



Counseling Guideline

Genetic counseling for hereditary breast and ovarian cancer





Motivation



What brings you to
genetic counseling
today?





Stages of consultation

1. Genetic background
2. Family pedigree creation
3. Decision-making
 - medical indication
 - personal decision
 - possible consequences





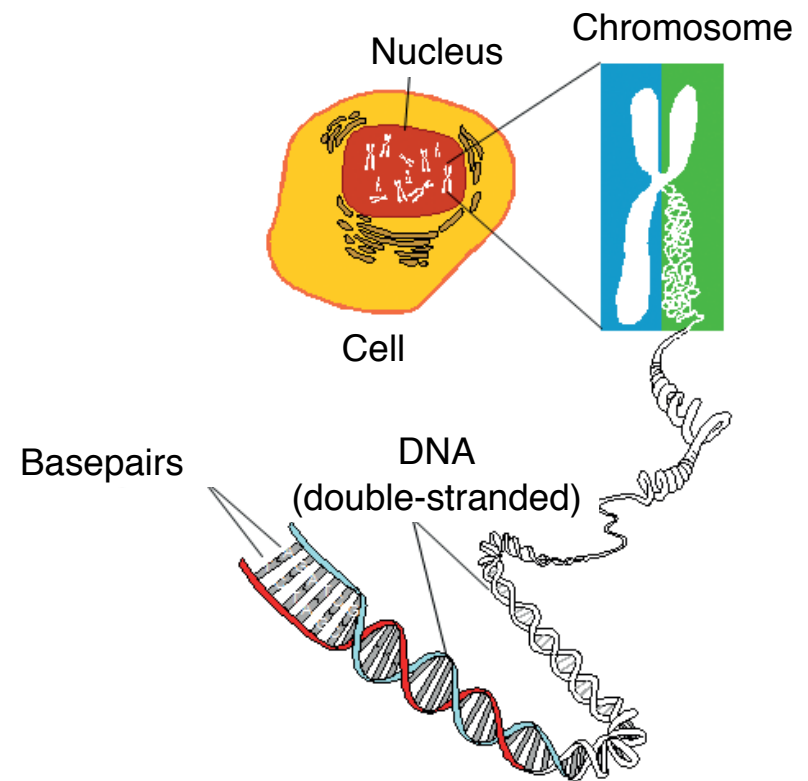
Breast and Ovarian Cancer

- One in eight women will develop breast cancer over the course of her lifetime (lifetime risk: 12.5%)
- One out of every 70 woman will develop ovarian cancer at some point in her lifetime (lifetime risk: 1-2%)



Genetic Background

- The complete genetic blueprint for your body is found in the nucleus of each cell in the form of DNA
- DNA = deoxyribonucleic acid (deoxyribonucleic acid)
- Gene: Sections of the DNA that determine features of the human body, e.g. blood type, eye color



BRCA1 and BRCA2

Since 1994 (*BRCA1*) and 1995 (*BRCA2*) were discovered



BRCA1 = BReast CAncer 1

BRCA2 = BReast CAncer 2

- *Repair genes
(Tumor suppressor genes)*
- *They are very important to avoid cancer*
- *Everyone has it (women and men!)*





BRCA1 and *BRCA2*

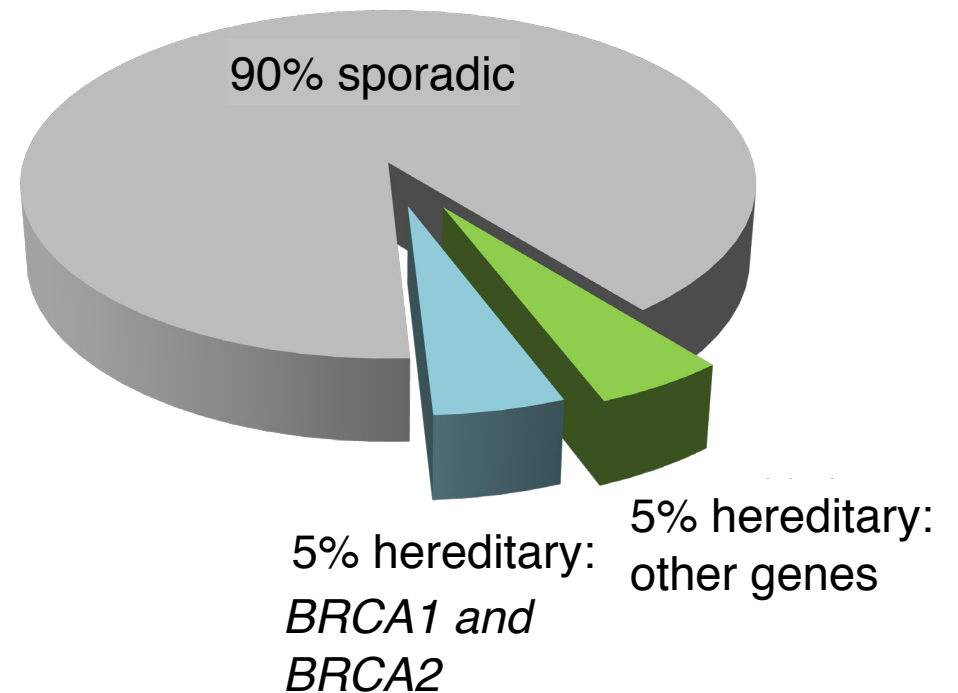
What happens with a change (= mutation) in one of these genes?

