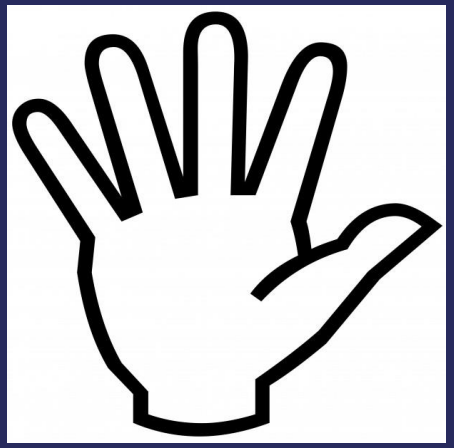
Erie St. Clair
Regional Cancer Program
in partnership with Cancer Care Ontario

Demystifying Cancer Genetics

HIGH RISK BREAST PATIENTS

Veronica Bryksa, MS, MS, CGC, CCGC
Certified Genetic Counsellor
Erie St. Clair Regional Cancer Program
Windsor Regional Hospital

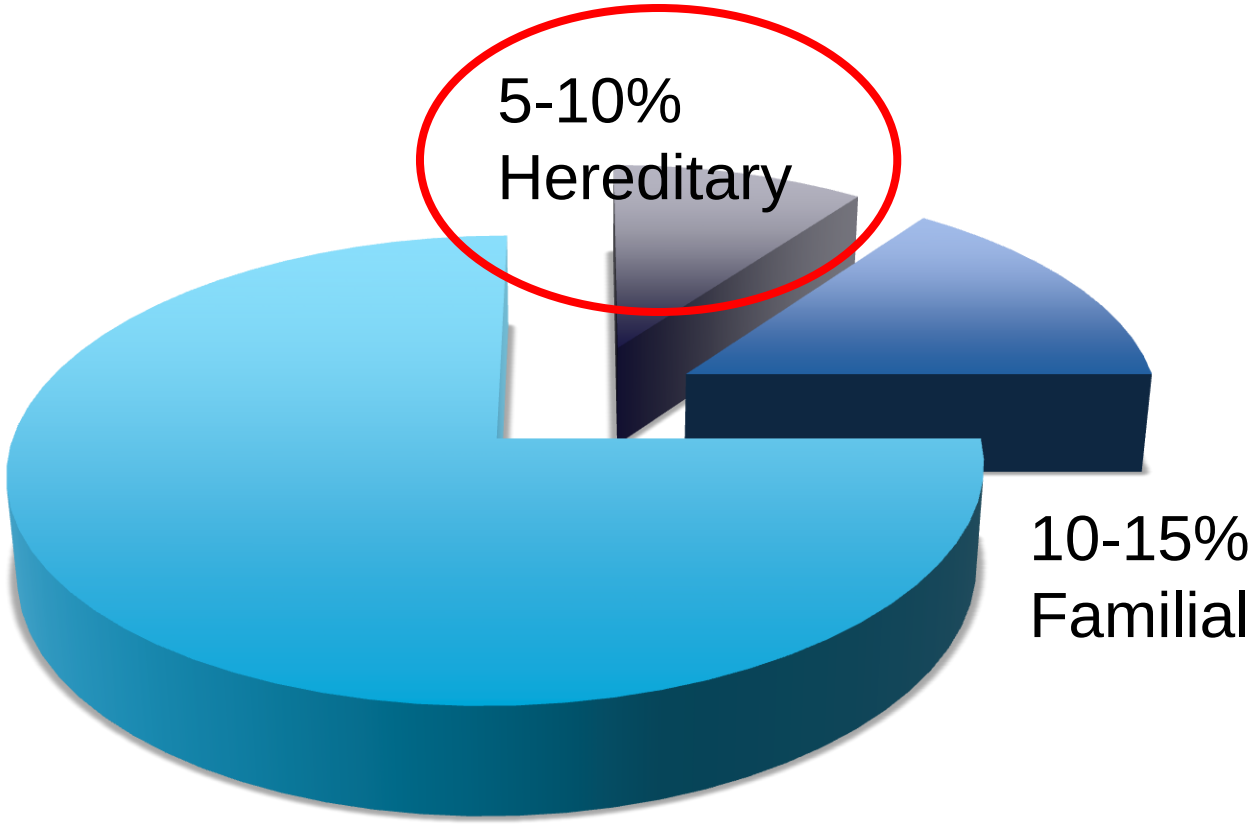
Who has talked to a patient about their family history of breast cancer?



Objectives

1. Hereditary breast cancer review
2. Discuss case examples
3. Explain who and how to refer
    - What to ask your patients
    - What forms to use
    - The genetic counselling/testing process

The breakdown…..



Hereditary Breast Cancer

- **BRCA1/2** - tumour suppressor genes
  - Cancer susceptibility

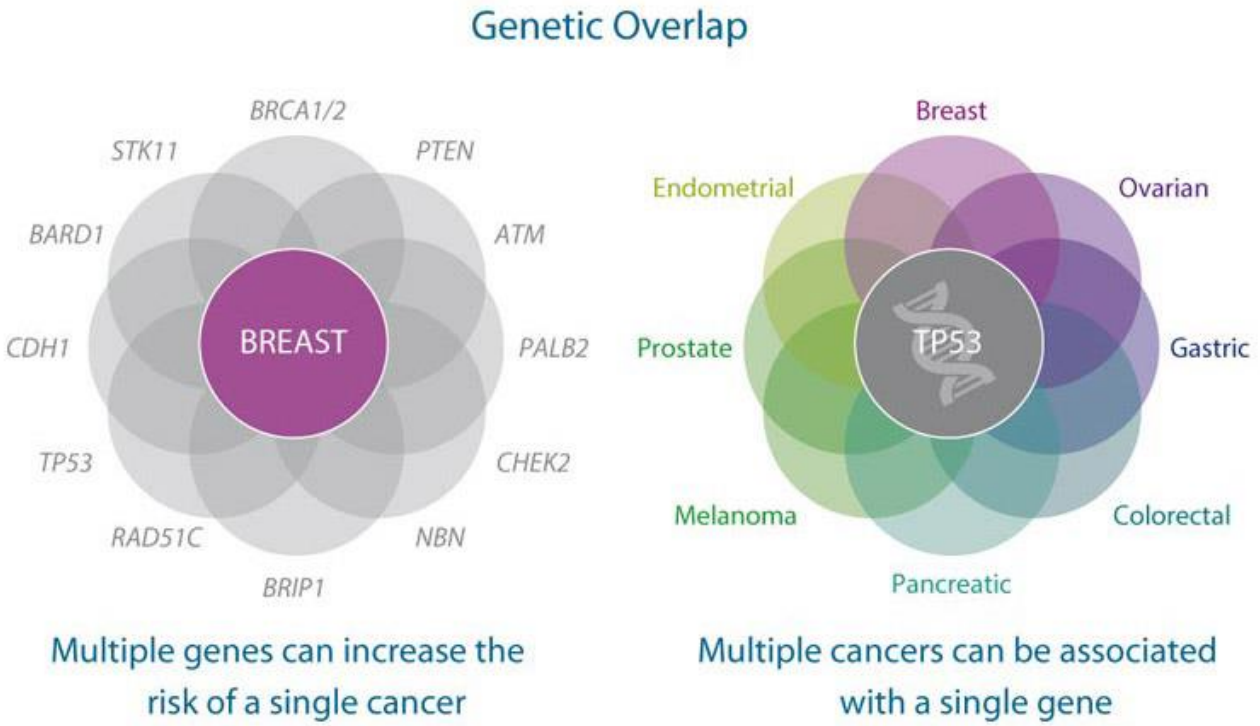
- OTHER breast cancer predisposition genes exist
  - We do test for them currently

