



# **Is it all in your GENES? Understanding the Risk for Breast Cancer**

**Genetic Counsellors  
Hereditary Cancer Clinic  
WRHA Genetics and Metabolism**

# Presenter Disclosure

- **Faculty:** [Kim Serfas/Heidi Rothenmund](#)
- **Relationships with commercial interests:**
  - **Grants/Research Support:** [none to declare](#)
  - **Speakers Bureau/Honoraria:** [none to declare](#)
  - **Consulting Fees:** [none to declare](#)
  - **Other:** [none to declare](#)

# Mitigating Potential Bias

- Not Applicable



# Learning Objectives

At the end of this session, participants will be able to:

- Differentiate between sporadic, familial and hereditary breast cancer
- List 3 “red flags” which may suggest an inherited predisposition to develop breast cancer in your patient and/or their family history
- Define the risk of developing breast cancer in carriers of a BRCA gene mutation
- Describe breast cancer risk reduction options and appreciate their benefits and limitations
- Explain the pros and cons of genetic testing for hereditary breast cancer



# What is a Genetic Counsellor?

Genetic counselling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease.

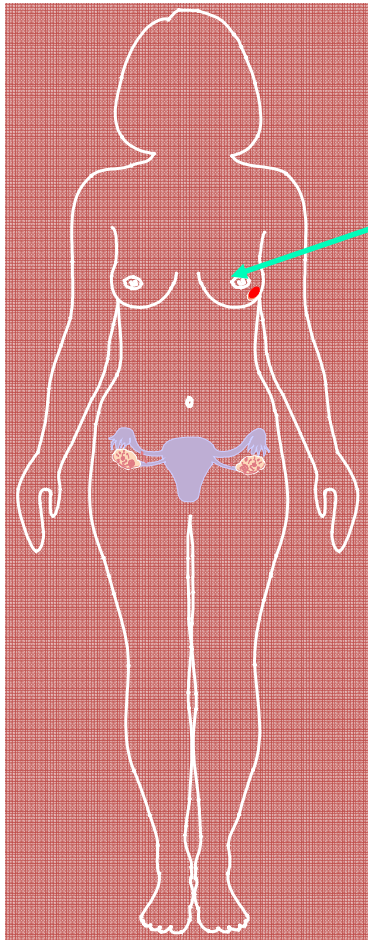
This process integrates:

- Interpretation of family and medical histories to assess the chance of a hereditary condition
- Education about inheritance, testing, management, prevention, resources and research
- Counselling to promote informed choices and adaptation to the risk or condition



