
JEWISH GENETICS
&
BREAST-OVARIAN
CANCER
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This Brochure presents some new information on Jewish Genetics & Breast / Ovarian Cancer. Medical References are provided.

It is recognized that further medical reports / knowledge may modify current concepts.

It is recognized that individual care will be modified by individual genetic, medical, & social needs.

Women are advised to keep informed, be responsible, & to discuss this and further information with knowledgeable physicians, surgeons, gynecologists, & oncologists.



(July 9, 2010)

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# Genetics

Critical new information on Jewish Genetics and Breast/Ovarian Cancers is not well known, either by most physicians or by the Ashkenazi Jewish community. The BRCA (BReast CAncer) mutations (three possible chromosomal mutations) occur in the population-at-large at an incidence of 1:800. Among Ashkenazi Jews, the incidence is 1:40.

## Incidence of BRCA Genetic Mutations

| Population at-large | Ashkenazi Jews |
|---|---|
| 1 :800 | 1 : 40 |

The normal healthy genes are considered tumor-suppressant genes. Everyone has two sets of these genes, one set inherited from each parent. In most individuals, each set is healthy. When an individual inherits one set with a mutation, it is comparable to having a car or bicycle with two brakes, one of which is not working. If the individual then loses function of the other, the normal gene, (for example, through damage from pesticides or industrial chemicals) then all "braking" control has been lost, and cancerous cell reproduction proceeds.

The BRCA genes (i.e. mutations) are present equally in males and females, and are passed equally from fathers or mothers to sons and daughters. In fact, men with a BRCA 2 mutation have a 6 % incidence of developing breast cancer.

# Risks with BRCA

Whereas the female population-at-large has a lifetime risk of breast cancer of 8-12%, women with one of these three BRCA mutations have a lifetime risk of breast cancer of 60-87%. Whereas the female population-at-large has a lifetime risk of ovarian cancer of 1%, women born with one of these mutations have a lifetime risk of ovarian cancer of 27-44%. Women born after 1940 with one of these BRCA mutations have a 67% risk of breast cancer before age 50.

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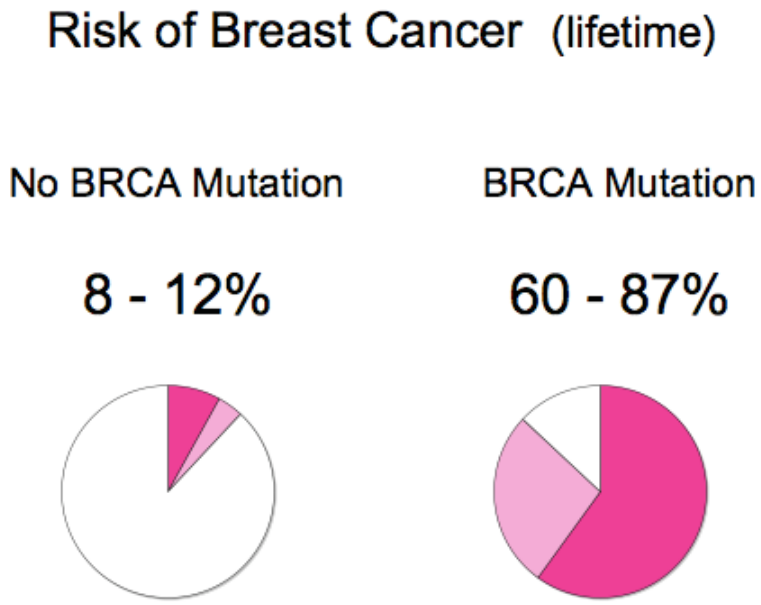


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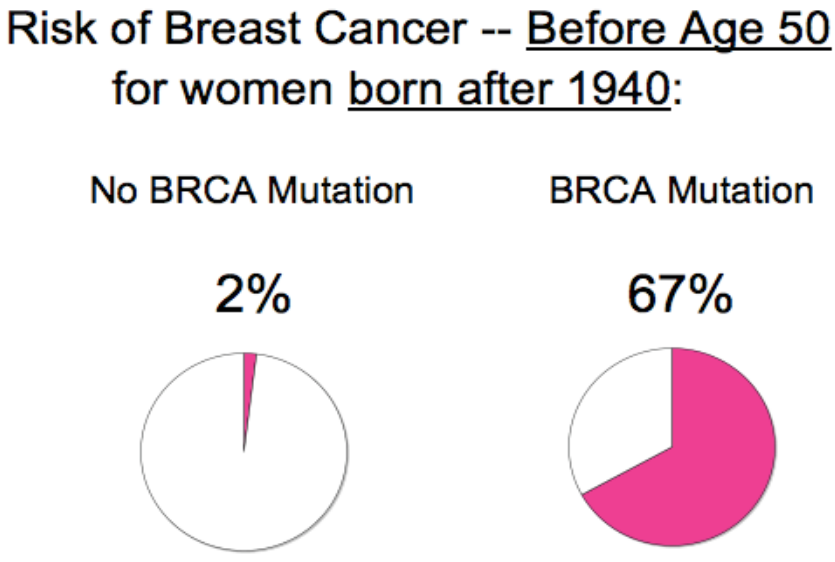


