Principles of genetic counselling pdf

URL of this page: Genetic Counseling (Medical Encyclopedia) Also in Spanish Some women inherit changes (mutations) in some genes that increases the risk of breast cancer (and possibly other cancers). Genetic testing can be done to look for mutations in some of these genes. While testing can be useful in some cases, not every woman needs to be tested, and the pros and cons should be carefully considered. When it comes to breast cancer risk, the most important inherited gene changes are found in the BRCA1 and BRCA2 genes. In women (and men) with one of these gene changes are said to have hereditary breast and ovarian cancer syndrome (GBOC). Women with changes in the BRCA gene significantly increase the risk of developing breast cancer, as well as increased risk of ovarian cancer, pancreatic cancer and possibly some other cancers. Men with the change in the BRCA gene are at an increased risk of breast cancer (although this risk is lower than in women to start with), prostate cancer, pancreatic cancer, and possibly some other cancers. If you have a family history of breast cancer, you have a higher risk of getting breast cancer yourself. Most women with a family history of breast cancer do not have an inherited gene change that significantly affects their risk. However, hereditary gene change is most likely in women with a strong family history of breast cancer, especially if family history also includes some other cancers such as ovarian, pancreatic or prostate cancer. The risk of developing inherited syndrome also depends on: the proximity of affected family members (Cancer in close relatives such as mother or sister is more concerned than cancer in further relatives, although it may also be important.) The number of family members affected by the age when your relatives have been diagnosed (younger age is more of a concern.) Given genetic counseling and testing for breast cancer risk If you have breast cancer or family history of breast cancer, your medical history is a breast cancer. As a first step, your doctor can use one of several risk assessment tools that are now available. These mathematical models use your family history and other factors to help give you and your doctor a better idea of your risk of a BRCA mutation. But the tools are not perfect, and each one can give different results, so doctors are still trying to figure out how best to use them. Regardless of whether one of these tools is used, your doctor may suggest you benefit from talking to a genetic or another health care professional who is trained in genetic counseling. They may review your family history in detail to see how likely you are to have family cancer syndrome such as HBOC. The consultant can also describe the genetic testing for you and explain that the tests may be able to tell you what can It's up to you to decide whether genetic testing is right for you. It is important to understand what genetic testing can and cannot tell you, and carefully weigh the benefits and risks of genetic testing before these tests are done. Testing can cost a lot and it cannot be covered (or can only be partially covered) by some health insurance plans. If you decide to get tested, a genetic counselor (or other health care professional) can also help explain what the results mean for you and possibly for other family members. To learn more about genetic testing in general, see Genetics and Cancer. Testing for BRCA gene mutations Some group of experts have developed guidelines for which women (and men) should consider genetic counseling and possibly testing for brCA gene mutations. These guidelines can be complex, and not all doctors agree with them, but overall they include two main groups of people: women who have already been diagnosed with breast cancer: Most doctors agree that not all women with breast cancer need genetic counseling and testing. But counseling and testing are likely to be helpful if: You have been diagnosed with breast cancer at a younger age (especially if you have triple negative breast cancer) you have been diagnosed with breast cancer a second time (not a repeat of the first cancer) you have an Ashkenazi Jewish origin You have a family history of breast cancer (especially at a younger age or in men) , ovarian cancer, pancreatic cancer, or prostate cancer Other groups of people: Genetic counseling and testing may also be recommended for other people who are at higher risk of BRCA mutations, including: People with a known family history of BRCA mutations women diagnosed with ovarian cancer or pancreatic cancer, or men diagnosed with breast cancer, pancreatic cancer, or high-grade or metastatic breast cancer , more than one family member with breast cancer, or breast cancer in a male family member People with a close family member with a history of ovarian cancer, pancreatic cancer, or metastatic prostate cancer testing for other gene mutations associated with the risk of breast cancer Mutations in one of the BRCA genes make up the majority of inherited breast cancer. But inherited changes in some other genes, including PALB2, CHEK2, ATM, PTEN (associated with Cowden syndrome), and TP53 (associated with Lee-Fraumeni syndrome) may also increase the risk of breast cancer. Testing for changes in these genes is carried out but in some situations this may be considered. As testing is conducted genetic testing can be done on blood samples or saliva, or from a tampon inside the cheek. Samples are sent to the laboratory for testing. There are many different possible mutations in the BRCA genes. Testing can be done to look for one (or several) specific mutations (s), or more extensive testing may be look for many different BRCA mutations. The approach to testing depends on the situation. For example, if someone is tested because they have a family member with a known BRCA mutation, testing can only focus on finding that particular mutation. In people of Ashkenazi Jewish origin, testing may focus on specific BRCA mutations that are most common in this group of people. But if there is no