Welcome: Uniting our Community

by Sue Friedman

As FORCE enters our 10th year, we’ve made astonishing strides as a community. We’re getting noticed, thanks to the continuous commitment and effort of our members and volunteers. As our outreach, donations and participation steadily grow we’re gaining more attention and more respect. Last December, for example, Time magazine ranked the FORCE-coined term “previvor” third on its “Top 10 Buzzwords of 2007.”

Acknowledgment of our mission is critical because it underscores the need for BRCA research—our best hope for better future options for cancer prevention, detection, risk management and treatment. This issue of Joining FORCEs highlights important new research for our community, including risk-reduction differences between BRCA1 and BRCA2 carriers, ovarian cancer survival in BRCA carriers, and the need for more information on Pre-implantation Genetic Diagnosis and other fertility options for high-risk women.

The need for more BRCA research is our call to action. FORCE is dedicated to the principle that we are important stakeholders in cancer research, and as such, we should have ongoing input into the direction of studies and access to research results. Our 2008 conference represents our community’s inaugural effort to establish hereditary cancer research priorities. I hope you’ll participate in this historic event by attending our conference. For more information or to register, visit www.facingourrisk.org/conference. Read more about the need for this session in the article on page 7.

Raising Awareness and Growing FORCE

by Barbara Pfeiffer

Question: How does an organization with one paid full-time staff member and a small office in Tampa provide life-saving information to thousands of people every day?

Answer: Through a dedicated and fast-growing Outreach community.

Starting with four groups launched at our 2007 conference, FORCE’s Outreach network quickly expanded to 26 groups. Forty-five coordinators now direct group efforts to leverage speaking, education and media opportunities.

In the last year, members shared their journeys with 150 students at Boston University Medical School, addressed a Master’s Level Genetics Counseling class at Fox Chase Center in Philadelphia, and spoke at a Limmud Day of Learning in San Diego. Our volunteers also reached out to other non-profits, hosting tables at Komen Races for the Cure, the LBBC Conference, and Run for Her. Members’ stories were featured on radio, the web, at synagogues, and in print articles from the Jewish Journal of Broward County to the Oregonian and Women’s Day.

We started our Outreach efforts to connect families who face hereditary breast and ovarian cancer with others in their situation. These face-to-face meetings remain an important part of our efforts, even as we’ve increased our focus to include media outreach, fundraising, and advocacy. Last year, we held over 75 meetings in coffee shops, hotels, and members’ homes, welcoming new members and staying in touch with old friends.

If reaching out to help others on behalf of our mission appeals to you, come to our 2008 Conference and meet our Outreach representatives at the roundtable networking breakfast. They’re looking forward to meeting you and signing up leaders for new groups!

View a list of our current Outreach groups at www.facingourrisk.org/support. Sign up today to make sure you receive information on activities in your area. Don’t see your city or state listed? Have time to volunteer? Please send email to barbarap@facingourrisk.org or stop by the volunteer table at our conference.
Setting a Hereditary Cancer Research Agenda

by Sue Friedman

Research is our best hope for better options for future cancer prevention, detection, risk management, and treatment. All cancer research is important, but we need to assure that a portion of research is dedicated to hereditary cancers.

FORCE is committed to promoting research specifically for hereditary cancer. We need dedicated studies because these cancers are different than sporadic cancers in many key ways. Our cancers tend to occur at a younger age, our risk management options are different, and our tumors may respond differently to treatment. Environmental influences may affect us differently than those without hereditary cancer syndromes, and we are more prone to second and even third cancer diagnoses. We share risk with relatives and may also pass our risk to our children.

Have you ever wondered what you can do to help cancer research, and particularly hereditary cancer research? FORCE is offering you an opportunity to make a difference. Attend our annual conference and participate in our “Setting a National Agenda on Hereditary Breast and Ovarian Cancer Research” session.

