ORDERING PHYSICIAN:

Dr. Tom Hanks
10 Parker Avenue
New York, NY

CLIENT:

Kim Wong
Sex: Female DOB: 01-02-1980
Barcode: 9876789806781384

TEST: Breast and Ovarian Cancer

REPORT DATE: 02-02-2015

CONFIDENTIAL

A PATHOGENIC MUTATION WAS IDENTIFIED IN BRCA1

This positive test means that you have a significantly greater risk of developing breast and ovarian cancer than that of the average US woman. **This result does not mean that you have a diagnosis of cancer or that you will definitely develop cancer in your lifetime.**

Variant Details:

BRCA1 c.181T>G (p.Cys61Gly)

Pathogenic Alternate name(s): C61G, chr17.GRCh37:g.41258504A>C

Transcript: ENST00000357654

Zygosity: Heterozygous

This is an established founder mutation in a population with the disease, with both in-vitro and in-vivo studies supporting a deleterious effect of the variant and reputable external databases also consistently report this variant as pathogenic. Variant occurs in a critical amino acid of a well established protein functional domain.

Other Genes Analyzed: ATM, BAP1, BARD1, BMPR1A, BRCA2, BRIP1, CDH1, CDK4, CHEK2, EPCAM, GREM1, MITF, NBN, PALB2, POLD1, POLE, PTEN, RAD51C, RAD51D, SMAD4, STK11, TP53

No pathogenic genetic variants were identified in the above genes.

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LAB DIRECTOR: _____(XXXX XXXX)

CLIA#XXXXXX CAP#XXXXXX

RISK:

BRCA-1 mutations increase the risk for developing breast cancer from up to 40% at 40 years of age to up to 80% by the age of 80. BRCA-1 mutations also increase the risk of developing ovarian cancer to up to 40% by the age of 60.

HOW DOES THIS RESULT IMPACT YOUR FAMILY?

- This mutation was most likely inherited from either your mother or your father. As such, your relatives on the side of the family from which the mutation was inherited may also have the same mutation.
- Your brother or sister have 50% chance of having inherited this mutation.
- Your children have a 50% chance of inheriting this mutation (irrespective of being male of female).

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PLEASE SPEAK WITH YOUR HEALTH CARE PROVIDER FOR REGULAR SCREENING GUIDELINES.

What happens when a patient tests positive in our breast cancer test?

There are a variety of management strategies available to patients who test positive for a disease-causing mutation These options include enhanced screening, risk reducing surgery, and medication to reduce risk. Following are the NCCN guidelines for management:

For women, increased screening includes the following:

- Increased breast awareness including self breast exam
- Routine clinical breast exam
- Breast MRI and mammogram starting at an early age
- Consider transvaginal ultrasound of the ovaries and CA-125 blood tests
- Consider full body skin exam
- Consider pancreatic cancer screening

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