Hereditary Breast Cancer: BRCA1/2

Features of HBOC:

- Breast cancer <age 35
- Ovarian cancer (non-mucinous, any age)
- Bilateral breast cancer or breast & ovarian cancer in same person
- Multiple cases of breast, ovarian, prostate, and/or pancreatic cancer in *close* relatives, on same side of family
- Breast cancer in males
- Breast or ovarian cancer in an Ashkenazi Jewish family
- A family with a confirmed *BRCA1* or *BRCA2* mutation

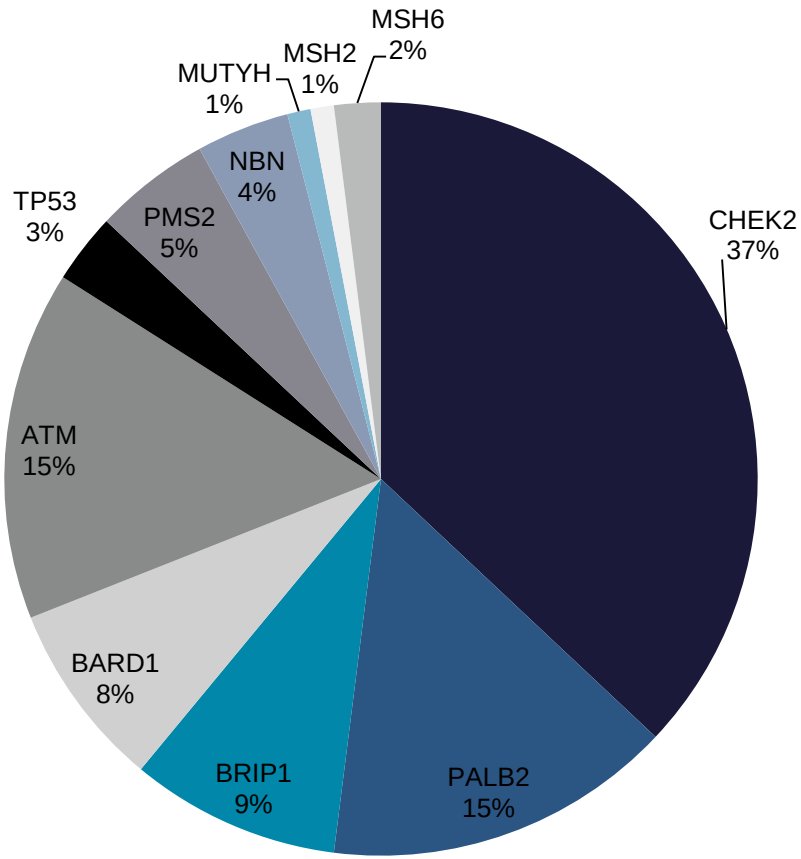
Multigene Panels



Source: http://www.myriadgenetics.eu/wp-content/uploads/2013/07/myRisk21.jpg

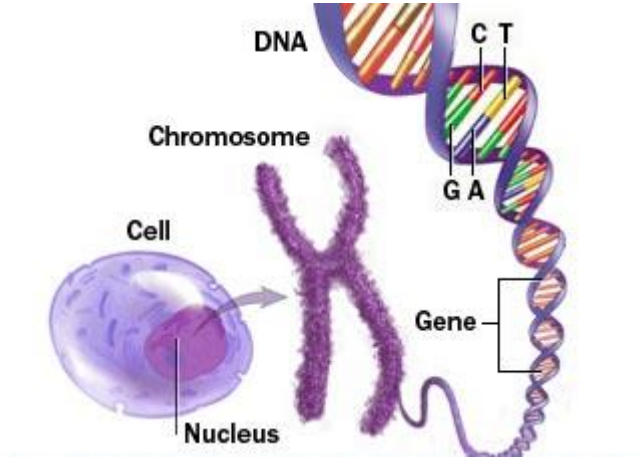
New approaches in breast cancer genetics

- 1781 hereditary breast cancer families
- 25-Gene Panel (MyRisk)
  - 13.5% had a mutation
    - 9.3%: BRCA1 or BRCA2
    - 4.2%: at least one other gene
  - 41% VUS rate



Allen, Tung et al ACMG 2014

Genetic Testing



Lifetime risk by cancer type	General population	BRCA1/2 gene mutation carrier	
		female	male
Breast cancer	11% or 1 in 9	45-65%	1.2-6.8%
Second primary breast cancer	-	47%	
Ovarian* cancer	1-2% or 1 in 70	11-39%	-
Pancreatic cancer	1% or 1 in 100	7% (BRCA2-specific)	
Prostate cancer	13% or 1 in 8	-	23-60%
Melanoma, bile duct, etc.	Variable	Increased above general population	