King, M.-C.: Science: vol. 302, p. 643, Oct. 24, 2003

# Risk Reduction-Mammograms & MRI

**High-risk women should begin having mammograms at age 25**

"28-65 % of the sharp decrease in breast cancer deaths from 1990 to 2000 was due to Mammograms".

National Cancer Institute study, in *The New England Journal of Medicine* 10-27-05

Misleading Controversy from U.S. Preventive Services Task Force in 2009. Based upon 7 prior studies, most in the **1970s or 1980s.** The only U.S.-based study began screening **in 1963**. (Wall St. Journal 12/9/09) However: **Accuracy of mammograms -- and advances in chemotherapy and radiation -- have improved tremendously in last 30-40 years. Therefore early and regular mammograms now save more lives!**

**MRI (Magnetic Resonance Imaging)------Better Than Mammograms**

Of 96 BRCA mutation carriers, MRI found 5 breast cancers, mammograms found 2 of the 5

*Journal of Clinical Oncology* vol. 19, no. 15; 2001: 3524-3531

In BRCA mutation carriers, MRI is more accurate than mammograms in early detection of breast cancer

*Journal of National Cancer Institute* 2001; 93:1095-1102

In high risk women, MRI detected 9/9 cases of breast cancer, Mammography detected 3/9

*Radiology* 2000; 215: 267-279

Of 45 breast cancer cases in high risk women, mammography detected 18, missed 27; MRI found 32, missed 13

*The New England Journal of Medicine* 2004; 351; 427-437

Of 969 women, MRI detected breast cancer in 30 who had normal mammograms and clinical exam

*The New England Journal of Medicine* 2007; vol. 356, no. 13: 1295-1303

**American Cancer Society Recommendation (***CA A Cancer Journal for Clinicians* **2007**;57; 75-89)

Annual Breast-Cancer Screening by MRI (and Mammograms) for women with

BRCA mutation, or

First-degree relative of BRCA carrier, but untested, or

20 % or greater life-time risk of breast cancer

# Risk Reduction - Chemoprevention & Surgery

**Prophylactic Tamoxifen** in the population-at-large reduces risk of developing breast cancer by **50** %. In several studies, Tamoxifen significantly reduced risk of breast cancer in women with BRCA1 or BRCA2 mutations. "Risk of contralateral breast cancer is reduced by **50** % in carriers of BRCA1 and BRCA2 mutation when Tamoxifen is used" (Narod et. al.:*The Lancet* vol. 356, 2000, 1876-1881). In one small study of BRCA patients, Tamoxifen demonstrated a **62** % risk reduction for BRCA2, but **no** benefit for BRCA 1 carriers. (King et. al.: *Journal of American Medical Association* vol. 286, no. 18, 2251-2256.) Benefit/risk profiles for Tamoxifen are more favorable for women below age 50.

Some folks worry that "Tamoxifen carries Risk of Blood Clots, Strokes, Uterine Cancer." However, look at the statistics - and compare with risk-reduction (above):

| | Tamoxifen |
|---|---|
| Blood Clots | 87 / 9,726 |
| Pulmonary Emboli | 54 / 9,726 |
| Uterine Cancer | 36 / 4,732 |
| Strokes | 53 / 9,726 |
| | 230 / 9726 |
| | 2.4% |

http://www.cancer.gov/ncicancerbulletin/NCI_Cancer_Bulletin_041806/page2bs.marketwatch.com/

**Prophylactic Oophorectomy** (removal of ovaries) post-child-bearing (not only reduces risk of ovarian cancer **96 %,** but also) **reduces risk of breast cancer 50 %** . (This procedure is often done as an out-patient --via a laparoscope.)

*The New England Journal of Medicine*: May 23, 2002, p. 1609
*The New England Journal of Medicine*: May 23, 2002, p. 1616
*The New England Journal of Medicine*: May 23, 2002, p. 1660

This oophorectomy risk reduction for breast cancer applies to the population-at-large, BRCA 1 carriers, and possibly BRCA 2 carriers as well.
*McCormick, Beryl: NCCN Breast Cancer Guidelines Update, NCCN 12th Annual Conference*

**Prophylactic mastectomy (& reconstruction)** provides **95 % - 97 %** risk reduction when complete mastectomy is performed. Reconstruction may be achieved with implants or muscle transposition, with results which are are generally well accepted. Many women have known only of mastectomy-reconstruction, and have been unaware of their other options for surveillance and risk-reduction.

