Hereditary Breast and Ovarian Cancer: BRCA1/BRCA2
CLINICAL PATHWAY

*The **Patient Clinical Pathway** is "the whole care pathway from identification, diagnostics, and multidisciplinary case discussions to surveillance and preventive surgery", so indeed a pathway in time, focusing on **HOW***

Periodic Review Recommended

HBOC is associated with a high lifetime risk of breast cancer (50-80%) and an increased risk of ovarian cancer, 20-60% (usually of a high grade serous type). Female carriers who have had breast cancer, have an increased risk of contralateral breast cancer.
Males with BRCA2 mutations have an increased risk of prostate cancer.

At time of diagnosis ALL patients (including carriers) should be seen in a genetics department for genetic counselling.

Periodic review should be undertaken by a specialist in BRCA1/2 (oncologist, surgeon, clinical geneticist, gynaecologist). Surveillance should be continued until at least 74 years of age and may be continued further after individual assessment

HBOC
Review Checklist—Adults (25+)

	WHAT TO LOOK FOR	WHEN TO REFER
BREASTS	MRI-breast from age 25-30y until 50-70y (upper limit depending on breast density and other clinical factors) Addition of annual mammogram from age 40 Addition of ultrasound should be considered if MRI is not possible or if requested by the radiologist Information on the possibility of prophylactic mastectomy including the pros (decreased risk of cancer) and cons (risk of surgery, cosmetic etc). Discussion at multidisciplinary team consisting of at least a representative from clinical genetics, oncology, breast surgery and possibly also plastic surgeon, radiologist and gynaecologist	In case of an abnormal mammography or MRI of the breasts and if signs or symptoms associated with breast cancer, refer to breast centre for investigation
OVARIES	Information on prophylactic bilateral salpingo-oophorectomy (BSO) between the age of 35-45 years including the pros (highly reduced cancer risk) and cons (long and short term side effects). BSO to be performed from 35-40y for BRCA1 and 40-45y for BRCA2 carriers Post-surgery, hormone replacement therapy is given until the age of 45-50 years unless there is a contraindication	Refer to gynaecologist familiar with BRCA between the age of 30-40 years Refer to diagnostic unit investigation if signs or symptoms associated with ovarian cancer
PROSTATE#	In some EU countries men with a BRCA2 mutation are offered PSA measurements every year via a general practitioner from the age of 40-45 years	Refer to urologist if PSA levels are increased Refer to diagnostic unit if signs or symptoms associated with prostate cancer.

PSYCHOLOGICAL BURDEN	Psychological problems are common but patients, both men and women, may be reluctant to talk about these issues and need encouragement.	Consider referral to an appropriate counselling service
PREGNANCY	Pre-natal diagnosis is usually not requested, but pre-implantation testing (PGT) is available. PGT relies on pre-pregnancy genetic work up and that the family fulfils the requirements for IVF.	Carriers (both male and female) who are planning pregnancy should be referred to clinical genetics
ANY OTHER NEW SYMPTOMS	Consider other possible complications. If the patient develops new symptoms that may be due to cancer, be generous with investigations as there is a small increased risk for other tumour types such as pancreas cancer for BRCA2 carriers	Refer to appropriate specialist

Guidelines differ in EU countries because of low level of evidence on surveillance outcome

Hereditary Breast and Ovarian Cancer (HBOC) Clinical Pathway

European Reference Network for rare or low prevalence complex diseases
Network Genetic Tumour Risk Syndromes (ERN GENTURIS)

Faculty: .....

Family name:

Given name(s)

Address:

Date of Birth: Sex: ☐ M ☐ F ☐ I

Annual Review Recommended

HBOC is associated with a high lifetime risk of breast cancer (50-80%) and an increased risk of ovarian cancer, 20-60% (usually of a high grade serous type). Female carriers who have had breast cancer, have an increased risk of contralateral breast cancer. Males with BRCA2 mutations have an increased risk of prostate cancer. At time of diagnosis ALL patients (including carriers) should be seen in a genetics department for genetic counselling.

AGE	DIAGNOSTIC APPOINTMENT	ANNUAL HBOC REVIEWS CARRIED OUT BY
25 - 70	Annual imaging	Periodic review should be undertaken by a specialist in BRCA1/2 (oncologist, surgeon, clinical geneticist, gynaecologist, radiologist).

Review Checklist — Adults 25+

Clinical Presentation:

..... ☐

..... ☐

..... ☐

Other symptoms:

.....

.....

Genetic counselling completed ☐

Date Completed: .....

Clinical diagnosis

.....

Genetic Test '+'ve ☐

Diagnosis Date: .....

General Health Check:

Please record the follow as soon as possible and then annually:

Height

.....

Weight

.....

Blood Pressure

.....

Notes:

.....

.....

.....

.....

Doctor: .....

Review date: .....

Faculty: .....

www.genturis.eu

WHAT TO LOOK FOR	WHEN TO REFER
BREASTS: MRI-breast from age 25-30y until age 55y. Additional annual mammograms (≥40y), additional ultrasound (only if MRI is not possible or if requested by a radiologist). Information on the possibility of prophylactic mastectomy including the pros (decreased risk of cancer) and cons (risk of surgery, cosmetic etc).	In case of an abnormal mammography or MRI of the breasts and if signs or symptoms associated with breast cancer, refer to breast centre for investigation. ☐ **Date Referred**: .....
OVARIES: Prophylactic bilateral salpingo-oophorectomy (BSO) to be performed from 35-40y for BRCA1 and 40-45y for BRCA2 carriers. Post-surgery, hormone replacement therapy is given until the age of 45-50 years unless there is a contraindication. Information on prophylactic BSO between the age of 35-45 years including the pros (highly reduced cancer risk) and cons (long and short term side effects).	Refer to gynaecologist familiar with BRCA between the age of 30-40 years Refer to gynaecologist if signs or symptoms associated with ovarian cancer ☐ **Date Referred**: .....
PROSTATE#: In some EU countries men with a BRCA2 mutation are offered PSA measurements every year via a general practitioner from the age of 40 years. # Guidelines differ in EU countries because of low level of evidence on surveillance outcome	Refer to urologist if PSA levels are increased Refer to diagnostic unit if signs or symptoms associated with prostate cancer. ☐ **Date Referred**: .....
PSYCHOLOGICAL BURDEN: Psychological problems are common but patients, both men and women, may be reluctant to talk about these issues and need encouragement.	Consider referral to an appropriate counselling service ☐ **Date Referred**: .....
PREGNANCY: Pre-natal diagnosis is usually not requested, but pre-implantation testing (PGT) is available. PGT relies on pre-pregnancy genetic work up and that the family fulfils the requirements for IVF.	Carriers (both male and female) who are planning pregnancy should be referred to clinical genetics ☐ **Date Referred**: .....
ANY OTHER NEW SYMPTOMS: Consider other possible complications. If the patient develops new symptoms that may be due to cancer, be generous with investigations as there is a small increased risk for other tumour types such as pancreas cancer for BRCA2 carriers.	Refer to appropriate specialist ☐ **Date Referred**: .....