**Including primary peritoneal and fallopian tube cancer*

Management of BRCA1/2 carriers: Females

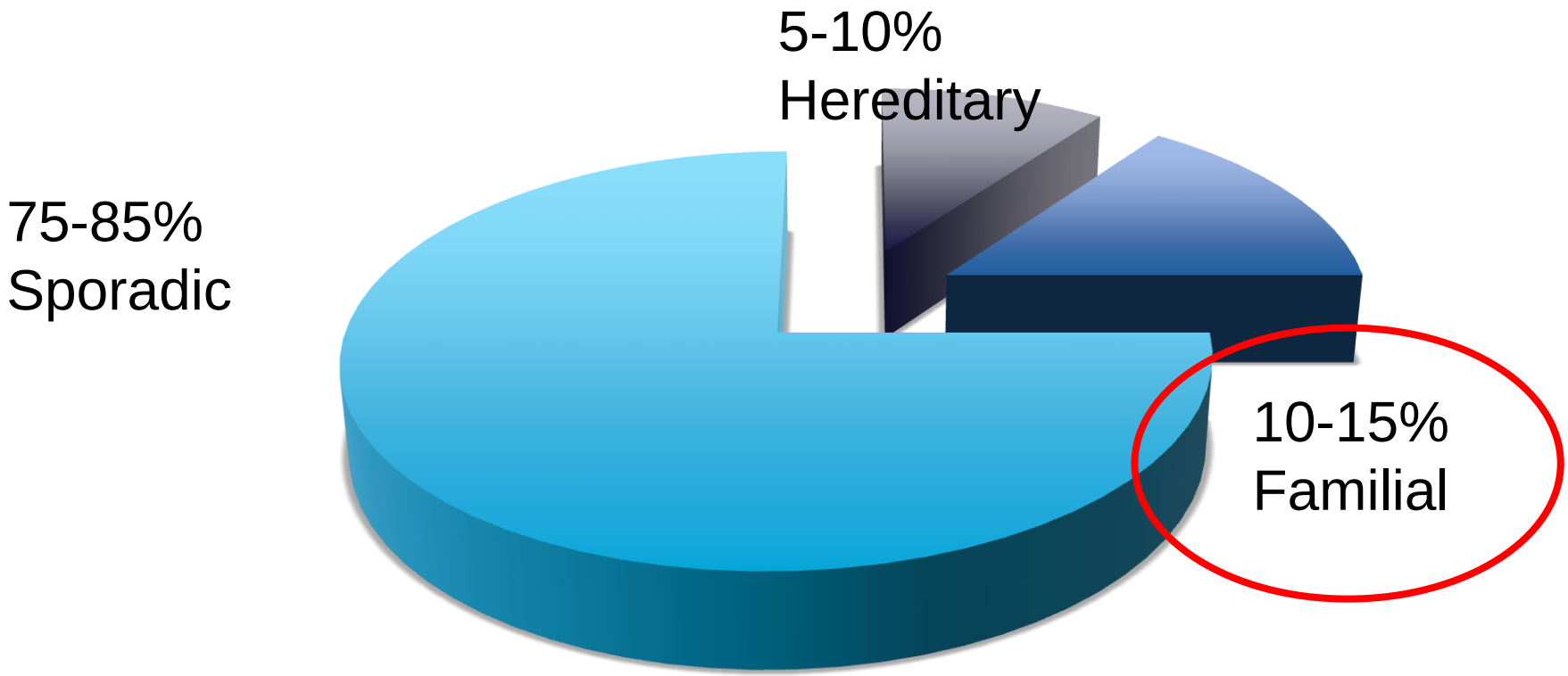
Intervention	Recommendation
Breast awareness	Starting at age 18y
Clinical breast exam	q.6-12mo, starting at age 25y
Breast MRI	q.12 mo, starting at age 25y **(can start earlier if family history warrants)*
Mammogram	q.12 mo, starting at age 30y
Risk-reducing BSO**	No age consensus, but typically between 35 and 40, or on completion of child bearing
Risk-reducing mastectomy	Discussion options with patient as needed

Management of BRCA1/2 carriers: Males

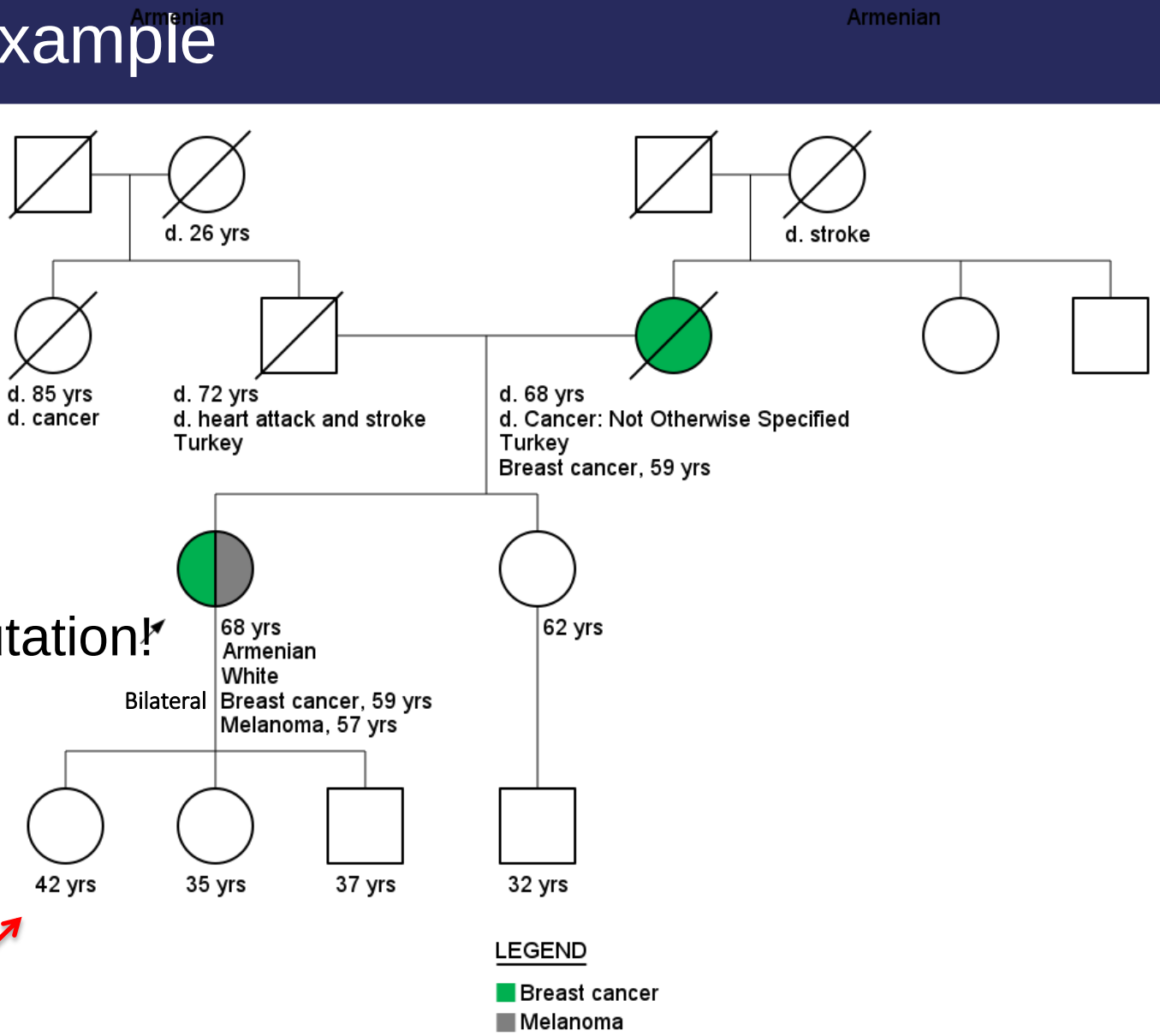
Intervention	Recommendation
Breast self-exam training and self-awareness	Starting at age 35y
Clinical breast exam	Starting at age 35y, q.12mo
Prostate cancer screening • Digital Rectal Examination • Prostate Specific Antigen (PSA)	Starting at age 40y
Physical examination	Starting at age 35. q.12mo

Other breast cancer susceptibility genes

Gene	Breast Cancer Risk and Management
CHEK2	**Increased risk of BC** • Screening: Annual mammogram and consider breast MRI with contrast age 40y • RRM: Evidence insufficient, manage based on family history
PALB2	**Increased risk of BC** • Screening: Annual mammogram and consider breast MRI with contrast age 30y • RRM: Consider based on family history


5-10%
Hereditary
75-85%
Sporadic
10-15%
Familial

Case Example



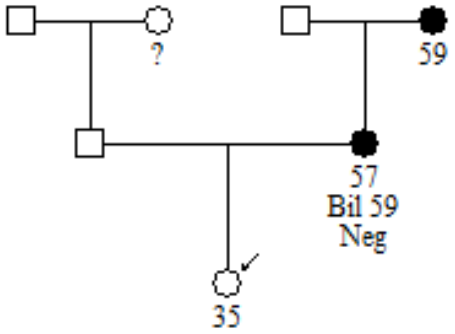
No gene mutation!