# Risks of a Second Breast Cancer

A woman with breast cancer and a BRCA mutation has a vastly increased risk of a second later cancer in the same or opposite breast. This is not a result of residual cancer cells "left behind" by the surgery. Rather it is a result of the remaining cells all being mutated, and therefore themselves susceptible to development of cancer.

## Risk of a REPEAT Cancer in Same Breast

| No Mutation | | BRCA MUTATION |
|---|---|---|
| 4.5 % | (5 yrs.)[1] | 14.9 % |
| 7.0 % | (5 yrs.)[2] | 14.0 % |
| 6.9 % | (10 yrs.)[1] | 22.0 % |
| 16.0 % | (10 yrs.)[2] | 30.0 % |
| 21.0 % | (12 yrs.)[3] | 49.0 % |

1. Journal of National Cancer Institute: vol. 91, p. 2112, Dec. 15, 1999
2. Eur. J Cancer 2004 May ;40(8):1105-8
3. The Lancet: vol. 359, p. 1471, Apr. 27,2002

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## Risk of CONTRALATERAL BREAST CANCER

| NO Mutation | | BRCA Mutation |
|---|---|---|
| 4.0 % | (5 yrs.)[4] | 31.0 % |
| 3.7 % | (5 yrs.)[1] | 14.8 % |
| ----- | | 16.9 % |
| 9.5 % | (10 yrs.)[1] | 27.0 % |
| ------ | | 29.5 % |
| 9.0 % | (12 yrs.)[3] | 42.0 % |
| 8.2 % | (? yrs.)[4] | 40.0 % |
| 6.1 % | (? yrs.)[5] | 24.5 % |

1Journal of the National Cancer Institute: vol. 91: 2112-2117, Dec. 15, 1999
3 The Lancet: vol. 359: 1471-1477, Apr. 27, 2002
4 Journal of Clinical Oncology: vol. 16: 1642-1649, 1998
5 The Lancet: vol. 351, Jan. 31, 1998: 316-321
9 Journal of Clinical Oncology: vol.22, no.12, June15, 2004: 2328-2335

## Reduction of Risk of Second Breast Cancer:

For women with a diagnosed breast cancer and a BRCA mutation, bilateral mastectomy as the initial procedure may reduce risk of a second breast cancer by 90-97 %.

The National Comprehensive Cancer Network in its 2007 Guidelines has now placed **prophylactic bilateral mastectomy for consideration as initial treatment for those women with a diagnosed breast cancer and a BRCA mutation**.

http://www.medscape.com/viewarticle/555758

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## Genetic Testing of The Cancer > Specific Risk Reduction Measures

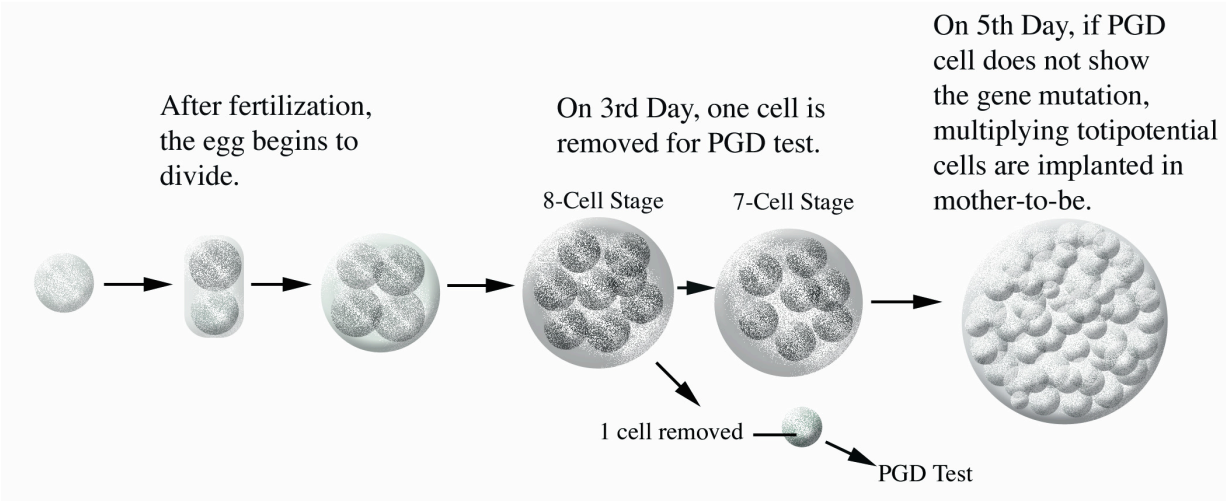
In recent years, a number of different tests examine the genetic mutations specific for the cancer cells of each individual person. First was the 70-gene test, called "MammaPrint", developed in the Netherlands. Another test which has been used is named Oncotype DX. Currently in use is the "21-gene PT-PCR" test. These tests, and others likely to follow, examine the specific cancer mutations of the specific person. (After extensive clinical studies), these tests now can predict which specific medications may best treat that individual's specific cancer.

