BRCA Genetic Testing:
Understanding the Physical, Emotional, and Financial Challenges

December 20th, 2016

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WELCOME

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Sharsheret is a national not-for-profit organization supporting young Jewish women and their families facing breast cancer. Our mission is to offer a community of support to women, of all Jewish backgrounds, diagnosed with breast cancer or at increased genetic risk, by fostering culturally-relevant individualized connections with networks of peers, health professionals, and related resources.
WHY POPULATION SCREENING?

- 1 in 40 Ashkenazi Jews carries a BRCA gene mutation
- Not every high risk family is easy to identify

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Population-based screening for breast and ovarian cancer risk due to \textit{BRCA1} and \textit{BRCA2}

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Contributed by Mary-Claire King, August 19, 2014 (sent for review July 17, 2014; reviewed by Anne M. Bowcock and Karl Skorecki)

In the Ashkenazi Jewish (AJ) population of Israel, 11% of breast cancer and 40% of ovarian cancer are due to three inherited founder mutations in the cancer predisposition genes \textit{BRCA1} and \textit{BRCA2}. For carriers of these mutations, risk-reducing salpingo-oophorectomy significantly reduces morbidity and mortality. Population screening for these mutations among AJ women may be justifiable if accurate estimates of cancer risk for mutation carriers can be obtained. We therefore undertook to determine risks of breast and ovarian cancer for \textit{BRCA1} and \textit{BRCA2} mutation carriers ascertained irrespective of personal or family history of cancer. Families harboring mutations in \textit{BRCA1} or \textit{BRCA2} were ascertained by identifying mutation carriers among healthy AJ males recruited

health burden to the target population; prevalence and attributable risk of disease due to the mutations are known; and effective interventions exist. However, one necessary piece of information remains unknown: What is the disease risk to mutation carriers ascertained from the general population, rather than carriers identified based on family history (13)?

Previous studies assessing cancer risks due to mutations in \textit{BRCA1} and \textit{BRCA2} ascertained carriers through high-incidence families (14), through a single index case with breast or ovarian cancer (3, 15) or through both affected and unaffected carriers (16). In a 1997 study of AJ volunteers, most index cases had no previous cancer diagnosis, but the percentage of index cases with

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Is it time to offer *BRCA1* and *BRCA2* testing to all Jewish women?

K.A. Metcalfe RN PhD, A. Eisen MD, J. Lerner-Ellis PhD, and S.A. Narod MD

It was 2007 when Women’s College Hospital first began to test for *BRCA1* and *BRCA2* mutations among all Jewish women in Ontario. On a research basis, testing was performed regardless of personal or family history of cancer for three recurrent Jewish founder *BRCA* mutations. To date, more than 7000 women have been tested, and the program remains active. Recently, two studies have supported the conclusion that population-based testing is a rational approach to identifying *BRCA* mutation carriers. In an Israeli study of 8105 unselected Jewish men and a British study of 1034 unselected Jewish men and women, more than one half of the 8105 and 1034 tested were found to have mutations.

Our experience is Ontario-based, but should be relevant for Jewish populations elsewhere. The utility of testing will depend to a large degree on the cost of the test provided and of the regional health care system. We do not argue that costs must be covered by a third-party payer (for example, the Canadian public health care system); it could be that the most efficient delivery of services will be based on direct-to-consumer testing offered by a private laboratory. Various scenarios should be explored.

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Dr. Mary-Claire King Proposes Population Screening in All Young Women for BRCA Mutations

By Caroline Halwick
February 10, 2015 (/issues/february-10-2015/)
Tweet this page (https://twitter.com/intent/tweet/?text=%22%3Cp%3EDr.%20Mary-Claire%20King%20Proposes%20Population%20Screening%20in%20All%20Young%20Women%20for%20BRCA%20Mutations%22&url=http://v10-2015/dr-mary-claire-king-proposes-population-screening-in-all-young-women-for-brc-mutations/)

It is not enough for Mary-Claire King, PhD, to have identified the germline BRCA1 mutation associated with hereditary breast and ovarian cancers. Her clinically applicable discovery is one of the world’s greatest in genetics and one for which she has been highly lauded.

But not one to rest on her laurels, Dr. King will not be satisfied, she says, until genetic screening for BRCA1 and BRCA2 is offered to all young women, regardless of family history of cancer. “To identify a woman as a carrier only after she develops cancer is a failure of cancer prevention,” she says.

Dr. King discussed her proposal at the 2014 San Antonio Breast Cancer Symposium, where she was honored with the American Association of Cancer Research Distinguished Lectureship in Breast Cancer Research. Dr. King is Professor of Medical Genetics and Genome Sciences at the University of Washington, Seattle.

