Breast Cancer Risk Assessment Tools for the GP

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13/09/2025

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THIS IS MODERN MEDICINE

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"To identify a women as a carrier only after she develops cancer is a failure of cancer prevention"

Most women develop breast cancer with no family history and only 5-10% of breast cancers are hereditary cancers

Identification of women at high-risk is important for the effective preventative strategy as Breast cancer risk affects families differently

We aim to bring a different level of care to high risk women

- Risk assessment tools
- Risk Category and screening guidelines
- Genetic testing
- Alternative to surgery
- Breast cancer risk and HRT

Over the years

1866
Pierre Paul Broca describes familial breast cancer in his book *Traité des Tumeurs*.

1926
Janet Lane-Claypon publishes *A Further Report on Cancer of the Breast, With Special Reference to its Associated Antecedent Conditions*.

1970s
Modern epidemiological studies demonstrate that breast and ovarian cancer risk is increased in women with a first-degree relative affected by the disease.

1974
Mary-Claire
begins her h
the genet
underpinnin
breast can

The Human
Genome Project

sequence of *BRCA2*
is reported.

sequence is completed.

1996
Myriad Genetics begins offering the first commercial diagnostic test for *BRCA1/2* mutations.

1998
BRCA1 and *BRCA2* are patented by Myriad Genetics.

2013
The US Supreme Court rules that naturally occurring genes may not be patented.

Present day
Patients are screened for a hereditary predisposition to cancer using multigene panels and NextGen DNA sequencing technologies.

Multiple risk models exist and they differ based on the risk factors included.

Tyrer-Cuzick

Personal Characteristics
Up to age 85
Body mass index
Ashkenazi Jewish heritage
Breast density

Family History
Breast and Ovarian Cancer
First, second-degree relatives
Age of onset
Bilateral breast cancer

Reproductive History
Age at menarche
Age at first live birth
Age at menopause
Use of hormone replacement therapy

Personal Medical History
Atypical ductal hyperplasia
Lobular carcinoma in situ
Ovarian cancer

Genetic Testing
BRCA1, BRCA2
Personal testing
First, second-degree relatives

Gail

Personal Characteristics
Age 35 - 85
Race/Ethnicity

Family History
Breast Cancer
First-degree relatives

Reproductive History
Age at menarche
Age at first live birth

Breast Disease
Breast biopsies
Atypical ductal hyperplasia

*Gail model can estimate risk for women without a BRCA1/BRCA2 mutation and no prior history of breast cancer, or ductal or lobular carcinoma in situ

BRCAPRO

Personal Characteristics
No age restriction
Ashkenazi Jewish heritage

Family History
Breast and Ovarian Cancer
First, second-degree relatives
Age of onset
Bilateral breast cancer
Male breast cancer

Claus

Personal Characteristics
Age 20 - 79

Family History
Breast Cancer
First, second-degree relatives
Age of onset

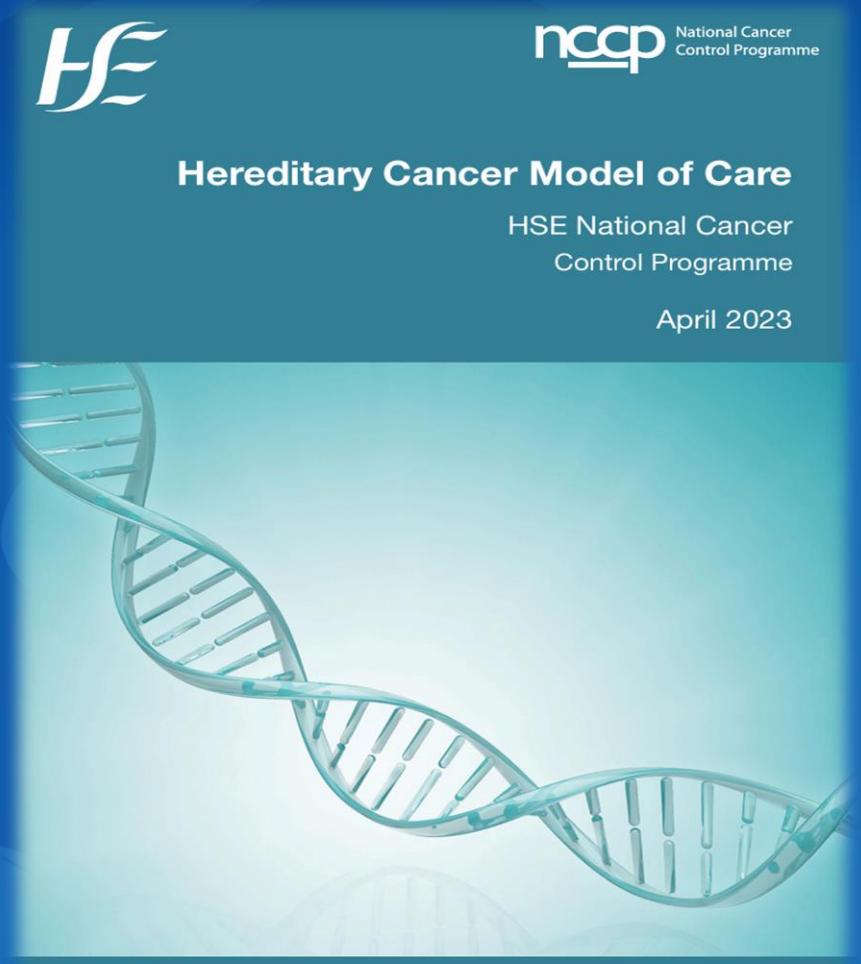
Risk Factor
Age at First Occurrence of Menstruation
Number of Children
Age of First Live Birth
Oral Contraception Usage
Hormone Replacement Therapy
Body Mass Index
Alcohol Intake (grams/day)
Age of Menopause
Mammographic Density
Height (cm)