Case Example

ID: 2804
Age is 35-yrs.
Age at menarche 14-yrs.
Age at first birth 31-yrs.
Premenopausal.
Height is 1.55 m.
Weighs 77 kg.
Never used HRT.

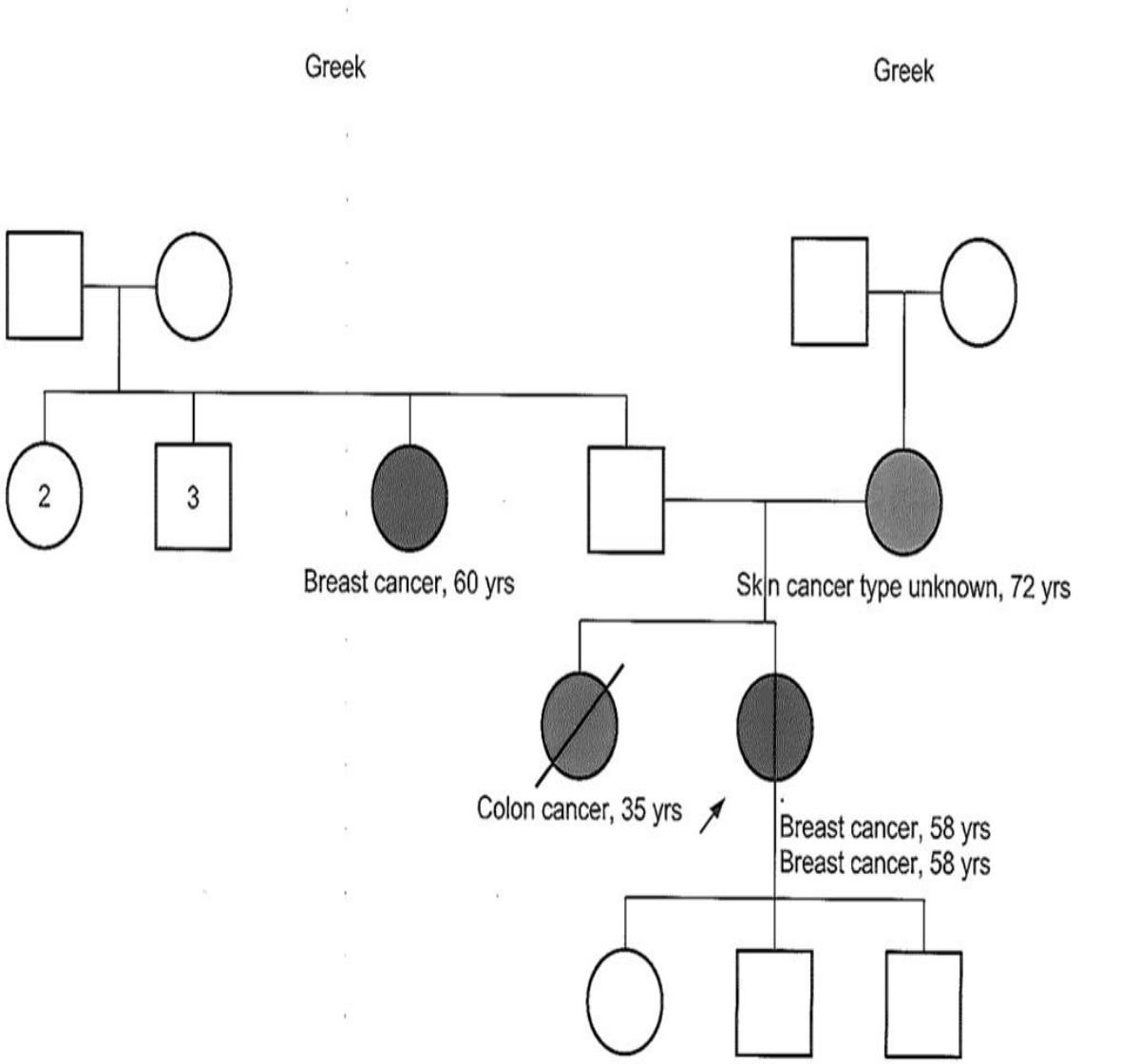
Risk after 45 years is 26.6%.
45 year population risk is 11.3%.
Lifetime risk is 30.7%.
Lifetime population risk is 13.2%.
Probability of a BRCA1 gene is 0.03%.
Probability of a BRCA2 gene is 0.07%.