We have invited representatives from the Centers for Disease Control and the National Cancer Institute to listen to and interact with us as we finally get a say in shaping the future of research for our community. The only way we can begin to advocate for the research we need is to show up in sufficient numbers and let the government agencies and researchers see that we are a united, focused and important community. Some organizations promote the belief that the hereditary cancer community is simply too small and too insignificant to warrant increased resources. We must prove them wrong!

Please join us in Tampa and be part of this historic conference, and particularly this session, as we help promote and shape the future of hereditary cancer research. For more information on setting a hereditary cancer agenda and the Joining FORCEs Conference visit the FORCE conference website at www.facingourrisk.org/conference.

Differences in Breast Cancer Risk Reduction for BRCA1 and BRCA2

by Margaret Snow, MD and Lisa Held

In 2002, two landmark studies, one by Dr. Noah Kauff at Memorial Sloan Kettering and another by Dr. Tim Rebbeck, lead investigator of the PROSE study, demonstrated the benefits of removing the ovaries of BRCA carriers. When women’s ovaries and tubes were removed before menopause, their breast cancer risk was reduced by as much as 75 percent.

New research involving several large cancer centers and recently published in the Journal of Clinical Oncology showed the level of breast cancer risk reduction may differ between BRCA1 and BRCA2 carriers. This study followed BRCA carriers who were deciding whether or not to remove both ovaries to lower their risk of breast cancer and ovarian cancer. It was one of only a few investigations that examined distinct risk reduction in BRCA2 carriers, separate from women with BRCA1 mutations.

BRCA2 study participants who had oophorectomy were 72 percent less likely to get breast cancer. For BRCA1 patients, the study suggested a 29 percent reduction in breast cancer risk. Although the reason for this difference is unknown, the authors suggest that ovary removal may be more protective for women with BRCA2 mutations because BRCA2 breast cancers are more
Testing BRCA Negative: Could It Be Cowden Syndrome?

by Sue Friedman

If your family is BRCA negative could the unexplained cancers among relatives be a result of Cowden syndrome? This inherited disorder is caused by a mutation in a gene called PTEN. It’s not always easy to diagnose a family with Cowden Syndrome because of the broad range of possible features.

People with Cowden Syndrome have increased risk for both benign (noncancerous) and cancerous growths. Types of cancers seen in families with Cowden Syndrome include breast cancer, thyroid cancer, and endometrial (uterine) cancer. Women who have a PTEN mutation may have a lifetime risk for breast cancer as high as 50 percent, and like individuals with BRCA mutations, premenopausal breast cancer (breast cancer diagnosed before age 50) may be more common. Women with Cowden Syndrome also have up to a 10 percent risk for developing uterine cancer. Men with the mutation may also be at elevated risk for male breast cancer. Both men and women with a PTEN mutation have a risk for thyroid cancer that may be as high as 10 percent.

Because some cancers are common to both BRCA and Cowden Syndrome families, the family medical history may appear similar prior to specific identification by genetic testing.

Differences in Breast Cancer Risk Reduction (continued)

likely to be estrogen receptor positive, and removing the ovaries before menopause leaves the body with little estrogen to stimulate cancer cells. Breast cancers in BRCA1 mutation carriers are usually estrogen and progesterone receptor negative.

Although dividing the study groups into BRCA1 and BRCA2 carriers produced specific results of each it did have one unfortunate result: it reduced the total participants in each group. Because few participants developed cancer during the research, the results included large “confidence intervals.” This means that although the data suggests that BRCA1 and BRCA2 carriers have differing levels of protection from oophorectomy, researchers could not conclude without question that the difference in risk reduction between the two groups was different. The study had another limitation: half of the women followed had their ovaries removed after age 45. Further research is needed to show whether oophorectomy before age 45 may offer more protection for BRCA1 carriers than the protection shown in this study; and

continued on page 8

Reference

Voices of FORCE

Each quarter, we’ll invite a FORCE member to share an insightful perspective, a valuable experience, or a touching story to help others who are dealing with issues of hereditary breast and ovarian cancer.