reason to suspect a specific gene change, testing is likely to look for many different mutations. Obtaining the results of genetic testing Before receiving genetic testing, it is important to know in advance that the results may or may not tell you about your risk. Genetic testing is not perfect. Tests may not give clear answers for some people. This is why meeting with a genetic consultant or cancer geneticist is important even before testing. The results of genetic testing may come back as: Positive for a mutation that has been tested on. If so, there may be steps you can take to help reduce your risk of breast cancer (or other cancers). If you have already been diagnosed with breast cancer, a positive result may affect your breast cancer treatment options. Negative for mutation (s) tested. It may be encouraging to learn that the test has not found a mutation that increases the risk. But it is important to understand that the results of genetic tests can not always guarantee that you do not take risks. For example, you may be more likely to have another mutation that you haven't tested for. No avail. In some cases, the test may not be able to tell for sure if you have a gene mutation. Positive version of unknown value (VUS). This means that the test has detected a change in the gene (option), but it is not clear if this particular change affects the risk. To learn more about these different types of test results, see what happens during genetic testing for cancer risk? The results of genetic testing can sometimes be complex or confusing, so it is important to navigate them with a genetic consultant or cancer geneticist professional. They can explain what they can mean to you (and possibly other family members). Direct to consumer genetic tests Some genetic tests are now available directly to the public, but there are some problems with these types of tests. For example, a test that looks for a small number of mutations in the BRCA1 and BRCA2 genes has been approved by the FDA. However, there are more than 1,000 known BRCA mutations. This means that there are many BRCA mutations that will not be detected by this test. A woman with a negative test result may suggest that she should not be its risk when in fact it may still have different BRCA mutations. Our section on genetics and cancer has more information about genetic mutations and When women and their doctors make breast cancer treatment decisions today, they have more tools than ever before to help them. This type of test can provide information about a woman's breast cancer and its risk for future cancers. As genetic testing has become more accessible and less expensive, more patients and physicians have started using tests to help them make informed treatment decisions. Many women with early-stage cancer can choose between breast preservation surgery and mastectomy. The main advantage of breast preservation surgery is that the woman holds most of her breasts. Studies have shown that this type of treatment gives women with breast cancer at least as good a chance of survival as a mastectomy that removes the entire breast. But some women who undergo genetic testing learn that they have a mutation that significantly increases the risk of second breast cancer. Women with this finding sometimes choose to reduce their future cancer risk by having both breasts removed - one that has cancer and one that doesn't. This is called a bilateral mastectomy. Only about 5% to 10% of breast cancer cases are considered hereditary, but having inherited mutations, especially in the BRCA1 or BRCA2 gene, can significantly increase the risk of breast, ovarian and other cancers. Who should have genetic counseling after a breast cancer diagnosis? The National Comprehensive Cancer Network (NCCN) has guidelines that recommend referral to a geneticist if a woman is at high risk of mutation. She can be considered a high risk if she has: a personal history of some cancers of a strong family history of some early childhood cancers when diagnosing the family history of the cancer-related genetic mutation Ashkenazi Jewish origin in combination with other risk factors In an ideal situation, a woman recently diagnosed with breast cancer, who is at high risk of mutation will meet with a genetic expert before deciding on treatment. Geneticists are trained in both genetics and counselling, and help women determine whether to be tested. If a woman is tested, a genetics expert is the best person to help her interpret the results. In fact, most newly diagnosed women first see a breast surgeon. The surgeon is in a key position to recommend her to a genetics expert if she meets the criteria in accordance with NCCN guidelines. In addition, the surgeon can actually order the test and then refer it to a genetics expert, move on to the results. The study shows genetic counseling is not used enough in the treatment of breast cancer But not all women or even all health professionals are aware of the guidelines, and many women who are at high risk of genetic mutation never meet with a genetic expert. Allison W. Kurian, MD, MSc, director of the Stanford Women's Clinical Cancer Genetics Program, studied this issue with researchers from other leading cancer centers in the United States. She said: There are gaps in how genetic testing is used in the treatment of breast cancer and it gets in the way of making the right treatment decisions. Kurian said she would like to see more involvement of genetic consultants in her research. Most guidelines say that anyone who receives genetic testing should consult a genetic counselor, and that certainly doesn't happen, she said. Kuryan and her colleagues conducted research to find out how women and their doctors use genetic testing to make treatment decisions after a new breast cancer diagnosis. In one study, they found that only about half of high-risk women received a genetic test. The study involved 2,529 women diagnosed with 0 to 2 breast cancer who responded to surveys 2 months after breast surgery. Of these, 773 (31%) had a higher risk of genetic mutation