- Eligible for OBSP high risk program
- Begin annual Mammograms and breast MRIs

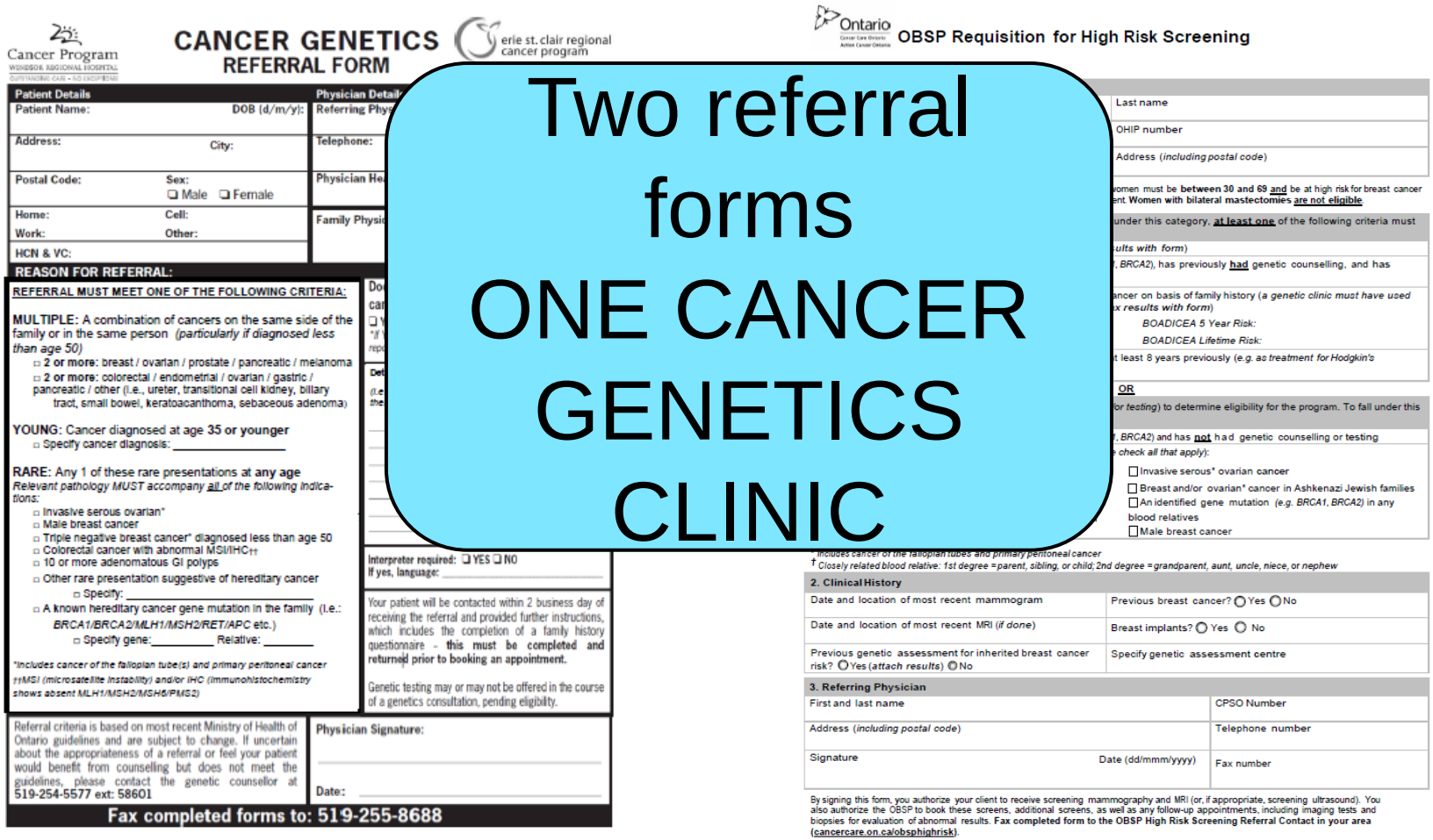


Case Example #2:

- Bilateral breast cancer at age 58
- +FHx of BrCa and colon Ca
- No gene mutation identified - BRCA1/2 normal
- Further testing revealed CHEK2 mutation



Ways to access genetics services in our area



CANCER GENETICS
REFERRAL FORM

erie st. clair regional cancer program

Cancer Program
WINDSOR REGIONAL HOSPITAL

Patient Details
Patient Name: DOB (d/m/y):
Address: City:
Postal Code: Sex: ❑ Male ❑ Female
Home: Cell:
Work: Other:
HCN & VC:

Physician Details
Referring Phys
Telephone:
Physician He
Family Physi

REASON FOR REFERRAL:

REFERRAL MUST MEET ONE OF THE FOLLOWING CRITERIA:

MULTIPLE: A combination of cancers on the same side of the family or in the same person *(particularly if diagnosed less than age 50)*
- ❑ **2 or more:** breast / ovarian / prostate / pancreatic / melanoma
- ❑ **2 or more:** colorectal / endometrial / ovarian / gastric / pancreatic / other (i.e., ureter, transitional cell kidney, biliary tract, small bowel, keratoacanthoma, sebaceous adenoma)

YOUNG: Cancer diagnosed at age **35 or younger**
- ❑ Specify cancer diagnosis: _____

RARE: Any 1 of these rare presentations at **any age**
Relevant pathology MUST accompany all of the following indications:
- ❑ Invasive serous ovarian*
- ❑ Male breast cancer
- ❑ Triple negative breast cancer* diagnosed less than age 50
- ❑ Colorectal cancer with abnormal MSI/IHC††
- ❑ 10 or more adenomatous GI polyps
- ❑ Other rare presentation suggestive of hereditary cancer
  - ❑ Specify: _____
- ❑ A known hereditary cancer gene mutation in the family (i.e.: *BRCA1/BRCA2/MLH1/MSH2/RET/APC* etc.)
  - ❑ Specify gene:_____ Relative: _____

*Includes cancer of the fallopian tube(s) and primary peritoneal cancer
††MSI (microsatellite instability) and/or IHC (immunohistochemistry shows absent MLH1/MSH2/MSH6/PMS2)

Interpreter required: ❑ YES ❑ NO
If yes, language: _____

Your patient will be contacted within 2 business day of receiving the referral and provided further instructions, which includes the completion of a family history questionnaire - **this must be completed and returned prior to booking an appointment.**

Genetic testing may or may not be offered in the course of a genetics consultation, pending eligibility.

Referral criteria is based on most recent Ministry of Health of Ontario guidelines and are subject to change. If uncertain about the appropriateness of a referral or feel your patient would benefit from counselling but does not meet the guidelines, please contact the genetic counsellor at 519-254-5577 ext: 58601

Physician Signature:

Date:

Fax completed forms to: 519-255-8688

Two referral forms
ONE CANCER GENETICS CLINIC

Ontario
Cancer Care Ontario
Action Cancer Ontario

OBSP Requisition for High Risk Screening

Last name
OHIP number
Address (*including postal code*)

…omen must be **between 30 and 69 and** be at high risk for breast cancer …ent. **Women with bilateral mastectomies are not eligible**

…under this category, **at least one** of the following criteria must …

…*ults with form*)
…, *BRCA2*), has previously **had** genetic counselling, and has …

…ancer on basis of family history (*a genetic clinic must have used …x results with form*)
BOADICEA 5 Year Risk:
BOADICEA Lifetime Risk:
…t least 8 years previously (*e.g. as treatment for Hodgkin's*

OR

…*or testing*) to determine eligibility for the program. To fall under this …

…, *BRCA2*) and has **not** had genetic counselling or testing
…*check all that apply*):
- ☐ Invasive serous* ovarian cancer
- ☐ Breast and/or ovarian* cancer in Ashkenazi Jewish families
- ☐ An identified gene mutation (*e.g. BRCA1, BRCA2*) in any blood relatives
- ☐ Male breast cancer

*Includes cancer of the fallopian tubes and primary peritoneal cancer
† Closely related blood relative: 1st degree =parent, sibling, or child; 2nd degree =grandparent, aunt, uncle, niece, or nephew

2. Clinical History

Date and location of most recent mammogram	Previous breast cancer? ◯ Yes ◯ No
Date and location of most recent MRI (*if done*)	Breast implants? ◯ Yes ◯ No
Previous genetic assessment for inherited breast cancer risk? ◯ Yes (*attach results*) ◯ No	Specify genetic assessment centre

3. Referring Physician

First and last name	CPSO Number
Address (*including postal code*)	Telephone number
Signature Date (dd/mmm/yyyy)	Fax number

By signing this form, you authorize your client to receive screening mammography and MRI (or, if appropriate, screening ultrasound). You also authorize the OBSP to book these screens, additional screens, as well as any follow-up appointments, including imaging tests and biopsies for evaluation of abnormal results. **Fax completed form to the OBSP High Risk Screening Referral Contact in your area (cancercare.on.ca/obsphighrisk).**

Cancer Genetics services in our area

Cancer Genetics Program

- **Females and males of all ages**
- Suggestive family history of **any form** of hereditary cancer

OBSP High Risk

- Women between ages of **30-69**
- Suggestive history of hereditary **breast and ovarian cancer** (see referral form)
- No acute breast symptoms
- Has NOT had a bilateral mastectomy

- Valid OHIP, Ontario Resident
- May or may not qualify for genetic testing
- May or may not qualify for high risk breast cancer screening
  - MM and brMRI

Cancer Genetics Referral: A closer look

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- ☐ Triple negative breast cancer* diagnosed less than age 50
- ☐ Colorectal cancer with abnormal MSI/IHC††
- ☐ 10 or more adenomatous GI polyps
- ☐ Other rare presentation suggestive of hereditary cancer
  - ☐ Specify: _____
- ☐ A known hereditary cancer gene mutation in the family (i.e.: *BRCA1/BRCA2/MLH1/MSH2/RET/APC* etc.)
  - ☐ Specify gene: _____ Relative: _____

**Includes cancer of the fallopian tube(s) and primary peritoneal cancer*

††MSI (microsatellite instability) and/or IHC (immunohistochemistry shows absent MLH1/MSH2/MSH6/PMS2)