The Impact of Genetic Testing without Genetic Counseling

For a Young Woman

by Lisa Held

A string of family cancer diagnoses and an uncle who worked for Myriad prompted a frenzy of genetic testing among the women in my family. The results were positive—BRCA2 positive.

Just months after I lost my mother to pancreatic cancer, I went with my aunt to her post-mastectomy appointment. At age 20, I had just discovered my own genetically-mutated condition the year before and was grappling with what it meant. Although we were obviously no strangers to cancer, my aunt and I knew nothing about BRCA mutations and hadn’t even considered what the next step should be for me. She thought that her oncologist might provide insight for me. Lisa just found out she’s BRCA2-positive, she explained to the oncologist. What’s your best advice for her?

The doctor, without hesitating, said, ‘Find a guy, get married, have kids right away, and then have your breasts and ovaries removed. It’s your only chance.’ You could hear a pin drop in the room. He stood to leave, then turned, as if remembering the most important point. ‘Oh! And make sure you tell him.’”

‘Hi!’ I asked, my voice shaking. ‘Yes, the guy you’re going to marry. You have to tell him right away.’

Suddenly, in the span of a moment, I didn’t recognize my own life. This man, this man who was going to suddenly marry me with the knowledge of my genetic fate, was going to understand that we’d have babies right away and then quickly slice off all feminine parts of me, where would I find him? And this pathway was willed to me, just like no one else really understands what they face each day. My greatest hope is that with the help of FORCE, all previvors will come to understand that they never need to feel as helpless as I did in that moment.

Lisa Held is a young BRCA2-positive pre-vivor. She lives in New York City and currently works for Facetng History and Ourselves, an international nonprofit organization. She is an aspiring writer and journalist. Lisa is also a coordinator for the newly-formed NY City FORCE Outreach Network.

I have long since abandoned the idea that being a pre-vivor means that my life is predetermined.

Share Your Story

Do you have something to say that may inform our readers or ease their experience? We invite you to share your reflections or personal story about dealing with the issues of hereditary breast or ovarian cancer. Tell us how you feel, how you cope, or what you’ve learned. Email stories of 500-550 words to info@facetingyourisk.org or mail to FORCE, 16057 Tampa Palms Blvd. W #373, Tampa, FL 33647. Please include your name and daytime telephone number so we can contact you if we decide to publish your story in a future issue.

Heather L. Shappell, M.S., CGC, is a genetic counselor and Founder of Informed Medical Decisions (www.informedmedicaldecisions.com), which provides genetic counseling with Board Certified Genetic Counselor by telephone. She is also an Advisory Board Member of FORCE.

Importance of Genetic Counselors

by Heather Shappell, MS, CGC

As a Board Certified Genetic Counselor, I’ve been invited to comment on this very personal story from a professional’s perspective. However, as a compassionate person, I feel the need to respond on an emotional level as well. The first thing I feel compelled to say is, ‘Lisa, I am so sorry that a health care professional responded to you in this careless and inappropriate way.’

BRCA testing is more than a ‘simple blood test.’ Stories like this one are the unfortunate result of well-intentioned health care providers who order genetic tests without considering the patient’s individual situation, and the various aspects of their lives that may be impacted by the results. As a genetic counselor who has focused on hereditary cancer for my entire career, I wish this was the only gut-wrenching story I had ever heard. Unfortunately it is not.

From my experience with a woman who was told by her gynecologist that she was BRCA1 positive, to the one question she was able to get answered about her breast surgery, to the one genetic counselor who has focused on hereditary breast and ovarian cancer a woman in her 20s is very different than the discussion with a woman in her 50s. The concerns and decisions women face vary substantially. It takes a skilled expert to help patients navigate their way through the many risk numbers, and screening and risk-reducing decisions they face because of their BRCA results. Individuals with BRCA mutations (and those who test negative) who feel confused about the meaning of their test results are encouraged to talk with an expert about their results and discuss what medical options they should consider based on their individual situation. It’s never too late to get the facts and the support you deserve!

