

Hereditary Breast and Ovarian Cancer syndrome: <i>BRCA1/BRCA2</i>		
CLINICAL PATHWAY		
<i>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</i>		
Periodic Review Recommended		
A woman with a <i>BRCA1</i> or <i>BRCA2</i> pathogenic variant has a high lifetime risk of breast cancer, up to 60-80% and an increased risk of ovarian cancer, up to 20-50%, usually of a high-grade serous type. Female carriers who have had breast cancer, have an increased risk for second primary breast cancer. Pancreatic, prostate cancer and male breast cancer risk is increased mainly in <i>BRCA2</i> carriers.		
All individuals identified as carriers of a pathogenic variant in <i>BRCA1/2</i> should be offered genetic counselling early in their patient journey.		
Periodic review should be undertaken by a specialist in <i>BRCA1/2</i> (oncologist, surgeon, clinical geneticist, gynaecologist). Surveillance should be continued until the person is in good health. Surveillance may depend on the level of the risk, which varies on which gene is involved, the family history of cancer and other non-genetic risk factors. Guidelines can differ in different EU countries. (Marmolejo et al, European Journal of Medical Genetics 2021, PMID 34606975)		
HBOC-<i>BRCA1/2</i>		
Review Checklist—Adults (25+)		
	WHAT TO LOOK FOR	WHEN TO REFER
BREASTS	<p>MRI-breast from age 25-30y until the woman is in good health. Addition of annual mammogram from age 40. Addition of ultrasound should be considered if MRI is not possible or if requested by the radiologist.</p> <p>Information on the possibility of risk reducing mastectomy including the pros (decreased risk of cancer) and cons (risk of surgery, cosmetic etc).</p> <p>Discussion at multidisciplinary team consisting of at least a representative from clinical genetics, oncology, breast surgery and possibly also plastic surgeon, radiologist and gynaecologist.</p>	<p>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.</p>
OVARIES	<p>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).</p> <p>BSO to be performed from 35-40y for <i>BRCA1</i> and 40-45y for <i>BRCA2</i> carriers. Post-surgery, hormone replacement therapy might be given until the age of 45-50 years, unless there is a contraindication.</p>	<p>Refer to gynaecologist familiar with BRCA between the age of 30-40 years.</p> <p>Refer to diagnostic unit investigation if signs or symptoms associated with ovarian cancer.</p>
PROSTATE	<p>In some EU countries, men with a <i>BRCA2</i> pathogenic variant are offered PSA measurements every year from the age of 40-45 years.</p>	<p>Refer to urologist if PSA levels are increased.</p>

		Refer to a diagnostic unit if signs or symptoms associated with prostate cancer.
PANCREAS	Pancreatic surveillance with endoscopic ultrasound or MRI can be considered in <i>BRCA2</i> carriers with family history, preferably under a research protocol.	Refer to a diagnostic unit if signs or symptoms associated with pancreas cancer.
PSYCHOLOGICAL BURDEN	Despite a short-term increased in anxiety scores when a pathogenic variant is identified, most studies show a good emotional response at mid and long term.	Consider referral to an appropriate counselling service, if increased anxiety or difficulty to cope with the genetic condition.
PREGNANCY	Pre-natal diagnosis is usually not requested. Pre-implantation genetic testing (PGT) is an option available in some European countries. 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 can be referred to clinical genetics, if reproductive counselling is requested.

Hereditary Breast and Ovarian Cancer (HBOC) *BRCA1 / BRCA2* Clinical Pathway



Faculty:

Family name:

Given name(s)

Address:

Date of Birth:

Sex: M F I


Periodic Review Recommended

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Periodic review should be undertaken by a specialist in *BRCA1/2* (oncologist, surgeon, clinical geneticist, gynaecologist, radiologist). Surveillance should be continued until the person is in good health. Surveillance may depend on the level of the risk, which varies on which gene is involved, the family history of cancer and other non-genetic risk factors. Guidelines can differ in different EU countries. (Marmolejo et al, European Journal of Medical Genetics 2021, PMID 34606975)

HBOC *BRCA1/2* Review Checklist — Adults 25+

<p>Clinical Presentation:</p> <p>..... <input type="checkbox"/></p> <p>..... <input type="checkbox"/></p> <p>..... <input type="checkbox"/></p> <p>Other symptoms:</p> <p>.....</p> <p>.....</p> <p>Genetic counselling completed <input type="checkbox"/></p> <p>Date Completed:</p> <p>.....</p> <p>Clinical diagnosis</p> <p>.....</p> <p>Genetic Test '+ve <input type="checkbox"/></p> <p>Diagnosis Date:</p> <p>.....</p>	<p>General Health Check:</p> <p>Please record the follow as soon as possible and then annually:</p> <p>Height</p> <p>.....</p> <p>Weight</p> <p>.....</p>	<p>WHAT TO LOOK FOR</p> <p>BREASTS: MRI-breast from age 25-30y until the woman is in good health. Addition of annual mammogram from age 40. Addition of ultrasound should be considered if MRI is not possible or if requested by the radiologist.</p> <p>Information on the possibility of risk reducing mastectomy including the pros (decreased risk of cancer) and cons (risk of surgery, cosmetic etc).</p> <p>Discussion at multidisciplinary team consisting of at least a representative from clinical genetics, oncology, breast surgery and possibly also plastic surgeon, radiologist and gynaecologist.</p> <p>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).</p> <p>BSO to be performed from 35-40y for <i>BRCA1</i> and 40-45y for <i>BRCA2</i> carriers. Post-surgery, hormone replacement therapy might be given until the age of 45-50 years, unless there is a contraindication.</p> <p>PROSTATE: In some EU countries, men with a <i>BRCA2</i> pathogenic variant are offered PSA measurements every year from the age of 40-45 years.</p> <p>PANCREAS: Pancreatic surveillance with endoscopic ultrasound or MRI can be considered in <i>BRCA2</i> carriers with family history, preferably under a research protocol</p> <p>PSYCHOLOGICAL BURDEN: Despite a short-term increased in anxiety scores when a pathogenic variant is identified, most studies show a good emotional response at mid and long term</p> <p>PREGNANCY: Pre-natal diagnosis is usually not requested. Pre-implantation genetic testing (PGT) is an option available in some European countries. PGT relies on pre-pregnancy genetic work up and that the family fulfils the requirements for IVF.</p>	<p>WHEN TO REFER</p> <p>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.</p> <p><input type="checkbox"/> Date Referred:</p> <p>Refer to gynaecologist familiar with <i>BRCA</i> between the age of 30-40 years.</p> <p>Refer to diagnostic unit investigation if signs or symptoms associated with ovarian cancer.</p> <p><input type="checkbox"/> Date Referred:</p> <p>Refer to urologist if PSA levels are increased.</p> <p>Refer to a diagnostic unit if signs or symptoms associated with prostate cancer.</p> <p><input type="checkbox"/> Date Referred:</p> <p>Refer to a diagnostic unit if signs or symptoms associated with pancreas cancer.</p> <p><input type="checkbox"/> Date Referred:</p> <p>Consider referral to an appropriate counselling service, if increased anxiety or difficulty to cope with the genetic condition.</p> <p><input type="checkbox"/> Date Referred:</p> <p>Carriers (both male and female) who are planning pregnancy can be referred to clinical genetics, if reproductive counselling is requested.</p> <p><input type="checkbox"/> Date Referred:</p>
<p>Notes:</p> <p>.....</p> <p>.....</p> <p>.....</p> <p>.....</p>		<p>Doctor:</p> <p>Review date:</p> <p>Faculty:</p>  <p>www.genturis.eu</p>	