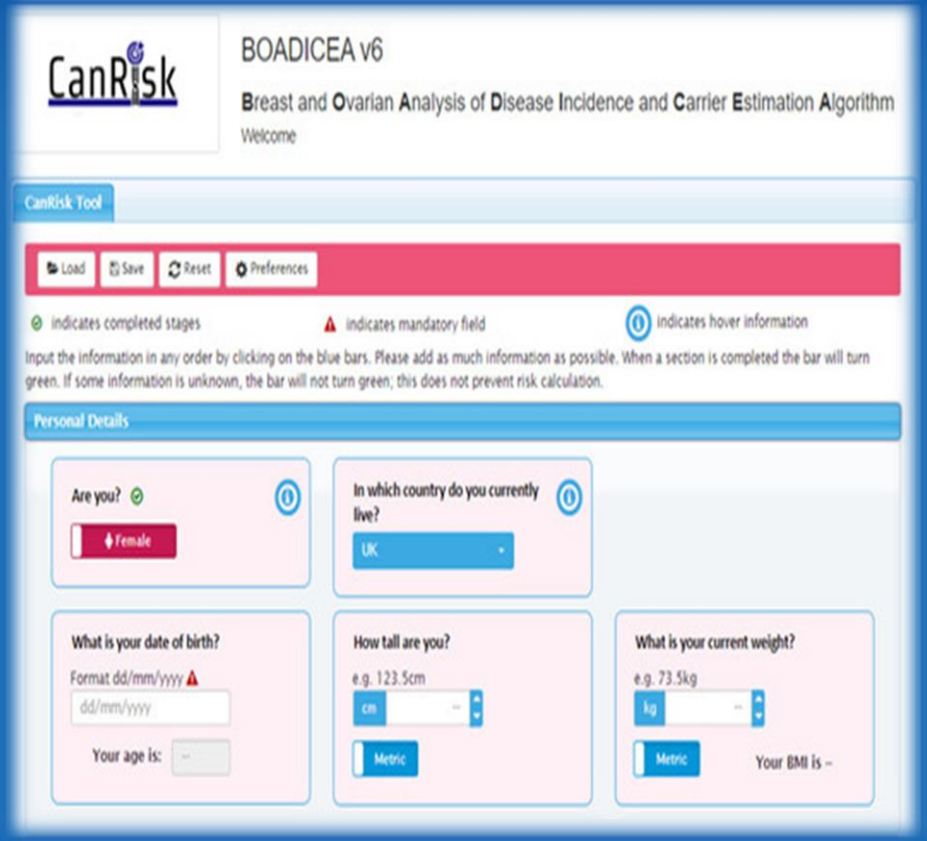
Breast Cancer Risk calculating models

Gail model	Claus model	BRCAPRO model	Tyrer–Cuzick model	BOADICEA model
• Age of the person • Age at menarche • Age at first live birth • Breast biopsies (AH) • Family history – First-degree relatives	• Age of the person • Age at menarche • Age at first live birth • Family history – First-degree relatives – Second-degree relatives	• Age of the person • Family history – First-degree relatives – Second-degree relatives – Third-degree relatives – Age at onset of breast cancer – Bilateral breast cancer – Ovarian cancer – Male breast cancer	• Age of the person • Body mass index • Age at menarche • Age at first live birth • Age at menopause • Hormone replace-ment therapy use • Breast biopsies (ADH, LCIS) • Family history – First-degree relatives – Second-degree relatives – Age at onset of breast cancer – Bilateral breast cancer – Ovarian cancer	• Age of the person • Family history – First-degree relatives – Second-degree relatives – Third-degree relatives – Age at onset of breast cancer – Bilateral breast cancer – Ovarian cancer – Male breast cancer

AH, atypical hyperplasia; LCIS, lobular carcinoma *in situ;* BOADICEA, breast and ovarian analysis of disease incidence and carrier estimation algorithm.

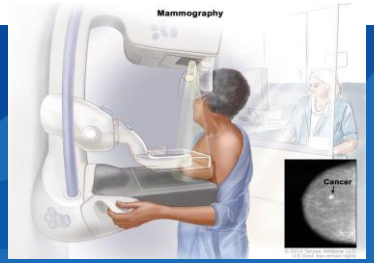
NCCP Breast Cancer Family Risk Assessment Group Family History GP Referral Guideline





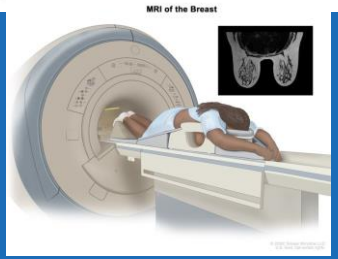
Risk Category and Screening Recommendation

Population Risk <17%	Moderate Risk ≥17% - <30%	High Risk ≥30% - <40%	Very High Risk ≥40%
NHS breast screening programme: 3 yearly mammograms 50-70 Health and lifestyle information Communication on future changes in risk	Moderate risk screening: Annual mammograms 40-49, then 3 yearly 50-70 Consider if family meets National Genomic Test Directory criteria for testing Health and lifestyle information Communication on future changes in risk Consider chemoprevention	High risk screening: Annual mammograms 40-59, then 3 yearly 60-70 Consider if family meets National Genomic Test Directory criteria for testing Health and lifestyle information Communication on future changes in risk Discuss risk management options including chemoprevention	Very high risk screening: Annual breast MRI 30-50, annual mammogram 40-70 Refer or ensure gene carriers under Clinical Genetics Discuss risk management options Refer to Very High Risk Screening Service (VHRS)
GP/COMMUNITY	BREAST UNITS	SECONDARY CARE CLINICAL GENETICS	CLINICAL GENETICS



