The Role of the Genetic Counsellor

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Agenda

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- What is genetic counselling?
  - What can and can't we do?
- The patient experience
- Consanguineous communities/families
- The future of genetics

Who are we?

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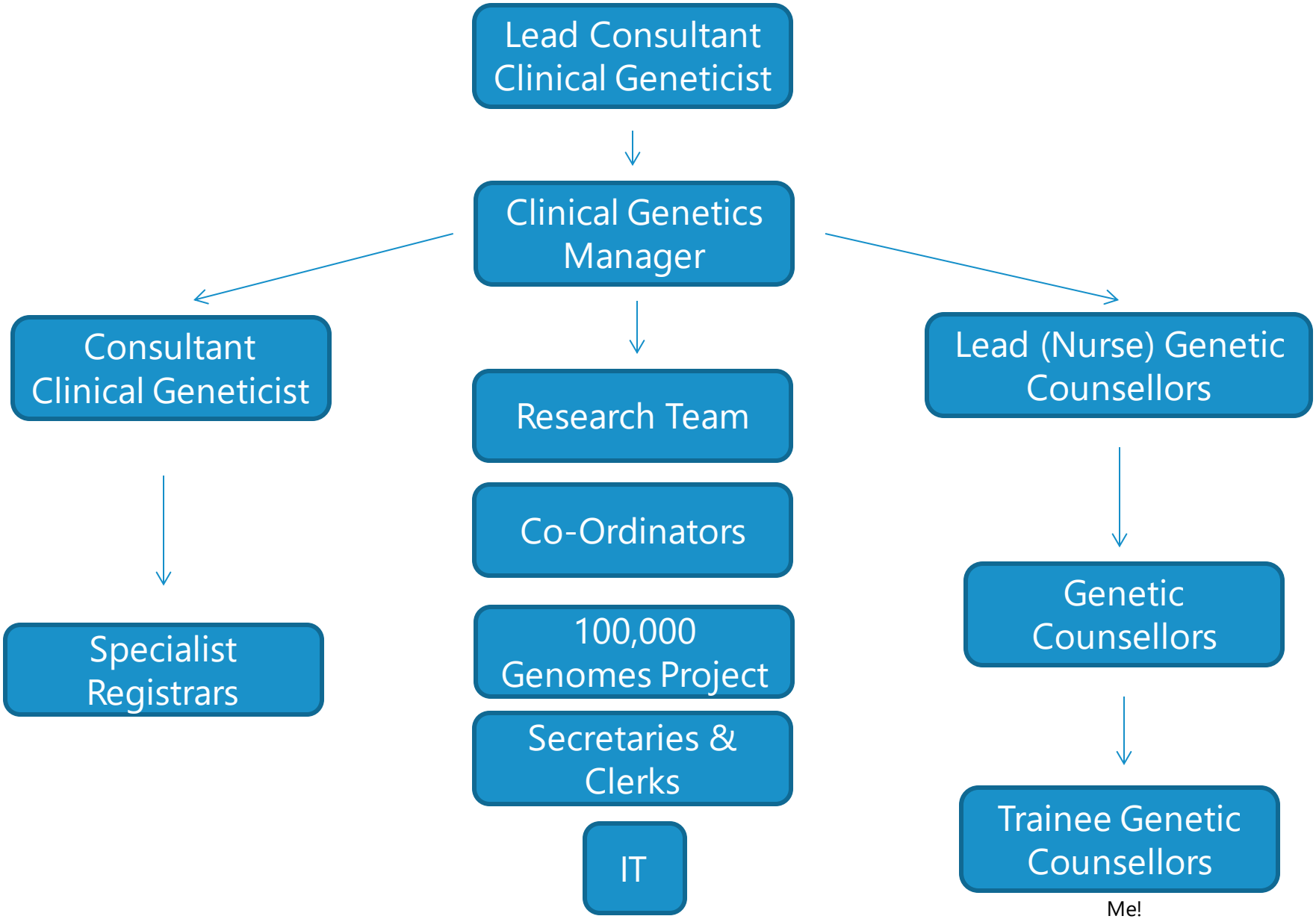

What do you actually do?