There now is an array of medications for hormonal and genetic control of malignancy. The oncologist now can prescribe what is more likely to be effective for each individual patient. The list of medications includes: Nolvadex ("Tamoxifen"), Trastuzumab ("Herceptin"), Anastazole ("Arimidex"), and Letrozole ("Femara").

In addition, there is an array of chemotherapy agents, from which the oncologist will prescribe those medications best suited for each person's cancer.

# Preimplantation Genetic Diagnosis

Preimplantation Genetic Diagnosis (PGD) now allows couples to have offspring free of the parental BRCA mutation. (A parent—either father or mother—with the mutant gene has a 50 % risk of passing that mutation to each offspring.) An in-vitro fertilization clinic harvests eggs from the mother-to-be and sperm from the father-to-be, and brings about fertilization on an agar plate. On the third day (when cell division has led to perhaps 6-15 cells), one of the cells is removed, placed in a test tube, and sent via FedEx to a lab in the U.S. Within 24 hours, it is known whether the cell is free of the mutation. If so, on day 5, the multiplying embryonic cells are implanted in the mother-to be --- with the (95-98 %) expectation that the offspring will be free of the cancer-causing mutation, (instead of only a 50 % chance).




Preimplantation Genetic Diagnosis is available for couples with a BRCA mutation. PGD can be secured at an in-vitro fertilization clinic near the couple's home, and within their own health network. Couples may discuss PGD advantages and disadvantages with their obstetrician and with physicians at the in-vitro fertilization clinic. National / International Laboratories performing BRCA PGD for IVF Clinics are:

Reproductive Genetics Institute
2825 North Halsted St.
Chicago, Illinois 60657
(773) 472-4900
www.reproductivegenetics.com

Genesis Genetics Institute
5555 Conner Ave.
Detroit, Michigan 48213
(313) 544-4006
www.genesisgenetics.org

## Genetic Testing in Non-Cancer Patient

Genetic Testing can determine if a woman has one of these 3 "Founder" BRCA mutations (and therefore may be at increased risk of developing breast / ovarian cancer). Genetic testing is indicated for:

1. An initial diagnosis of breast cancer in a woman below 50.
2. Ovarian Cancer at any age.
3. Having a blood relative (on maternal or paternal side of the family) with breast cancer before age 50.
4. Having a blood relative (on maternal or paternal side of the family) with ovarian cancer at any age.

The test can be taken at a lab, hospital, or doctor's office. The blood is put in a special tube and sent to Myriad Genetic Laboratories, 320 Wakara Way, Salt Lake City, Utah 84108-9930. For information on current costs, Myriad Genetic Laboratories may be reached at 1 (800) 469-7423 or at **www.myriadtests.com** (**email: BRCA@myriad.com**). Be specific in your request. The charge for the "Ashkenazi Multisite 3" is a fraction of the cost of a "Comprehensive" genetic test for BRCA mutations. (Mutations other than these 3 BRCA may also cause inherited predisposition to breast cancer).

Routine genetic testing results are available in three to four weeks. However, for women with an initial diagnosis of breast cancer, rapid results are available in 10 days. If a BRCA mutation is found, the patient may then decide to proceed with bilateral mastectomy (and reconstruction).

Prior to testing, consultation with a Genetics Counselor is recommended (1) to address patient's concerns and emotions, and (2) to further explain implications and options of possible test results.

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The Genetic Information Non-Discrimination Act of 2008 ("GINA"), signed into law May 21, 2008:

(1) Prohibits health insurance companies from using genetic data to set premiums or determine enrollment eligibility.

(2) Prohibits employers from denying employment, promotions, or health coverage based upon genetic tests.

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With respect to life insurance and disability insurance: (1) actuarial considerations of adverse selection raise issues of legitimate concern, and therefore (2) for these policies, eligibility and premium determination are decided by the insurance companies.

## Why Test ? What Would I Do ?

Mammograms, annual, – starting at age 25 yrs.

Breast MRI, annual, ---at a facility

With dedicated "breast coil"

Experienced in timing sequence of contrast agent

Able to perform MRI guided needle sampling &/or wire localization

Consider Chemoprevention (e.g. Tamoxifen)

Consider Oophorectomy

Between 35 & 40 yrs.

