis is more than simply having a test. It carries with it many emotions and further decision-making. The outcome of a diagnosis can have consequences for other family members, which a genetic counsellor will discuss and support a patient to process.

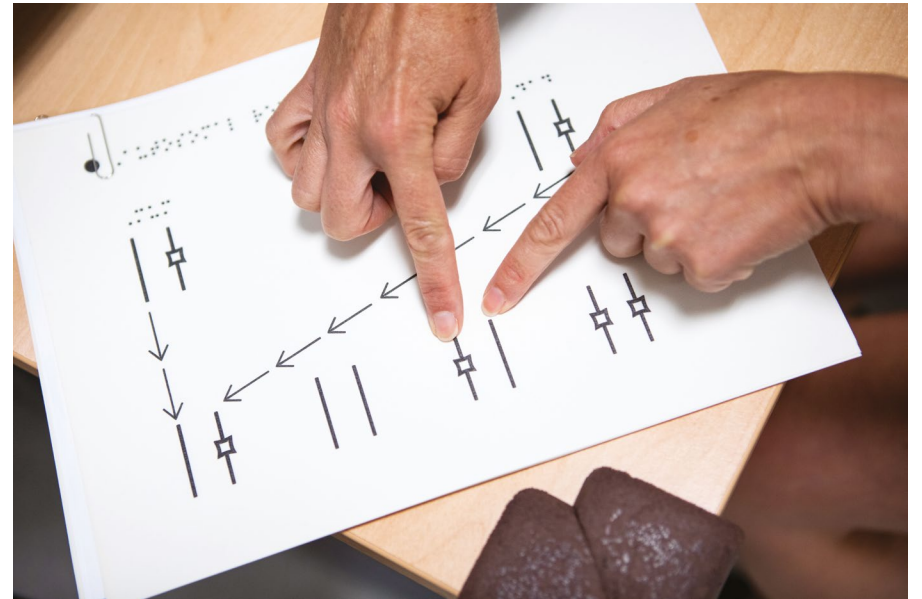
Link to the film: bit.ly/empoweringpeople-georgina

Portrait and environment

Chrystal Ding, 2019

This portrait of Georgina was captured at the Manchester Centre for Genomic Medicine. Georgina specialises in eye genetics and works with a range of patients and families with conditions that affect vision and may be inherited. She works with many patients who are partially sighted or blind. To enable complex and important information to be communicated she uses tools such as a pedigree, whose features showing inheritance patterns are raised and distinct from each other so that they can be felt through touch.





CHRISTINE PATCH

Helping patients cope with bad results

Professor Christine Patch is Principal Staff Scientist for Genomic Counselling at the Wellcome Genome Campus, in the Society and Ethics Research team. She is also the Clinical Lead for Genetic Counselling at Genomics England, the organisation leading the 100,000 Genomes Project.

Here, Christine shares how genetic counsellors support individuals to make decisions that are right for them, and to use their blend of skills to help people process an unexpected or unwanted result so that they feel empowered to take their next steps.

Link to the film: bit.ly/empoweringpeople-christine



Portrait

Chrystal Ding, 2019

Christine started her career as a nurse and has worked in genetics since the 1980s. She has been involved in direct patient care as well as leading, developing and managing clinical genetic services. She is passionate about combining the science of genomics with the art of working alongside patients and their families coming to terms with the impact of genomic information in their lives. With the rise of genomic testing, and the insights and uncertainties that this brings, Christine's work champions responsible uses of the technology in delivering effective patient and family-focused health services.

Integrating with the healthcare pathway

Genetic counsellors work alongside other healthcare professionals, from managing referrals to sharing results and planning next steps. Genetic counsellors are often part of large multidisciplinary healthcare teams involving medical doctors, clinical scientists, nurses, pathologists, and radiologists. As genetic testing becomes more and more routine, and spreads across whole healthcare settings, the importance of an integrated and upskilled healthcare system and practitioners becomes even more important.

Genomic (as opposed to genetic) testing involves casting a wide net by exploring many genes in one go. This can mean that unexpected additional results might emerge. It will be important for healthcare systems to provide the guidance and support that enables individuals to manage these uncertainties and make choices that are right for them.



Phlebotomy kit and spit sample tubes

To have a genetic test a person must supply their DNA. This can be done by a blood or saliva sample. It's most commonly done via blood as better quality DNA samples are usually obtained this way.

Phlebotomy is the process of drawing blood for testing. Some genetic counsellors are trained to take a blood sample from a patient in their clinic using kits such as this one. However, many hospitals will have special clinics where blood is taken. If a person cannot give a blood sample then a saliva sample can be collected in tubes like these.

GEMMA CHANDRATILLAKE

An important part of multidisciplinary teams

A trained genetic counsellor, Dr Gemma Chandratillake is Education & Training Lead for the East of England NHS Genomic Medicine Centre. She is also Course Director for the Institute of Continuing Education's Genomic Medicine programme.

In this film, Gemma explores how genetic counsellors work as part of multidisciplinary teams in the healthcare system and the growing need to train other healthcare professionals in genomics, enabling them to understand genetic results and their implications for a person's healthcare pathway.

Link to the film: bit.ly/empoweringpeople-gemma

Portrait

Chrystal Ding, 2019

As an educator and researcher Gemma spends much of her time working with other healthcare professionals to understand training needs and knowledge gaps, and then developing and delivering the tools and teaching required to meet them. As genetic and genomic testing become more widespread and integrated with other clinical areas, there is a growing importance to have a healthcare workforce that understands genomics and can consider the implications of having these tests.