Over time, as Lisa realized, most people who are tested begin to feel that having this information and taking charge of their cancer screening and risk-reducing plans is empowering. Thankfully, this happens even after they encounter uninformed health care providers who make careless comments along the way.

With organizations like FORCE, this community (genetic counselors included) can take some comfort in knowing that individuals with BRCA mutations have a resource for education and support that can help them deal with and share these negative experiences. More importantly, FORCE gives them a chance to reach out and talk to others facing similar concerns and issues. The importance of this type of camaraderie and support is obvious in Lisa’s story.

Lisa has long since abandoned the idea that being a pre-vivor means that my life is predetermined.
BRCA1/2 Carriers with Ovarian Cancer Show Better Survival Rates

by Lisa Held and Margaret Snow, MD

A study conducted in Israel and published in the January 2008 issue of the Journal of Clinical Oncology showed that ovarian cancer patients with a BRCA1/2 mutation had higher five-year survival rates than women without a BRCA mutation. BRCA carriers diagnosed with ovarian cancer at stage III or IV were much more likely to live beyond five years than women without mutations. Researchers don’t know if this is due to traits in the actual ovarian cancer tumors and/or a better response to chemotherapy.

The study was conducted between 1994 and 1999 and the women were followed for survival until 2003. All 605 participants were Ashkenazi Jewish and all were tested for BRCA mutations. Among these women, 213 (35.2 percent), carried a BRCA1 or BRCA2 mutation. This high percentage of carriers emphasizes the importance of genetic counseling and testing for any Jewish women with ovarian cancer.

Women with a BRCA mutation diagnosed at stage III or IV had a 28 percent reduced risk of dying than women with similar stage cancer and no mutation. There was no significant difference in survival rates among women diagnosed at stages I to II; however, being a BRCA carrier increased median survival by 16 months.

This is not the first study to conclude that ovarian cancer patients with BRCA1/2 mutations have improved survival rates compared to those without the mutations, but it is the largest and the first to follow women prospectively for over 6 years. Researchers acknowledged several other studies with similar results.

Because of research methods, in some cases the application of these results is limited. For instance, the findings of this study may not apply to women who have BRCA mutations other than the three most commonly found in people of Jewish descent. Combined with previous research, this study shows an extremely promising trend.

Although the study did not examine why women with mutations survive longer, the researchers speculated that hereditary tumors may be biologically different and/or may be more sensitive to chemotherapy typically given for ovarian cancer.

Whatever the reason, survival rate differences for BRCA carriers is an important discovery with real implications. These results suggest that it may be beneficial to treat BRCA and non-BRCA tumors differently. Because BRCA carriers with ovarian cancer survive longer, they may be at high risk for breast cancer, emphasizing the importance for these women to consider their options for managing breast cancer risks.

Dr. Margaret Snow is a pre-vivor and a Physical Medicine and Rehabilitation physician who enjoys golfing and photographing birds. She serves as FORCE’s West Michigan Outreach Coordinator.

Could It Be Cowden Syndrome? (continued)

exam results and family medical history, including cancer, to help determine if there may be a PTEN mutation in the family.

Like those who have a BRCA mutation, individuals with a PTEN mutation have a 50 percent chance of passing the gene mutation along to their children. Genetic testing can identify this syndrome; however, because of the complexity associated with diagnosis, families with an increased incidence of cancer should consult with a cancer genetics expert to determine if Cowden Syndrome may be the cause. Families who have the above cancers and who test negative for a BRCA mutation should also consult with genetics experts to determine if testing for a PTEN mutation may be appropriate.

Learn more about Cowden Syndrome in “Making Sense of Uninformative Negative Tests” at our Joining FORCES conference.
Preimplantation Genetic Diagnosis (PGD) is a medical procedure that allows people who carry a disease-causing hereditary mutation to have children who are free from the specific mutation that causes the disease.

The procedure for PGD includes in-vitro fertilization (IVF)—a fertility treatment where the women’s eggs are removed and fertilized in a test tube. When the embryos reach a certain size, one cell is removed and is tested for the hereditary disease in question. Once the genetic status is determined, the parents can decide which embryos they want implanted.