Or upon completion of child-bearing

Consider Mastectomy (& Reconstruction)

especially breast cancer patients

Transvaginal Ultrasound & CA-125

Consider Preimplantation Genetic Diagnosis

Notify Daughters & Sisters (& Sons & Brothers)

They have 50 % Risk of BRCA Mutation

After age 20, they should consider testing

If they are positive, they could consider Surveillance & Risk Reduction

## Challenge for Medical Care

In the 21st century, there is no place for Paternalism,
where physicians decide what women should do.

In the 21st century, there is no place for Maternalism,
where anyone decides what women should know.
*Bernard A. Lublin, M.D.*

In a Survey of Practicing U.S. Physicians:

May physicians, in considering their religious and moral beliefs, avoid disclosing all medical options to patients?

7-21 % > YES

May physicians, in considering their religious and moral beliefs, avoid referring patients to another physician who would provide more information or the desired procedure?

12-35 % > YES

*New England Journal of Medicine* 356: pp. 593-600, Feb. 8, 2007

**NCCN (National Comprehensive Cancer Network)**
**2010 Physician Guidelines**

Breast Cancer Screening and Diagnosis
http://www.nccn.org/professionals/physician_gls/PDF/breast-screening.pdf

Genetic / Familial High-Risk Assessment: Breast and Ovarian:
http://www.nccn.org/professionals/physician_gls/PDF/genetics_screening.pdf

Breast Cancer Risk Reduction:
http://www.nccn.org/professionals/physician_gls/PDF/breast_risk.pdf

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- Peelen, van Vliet, Petrij-Bosch, et. al.:A High Proportion of Novel Mutations in BRCA1 with Strong Founder Effects among Dutch and Belgian Hereditary Breast and Ovarian Cancer Families: Am. J. Hum. Genet. 1997 May; 60 (5) 1041-9 and 1013-20.
- Thorlacius, Struewing, Haartge, et. al.: Population-based Study of Breast Cancer in Carriers of BRCA 2 Mutation: Lancet, Oct 24, 1998, 352 (9137) pp. 1337-9


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**On the Risk of a Repeat Cancer in the Same Breast**


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**On The Risk of Later Contralateral Breast Cancer among BRCA+ Women Who Have Had an Initial Breast Cancer**


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**Risk-Reducing Oophorectomy in Women with a BRCA1 or BRCA2 Mutation**


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**On Alleviating Menapausal Hot Flashes with Selective Serotonin Reuptake Inhibitors**

www.brighamandwomens.org/patient/menopauseqanda.asp

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**Mammography and MRI Comparison**


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- CA A Cancer Journal for Clinicians vol. 57; pp. 75-89, 2007
- http://www.cancer.gov/ncicancerbulletin/NCI_Cancer_Bulletin_040307/page2

May 4, 2007

My name is Karen. I am Dr. Bernie Lublin's niece. Six years ago when I was 33 years old, I was diagnosed with breast cancer and a couple of weeks later, I tested positive for the BRCA1 mutation. I had a bilateral mastectomy with reconstruction, 6 months of aggressive chemotherapy followed by a complete hysterectomy (ovaries and uterus) which was performed laparoscopicly. I made all these medical decisions with one single goal: to live to be 90+ years old. The quality of my life has not been compromised. I have a wonderful loving family and many friends. I lead a really fun life as a happily retired attorney and stay-at-home mom of three young children. I am very happily married. When I was considering these "female surgeries", I asked a fellow breast cancer survivor for advice. Like me, she is young. She said to me, and I agree, that losing one's breasts, ovaries and uterus is not like losing an arm, or your sight, or your hearing. If I could not hug, see or hear my children and others, I believe that I may feel that the quality of my life has been greatly compromised. As I get older, I realize that everyone has their problems (medical or otherwise) and we all just have to learn to adjust. It has been an adjustment for me to accept my new breasts and general lack of female body parts, to accept the finality of having no more children, and to accept the lack of estrogen in my body and the early onset of menopause and the troubleshooting of menopause symptoms. These are really just inconveniences when you look at it, and I have to see a lot of doctors from time to time to troubleshoot these inconveniences. What would truly interfere with the quality of my life would be more cancer. I have had quite enough of that. My surgeries gave me piece of mind that I am cured of my genetic disorder and I would make the same surgical decisions again today.