NICE National Institute for Health and Care Excellence

Improving health and social care through evidence-based guidance



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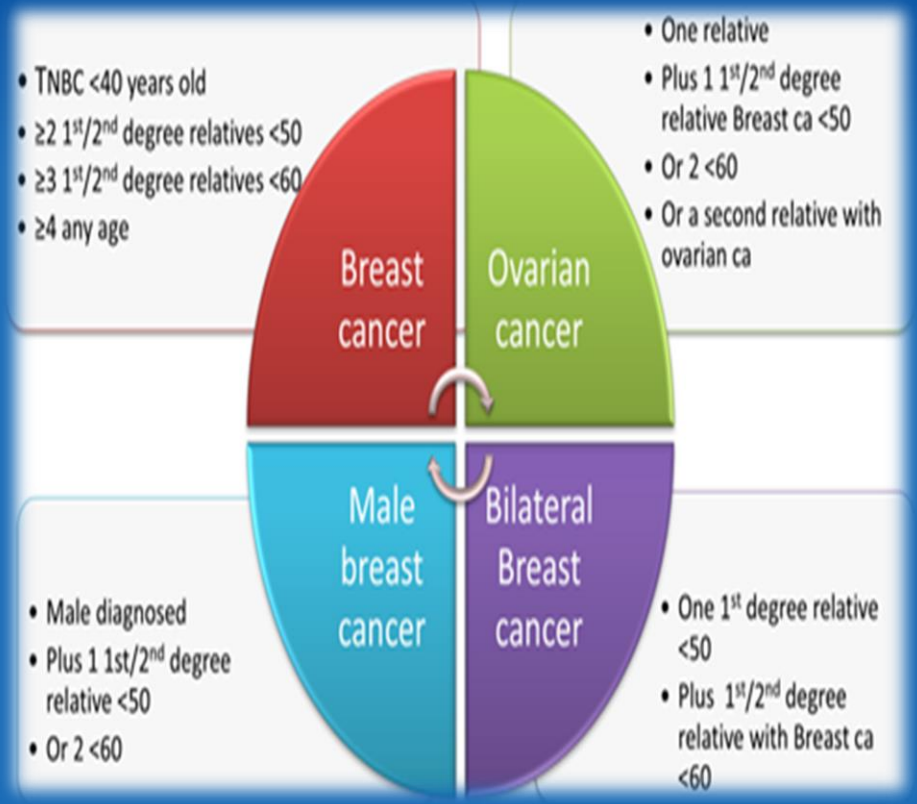
THE RISK GROUP REQUIRED REFERRAL

- **Every women can be referred for assessment**
- People without a personal history of breast cancer can be cared for in primary care provided that none of the following are present in the family history:

1. **bilateral breast cancer**
2. **male breast cancer**
3. **ovarian cancer**
4. **Jewish ancestry**
5. **sarcoma in a relative younger than age 45 years**
6. **glioma or childhood adrenal cortical carcinomas**
7. **complicated patterns of multiple cancers at a young age**



