

KNOWING YOUR GENETICS, THERE ARE SO MANY BENEFITS

Clot Risk Genes (1 in 20 people)

Factor V Leiden and Factor II Prothrombin gene changes have a 4-to 7-fold increased risk of deep vein thrombosis and pulmonary embolism. The combined contraceptive pill and HRT have increased clot risks so knowing you have a clot risk gene means you can make safer choices. For long-distance flights - simple blood thinning tablets.

Haemochromatosis (1 in 150 people)

Genetic recessive disorder causing iron overload, leading to cirrhosis, liver cancer & affecting the heart. With early diagnosis & regular blood removal- the condition will not affect your life expectancy. **Only 1 in 20 people with haemochromatosis know they have it.**

Familial Hypercholesterolemia FH (1 in 250 people)

FH results in very high levels of LDL - bad cholesterol, causing early heart attacks and strokes. Finding out means you can take highly effective medicines. **9/10 people are unaware that they carry these gene changes.**

Reassurances

Protection by the ABI Code of Insurance: insurance companies will not ask for your genetic test results, other than if you have a pre-existing condition with an associated gene in the UK. We only test for genes which are “actionable” meaning you can do something positive. **We have five multi-skilled consultant geneticists.** **You can use a pseudonym** - be anonymous.

Genetic Reproductive Options

Pre-Implantation Genetic Testing (PGT):

If the genetic condition is serious enough, you can stop it going through to the next generation by IVF & selecting embryos without the condition.

1 in 350 babies have a recessive disorder

Conditions can vary from Tay Sachs disease, typically fatal by ages 4-5 with progressive neurodegeneration, to cystic fibrosis (1 in 25 of us carry a change in this gene).

Genes come in pairs and for a recessive disorder to develop both parents must be carriers of the same recessive gene. If you have a carrier match we can then advise you about reproductive options. There are over 2,000 recessive disorders.

CARRIER TESTING is available to all couples.



HOW TO BOOK A CONSULTATION

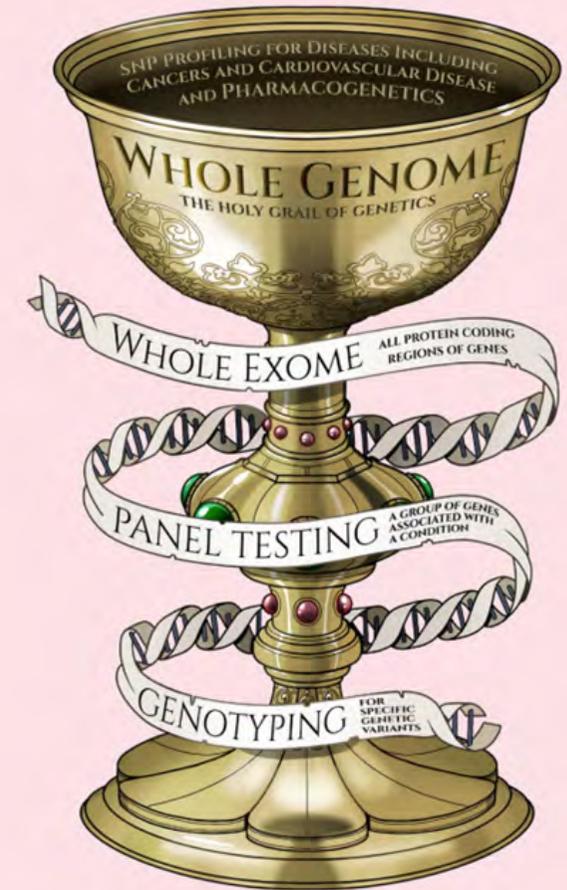
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WOMEN'S HEALTH

THE LONDON GENETICS CENTRE

Our aim is to stop suffering and save lives

Genetic testing aims to drop overall cancer mortality by 1/5th - 1/7th

We find those at increased risk through two techniques: **Single Risk Genes** and **Polygenic Risk Scores**

Breast Cancer (1 in 8 women)

10% of cases are due to inherited gene changes, eg *BRCA1*, *BRCA2* (1 in 277 to 1 in 381 carrier frequency; 1 in 40 with Ashkenazi Jewish ancestry), & 10 other risk genes.