David Kettering's case

David is a man in his 30s with stage four bowel cancer. This means the cancer is advanced and has spread from the bowel to other organs in his body and he knows his condition is terminal. After undergoing many treatments David has decided to discontinue these as he'd rather spend his remaining time with his family at home, instead of in hospital. He has been referred for genetic counselling by his senior specialist nurse in order to understand whether his cancer is an inherited type. This feels especially important to David as his mother died from a uterine cancer when she was 45. David is particularly concerned to find out whether his three young children are at risk of developing it, and if so, what bowel screening can they access as children.

David was dealing with the difficult realisation that he would not live long enough to see his children grow up. His mother's cancer diagnosis and early death was discussed with the genetic counsellor and was a strong factor in his decision to have a diagnostic genetic test. His test revealed an inheritable gene alteration. Whilst he was keen for his children to be tested, David worked with his genetic counsellor to find the right ways to communicate the options with his wife and children for when they felt it was the right time for them. The genetic counsellor also identified other family members who could also be at risk from developing bowel cancer.

REFLECTIONS



Genomic technology is sweeping across society. 2016 was declared “Generation Genome” by the UK’s Chief Medical Officer, Dame Sally Davies, who said in her opening statements of that year’s annual report: “Genomic medicine has the potential to save costs and improve quality of care by targeting treatment, maximising benefit and reducing side effects. For patients with rare diseases, it can shorten their ‘diagnostic odyssey’ helping to identify therapeutic options faster and improve outcomes.”

With the explosion of availability of genetic testing, both in clinical settings and via the consumer market through companies such as 23andme and Ancestry.com, genomics is officially now relevant to all of us. Even if you choose to never partake in genetic testing yourself, the moment a biological relative has, information you are linked to becomes available in a database somewhere. The transition into the genomic era means that the definition of what constitutes a “patient” will likely have to shift as well, from the individual to the biological family. Genetic counsellors are specially trained in how to make sense of a genetic test and how these tests impact a family. As a healthcare profession, whose practitioners have expertise both in clinical genetics and in counselling, genetic counsellors have a distinct point of view on the genomic healthcare revolution taking place in the UK and elsewhere. Research coming out of the Society and Ethics Research group at the Wellcome Genome Campus, from how to both counsel and illustrate transgender people on family trees in a culturally sensitively and inclusive manner, to whether an element of genetic counselling can be automated via chatbot technology in order to keep up with growing demand, to how doctors should balance duties of confidentiality with duties to warn, genetic counsellors will play a vital role in shaping the future of healthcare in the genomic era.

Professor Anna Middleton
Head of Society and Ethics Research
Wellcome Genome Campus



From 2017-2019 I spoke to 23 people for Genetopia, a book of photographs, interviews, and personal documents through which these individuals told the stories of their journeys with DNA testing - why they did it, what they found, and what they did with that information.

Of the 23 people I interviewed, only those who had been referred to a genetic counsellor by a doctor had ever heard of one. For others, any information raised by their DNA test results was theirs to Google, information that included newfound family relations and hereditary conditions. The individuals were often surprised to learn that there are people in the world trained to provide exactly the support and answers that they might have been looking for. More than a few wished they had known earlier.

In my documentary photography practice I often focus on issues of trauma and identity. In 2019 I have been working on a project funded by the Rebecca Vassie Memorial Trust, which looks at counselling in a very different context: that of genocide survivors in rural Rwanda. Though the two projects might seem worlds apart, one thing that is emerging as a common thread is how indelibly linked trauma, identity, and healing are. And genetics occupies the particularly complex space between the science of who we are and our lived experience of the world. How we absorb and take on information relating to our identity dictates our experience of it, and often sends us off in one direction or another.

Before I visited Rwanda I couldn't even imagine what counselling would look like there. Where would it take place? Who would be involved? What does 'counselling' mean and do in that situation? Similar questions came to mind when I thought about genetic counselling. Empowering People was a way of accessing those spaces and the people who work in them to get a better understanding of what it is they do, and why it is important.

At its core, genetic counselling - like any form of counselling - is a very human-centric experience, and it was in the details that this came through for me. In Guy's and St Thomas' Hospital, London, genetic counsellor Amy Goldman explained to me how she used to place the tissue box on the edge of the table to be easy to reach, but over time realised that it could lead patients to expect bad news where there wasn't any, and subsequently the box was placed further out of sight. In St Mary's Hospital, Manchester, there are special provisions for ophthalmic genetic counselling where patients might be visually impaired. In many of the clinic rooms chairs are arranged with arms touching, mimicking where their occupants hold hands. And where there are children, the bright shock of toys that translates any clinical setting into a room for family.

At the heart of any counselling relationship is the connection between the counsellor and the person or persons in their care. In many ways similar to photography, attention is key - attention to detail, attention to the individual, and to the space in which the relationship is forged. The photographs taken for this exhibition are an attempt to show some of these aspects of the care involved in the handling and navigating of our genetic information by genetic counsellors.

Chrystal Ding

Documentary Photographer / Writer

www.chrystalding.com



**Who owns
a genetic
test result,
an individual
or family?**

WELCOME GENOME CAMPUS

CONNECTING
SCIENCE