- Gene modification is innate
- Impaired repair function
 - Strong risk increase for breast and ovarian cancer (and low risk of other cancers)



Genetic breast cancer

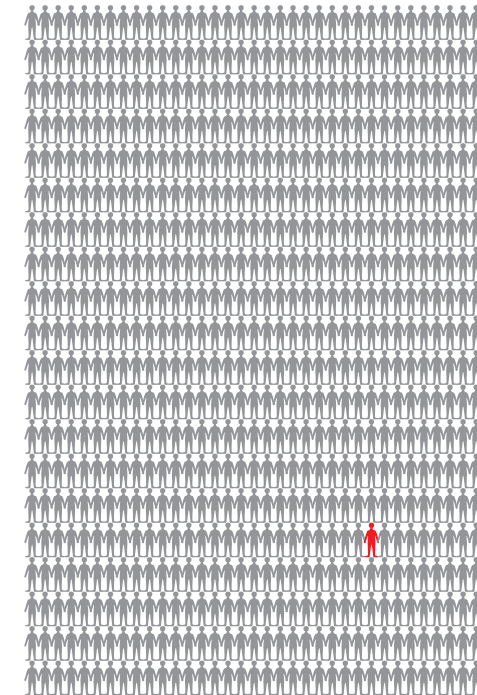
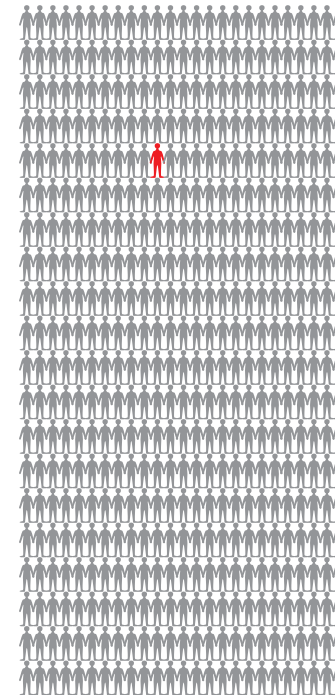
- 90-95% sporadic breast cancer
- 5-10% of all breast cancers are inherited, the causes are genetic changes