PGD has been used for hereditary diseases such as cystic fibrosis and Huntington’s disease for over a decade. Recently, there has been some debate among the medical community regarding its use for selecting embryos free of gene mutations such as BRCA1/2. Central to this debate is the issue of whether or not this procedure should be used for mutations such as BRCA which don’t cause cancer until adulthood, where not everyone who carries the trait will get cancer, and where there are options available for preventing cancer (such as chemoprevention and surgery).

Previously, this debate has taken place absent input from the BRCA community. Researchers from Moffitt Cancer Center in Tampa teamed up with FORCE to survey FORCE members about their knowledge of and attitudes about PGD. Results from this survey will be used to better address the information needs and design educational materials on PGD for couples who have been affected by hereditary breast and ovarian cancer and who are interested in this technology.

Findings from the survey include:
• 79% of the respondents had never heard of PGD.
• 29% thought PGD could be used for the wrong reasons such as creating “designer babies.”
• 2% were shocked and offended knowing PGD existed, feeling that if their parents had used PGD, they would not exist.
• 53% of the women surveyed felt PGD was an acceptable option to combat hereditary disease.
• 33% would consider using PGD themselves.

This research is important because the medical community often develops educational materials and interventions for a particular group, without consulting that group on their needs and wants. This survey makes it clear that most people who might choose PGD are unaware of this option. More discussion is needed on the topic.

The research article will be published in an upcoming issue of Fertility and Sterility. Information on PGD will be presented at the 2008 Joining FORCES Conference May 16-17 in Tampa, Florida. Visit www.facingourrisk.org/conference for more information.

“Case studies of living with cancer help to understand the value of our research.”

The Moffitt Fertility Preservation Research Group in the Health Outcomes and Behavior Program of Moffitt Cancer Center is focused on examining the psychosocial and communication issues related to reproductive health among at-risk individuals.

More Information About PGD

Websites
www.emedicine.com/med/topic3520.htm
www.rhtp.org/fertility/pgd/default.asp
www.dnapolicy.org/images/reportpdfs/PGDDiscussionChallengesConcerns.pdf
www.ihr.com/infertility/provider/preimplantation-genetic-diagnosis

Scientific Journal Articles Discussing PGD


Parenting Options and Fertility After Cancer
to prove that the difference in risk reduction in BRCA1 compared to BRCA2 carriers was not random.

As expected, the study also found that removing their ovaries protected women with BRCA1 mutations against ovarian cancer. After oophorectomy, women with BRCA1 mutations were much less likely to develop ovarian cancer compared to women who did not have surgery. Even after removing the ovaries, a risk remains for primary peritoneal cancer, a cancer of the abdominal lining that behaves like ovarian cancer and is treated the same way.

Although removing the ovaries and tubes is expected to offer similar protection from ovarian cancer in BRCA2 carriers, in this study the number of BRCA2 carriers with ovarian cancer was too small to show a difference.

The authors of the study support the practice of offering ovarian removal as a cancer prevention strategy both for BRCA1 and BRCA2 carriers.

What’s New @ FORCE

Our website redesign will be unveiled soon. Visit www.facingourrisk.org to see our fresh look with easier navigation, find a support group near you, or read stories shared by FORCE members.

FORCE Receives Spirit of Humanity Award
We are proud to be the 2008 recipient of the Spirit of Humanity Award. Presented to FORCE by the DeBartoloFamily Foundation, the award honors non-profit organizations that provide extraordinary assistance to Tampa Bay residents with special needs. Since 2002, the award has acknowledged the community service of 25 organizations. We appreciate the recognition of our continuing efforts, and the very generous accompanying grant. See www.debartolofamilyfoundation.com/grants to learn more about the DeBartolos and the great work they do.

Help FORCE Go Green
Want to save some trees? Help FORCE save dollars? To receive an electronic version of this newsletter rather than a print copy e-mail us at: newsletter@facingourrisk.org. Include your name and city and state in the e-mail.