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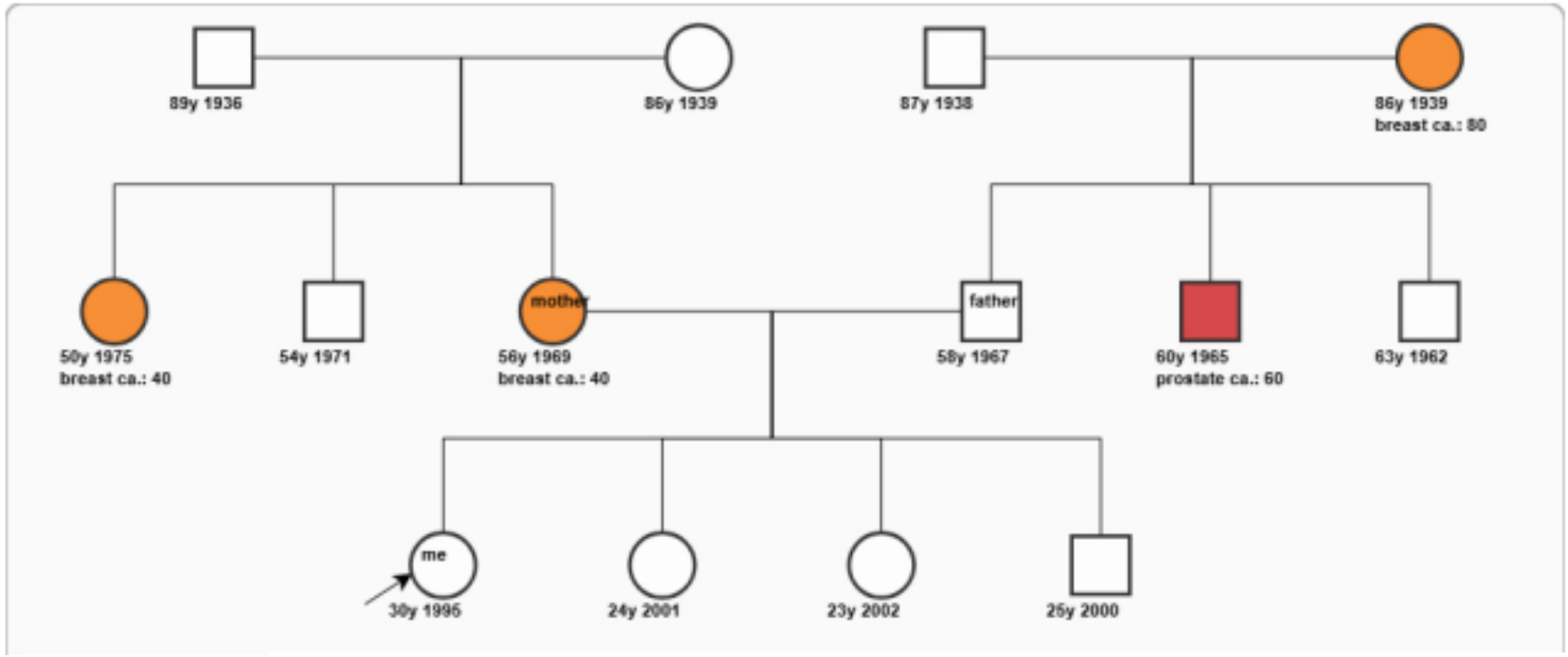
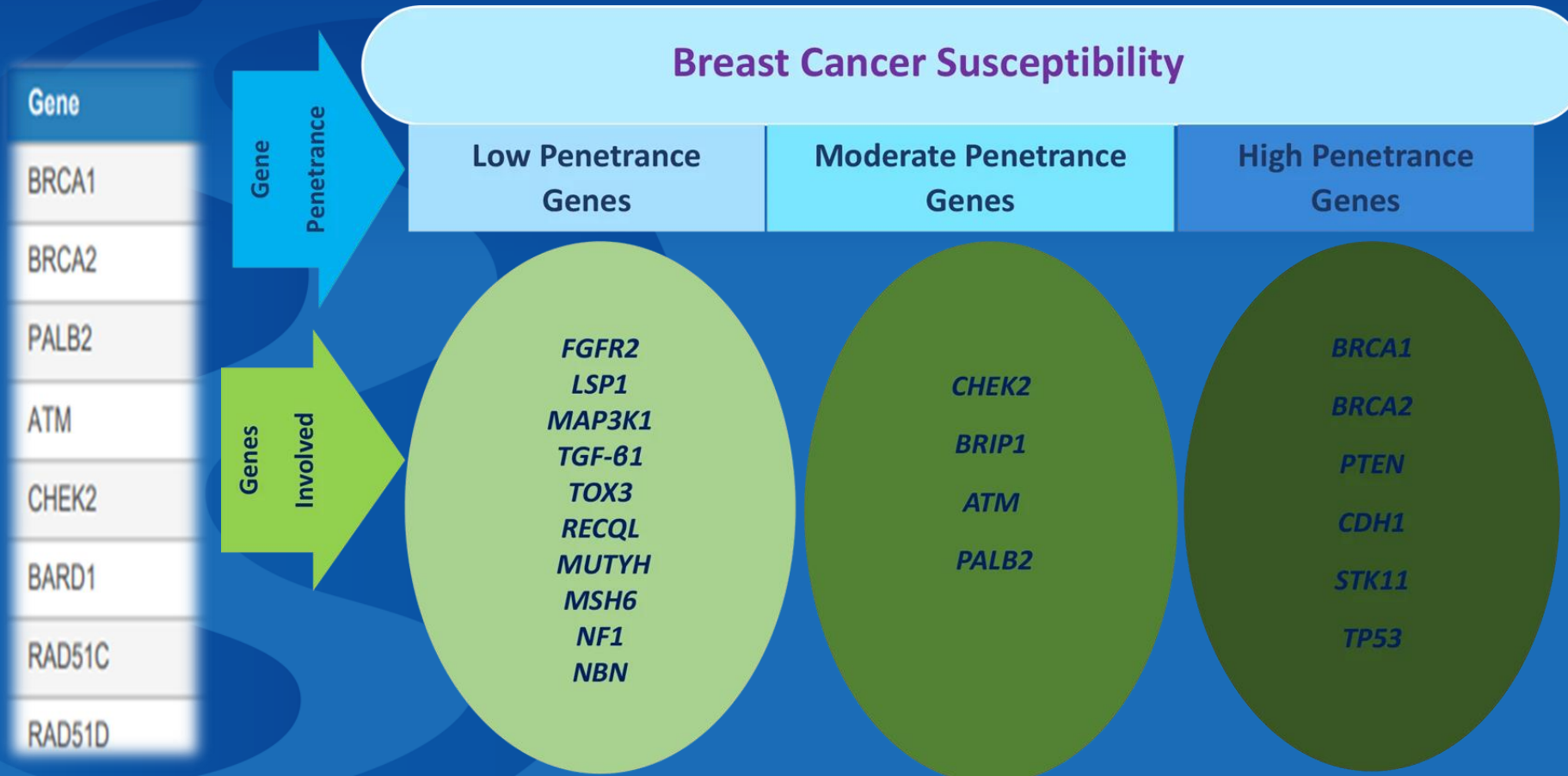


Cancer	Colour
Breast	
Contralateral breast	
Ovarian	
Prostate	
Pancreatic	

	Near population risk (a)	Moderate risk (b)	High risk (c)
Risk between ages 20 and 80	Less than 17%	**17% or greater but less than 30%**	30% or greater
Risk between ages 40 and 50	Less than 3%	**3% or greater to 8%**	Greater than 8%