Frequency of BRCA1 / 2 mutations in the population

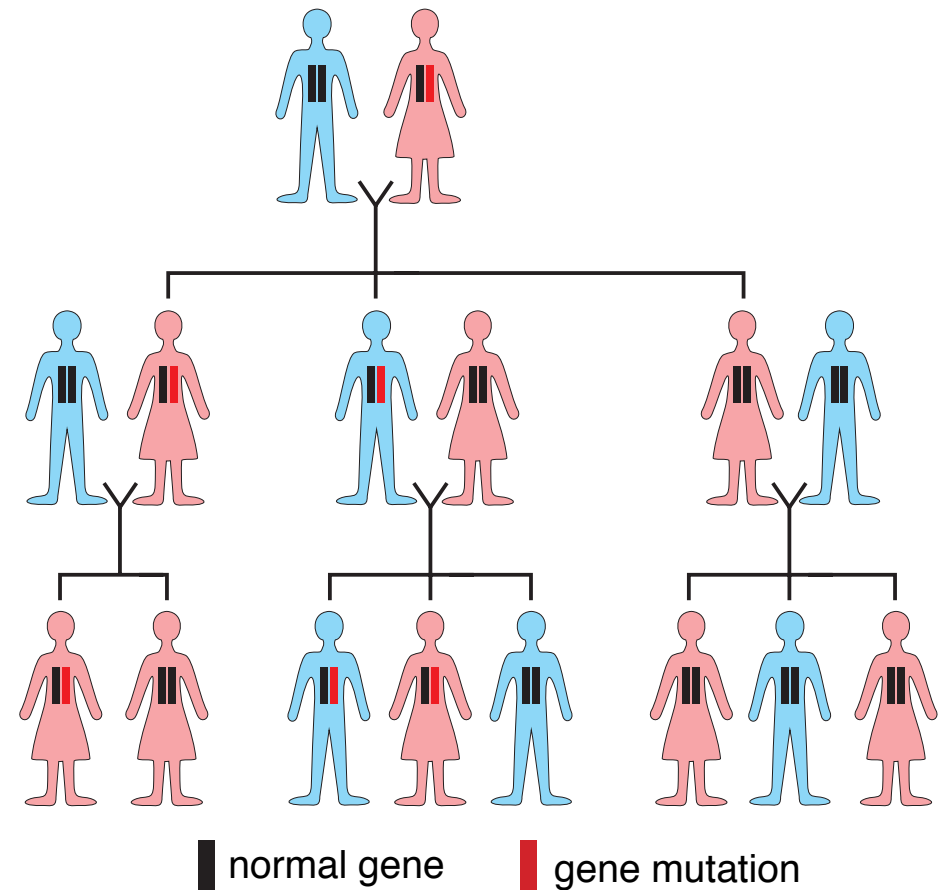
Rare!

- BRCA1:
1 out of 500 people affected
- BRCA2:
1 of 700 people affected

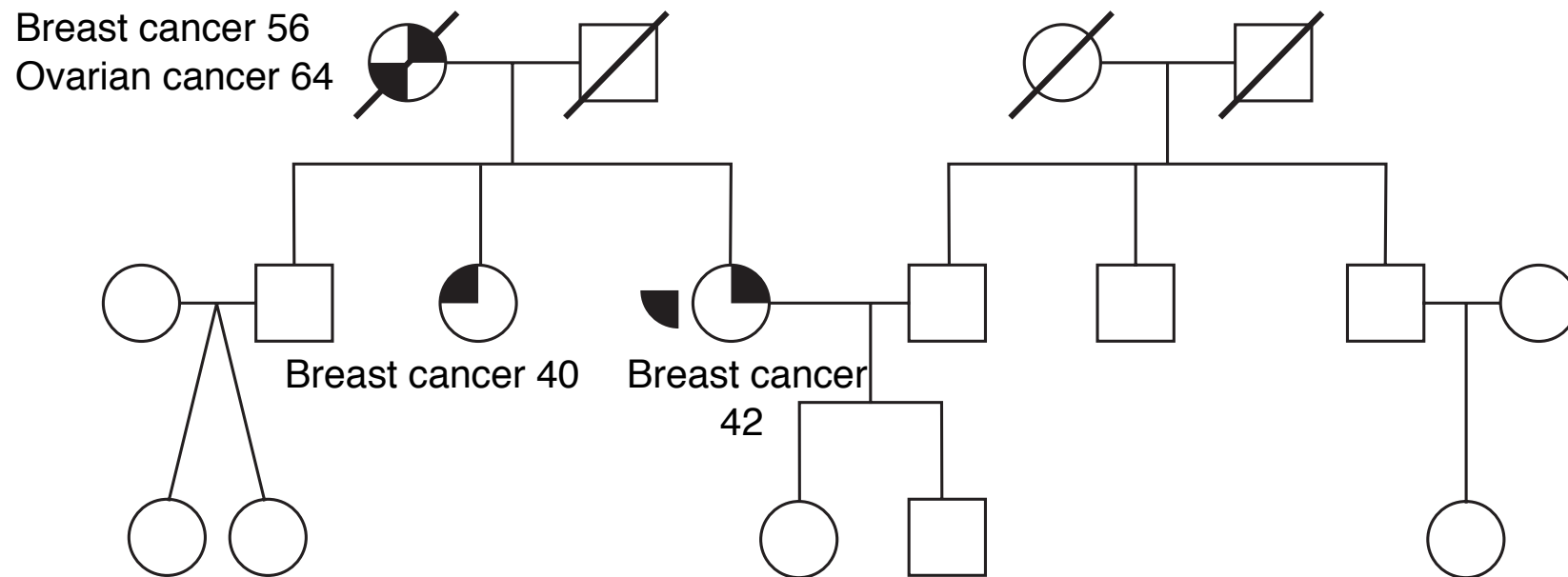


Heredity

- 50% chance of inheriting the altered gene → it is the same for every child
- Regardless of gender
- Non-carriers cannot pass on genetic change to children
→ the altered gene cannot skip a generation!