I would like to share with you "my story". I found the lump myself. My OBGYN had not found it 4 months prior in her clinical breast exam. I truly believe that if I had not found the lump myself, I would be dead today or dealing with metastatic disease. I was not scheduled to see the OBGYN for my next breast exam for 8 more months. I was way too young to be recommended for mammography. I had no breast cancer in my family and did not consider myself at high risk. I had been told 5 years earlier by my OBGYN not to worry that my father's mother had been stricken with ovarian cancer because the risk of ovarian cancer would not travel to me through my dad's family. This is now known to be false. I believed my doctor and never thought about it again. I had no knowledge of BRCA mutations and how they lead to early onset breast and ovarian cancer.

So why did I find the lump? I was playing on the floor with my three daughters, a 3 year old and twins that were 10 months old. I was feeling understandably lousy since my twins never slept. My breasts were aching, also understandably since the bras probably needed changing post childbirth. Nonetheless, something seemed not right. I felt off and toxic. So I decided to listen to what my mother had taught me – I immediately left the kids and did a self-breast exam in the shower as

I did about every 3 or 4 months. I listened to my mom! Who listens to their mom? Who actually does self-breast exams when they are 33 or younger, if ever, particularly when they have no reason to think they are at risk of imminent breast cancer? I did. I found a lump, smallish, but there. What to do then? It was Saturday (of course). These things don't happen on a Tuesday morning. Bear with my story that follows because it teaches a lesson: you must push, push, push, to expedite the things you need. So I called the OBGYN office on Saturday and said it was a medical emergency. A doctor called back and explained that women my age, post childbirth, have lumpy breasts. This is not an emergency. Please schedule an appointment in two weeks, mid-menstrual cycle when my breasts would be less lumpy. I said OK. Monday morning, I squeezed my breast and blood oozed-out. Without an appointment, I drove 30 minutes to the OBGYN office and shocked the receptionist with my insistence to be seen (immediately). The waiting room was empty but since I was crying and refused to leave, they found a doctor for me who did an exam, told me my lump seemed like a normal lump and she set me up for a diagnostic mammogram with ultrasound just to be sure. Still due to my tears and obvious panic, they scheduled all this ASAP (a couple of days later). The mammogram and ultrasound revealed I needed a needle biopsy. I said "do it now". Two days later I was diagnosed with breast cancer. My radiologist was like an angel who took me under her wing and scheduled me a quick appointment with a way overbooked and very well respected surgeon at the same hospital.

To decide what type of surgery to have, I ultimately consulted with 4 renown oncological breast surgeons in my metropolitan city, one genetics counselor, 1 gynecological oncologist, 3 medical oncologists, 2 reconstructive surgeons and my family did much internet research on my behalf. My point is: I spent several weeks searching for information and pulling teeth and pulling strings to get access to information and to reach a decision on how to proceed surgically. I waited endlessly it seemed (somewhere around 2 weeks to receive the "rush" results of my genetic testing).

As I said, I met with four oncological breast surgeons. Why so many? Sometimes in your gut, you know you are not getting all the information you need to make a reasonable decision. At first, I only had Surgeon # 1 scheduled. Prior to that appointment, my uncle (Dr. Lublin) told me: "I've done some research and I am pretty confident you have a genetic disorder that predisposes you to breast cancer and this is why you got cancer so young and this is why your grandmother was stricken with ovarian cancer, also young. This is a common genetic mutation among Ashkenazi Jews." He told me to make sure I discuss this with my surgeon. He sent me two research articles. I read them. I brought them to my appointment and then I forgot to discuss them. Through my tears, and being paralyzed by fear, neither myself nor my husband or parents were my best advocates at the moment. The well respected, very compassionate and well spoken surgeon recommended a lumpectomy followed by radiation and he said that I would probably need chemotherapy as well. He believed my prognosis for a total cure was very good. We left relieved. My uncle was furious. I forgot to ask my genetics questions and the surgeon did not mention the issue himself, despite the fact that I mentioned that my grandmother had ovarian cancer.

By pulling strings of the wealthy and well-connected, I was able to get an appointment a few days later at a teaching hospital with surgeon #2. I was prepared again to discuss my uncle's concerns. I did not need to speak.