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A Genetic Counsellor will:

- 1. Identify the needs of the individual or family and use an **empathic approach** when giving genetic counselling
- 2. Collect, select, interpret, confirm and analyse information (including family and medical history, pedigree, laboratory results and literature) relevant to the delivery of **genetic counselling for individuals or families**
- **3. Help people understand and adapt to the medical, psychological, social and familial implications of genetic contribution to disease**
- 4. Assess the chance of disease occurrence or **recurrence**
- 5. **Provide diagnostic information** to clients based on family and/or medical history and/or genetic testing
- 6. Provide **education** about inheritance, testing, management, prevention, resources and research to relevant individuals or families **and other healthcare professionals**
- 7. **Promote informed choices and psychological adaptation** to the condition or risk of the condition
- 8. Apply expert knowledge to facilitate the individual or family to **access the appropriate healthcare resources**, including a medical diagnosis and resources for management of the condition.

GCRB – What is genetic counselling? (2017) Available at: http://www.gcrb.org.uk/

What do you actually do?

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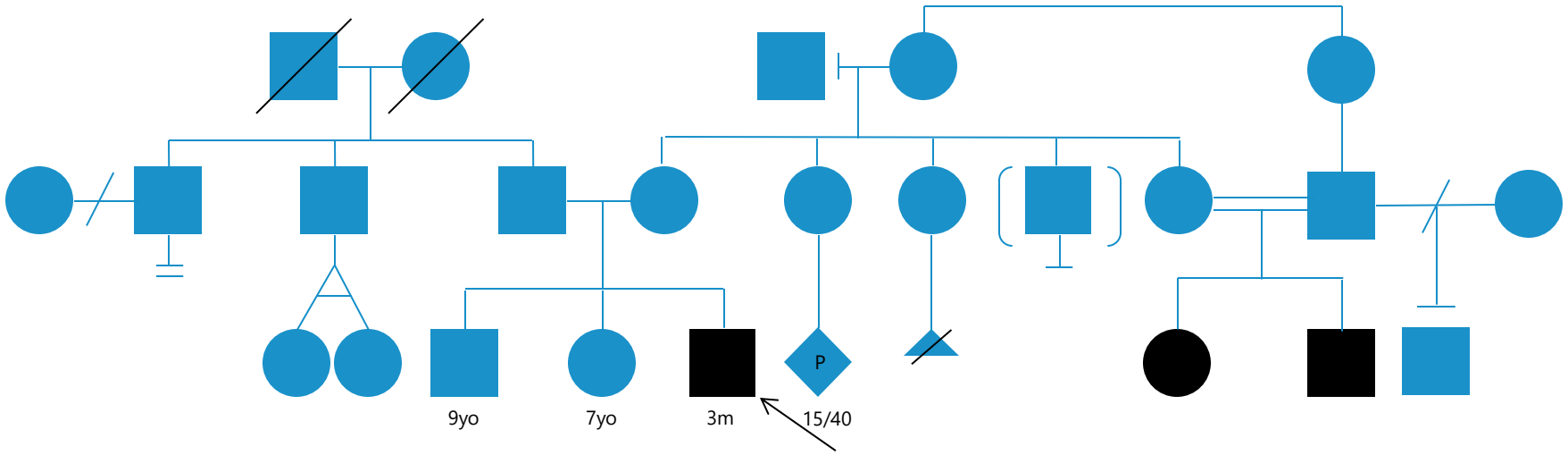
Scenario: a couple have a child affected with cystic fibrosis (CF) and want to know the chance of having another child affected with the condition.



Problems:
Is all of this information accurate?
Who should be tested first?
What about estranged relatives?

What do you actually do?

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- Provide information about genetic contribution to disease and offer tests
  - Diagnostic
  - Carrier/Predictive
  - Prenatal
- Convey complex information in lay language
  - What is a genome? A chromosome? A gene? DNA? Types of inheritance? Mosaicism? X-inactivation?
  - "Mutations"?
  - VUS?
  - Negative test result?

