UCSF Helen Diller Family Comprehensive Cancer Center

Hereditary Cancer Clinic

(415) 353-9797

BRCA1 Mutation Positive Guidelines

Breast Cancer

Education

- Breast self-awareness beginning at age 18
- Seek medical evaluation for changes in the breast
- The majority of breast cancers in women with a BRCA1 mutation are "triple-negative" cancers, and these are more likely to require treatment with chemotherapy

Surveillance

- Clinical breast exam every 6 months beginning at age 25
- Breast MRI once a year beginning at age 25; Use mammogram, if MRI not available
- Add mammogram once a year beginning at age 30, separated 6 months from MRI
- After risk-reducing mastectomy, only clinical breast exam is recommended (no routine imaging is indicated)

Prevention

- Tamoxifen and other preventive medications have been shown to significantly reduce breast cancer risk in women at high risk, however, the data on women with genetic mutations is limited. Women can elect to consult with an oncologist about the potential benefits and risks of preventive medications. Women who choose to take a preventive medication require ongoing surveillance with breast imaging and clinical breast exam.
- Risk-reducing mastectomy with or without reconstruction is a preventive option that reduces breast cancer risk by >90%
- Breast cancer risk may also be reduced in women who have a risk-reducing salpingo-oophorectomy before menopause

Men

- Breast self-awareness and clinical breast exam every year beginning at age 35
- Breast cancer risks in men with a BRCA1 mutation are lower than the risks in women, so there is no current recommendation for preventive medications or surgery

Based on NCCN Guidelines and UCSF Center for BRCA Research Steering Committee
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Ovarian Cancer

Education

- Seek medical evaluation for severe and/or persistent (>2 weeks) of abdominal bloating, abdominal or pelvic pain, changes in bowel habits, frequent urination, or early satiety (feeling full easily)
- BRCA1 mutations also increase the risk for fallopian tube cancer and primary peritoneal cancer

Surveillance

- Transvaginal ultrasound and serum CA 125 may be considered beginning at age 35 with regular surveillance until risk-reducing salpingo-oophorectomy. The benefits of ovarian cancer screening are limited. Women should discuss the benefits and risks of ovarian cancer screening with a health care provider knowledgeable on ovarian cancer
- After risk-reducing salpingo-oophorectomy, there is a small risk of primary peritoneal cancer that remains after surgery though no routine screening is recommended for this cancer. Women should have an annual pelvic exam and symptom evaluation following surgery (as well as Pap smear screening as needed)
- Bone density screening is recommended two years after risk-reducing salpingo-oophorectomy with follow-up testing based on the initial results

Prevention

- Oral contraceptives (if not contraindicated) reduce the risk of ovarian cancer by approximately 50% when taken for 3-5 years
- Risk-reducing salpingo-oophorectomy is recommended between age 35-40, when childbearing is complete or not desired. Risk-reducing salpingo-oophorectomy reduces ovarian cancer risk by >90%. Surgeons performing this surgery should follow the high-risk protocol for surgery and pathology

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Prostate Cancer

Surveillance

- Digital rectal exam and serum PSA beginning at age 40 once a year
- Refer to GU oncology for PSA >2.0

Pancreatic Cancer

Education

- Seek medical evaluation for jaundice, severe and/or persistent (>2 weeks) upper abdominal and/or mid-back pain, unexplained weight loss, or poor appetite

Prevention

- Avoid smoking

Surveillance

- Individuals with a BRCA1 mutation and family history of pancreatic cancer may be candidates for pancreatic cancer screening
- Individuals with a BRCA1 mutation and personal history of pancreatitis may be candidates for pancreatic cancer screening
- If screening is indicated, annual pancreatic cancer screening with either endoscopic ultrasound (EUS) or MRI should begin at age 50 or 10 years earlier than the youngest diagnosis in the family, whichever comes first

Family Members

- Inform siblings, adult children, relatives on the appropriate side of the family
- Offer counseling and consider genetic testing for relatives over 18-25 years
- Kintalk.org- an educational and family communication site for individuals and their families with hereditary cancer conditions

http://kintalk.org/tips-for-sharing-your-genetic-information/

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Reproductive Counseling

- Each child has a 50% chance of inheriting mutation. The mutation is *not* known to cause significant cancer risk under age 25
- If planning to have biological children, recommend testing of partner. If both partners have a BRCA1 mutation, each child has a 25% chance of inheriting both mutations, which causes Fanconi Anemia
- Some data suggest early ovarian aging in women with BRCA1 or BRCA2 mutation
- Women who are choosing RRSO but would like to consider future childbearing can consider oocyte or embryo cryopreservation
- Discuss option of prenatal testing
- Discuss option of preimplantation genetic diagnosis (PGD): When couples do in-vitro fertilization (IVF), it is possible to select embryos that do not have the mutation

Resources

Support Organizations:

- UCSF Psycho-Oncology
- Kintalk
- Facing Our Risk of Cancer Empowered
- Be Bright Pink

Documentary:

- *In the Family* by Joanna Rudnick

Books:

- *Positive Results* by Joi L. Morris and Ora Karp Gordon
- *Previvors* by Dina Roth Port

Genetic Counseling Follow-Up

Recommend genetic counseling follow-up every 1-2 years or as needed to review most current guidelines and research in personal and family context

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