The surgeon took one look at me, sitting there with my mommy and daddy and he had it all figured-out. He asked: "How old are you?" "33" I said. "Where are you from?" "Virginia" I said. "No, where are your ancestor's from?" "Russia, Poland", I said. "Are you Jewish?" "Yes", I said. "Any breast cancer in your family?" "No". "Any ovarian cancer". "Yes, my grandmother". He noted a few points in my biopsy results "possible medullary carcinoma and estrogen negative tumor receptors." The red flags were waving high. Then he said he would discuss surgery with me after I met with the genetic counselor. He saw I was desperate to see her at that moment. He got her on the phone and sent me immediately upstairs to her. After discussing matters with her, she concluded that she was more than 95% confident that I had a BRCA mutation. She said that even if my genetic testing came back negative, she was confident that I had a (yet to be determined) BRCA mutation. She recommended that I assume that my genetic results were positive for a mutation even if they came back negative. Then she explained what my uncle is explaining (how to cure me): while lumpectomy and radiation would have been fine for curing my then current cancer, it would have done nothing to protect me from probable future new cancers in my breasts and ovaries. The only way to greatly reduce my risk (by more than 90%) of new cancers was a total bilateral mastectomy and an oophorectomy (with a total hysterectomy being even more preferable). The wise genetics counselor introduced me immediately to my future gynecological oncological surgeon who happened to be sitting in his office and happy to talk to me. He explained that it is recommended that, I receive transvaginal ultrasounds and pelvic exams every three months, and that I have a complete hysterectomy by the time I was 35 unless I was not done with childbearing. Ultrasounds and pelvic exams are poor ways to predict early stage ovarian cancer. Several hours later, I was back in Surgeon #2's office. He emphasized that if I am only interested in curing my current cancer, lumpectomy and radiation is just as effective as mastectomy. He asked if this were enough for me. He said I needed to look into my heart to determine what I really wanted – a cure for all my genetic woes or just a short term fix. He said that for some women, they could not have a good quality of life without their real breasts. It would negatively impact their sense of womanhood and sexuality. They would rather take their chances. I could tell he wanted me to choose – a long term cancer free life (particularly since I was so young). I said I wanted him to do the bilateral mastectomy, but I wanted to wait for my genetic results to be positive for sure. I was very confused about what to do if the results were negative. Could I really treat negative results as though they were positive?

I went home and called Surgeon #1 who was stunned by my day with Surgeon #2 and his team. I updated him. He said it was highly unlikely that I have a BRCA mutation since there was no breast cancer in my family, and even if I did have such a mutation, he did not recommend the bilateral mastectomy. It was not the standard of care, period. It was not called for. He would do the surgery to make me happy but with reluctance. I needed more information. How could two well respected surgeons give such polar opposite opinions. Surgeons #3 and #4 both agreed that the

standard of care called for lumpectomy and radiation, not bilateral mastectomy. They explained that it takes too long to get the result of genetic testing and that it was better to get a quick fix and deal with genetics at my convenience. I explained that the two plastic surgeons I had consulted with clearly explained that it's difficult to do reconstruction once the breast has undergone radiation. They understood but the standard of care is to focus on the cancer I had at that time. They would do bilateral mastectomy but with reluctance.

Then something amazing happened: Surgeon #4 called me the next day to tell me he had changed his opinion. He felt in his gut that my instincts were right and that he would have the bilateral mastectomy if he were me. He said that doctors feel duty bound by the "standard of care", but in my case, he thought I had everything well thought-out and I should go protect myself, my life, and my best chance for a nice breast reconstruction. Ultimately, I learned I tested positive for BRCA1. I was relieved. At least my surgical decisions were now clear to me: immediate bilateral mastectomy with reconstruction, and a total hysterectomy 1½ years later (at age 34).

So why have I told you all this: you need to do two things when faced with medical decisions – go with your gut and get enough information to make a well reasoned medical decision. Do not go to just one doctor. Always get several opinions. Even great doctors often have outdated information or biases and it is confusing to sort all that out.

I made the decisions that were right for me. I found the "standard of care" at the time to be way outdated. I felt I had a genetic disease which needed an immediate cure. I needed surgery to save my life and sanity. I would do it again.

Karen

Ed. Note:
The "Standard of Care" has now been changed. As of March, 2007, The National Comprehensive Cancer Network no longer says that outcome results are the same whether women choose lumpectomy-radiation or mastectomy. After extensive case reviews at multiple leading medical centers, the National Comprehensive Cancer Network (of 21 leading cancer centers in the United States) now recommends that women with a BRCA mutation and an initial breast cancer consider bilateral prophylactic mastectomy (as initial treatment).

November 28, 2006

To whom it may concern,

My name is Debbie Horwitz, I am 34 years old and was diagnosed with Stage 1 breast cancer two and a half years ago in June 2004. My reason for writing this letter is to begin the discussion about women, like me, who are positive for either the BRAC1 or BRAC2 mutation and what this means for us regarding surgery and treatment.

At the time of my diagnosis I had not had gene testing. I am an Ashkenazi Jew and have a very strong family history of breast cancer. My mother died of breast cancer at the age of 39 and my maternal grandmother had breast cancer but died of uterine cancer. I opted not to have gene testing in my twenties, but decided instead to up my GYN appointments and do regular breast exams as well as mammograms. I feared that if I tested positive that young for the mutation, I would be faced with too many decisions that I was unable to face at that particular point in my life. However, what I did know for sure at that time was that if I ever was diagnosed with breast cancer, I would have a double mastectomy in order to reduce my chances of recurrence and also to reduce my worrying about cancer in the other, healthy breast.