When in doubt, call Veronica at 519-254-5577 ext: 58601

Fax to 519-255-8688

Ontario Breast Screening Program

HIGH RISK SCREENING

Annual Breast MRI and Mammography as early as age 30

Category A

- Booked for MRI and mammogram without needing a genetics assessment

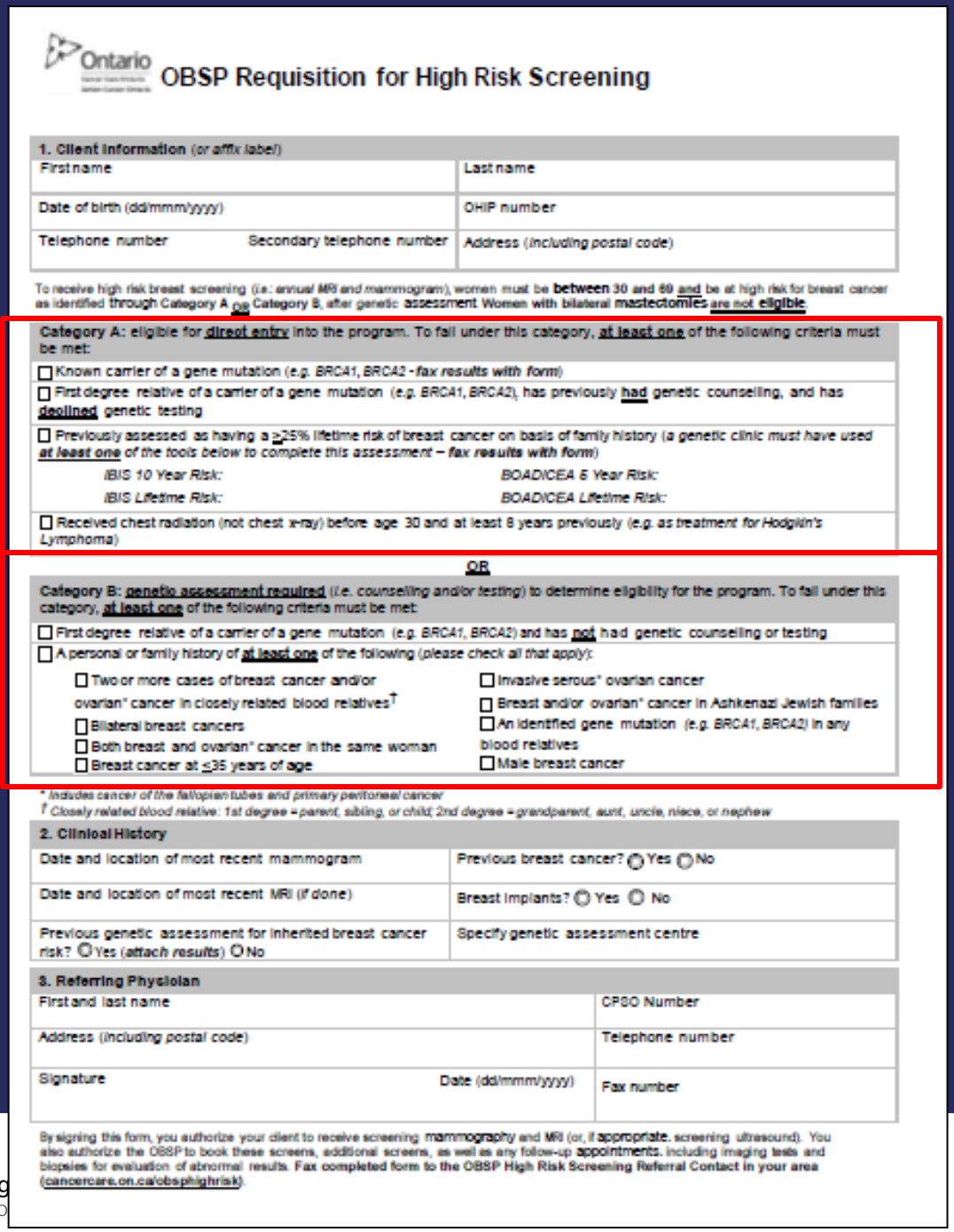
Category B

Referred to genetic counsellor for further assessment.

- Eligibility for MRIs is determined/coordinated

Category A

Category B



Ontario

OBSP Requisition for High Risk Screening

1. Client Information (or affix label)

First name	Last name
Date of birth (dd/mmm/yyyy)	OHIP number
Telephone number Secondary telephone number	Address (including postal code)

To receive high risk breast screening (i.e.: annual MRI and mammogram), women must be **between 30 and 69 and** be at high risk for breast cancer as identified through **Category A OR Category B**, after genetic assessment. **Women with bilateral mastectomies are not eligible.**

Category A: eligible for **direct entry** into the program. To fall under this category, **at least one** of the following criteria must be met:

- ☐ Known carrier of a gene mutation (*e.g. BRCA1, BRCA2 - fax results with form*)
- ☐ First degree relative of a carrier of a gene mutation (*e.g. BRCA1, BRCA2*), has previously **had** genetic counselling, and has **declined** genetic testing
- ☐ Previously assessed as having a ≥25% lifetime risk of breast cancer on basis of family history (*a genetic clinic must have used **at least one** of the tools below to complete this assessment – **fax results with form***)
  - *IBIS 10 Year Risk:*
  - *IBIS Lifetime Risk:*
  - *BOADICEA 5 Year Risk:*
  - *BOADICEA Lifetime Risk:*
- ☐ Received chest radiation (not chest x-ray) before age 30 and at least 8 years previously (*e.g. as treatment for Hodgkin's Lymphoma*)

OR

Category B: **genetic assessment required** (*i.e.* counselling and/or testing) to determine eligibility for the program. To fall under this category, **at least one** of the following criteria must be met:

- ☐ First degree relative of a carrier of a gene mutation (*e.g. BRCA1, BRCA2*) and has **not** had genetic counselling or testing
- ☐ A personal or family history of **at least one** of the following (*please check all that apply*):
  - ☐ Two or more cases of breast cancer and/or ovarian* cancer in closely related blood relatives†
  - ☐ Bilateral breast cancers
  - ☐ Both breast and ovarian* cancer in the same woman
  - ☐ Breast cancer at ≤35 years of age
  - ☐ Invasive serous* ovarian cancer
  - ☐ Breast and/or ovarian* cancer in Ashkenazi Jewish families
  - ☐ An identified gene mutation (*e.g. BRCA1, BRCA2*) in any blood relatives
  - ☐ Male breast cancer

* *Includes cancer of the fallopian tubes and primary peritoneal cancer*

† *Closely related blood relative: 1st degree = parent, sibling, or child; 2nd degree = grandparent, aunt, uncle, niece, or nephew*

2. Clinical History

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Date and location of most recent MRI (if done)	Breast implants? ◯ Yes ◯ No
Previous genetic assessment for inherited breast cancer risk? ◯ Yes (*attach results*) ◯ No	Specify genetic assessment centre

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Signature Date (dd/mmm/yyyy)	Fax number

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OBSP Requisition for High Risk Screening