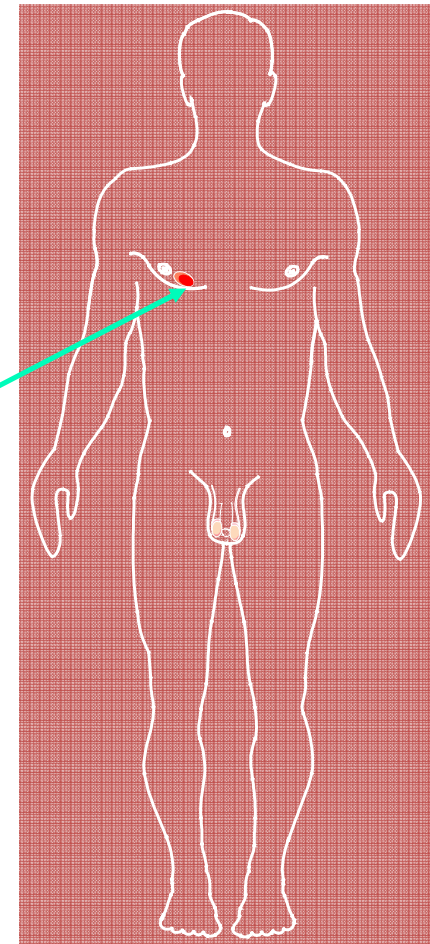


# Breast Cancer in the General Population



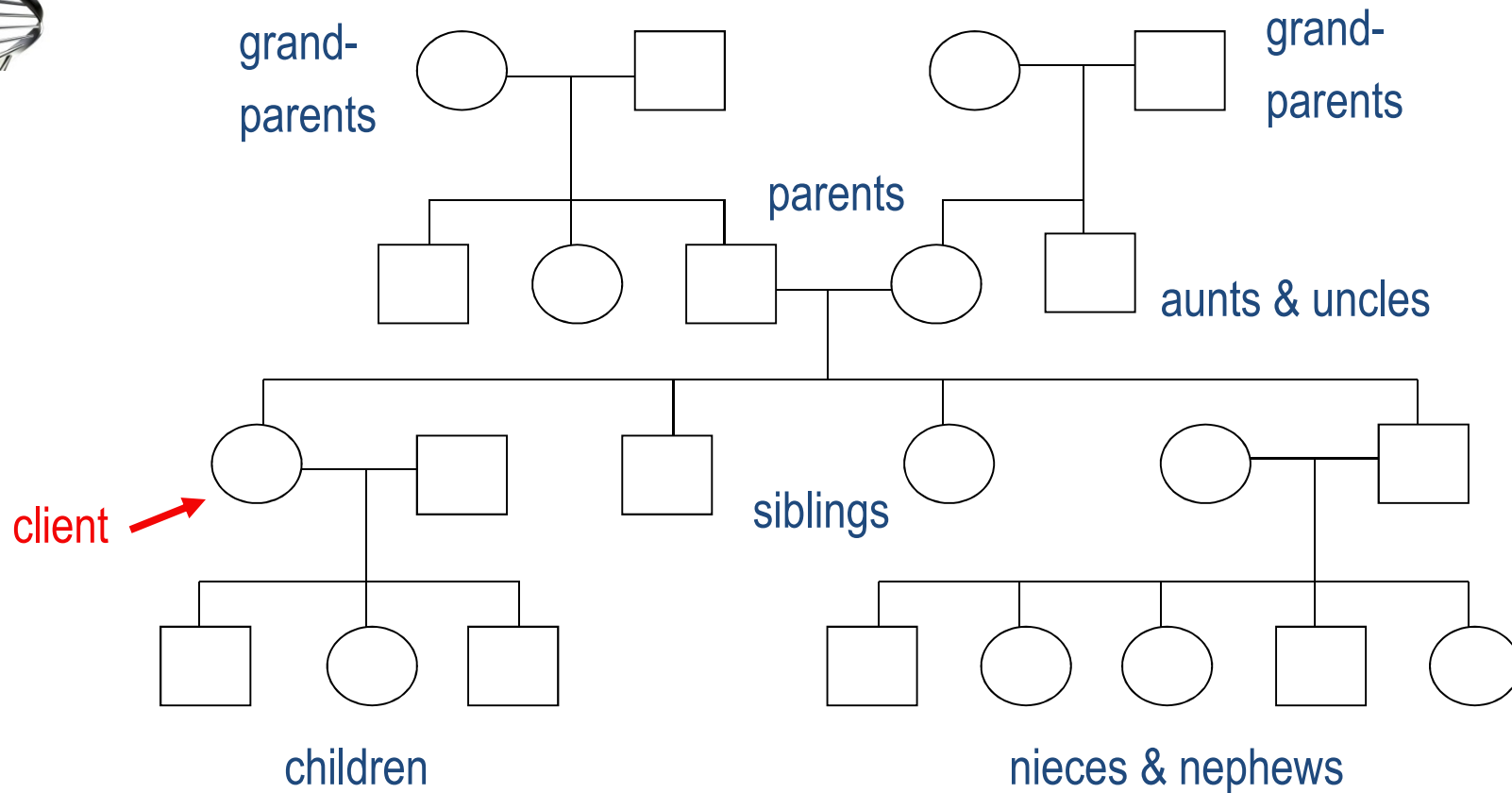
breast cancer  
(11-12%)

male breast cancer  
(.001%)





# Family History: organized in a pedigree



Less concerned when client reports a history of breast cancer in her cousins (1<sup>st</sup> or 2<sup>nd</sup>) or in her great grandma or her great aunts, even less concerned if she reports breast cancer in her sister-in-law or neighbour.