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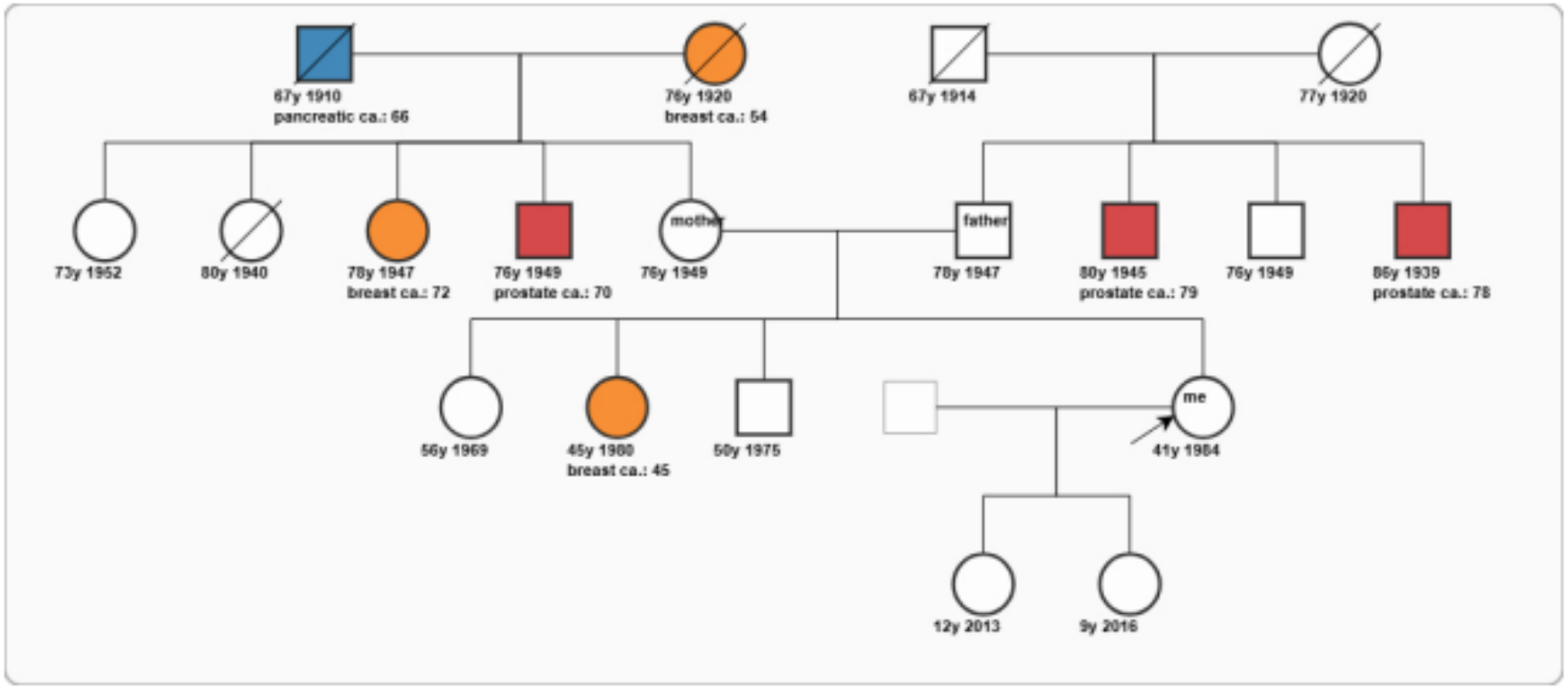
Genetic Mutations of Potential Relevance in Breast Cancer (BC) Development[4,5]

Affected Gene	Associated Risk & Syndromes
BRCA1* and *BRCA2	7 in 10 chance for BC
ATM	High BC risk, *ataxia-telangiectasia*
TP53	Rare BC cause; *Li-Fraumeni syndrome*
CHEK2	2-fold risk increase for BC
PTEN	Increased BC risk; *Cowden syndrome*
CDH1	Invasive lobular BC; *Hereditary diffuse gastric cancer*
STK11	Increased BC risk; *Peutz-Jeghers syndrome*
PALB2	Increased risk of BC