Category A: eligible for **direct entry** into the program. To fall under this category, **at least one** of the following criteria must be met:

- ☐ Known carrier of a gene mutation (*e.g. BRCA1, BRCA2 - fax results with form*)
- ☐ First degree relative of a carrier of a gene mutation (*e.g. BRCA1, BRCA2*), has previously **had** genetic counselling, and has **declined** genetic testing
- ☐ Previously assessed as having a ≥25% lifetime risk of breast cancer on basis of family history (*a genetic clinic must have used **at least one** of the tools below to complete this assessment – **fax results with form***)

  IBIS 10 Year Risk: *BOADICEA 5 Year Risk:*

  IBIS Lifetime Risk: *BOADICEA Lifetime Risk:*

- ☐ Received chest radiation (not chest x-ray) before age 30 and at least 8 years previously (*e.g. as treatment for Hodgkin's Lymphoma*)

OBSP Requisition for High Risk Screening

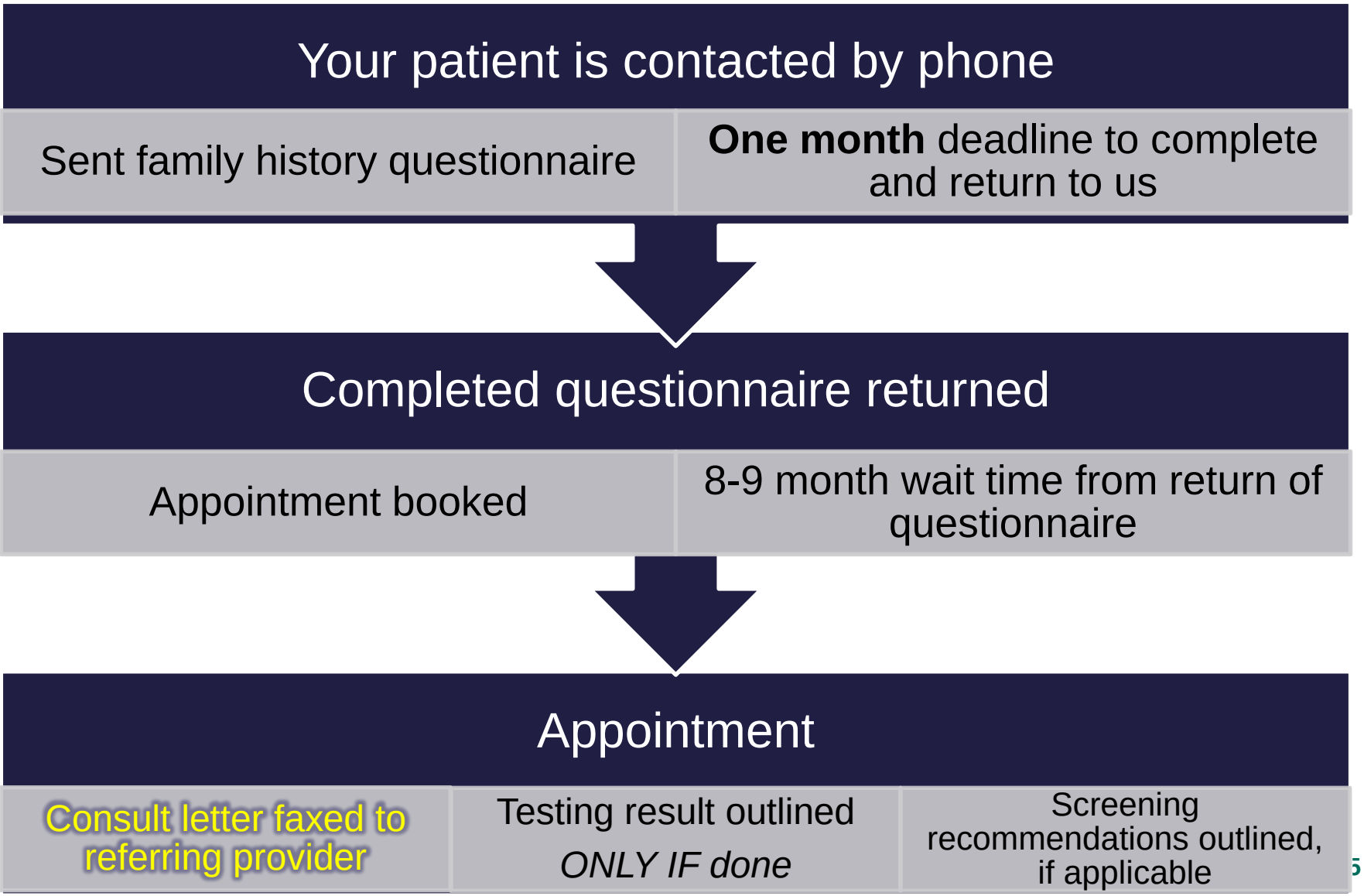
Category B: genetic assessment required (*i.e. counselling and/or testing*) to determine eligibility for the program. To fall under this category, **at least one** of the following criteria must be met:

- [x] First degree relative of a carrier of a gene mutation (*e.g. BRCA1, BRCA2*) and has **not** had genetic counselling or testing
- [x] A personal or family history of **at least one** of the following (*please check all that apply*):
  - [ ] Two or more cases of breast cancer and/or ovarian* cancer in closely related blood relatives†
  - [ ] Bilateral breast cancers
  - [ ] Both breast and ovarian* cancer in the same woman
  - [x] Breast cancer at ≤35 years of age
  - [ ] Invasive serous* ovarian cancer
  - [ ] Breast and/or ovarian* cancer in Ashkenazi Jewish families
  - [ ] An identified gene mutation (*e.g. BRCA1, BRCA2*) in any blood relatives
  - [ ] Male breast cancer

* *Includes cancer of the fallopian tubes and primary peritoneal cancer*

† *Closely related blood relative: 1st degree = parent, sibling, or child; 2nd degree = grandparent, aunt, uncle, niece, or nephew*

After the referral: Genetics or Category B OBSP



Cancer Genetics Program: History

- Established in 2013 to increase access to cancer genetics services in the ESC LHIN
  - In-person appointments, WRCC
  - Phone appointments during and after business hours (8am-4pm)