What factors do you think may indicate a woman is at higher risk of breast cancer?





# Risk Factors for Breast Cancer

- Gender
  - Age
  - Age of menarche
  - Having children
  - Breast density
  - Benign breast conditions
  - Previous chest radiation
  - Being overweight
  - Alcohol
- Personal history of breast cancer
  - Family history of cancer
  - Race and ethnicity
  - Inherited genetic mutation



What percentage of breast cancer is genetic?

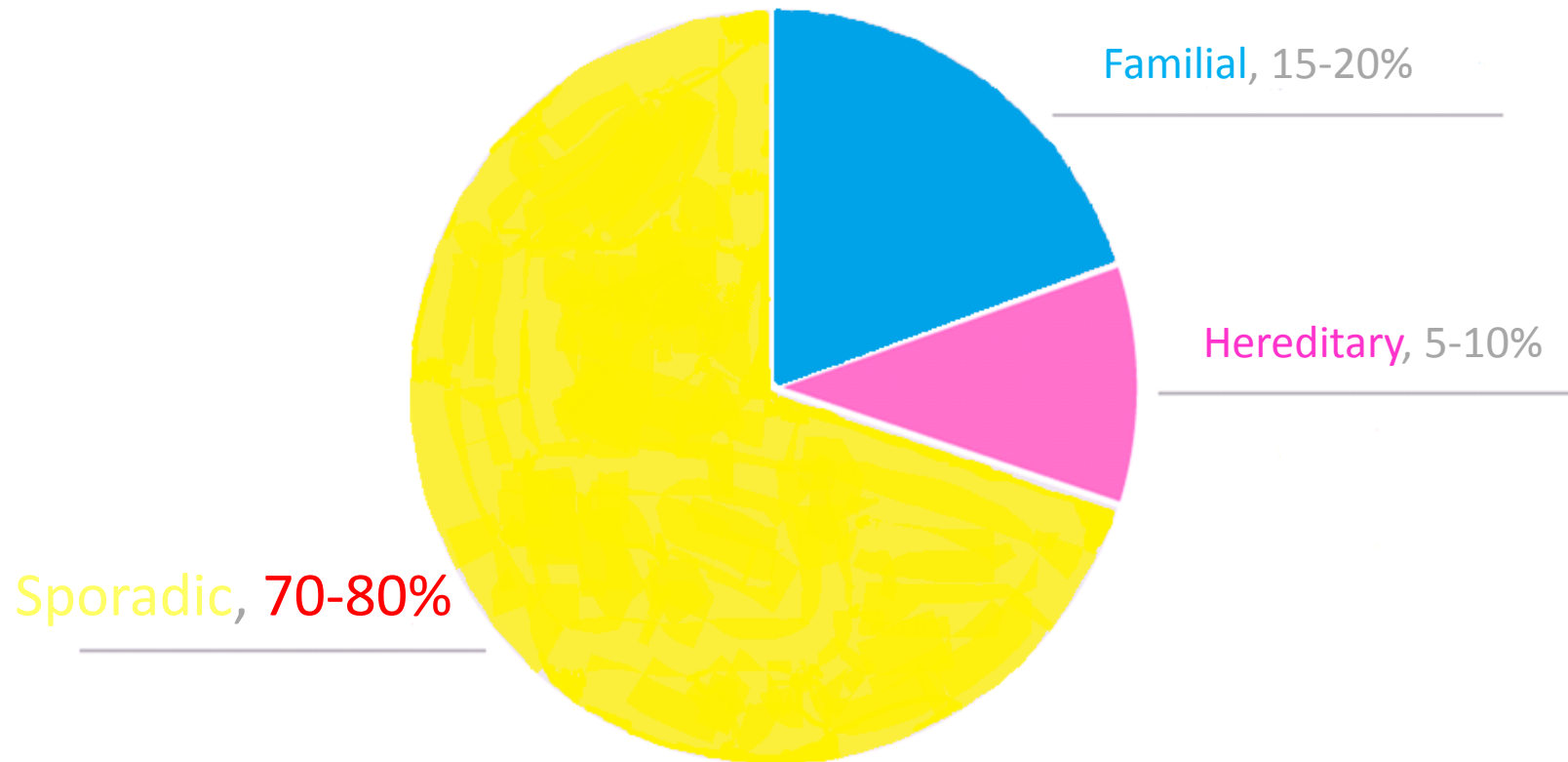
All Cancer is  
Genetic



However, most  
cancer is **NOT**  
Hereditary



# Causes of Breast Cancer

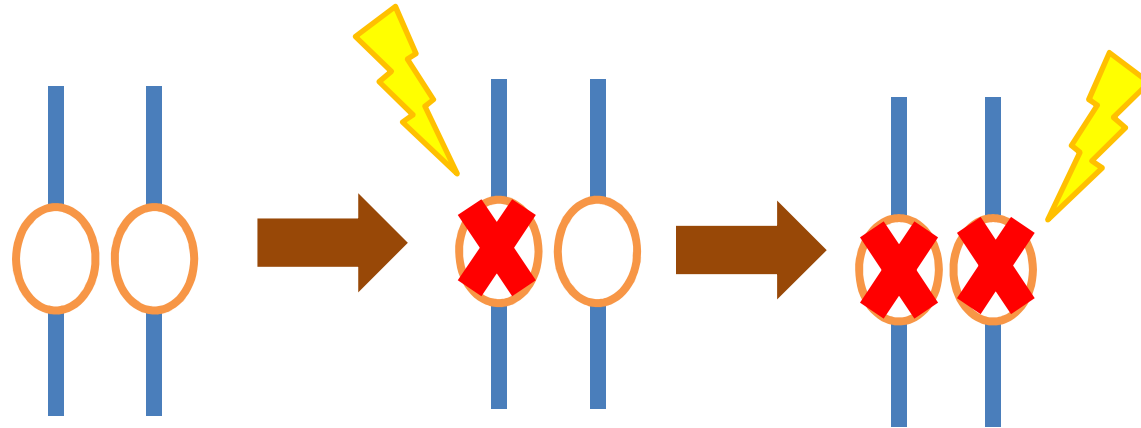


Most cancers are sporadic!

