Program caring for women at familial and genetic risk for breast cancer

iPAAC INNOVATIVE PARTNERSHIP FOR ACTION AGAINST CANCER

Co-funded by the Health Programme of the European Union

TYPE	STATUS	LAST UPDATE		
Program	Fully implemented and ongoing	October 2021	ITALY ● REGIONAL	Breast cancer

PROBLEM & OBJECTIVE

PROBLEM About 20% of breast cancers occur in women with high familial risk and about 5-10% occur in women with known hereditary genetic mutations. These women may benefit from intensive surveillance and prophylactic interventions.

OBJECTIVE

- To identify women at increased familial and genetic risk of breast and ovarian cancer living in the Emilia-Romagna region.
- To offer them appropriate assessment of familial and genetic risk, including eventually genetic counselling and genetic testing for known mutations.
- To offer appropriate surveillance and prophylactic measures to women at increased risk.

CONTACT

Coordinamento screening
Regione Emilia-Romagna
Health Authority
https://salute.regione.emilia-romagna.it/screening/tumori-femminili/screeningmammografico/rischio-eredo-familiare

KEY COMPONENTS / STEPS

- Radiographers collect systematically family history with a standardized tool from all women attending mammographic screening program. As well GPs and clinicians involved in breast cancer care use the same tool.
- Women with a risk score higher than the average are referred to a first genetic counselling in the spoke genetic clinic where Tyrer-Cuzick model is applied, if women are eligible for genetic test, they are finally referred to the hub genetic clinic where decision about genetic testing is discussed.
- In this multi-step process women are stratified according to their risk that define the surveillance protocol, i.e. starting age, interval and which test (mammography, ultrasound and MRI).

KEY CONTEXTUAL FACTORS

- In Emilia-Romagna population-based screening invite all women aged 45 to 74 for mammographic screening. (add information on screening coverage in the region?).
- Women with familial and genetic risk have high lifetime risk of cancer, and in particular high risk of early onset, before the screening target age.
- Guidelines are consistent in recommending risk-stratified surveillance protocol modulating age to start, frequency and type of imaging (including MRI in some groups) and eventually prophylactic surgery (bilateral mastectomy, salpingectomy and ovariectomy). Nevertheless, specific surveillance protocols are not harmonized at international and national level.

MAIN IMPACTS / ADDED VALUE

- Since 2012 in the first 5 years of activity, 660,040 women were firstly screened for risk, 18,155 (2.8%) were eligible for spoke evaluation, plus 4,134 (0.7%) among 586,362 rescreened women, but only 5618 participated (25.2%). Other 2,260 and 3,798 women were referred to spoke from GPs and other physicians, respectively. 4,627 (39.7%) were referred to the hub, 2815 (60.8%) accepted and other 2871 were referred directly to the hub. Out of 5,686 hub visits 2,431 (42.8%) were referred to genetic test and 544 (23.2%) were BRCA1 or 2 mutated, while 89 tests were still in process.

LESSONS LEARNED

- To our knowledge, there are few population-based multistep programs aimed to identify individuals with high familial risk, including those with BRCA1/2 mutations to offer them an intensive surveillance program.
- The proposed model is feasible and effective in identifying high risk women. Nevertheless its acceptability is low as showed by the low participation rate in counselling clinics by women who are identified as at potentially high risk by screening tools and by the low referral from general practioners. An effective communication is a critical point to improve participation.
- Only a small proportion of women referred to the hub actually has the specific conditions to undergo a genetic test.

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