

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

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	НВОС				
Review Checklist—Adults (25+)					
	WHAT TO LOOK FOR	WHEN TO REFER			
	λ				
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).	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			
	Discussion at multidisciplinary team consisting of at least a representative from clinical genetics, oncology, breast surgery and possibly also plastic surgeon, radiologist and gynaecologist				
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			
		Refer to urologist if PSA levels are increased			
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 diagnostic unit if signs or symptoms associated with prostate cancer.			

Networks

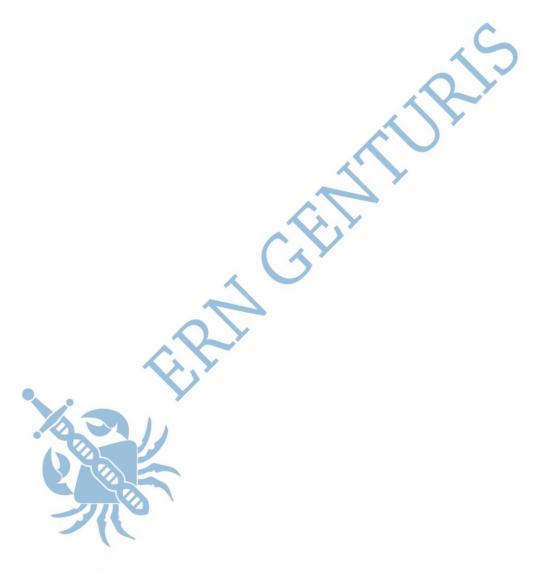


Genetic Tumour Risk Syndromes (ERN GENTURIS)



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	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 who are planning probe referred to clinical parts).	
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)	

	www.genturis.eu	
Family name:		
Given name(s)		
Address:		

Sex:

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Faculty:	

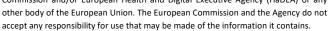
Annual Review Recommended

Date of Birth:

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 CA	RRIED 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:	General Health Check	WHAT TO LOOK FOR	WHEN TO REFER		
	Please record the follo soon as possible and the annually:	additional ultrasound (only if MRI is not possible or if requested by a radiologist).	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.		
Other symptoms:	Height	Information on the possibility of prophylactic mastectomy including the pros (decreased risl of cancer) and cons (risk of surgery, cosmetic etc).	□ Date Referred:		
Genetic counselling completed		OVARIES: Prophylactic bilateral salpingo- oophorectomy (BSO) to be performed from 35 40y for BRCA1 and 40-45y for BRCA2 carriers.	,		
Date Completed:	Weight	Post-surgery, hormone replacement therapy is given until the age of 45-50 years unless there is a contraindication.			
Clinical diagnosis	Blood Pressure	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).	☐ Date Referred:		
Diagnosis Date:		PROSTATE#: In some EU countries men with a BRCA2 mutation are offered PSA measurements every year via a general	Refer to urologist if PSA levels are increased		
Notes:		# Guidelines differ in EU countries because of low level of evidence on surveillance outcome	Refer to diagnostic unit if signs or symptoms associated with prostate cancer.		
		PSYCHOLOGICAL BURDEN: Psychological problems are common but patients, both mer and women, may be reluctant to talk about these issues and need encouragement.	Consider referral to an appropriate		
	1	PREGNANCY: Pre-natal diagnosis is usually no 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.	planning pregnancy should be referred to clinical genetics		
Doctor:		ANY OTHER NEW SYMPTOMS: Consider other	· · · · · · · · · · · · · · · · · · ·		
Review date:		possible complications. If the patient develops new symptoms that may be due to cancer, be			
Faculty: www.genturis.eu		generous with investigations as there is a sma increased risk for other tumour types such as			







pancreas cancer for BRCA2 carriers.



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