- **Earlier/more frequent breast screening**
- **Adjust lifestyle factors** - alcohol, weight, exercise, consider increase risk associated with HRT and oral contraception.
- **Preventive - 98% risk reduction risk-reducing breast surgery** for those at highest risk

Ovarian Cancer (1 in 50 women)

20% of cases caused by inherited gene changes eg *BRCA1*, *BRCA2*, Lynch syndrome (9 ovarian cancer genes).

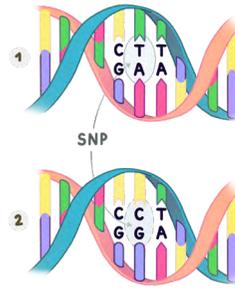
- **Preventive surgery reduces risk by 95%** for those with these gene changes.
- **Drop the overall ovarian cancer mortality by nearly 20%**. Screening does not reduce mortality.
- **Hormones** - oral contraception reduces the risk of ovarian cancer by 30-50%.

Bowel Cancer (1 in 18 women)

10% is due to inherited genetic changes, such as Lynch syndrome.

- **Colonoscopies** reduce the risk of bowel cancer mortality by 70% for 10 years after.
- **Medications** - Aspirin reduces the risk of bowel cancer in Lynch syndrome carriers by up to 50%.

Knowing you have Lynch syndrome improves mortality by 25% and can inform your relatives to take action to reduce risk.



WHAT IS A POLYGENIC RISK SCORE (PRS)

Polygenic risk scores (PRS) are a major separate way of predicting risk. We hope to save more lives using PRS risk stratification in addition to single risk gene testing.

A SNP is when the 4 organic bases - C, T, G, A - swap, as per the diagram. Scientists add up the known positions of these SNPs and their associated weight of increased risk to form a PRS. Common diseases available for such as prostate, breast, bowel and ovarian cancer, as well as non cancer hypertension, coronary artery disease risk, etc.

Single Risk Genes

Polygenic Risk Scores

The PRS includes 12 conditions in women, importantly **osteoporosis and early menopause risk.**



BREAST CANCER (12 genes)

A *BRCA1* or *BRCA2* gene change confers up to an 80% lifetime risk of breast cancer.

(higher risk stratum gives)
3-fold risk

Informs on screening intervals and age of commencement.



CARDIAC DISEASE (81 genes)

Cardiomyopathy, QT prolongation, familial hypercholesterolemia (FH). Aorta

3-fold risk

Coronary Artery Disease.
LDL Cholesterol,
Hypertension PRS

If know you have a high coronary disease PRS, take action and avoid having bypass surgery in your 50's.



BOWEL CANCER (20 genes)

Lynch Syndrome confers up to a 50% lifetime risk of bowel cancer as well as uterine cancer in women. Polyposis syndromes include FAP and MAP.

3.8-fold risk

FINDS those who need earlier and more frequent screening.



OVARIAN CANCER (9 genes)

BRCA1, *BRCA2*: confers up to a 60% lifetime risk of ovarian cancer. Lynch syndrome: confers up to a 50% lifetime risk of uterine cancer & a 20% risk of ovarian cancer.

2-fold risk

Ovarian PRS has a weaker effect whereas the higher-risk genes are more important.



OSTEOPOROSIS

3-fold risk

Informs on DEXA scan and management strategies.



PHARMACOGENOMICS (120+ drugs)

Pharmacogenes **INFORMING YOU** of how you will metabolise Clopidogrel, Codeine, Metoprolol, antidepressants and **hundreds of other medicines.** Personalised adjusted genetic drug prescription is now the ideal.



Learn more about genetic testing pathways.



Learn more about our story here.