- Medical Genetics affiliation LHSC
  - Monthly cancer geneticist clinic

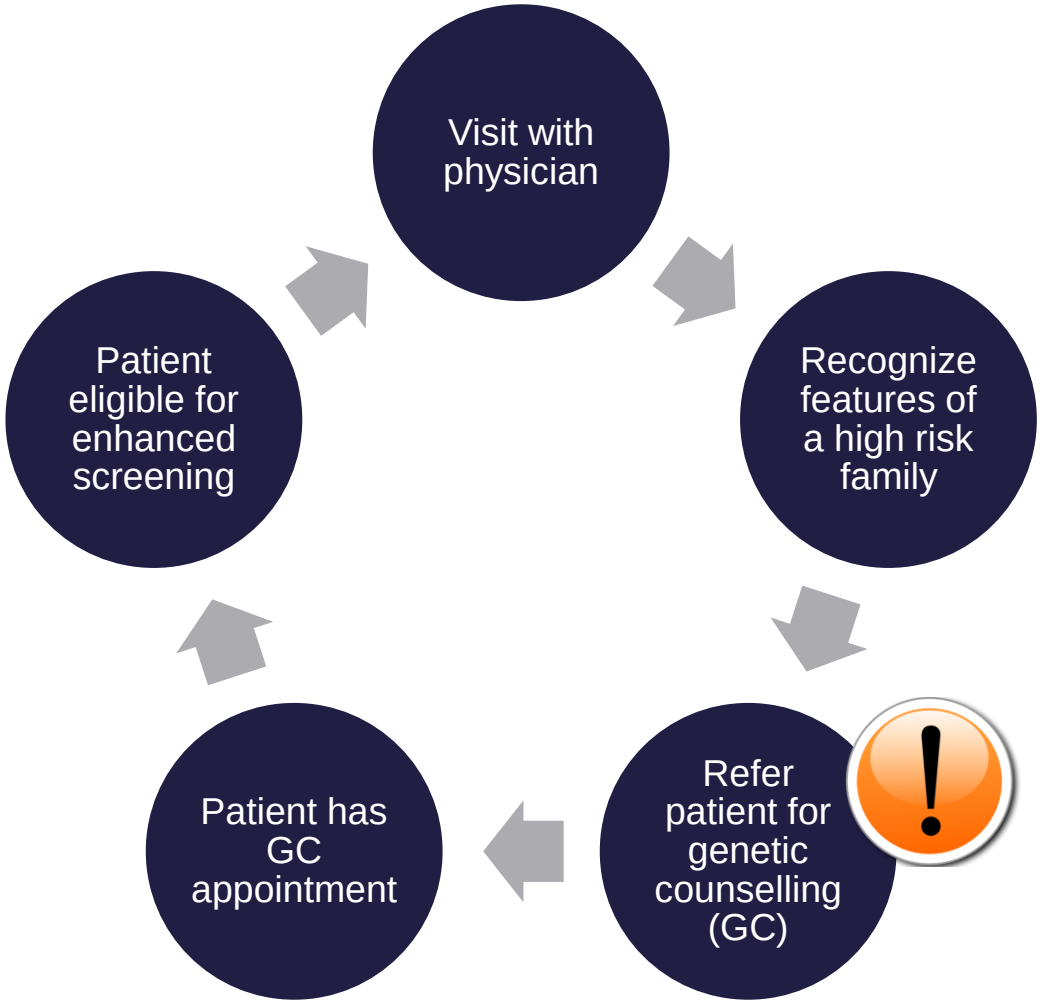
Veronica Bryksa, MS, MS, CGC, CCGC

Elana Wishnefsky, MS, CGC

Jack Jung, MD, FRCPC, FCCMG

Your important role as PCPs

Recognize
Educate
Refer
Coordinate screening



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Regional Cancer Program
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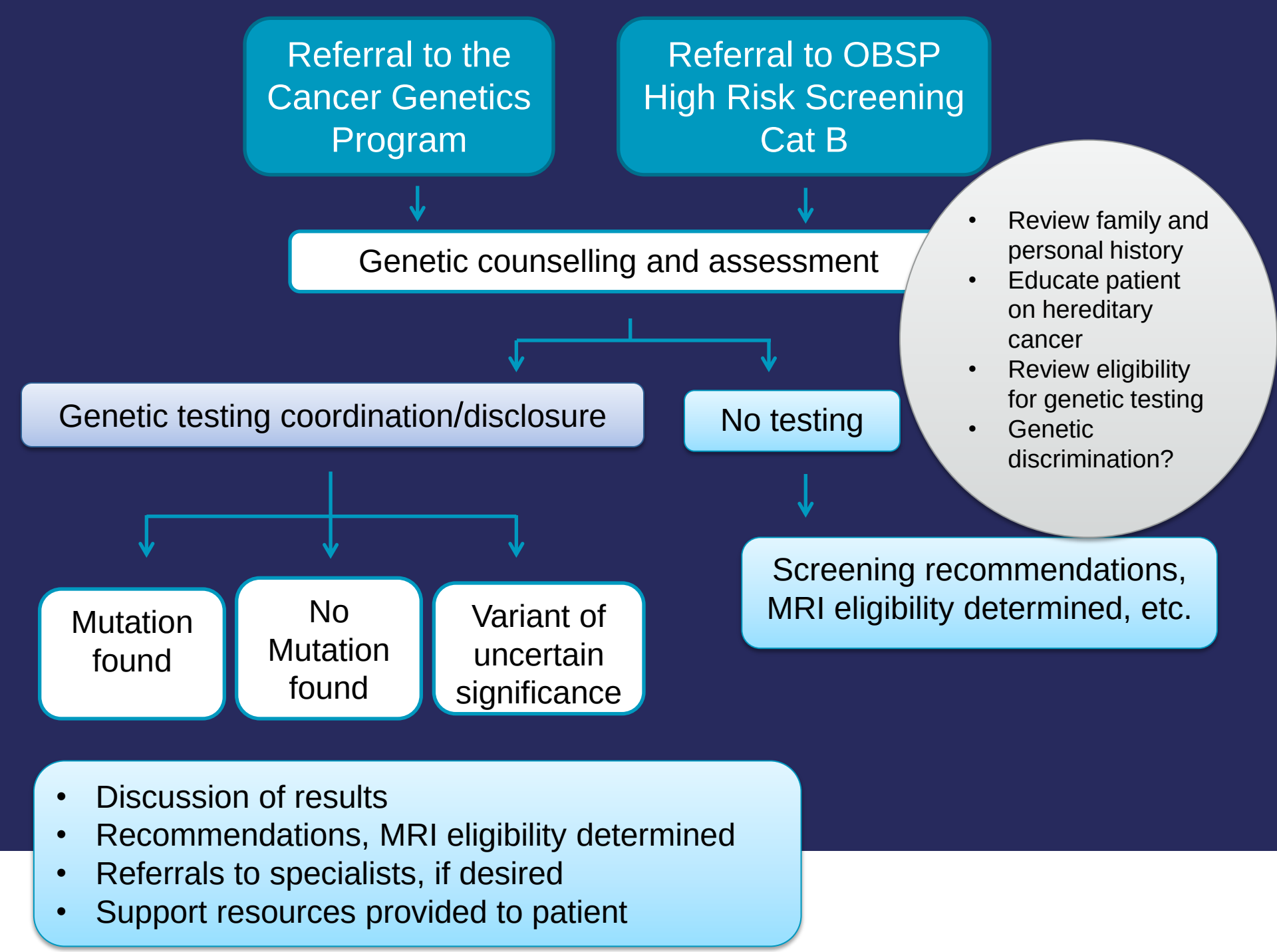
THANKS!

QUESTIONS??

Call us!! 519-254-5577

- Veronica/Genetic Counsellor: x58601
- Dina/Genetics Secretary: x58620


Referral to the Cancer Genetics Program
Referral to OBSP High Risk Screening Cat B
Genetic counselling and assessment
Review family and personal history
Educate patient on hereditary cancer
Review eligibility for genetic testing
Genetic discrimination?
Genetic testing coordination/disclosure
No testing
Mutation found
No Mutation found
Variant of uncertain significance
Screening recommendations, MRI eligibility determined, etc.
Discussion of results
Recommendations, MRI eligibility determined
Referrals to specialists, if desired
Support resources provided to patient