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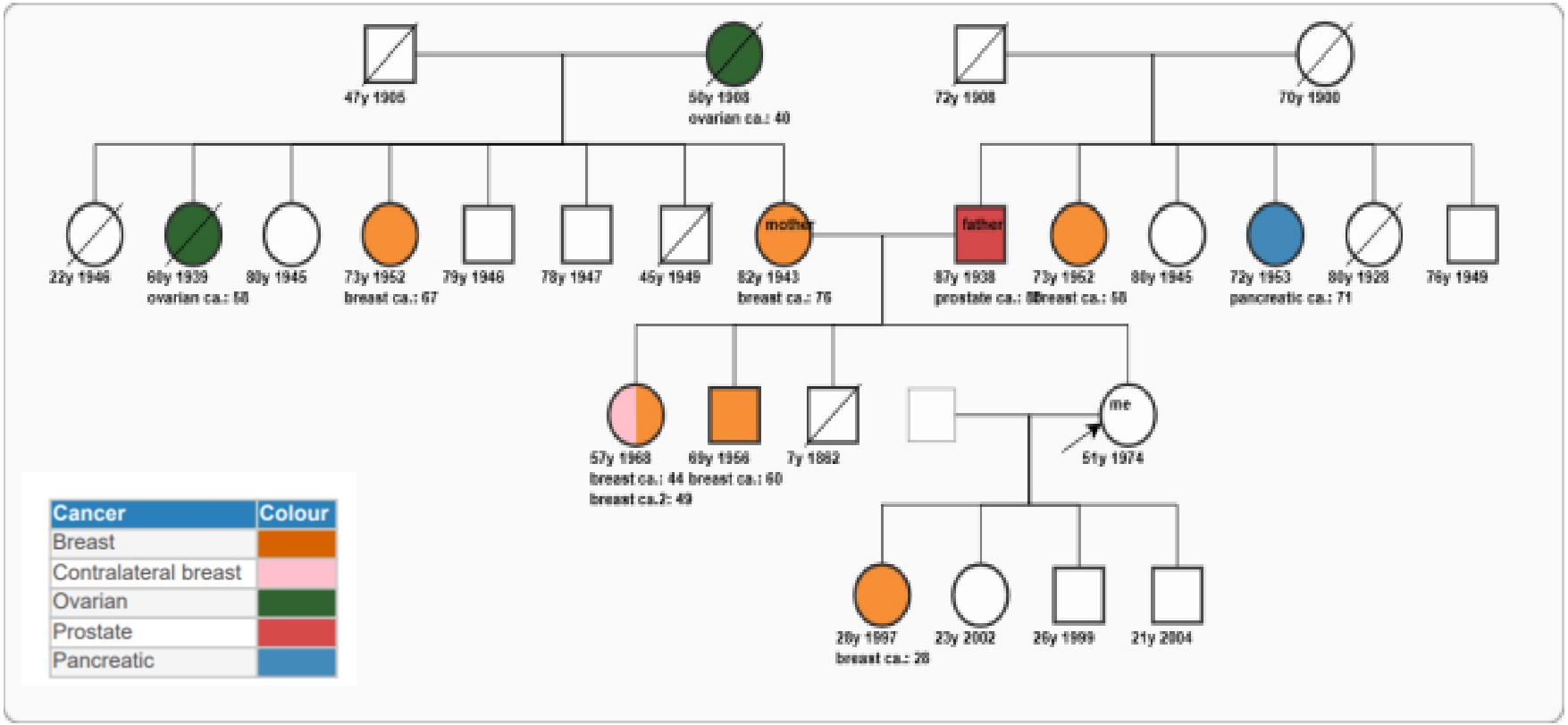

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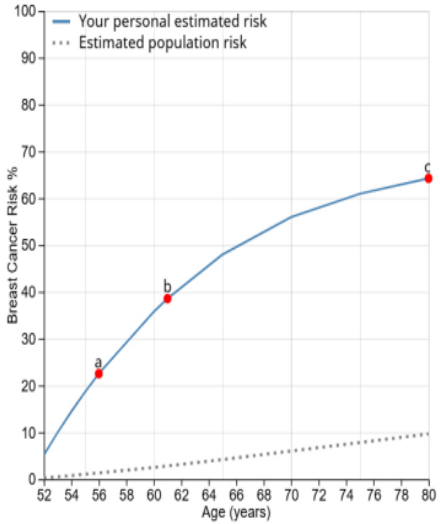
Your Breast Cancer Risks Compared to the Rest of the Population

The graph represents your risk of developing breast cancer between now and the age of 80 years compared to the population. In other words, the graph shows your personal risk of developing breast cancer compared to the average risk of women in the population.



Graph key:

Label	Your estimated breast cancer risk
a	Next 5 year risk is 22.5%
b	Next 10 year risk is 38.6%
c	Risk between now and the age of 80 is 64.3%

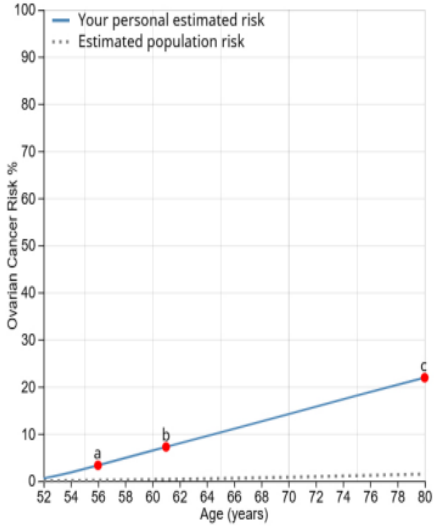
Below is an alternative way of visualising your risk of developing breast cancer between now and the age of 80 years compared to the average population risk.

Your risk: 64.3% **Population risk: 9.7%**

CanRisk Report: bbcfamilyhistory

Your Ovarian Cancer Risks Compared to the Rest of the Population

The graph represents your risk of developing ovarian cancer between now and the age of 80 years compared to the population. In other words, the graph shows your personal risk of developing ovarian cancer compared to the average risk of women in the population.



Graph key:

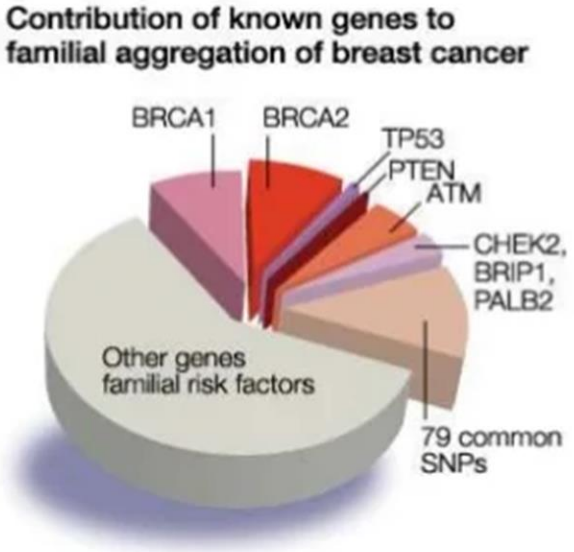
Label	Your estimated ovarian cancer risk
a	Next 5 year risk is 3.3%
b	Next 10 year risk is 7.2%
c	Risk between now and the age of 80 is 21.9%

Below is an alternative way of visualising your risk of developing ovarian cancer between now and the age of 80 years compared to the average population risk.

