Guidelines for hereditary breast and/or ovarian cancer syndrome diagnostic testing criteria

Woman with breast cancer + one or more of the following

- diagnosed ≤ 40 yrs
- diagnosed < 50 yrs and one relative with bilateral, or ovarian, or breast < 50, or male breast cancer
- bilateral breast cancer and both diagnosed < 50 yrs
- ovarian cancer, any age
- triple negative breast cancer < 60 yrs
- three individuals with breast cancer, one is a first degree relative (FDR) of the other two (excluding male transmitters) and one diagnosed < 50 years
- individual of ethnicity associated with higher frequency of specific mutations (eg, Ashkenazi Jewish): eligible for founder mutation testing
- other family situations (eg multiple pancreatic cancer) with a priori chance of mutation >10% according to BRCAPRO or Evans criteria or Manchester score
- test more than one affected relative if criteria remain positive after excluding the negative case as a phenocopy

Woman with high grade serous or papillary epithelial ovarian cancer at any age (excludes borderline, low grade and mucinous ovarian cancer)

Male with breast cancer

Individual with pancreatic cancer at any age with ≥ 2 FDR excluding male transmitters with breast where one diagnosed <50 or bilateral, or ovarian, or 2 more pancreatic cancer at any age

Family history

- first degree unaffected relative of any of the above on a case by case basis
- testing of unaffected family members should only be considered when no affected family member is available and then the unaffected family member with the highest probability of mutation should be tested

The above mentioned guidelines were prepared by an ad hoc working group of the BeSHG/College on HBOC testing criteria (12/01/18), and reviewed and approved by the College of Genetics and Rare Diseases (12/01/18).