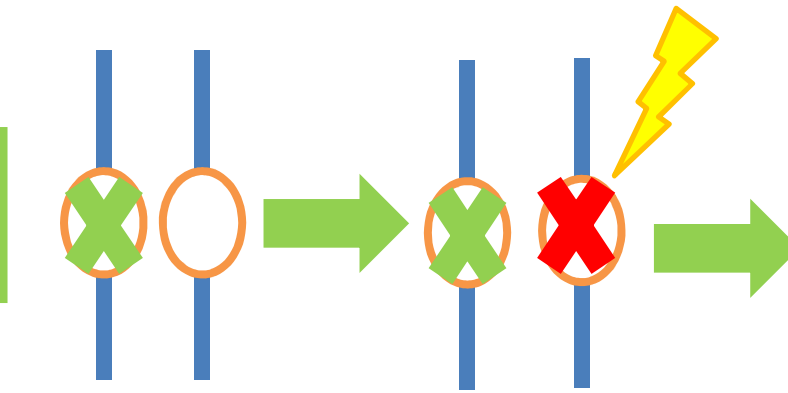


# 2-Hit Knudson Hypothesis

Sporadic cancer

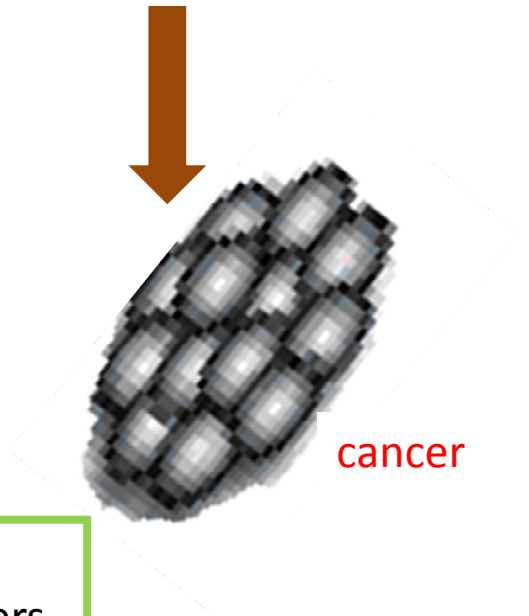


Hereditary cancer



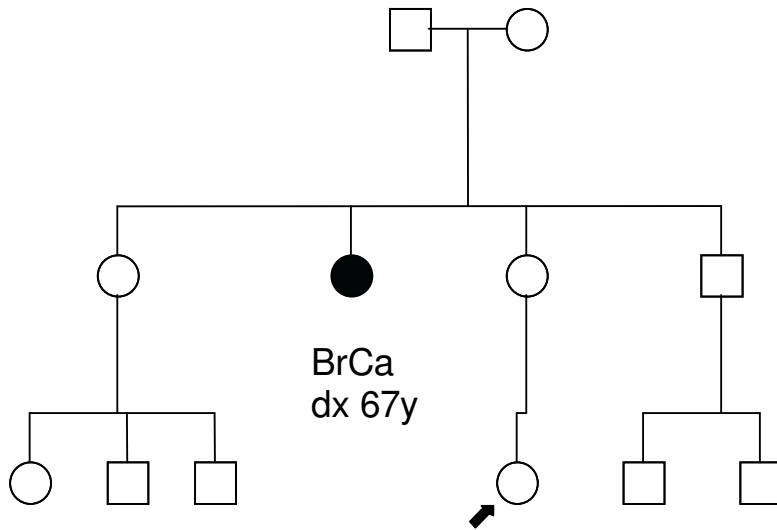
50/50

Younger diagnoses  
Multiple primary cancers  
Multiple cases in Family