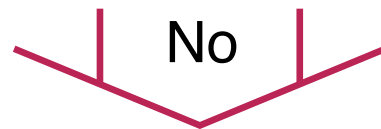
Create your family tree



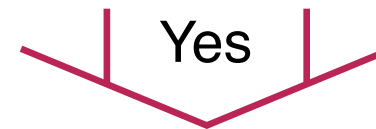


Medical Indication

- Maternal or paternal line: type and number of cancers, age of onset
- Presence of a familial high-risk constellation?



Not high risk;
No genetic testing necessary



High risk;
Offer genetic testing





Possible results I

(if no genetic mutation was found in the family)

Results	Risk
no mutation	unknown
mutation	BRCA1: 85% BC, 53% OC BRCA2: 84% BC, 27% OC
unclassified variant	unknown





Possible results II

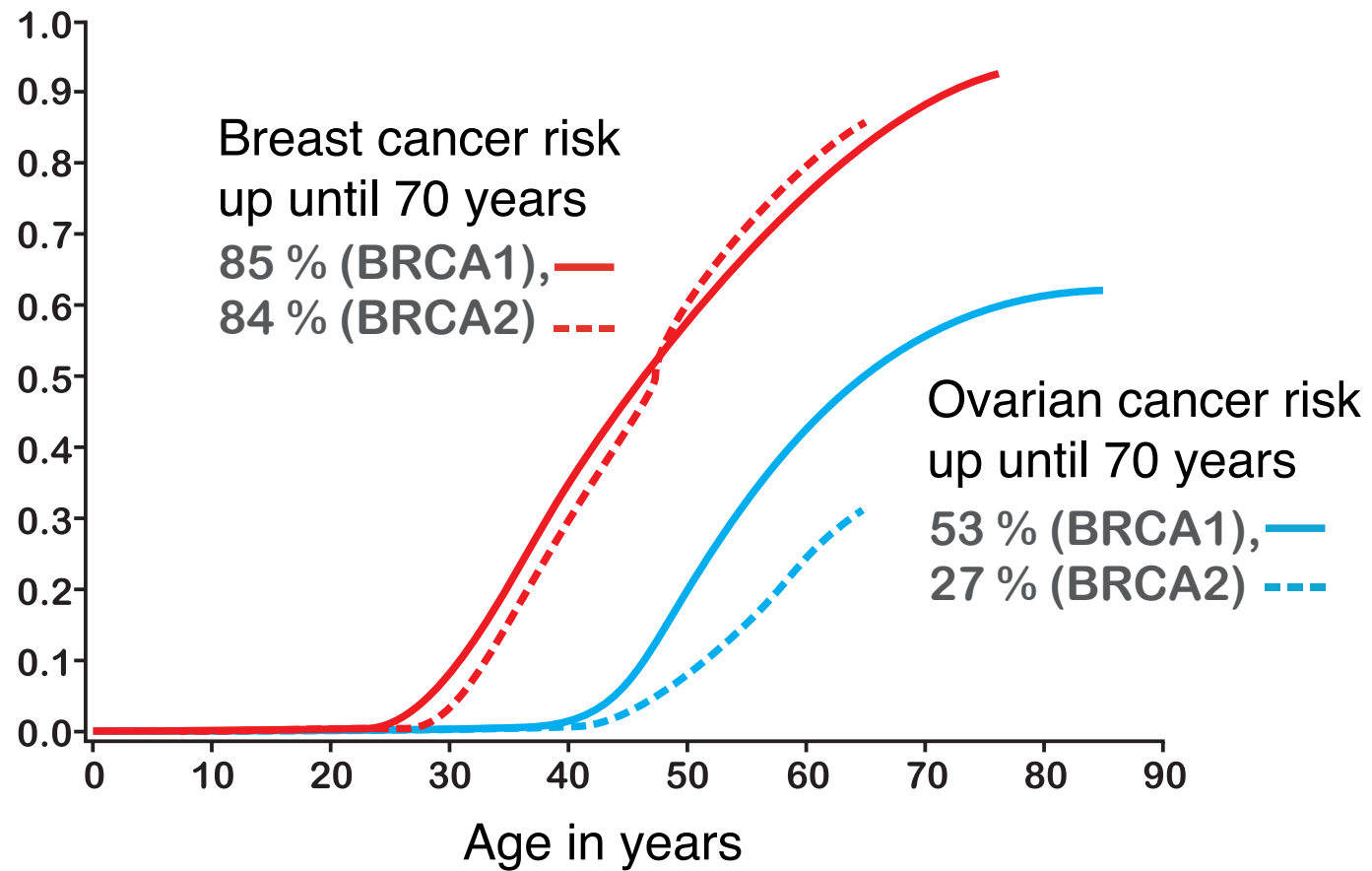
(if genetic mutation was found in the family)