Your risk: 21.9% **Population risk: 1.5%**

Genetics of breast cancer

- BRCA1
- BRCA2
- TP53
- PTEN
- PALB2
- STK11
- CDH1
- ATM
- CHEK2



(Prat A et al., 2011)

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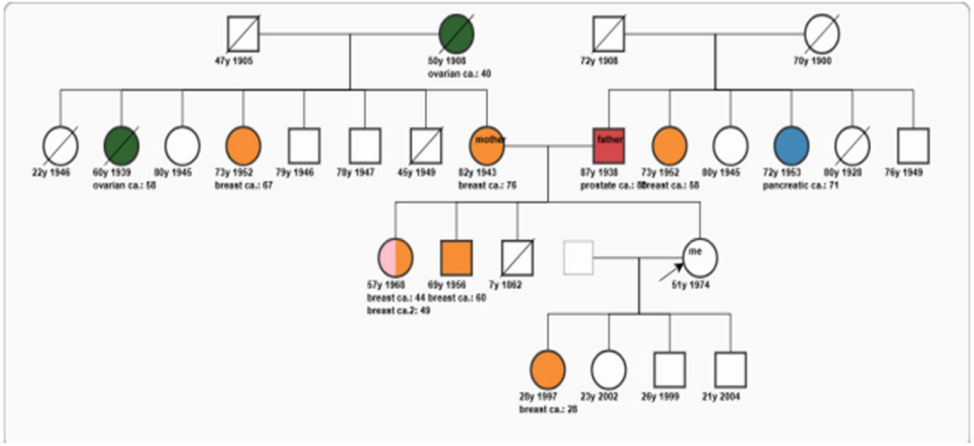
BRCA1

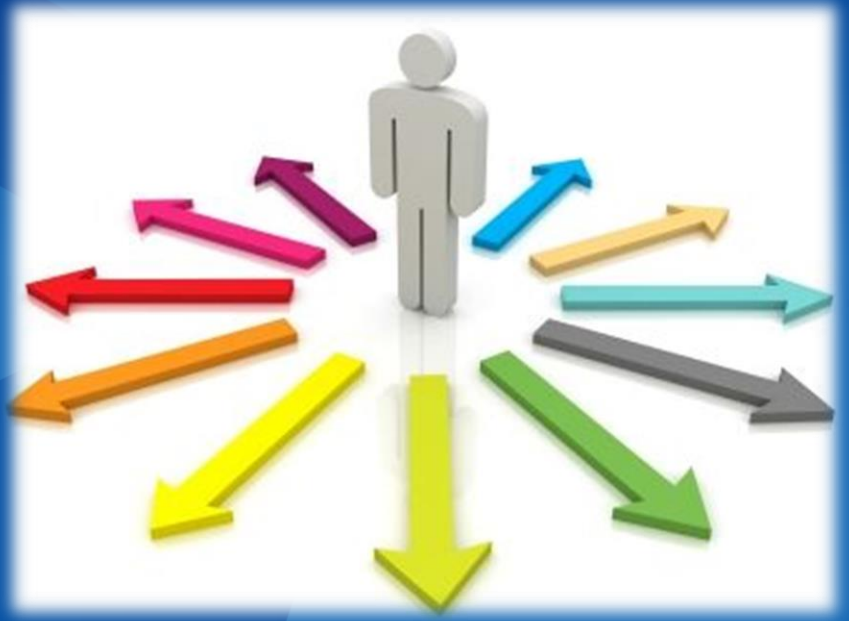
- **60%-72%** risk of developing breast cancer
- Up to **1.2%** of male breast cancer
- Associated with **39%-58%** of ovarian cancer
- Also associated with **pancreatic** and **prostate** cancer risk

BRCA2

- **55%-69%** risk of developing breast cancer
- Up to **7.1%** of male breast cancer
- Associated with **13%-29%** of ovarian cancer
- Also associated with **pancreatic cancer, prostate cancer, and melanoma** risk



Gene	Pathogenic Variant Carrier Probability
BRCA1	64.34%
BRCA2	18.85%
BRCA1 or BRCA2	83.19%
PALB2	1.54%
CHEK2	0.34%
ATM	0.12%
BARD1	0.03%
RAD51D	0.03%
RAD51C	0.02%
BRIP1	0.02%



Risk reduction modality

Less Cancer, Less treatment, Less death

- **Risk reducing Surgery**
  Personal preferences, timing

- **Alternative to surgery**
  Intensive screening
  Chemoprevention / Medication
  Modifiable lifestyle choices (Smoking, Weight, Alcohol, Exercise, Diet)

- Life expectancy vs quality of life




Chemoprevention and cancer risk reduction

- Age and general health
- Less effective than surgery

- Personal Choice / LCIS
- Tamoxifen / AI (Examestane/ anastrazole)
- To reduce the risk of ER positive invasive breast cancer

- It is not recommended in Male BRCA carrier
- Tamoxifen reduce risk in BRCA 2 but not BRCA1 carriers

- Risk reduction benefit continues for at least 10 y

BSO and cancer risk reduction

- Completed childbearing
- Gynecological oncologist
- Decrease risk of ovarian cancer and mortality if <50
- Earlier in BRCA 1
- Menopause / HRT
- Bone disease
- ? Hysterectomy (benefit vs surgery risk)
- Salpingectomy alone (Ovarian and breast reduction risk)