With all of that being said, when I was diagnosed in 2004, it seemed I had a very different opinion of how to treat my breast cancer then any of my physicians did. I just assumed they would agree that taking both breasts, given my family history, would be the best scenario for me. Instead, my surgeon said "I can cure this cancer in your right breast with a lumpectomy and it would give you the same cure rate as if I took that breast off." I immediately replied "what about the healthy breast? Can you assure me I won't develop cancer in that breast?" It was clear to me that was not his concern at the time, nor was having me gene tested. He felt that we needed to deal with the lump in the right breast and he also stated that I was young and would not want all of the issues that would come along with a double mastectomy. I was shocked. My father who was in the room is also a physician. He agreed with what my surgeon was saying as did my fiancé. It was a convincing and compelling argument on my surgeon's part, but at the end of the day- I would be the person living with the decisions that were being made.

I went home that night and knew that the only way for me to have peace in all of this is to have the bilateral mastectomy. I was angry that my surgeon did not want that for me because quite honestly, it was a very difficult decision to be making by myself. For the next several months following my bilateral mastectomy I continued to wonder and stew over why my surgeon did not have me gene tested right there at diagnosis to gather more information on my treatment? I wondered why he didn't understand that living would out weight any aftermaths that could follow a double

mastectomy such as: intimacy issues, breast feeding, feelings of loss, etc… I realized I felt cheated. Even though I went ahead and chose the surgery that was best for me to live with, my surgeon chose something else and that has never sat right with me.

So, why bring this up for discussion? Perhaps this is just one case where the surgeon and patient disagreed. I wish that were true. Today, I work full time with young breast cancer patients on a project I created called Myself: Together Again. The project and book were created to empower women through the breast reconstruction process following double mastectomy surgery by showing them pictures of the "process" in order to help them cope. Process pictures are what women need- not simply before and after pictures. Before and after pictures do not tell the real story of reconstruction, and reconstruction itself is a process. As I have worked with these women I am hearing more and more stories like mine where women were "spared" the bilateral mastectomy because their physicians felt it was too much too soon. Fortunately, most of these women received second and third opinions, or like me, just knew what they had to do. Like me these women were angry that they had to be their own advocate in such a terrible situation. They felt more or less abandoned by their physicians and they felt wrong for going against medical advice.

I am choosing to write this letter because we have to bring this issue to the surface. Physicians need to know that sparing women like us is not what we need. We are capable of making decisions for ourselves but we need the medical community to not just give medical facts, but to actually put themselves in our shoes and think about what it is like as a BRAC1 or BRAC2 patient to live with a mutation in your DNA that increases your risk of cancer and then take it a step further and think about what choices we feel like will bring us peace of mind. It is not just about curing the cancer a physician finds. It is about helping that patient be able to accept what lies ahead in his/her life and how the treatments and surgeries that are chosen contribute to overall happiness for that patient.

Today, I can't say I don't live with some regrets that I don't have my breasts. Life is different, I am different. I can say however that I have peace in my life with the decision I have made to have both breasts removed and I feel grateful to not have the fears everyday of what is growing in what would have been my healthy breast. Women deserve peace of mind as much as they deserve to be cured. I want the medical community to wake up to this notion and start treating these patients, (especially the BRAC1 and BRAC2 patients), differently because their risks are greater than those patients without mutations.

In closing, as I read through my genetic report, (I finally had gene testing once I was through my treatments), I read that "women with the BRAC1 mutation that I have, have a 64% chance of being diagnosed with a completely new (second primary) breast cancer." So I wonder, how was it that we did not know this information or care to get this information at the time of my diagnosis? How would I feel today if I went with the lumpectomy and then later had gene testing only to find out

about this risk of a second breast cancer? Fortunately, I do not have to worry, but many women feel cheated when they get to this point and I think that it is only fair for the medical community to acknowledge that on top of all we have been through, we could have handled knowing this information earlier. Also, we could really benefit from recommendations about a double mastectomy even if we chose otherwise. What I am saying is don't make us wait until the new cancer develops to realize that we could have chosen a more drastic approach the first time, but you, the surgeon, did not want us to face that.

I hope this letter helps in some small way open the discussion about BRAC1 and BRAC2 women needing the medical community to advise them of the benefits to a bilateral mastectomy instead of all of the reasons why they should not choose that route. Trust in these women that they will do what is right for them- all you can do is help them know all of the information and that is what we are not getting from surgeons today.

Thank you for being open to this discussion and I can be reached by e mail at info@myselftogetheragain.org

Sincerely,

Debbie Horwitz