Results	Risk
no mutation	General population
mutation	BRCA1: 85% BC, 53% OC BRCA2: 84% BC, 27% OC
unclassified variant	unknown



BRCA disease risk

Disease risk





Possible medical and psychosocial consequences

Early detection

Prevention



Screening program

Type of investigation	From 18y	From 25y	From 35y	if necessary
Gynecological check	Once a year			
Medical breast examination	Once a year			
Breast ultrasound				x
Mammography			Once a year	
Breast MRI		Once a year		
Vaginal ultrasound			Once a year	
Tumor marker			Once a year	

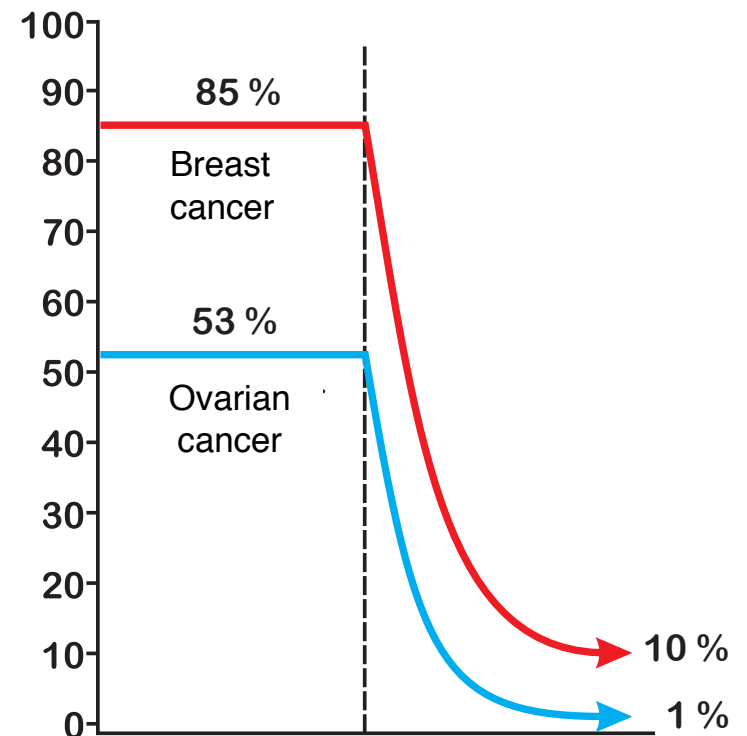
Guideline of the Austrian Society for Senology, Austrian Society for Obgyn., Austrian Society for Surgery, Austrian Society for Radiology



Preventive Surgeries

- Preventive removal of the breast tissue (prophylactic mastectomy)
- Preventive removal of ovaries and fallopian tubes (prophylactic ovariectomy, prophylactic bilateral salpingo-oophorectomy)

Risk of developing cancer, %





Necessary considerations before an investigation:

- How would you integrate an unfavorable result into your life?
- Which consequences would you presumably draw (e.g. early detection / preventive surgeries etc.)?
- Who would you tell about the gene mutation?
- How would you handle the information that your children might be affected by the hereditary disease risk?





Further process

- Invitation to disclosure of test results
- Right to know i.e. to decide if you want to know the result or not





For more information:

www.brustgenberatung.at



ZENTRUM für
Familiären Brust-
und Eierstockkrebs