# Sporadic Cancer

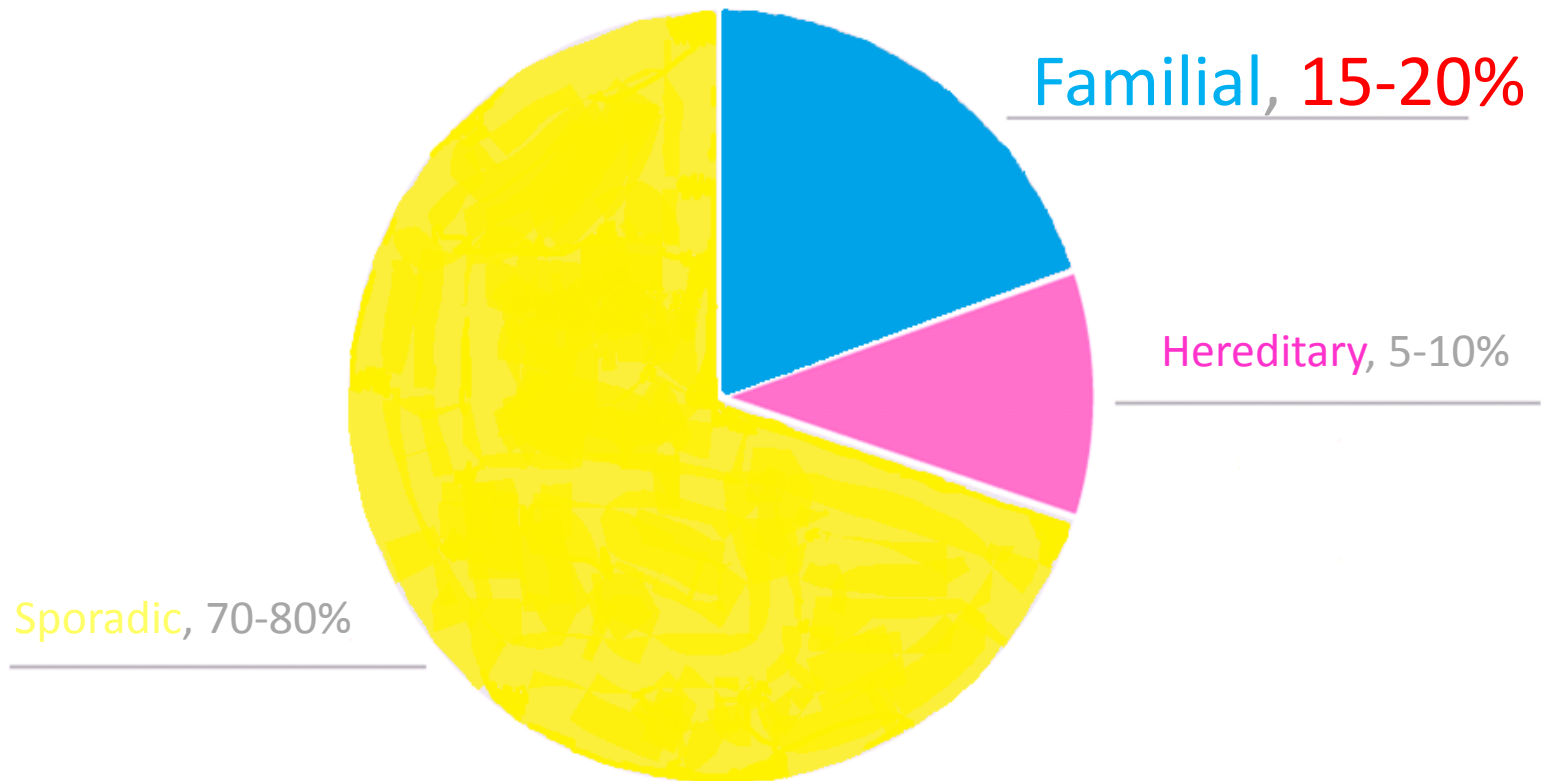


- majority of cancer cases
- **single** diagnosis in the family
- onset later in life
- inherited risk unlikely
- should not be referred to Hereditary Cancer Clinic

This women is likely at population risk and she should have mammograms every other year starting at age 50.