than average. About 81% of high-risk women reported that they wanted to be tested, and 71% said they talked to their doctor about testing. However, only 52.9% actually received genetic testing. Of the high-risk women tested, 61.7% met with a genetic counselor. The most frequently cited reason not tested was: My doctor did not recommend it. The study was published February 7, 2017 in the Journal of the American Medical Association. According to Kuryan, among women who absolutely met these criteria, only half of them even received testing. It was one problem that people who really need to get genetic testing don't get it. Why should they get it? There are many reasons, but perhaps the most important reason is that for a breast cancer patient, if she has a genetic mutation in BRCA 1 or 2, she has a high risk of ovarian cancer that can kill her. When we don't test these patients and find these mutations, we basically miss the chance to save lives. Breast Cancer Surgeon Cletus A. Arciero, MD FACS Winship of Emory University Cancer Institute, which was not involved with the study, agrees that health professionals should adhere to NCCN guidelines for who should be tested, and that all women tested should have the benefit of genetic counseling. I think the research shows what's been going on for a long time. The world of genetics is developing rapidly. The cost is incredibly affordable, even for those without insurance, and we are testing so many more genes than in the past that a lot of information can be behind in the educational part for doctors, Arciero said. Arciero says that surgeons and medical oncologists may be trying to provide new genetic technology for their patients, but may not have the genetic practice to designate their patients. We just need more genetic consultants, he says. Patients may be unwilling or unable to travel long distances to see genetics in another city, or they may be concerned about the delay in treatment. While Arciero says patients - even those with aggressive cancer - can usually safely wait up to 2 months before starting treatment, most are understandably eager to start treatment as soon as possible. Concerns about the timing and use of genetic testing results Even when women receive genetic testing, there are often questions around when they receive screening and how their doctor interprets the information, according to a second study by Kurian and her colleagues. Researchers used the same dataset for the study, published April 12, 2017 in the Journal of Clinical Oncology. They found that a significant proportion of women who undergo genetic testing do not receive all the information they need to make an informed decision about surgery. In this study, 666 women reported that they had received genetic testing. Of this group, 393 (59%) of those in the group said they would like to be met the criteria for an increased risk of genetic mutation. Of the 393 high-risk women who received genetic testing: 27% were not tested until after breast surgery, eliminating the possibility that they could use the results to decide treatment. 57% discussed their results with a genetic consultant. Others discussed their results with a surgeon or medical oncologist. The study identified one area of particular concern: the management of the result of genetic testing, called variants of uncertain value (VUS). Unlike mutations in the BRCA gene that increases the risk of breast cancer, VUS is known to not increase the risk of breast cancer - or any other cancer - and genetic experts say there is no medical reason for a woman with VUS to undergo a bilateral mastectomy. However, the study found that up to half of surgeons do not recognize the differences; they reported no difference in their management of women with BRCA mutations known to raise the risk of breast cancer against VUS. The study found that women with VUS often underwent a bilateral mastectomy. Kurian said: We have seen high levels of clinicians say they will treat an uncertain genetic outcome in the same way as a positive result for the mutation. It was amazing and disturbing. The misconception that one of these uncertain outcomes should be considered a harmful mutation can lead to too many mastectomies. It's an education issue for patients and doctors, Arciero said. He says patients are more likely to be alarmed if they receive the news that they have a VUS mutation in the BRCA 1 or 2 gene. He says the greater availability of genetic advisory resources could help pull clinicians out of the genetic process He says doctors without genetic resources may be trying to help their patients by ordering tests, but if they don't fully understand the results, or if they understand them but don't have skills to advise patients about them, they do not serve the patient well. What can be done by the medical community recognizes that there is a shortage of genetic consultants in the US. Professional genetic organizations, including the American Council on Genetic Counseling and the American College of Medical Genetics and Genomics, are taking steps to expand their curriculum. At the same time, the study authors propose to improve education for all physicians as well as systems for accelerated the greatest risk of genetic counseling patients. Arciero says surgeons and oncologists can help close the gap by learning more about genetic testing and counseling by participating in local medical organizations, reading medical journals, and attending national conferences. Women who have faced the decision to have breast cancer surgery are advised to have a frank conversation with their health team. Arciero says that women newly diagnosed with breast cancer almost always have time to think through all the options before deciding on the best type of surgery for their individual situation. Kuryan recommends these questions to ask the doctor: Should I get a genetic test? Is that going to help me? Could you see me as a genetics expert? Expert? principles of genetic counselling ppt. principles of genetic counselling pdf. ethical principles of genetic counselling