What do you actually do?

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- Helping patients to make informed decisions
  - Is testing appropriate – clinically, emotionally? Timing?
  - Autonomy; non-directive; confidentiality
  - **Consent**

- Offer/initiate testing if appropriate
  - Consent & phlebotomy
  - Organising samples/consent/information sharing

What do you actually do?

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- Explanation of results
  - Organising follow-ups
  - Making new referrals, e.g. breast/gynae
- Support
  - Counselling
    - Dealing with grief/loss/death, attachment, family dynamics, guilt, depression/MH issues, anxieties/"cancer worry"
  - Helping to communicate information through families
  - Further referrals, e.g. Cavendish centre, or therapy

Patient Experiences: Scenario 1

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"I would be grateful if you could see this pleasant 40 year old lady, who has recently been diagnosed with a stage 4 triple-negative breast cancer. She says her mum had breast and ovarian cancer at 42 and died at 63. She has 3 young daughters and is worried about their risks of developing cancer."

Patient Experiences: Scenario 1

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- Does the patient actually want to be tested?
  - What is the motivation to seek testing?
  - What makes them feel the most safe? How do they cope with threats? (aunty's burglar)
- What do they understand about the reason they are in genetics?
  - "GP sent me" vs "I really want to be tested"
- Is now the right time to consider testing?
  - Fast-tracks, life events etc
  - Age of daughters?

Patient Experiences: Scenario 1

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- What kind of support do they have?
  - Alone?
  - Preparedness for other relatives
- Results
  - Relief, empowerment, being looked after
  - Grief, anxiety, depression, guilt
  - Running out of the room vs not leaving the room vs not caring in 2 weeks vs not moving in 16 weeks
- Testing not available
  - Low/moderate risk – still have to live with "risk"
  - Need to test someone else, e.g. living affected relative
  - No one alive to test

Patient Experiences: Scenario 1

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- NB: not intended to convince people not to seek testing, but to challenge whether it is right for them, at that time
- Anecdotally – very few BME patients?
  - Stigma?
  - Barriers in accessing healthcare?
  - Minority groups = fewer patients?
- Self-selecting group of people motivated to test

Patient Experience: Scenario 2

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- 4 year-old boy with autism, learning difficulties, speech delay, mild dysmorphic/facial features
- Baseline genetic testing performed by paediatrician
  - Helpful diagnosis?
  - Variant of uncertain significance?
- Parents tested
  - One parent has same thing/similar problems
  - Neither parent has same variant
  - Chance of happening again?
- Further testing in the family?

Consanguinity / Close-Knit Communities

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- As standard – **always** ask about consanguinity
- BRCA & Fanconi Anemia - Ashkenazi Jewish
- Sickle Cell Anemia – Asian & African American
- Cystic Fibrosis – Caucasian/British
- Haemochromatosis – Traveller community
- Bowel cancer – recessive genes

Consanguinity / Close-Knit Communities

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- Similar consultation to any referral but may alter management or testing protocol
- Usually either referred due to new diagnosis **or** as follow-up genetic testing
- Support from then on for the family but not usually before
- "Counsellor"
  - Little capacity for long-term support
  - Rarely – home visits
- "I do not need to see genetics because my partner and I are not related"

Communicating Genetics

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- Wellcome Genome Campus - https://societyandethicsresearch.wellcomegenomecampus.org/
  - Music of Life
    - https://vimeo.com/album/5216273
  - Socialising the Genome
    - https://vimeo.com/album/4647208
- Online courses (e.g. HEE Whole-Genome Sequencing)
  - https://www.futurelearn.com/courses/whole-genome-sequencing?utm_source=gep-website&utm_campaign=wgsfl&utm_medium=referal

The Future of Genetics

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- Laboratory reorganisation
- Genomic testing
  - 100,000 Genomes Project results
- "Mainstreaming genetics"
- Likely to find more
  - More diagnoses?
  - More uncertainty?
  - Role of genetics services?