# Causes of Breast Cancer

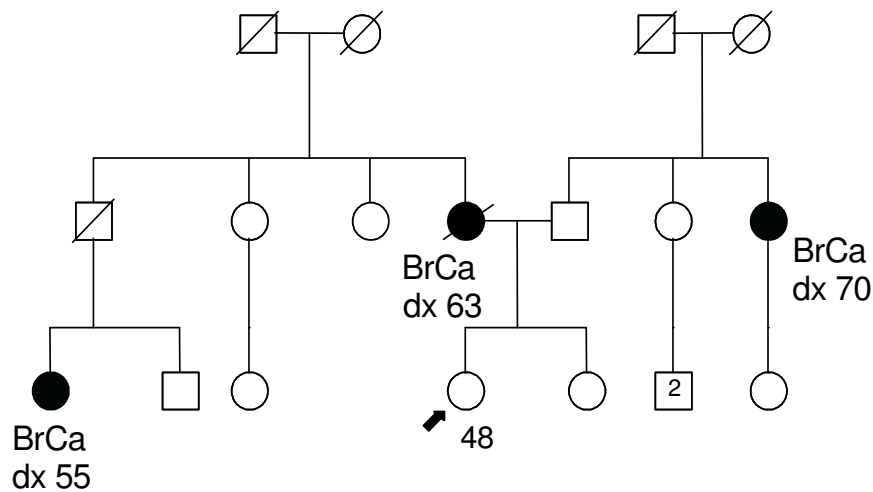


Familial = Multifactorial



# Familial Cancer

## Cluster of Cancer within Families



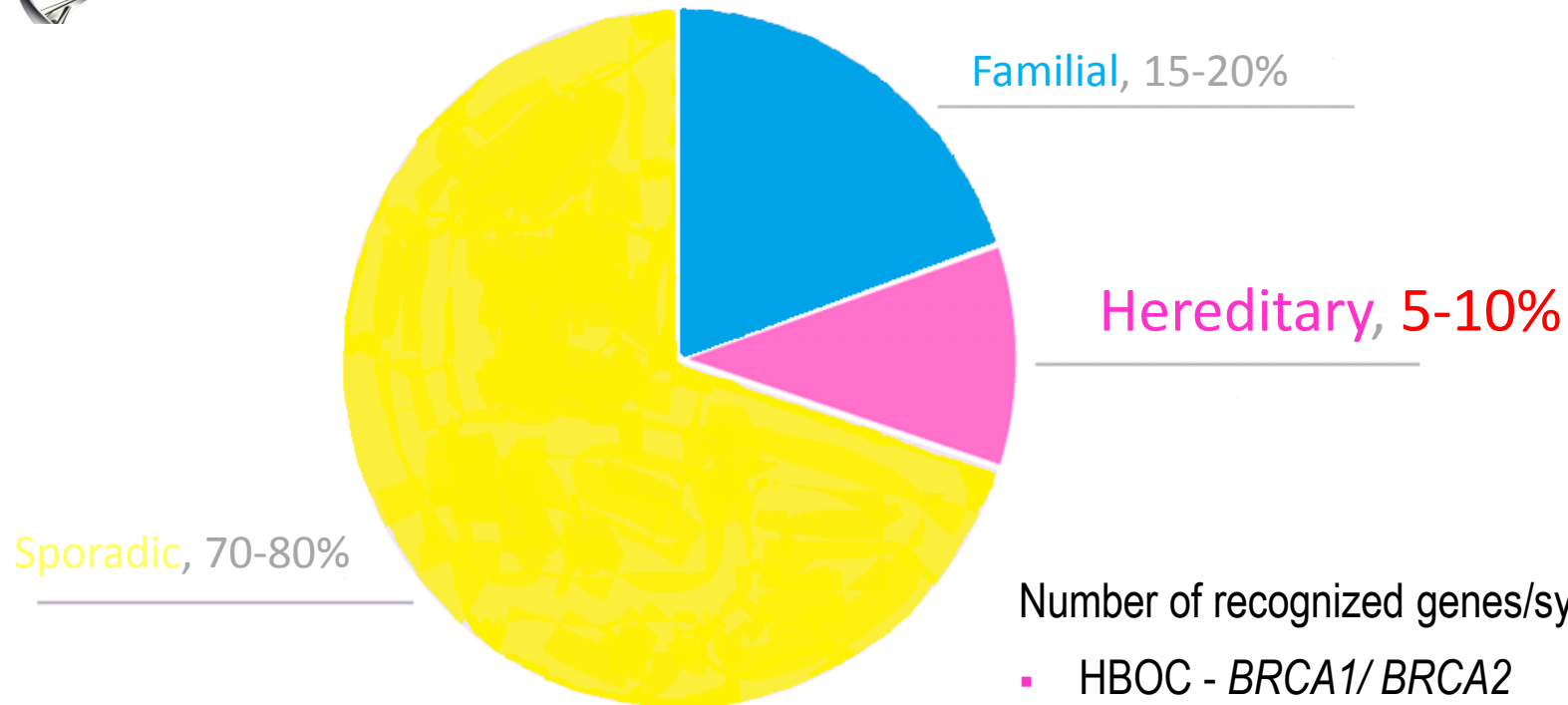
- 2 or more affected relatives on same side of the family
- **later onset**
- unilateral
- unclear pattern of inheritance
  - ? chance alone
  - ? common environment
  - ? genetic factors

This woman is likely at slightly higher risk than other women in the general population. She should have yearly mammograms starting at age 50.





# Causes of Breast Cancer



Number of recognized genes/syndromes

- HBOC - *BRCA1/ BRCA2*
  - *CHEK2, BRIP1, PALB2 etc*
- Li- Fraumeni – *TP53*
- Cowden syndrome- *PTEN*
- Diffuse Gastric Cancer - *CDH1*
- Peutz-Jeghers – *STK11*
- Ataxi Telangiectasia – *ATM*