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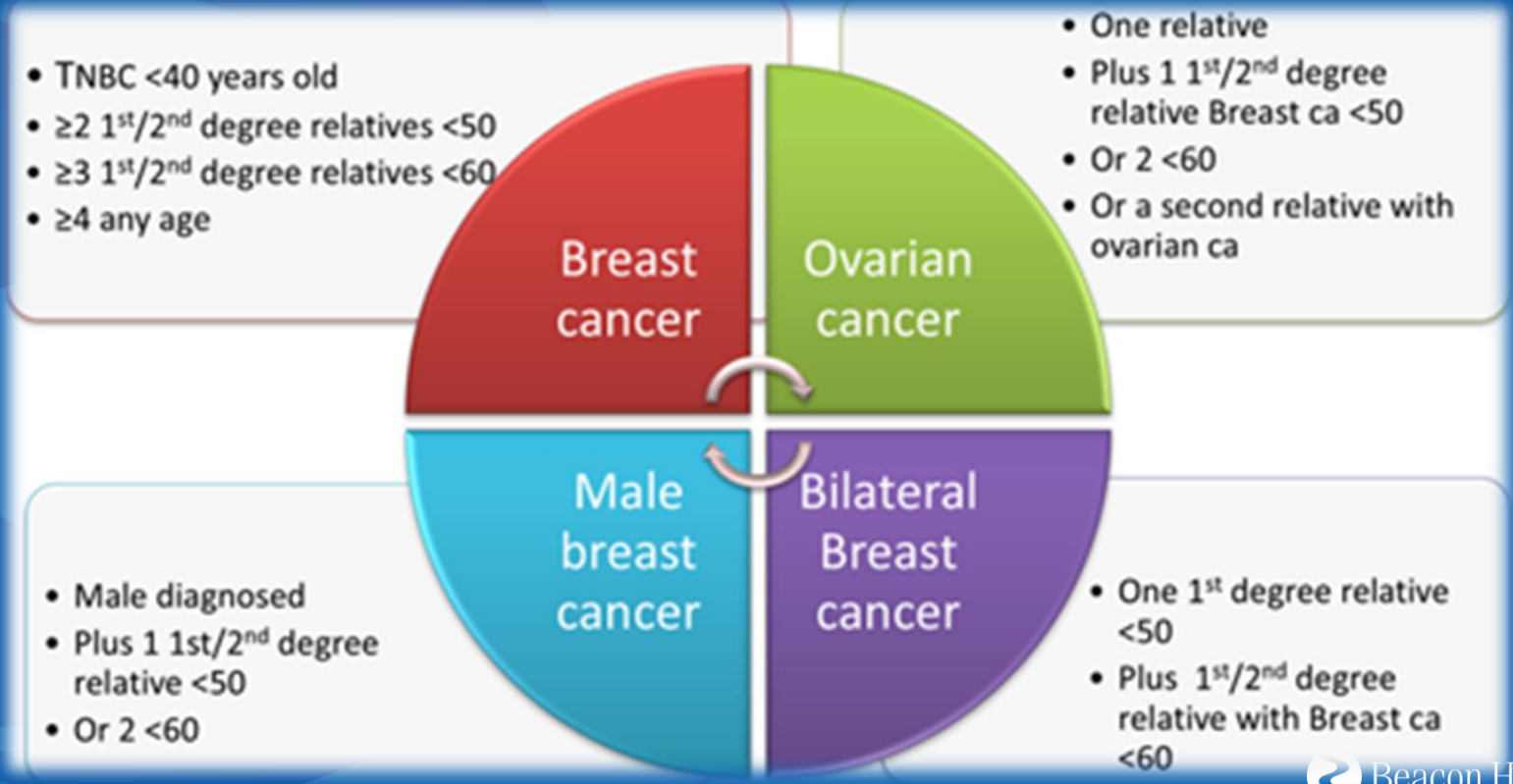

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HRT – understanding the risks of breast cancer

A comparison of lifestyle risk factors versus Hormone Replacement Therapy (HRT) treatment.
Difference in *breast cancer incidence per 1,000 women aged 50-59.* Approximate number of women developing breast cancer *over the next five years.*

Baseline

23
cases of breast cancer diagnosed per 1,000 of the UK general population over the next 5 years

An additional....

24 cases in women who are overweight or obese (BMI equal or greater than 30)

5 cases in women who drink 2 or more units of alcohol per day

4 cases in women on combined HRT

3 cases in women who are current smokers

Reduction

7 fewer cases in women who take at least 2½ hours moderate exercise per week

4 fewer cases in women on oestrogen only Hormone Replacement Therapy (HRT)

https://www.womens-health-concern.org/wp-content/uploads/2019/10/WHC-UnderstandingRisksofBreastCancer-MARCH2017.pdf

Beacon Breast Centre Patient Focused Service

New Referrals-

Breast cancer risk assessment
Breast awareness, life style advise
Medium and high risk pathway

Existing high risk patient-

reassess regard risk
imaging need/ genetic testing
risk reducing life style education
chemoprevention/ RR surgery

Confirmed Mutation patient-

regular clinical reviews
imaging surveillance,
RR surgery,
Other cancer screening

High risk cohort other than FHx

chest wall radiotherapy

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5-10% of breast cancer ~ 60% are high-risk group

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https://ibis.ikonopedia.com/
https://www.canrisk.org/
https://www.nice.org.uk/guidance/cg164

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Thank you

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