“Here’s what an evolutionary biologist thinks we should do next about BRCA1 and BRCA2,” Dr. King began. “My proposal is that we offer population screening for unambiguously damaging mutations in these genes to all women at about age 30. In other words, we move beyond testing only women in severely affected families to testing women regardless of family history of breast or ovarian cancer—who can then undertake preventive action if they learn they carry a mutation.”

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BRCA GENETICS SYMPOSIUM

Sharsheret’s Executive Director Elana Silber Shares the patient perspective at a research conference in Israel

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INCOMPLETE FAMILY HISTORY

- Holocaust
- Few siblings
- Family members are out of touch
- Preponderance of male family members
- Paternal inheritance

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ADVANTAGES OF POPULATION SCREENING

• Finding more women at high risk
• Offering them unique medical management to reduce cancer risk
  – Enhanced screening
  – Risk reducing surgery
• Opportunity to save lives

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HIGH RISK SCREENING

• Annual mammogram
• Annual breast MRI
• Annual breast sonogram
• Biannual breast exam

• Pelvic exam
• Transvaginal sonogram and CA-125
• Ovarian cancer screening has not been proven to increase survival

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RISK REDUCING SURGERY

• Breast
  – Bilateral mastectomy before the diagnosis of cancer
  – Often includes breast reconstruction

• Ovarian
  – Bilateral salpingo-oophorectomy (ovaries and tubes)
  – May consider hysterectomy

• Both procedures offer significant reduction in the chance of ever developing cancer

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DISADVANTAGES OF POPULATION SCREENING

- Individuals without personal or family history of breast, ovarian or pancreatic cancer may not meet insurance guidelines for testing
- The cost of genetic counseling and testing may be several hundred to several thousand dollars

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WHO WILL BENEFIT FROM SURGERY?

- The risk of cancer is not 100%
- The age of onset is not predictable

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SURGICAL COMPLICATIONS

• Recovery from bilateral mastectomy with reconstruction takes weeks or months
• Reconstructed breasts often look fabulous once you are healed, but sensation can be significantly reduced
• Reconstruction of the nipple often involves tattooing, which some may find problematic according to Jewish law
• Rarely, one may develop breast cancer even after prophylactic surgery

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SURGICAL COMPLICATIONS

• Guidelines suggest age for removal of ovaries and tubes
  – *BRCA1*: ages 35-40
  – *BRCA2*: ages 40-45

• Loss of ovaries leads to immediate menopause

• Risks of early menopause (age 45 or younger)
  – Loss of fertility, hot flashes, vaginal dryness, loss of sex drive, and osteoporosis
  – Possible neurologic or cardiovascular risks, studies are not conclusive

• Rarely, one may develop primary peritoneal cancer, which is very similar to ovarian cancer, even after prophylactic surgery

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LOSS OF FERTILITY

• May result from removal of the ovaries and tubes
• May result from a diagnosis with cancer
• May result from the mutations themselves, which may cause an increased chance for premature ovarian failure

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BRCA TESTING AND PRENATAL SCREENING

• Carrier screening has no implications for one’s own health, only implications for potential future offspring
• Being a BRCA carrier has a significant impact on one’s own health and requires a different kind of informed consent

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WHAT’S NEXT?

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**GENETICS FOR LIFE**

- **Sharsheret’s Genetic Counselor** – Speak one-on-one with our experienced genetic counselor about your family history, concerns about cancer risk, and the implications of genetics for you and your family.

- **Free Family Conference Call** – Let us help you communicate with your family about your hereditary cancer risk and proactive steps you can take.

- **Your Jewish Genes** – Read about what’s Jewish about hereditary breast and ovarian cancer in our educational booklet series.

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EMOTIONAL CONSIDERATIONS

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IMPACT OF POPULATION TESTING

Every Family has a History.....
What's Yours?

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TRACING FAMILY HISTORY

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KNOWN FAMILY HISTORY

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NO FAMILY HISTORY

BRCA+

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DIFFICULT DECISIONS

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So that's the first 10. Who's up for the next 603?

JEWISH LAW

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LOST AND FOUND

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COMMUNICATING RESULTS

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HOW WE CAN HELP

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PERSONAL STORY

Miryam, a Sharsheret peer supporter, shares her personal story about navigating the emotional impact of cancer.

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To ask a question, please dial *1 or enter your question into the chat box.

Questions will be addressed in the order received.

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You will be able to access the transcript and audio of the webinar at:

http://www.sharsheret.org/resources/transcripts

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