



August 12, 2011

Greg Bizette, MD
1000 Ochsner Blvd.
Covington, LA 70433

Re: Ms. Debra Dammon

Dear Dr. Bizette:

Ms. Debra Dammon is 56 years of age and has a family and personal history of cancer. She comes to Genetics Clinic for genetic counseling. Ms. Dammon has previously had BRCA1 and BRCA2 gene testing. There was a deleterious mutation in the BRCA2 gene (5804del4).

Family History:

Ms. Dammon is one of five daughters born to her parents. Ms. Dammon was diagnosed with breast cancer and most recently diagnosed with brain cancer. She also reports some cystic changes of her pancreas. One sister had breast cancer at age 30. Two other sisters have had lung cancer. One sister is generally healthy. Ms. Dammon has one son and a grand-daughter who are healthy.

Ms. Dammon's mother was diagnosed with lung cancer and died at age 49. Her father was 63 years of age when diagnosed with stomach cancer. He also had renal cancer. Ms. Dammon's father had three sisters and four brothers. Two of the sisters had breast cancer. The other may have had ovarian cancer. Three of the four brothers had cancer; GI, pancreas, and stomach. The remaining brother died from heart disease.

Ms. Dammon has a number of paternal first cousins who have had cancer. One is a male cousin reported with a BRCA mutation. There are two other paternal relatives with BRCA mutation that have not yet had cancer. There are a number of relatives at risk to inherit the BRCA mutation but they have not been tested.

Genetic Counseling:

Genetic considerations were discussed with Ms. Dammon and her husband. Note is made that the medical record indicates a BRCA1 mutation. However, the laboratory report indicates that Ms. Dammon has a deleterious mutation involving BRCA2. In addition to the high risk for breast and ovarian cancers, mutations of BRCA2 have a slightly greater predisposition for other cancers including pancreatic cancer, prostate cancer, and melanoma among others. Lung cancer most often occurs

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sporadically and is unrelated to the BRCA2 mutation. It is likely that many of the cancers present in this family are due to the influence of the deleterious BRCA2 mutation.

In general, I explained that when a deleterious mutation of BRCA1 or BRCA2 genes is identified in a family then we can extend studies to family members at risk to inherit the mutation. Individuals with the mutation would have a risk for cancer and a risk that the gene could be transmitted to offspring. Women in the family with the mutation remain at high risk for the occurrence of breast and ovarian cancers. They can consider prophylactic surgeries (removal of breasts and ovaries). Those electing not to have surgery should have increased surveillance. Surveillance should occur every six months with imaging to determine that breast and ovarian tissues remain healthy. Of course, individuals who do not inherit a known mutation can be reassured that their risks remain that of the general population.

Screening studies for other cancers associated with BRCA mutations have not been well established. It seems appropriate to include yearly study of the pancreas by abdominal ultrasound for those who have a BRCA2 mutation. Ms. Dammon has been diagnosed with pancreatitis and pseudocyst of the pancreas. She should continue with medical care and studies as established. Skin examination for melanoma including yearly eye exam would be appropriate.

Dr. Bizette, thank you for the opportunity genetic counseling to Mr. and Mrs. Dammon. They will contact family members at risk to inherit the BRCA mutation to notify them that they should consider testing for this defect. If I can assist in this endeavor, I would be happy to do so. Otherwise, follow-up in Genetics Clinic remains at your discretion and that of the family. I certainly hope Ms. Dammon does well and recovers from her difficulties.

Cordially yours,



Duane W. Superneau, M.D.
Our Lady of the Lake Genetic Services

DWS/gh

Cc: Ms. Debra Dammon
117 North Queens Drive
Slidell, LA 70458

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St. Tammany Parish Hospital
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Comprehensive BRCAAnalysis®
BRCA1 and BRCA2 Analysis Result



MYRIAD

PHYSICIAN	
Greg Bizette, MD	
Ochsner Clinic	
1000 Ochsner Blvd	
Covington, LA 70433	

SPECIMEN	
Specimen Type:	Blood
Draw Date:	Apr 08, 2009
Accession Date:	Apr 10, 2009
Report Date:	Apr 16, 2009

PATIENT	
Name:	Dammón, Debra
Date of Birth:	Nov 30, 1954
Patent ID:	
Gender:	Female
Accession #:	00483997-BLD
Requisition #:	902244

Test Results and Interpretation

POSITIVE FOR A DELETERIOUS MUTATION

Test Performed	Result	Interpretation
BRCA1 sequencing 5-site rearrangement panel	No Mutation Detected No Mutation Detected	No Mutation Detected No Mutation Detected
BRCA2 sequencing	5804del4	Deleterious

Analysis consists of sequencing of all translated exons and immediately adjacent intronic regions of the BRCA1 and BRCA2 genes and a test for five specific BRCA1 rearrangements

The results of this analysis are consistent with the germline BRCA2 mutation 5804del4, resulting in premature truncation of the BRCA2 protein at amino acid position 1861. Although the exact risk of breast and ovarian cancer conferred by this specific mutation has not been determined, studies of this type of mutation in high-risk families indicate that deleterious mutations in BRCA2 may confer as much as an 84% risk of breast cancer and a 27% risk of ovarian cancer by age 70 in women (Am J Hum Genet. 62:676-689, 1998). Mutations in BRCA2 have been reported to confer a 12% risk of a second breast cancer within five years of the first (J Clin Oncol 17:3396-3402, 1998), as well as a 16% risk of subsequent ovarian cancer (J Natl Cancer Inst 91:1310-1315, 1999). This mutation may also confer up to a 6% risk of male breast cancer by age 70 and 20% risk of prostate cancer by age 80 (J Natl Cancer Inst 91:1310-1315, 1999), as well as increased (albeit low) risks of some other cancers. Each first degree relative of this individual has a one-in-two chance of having this mutation. Family members can be tested for this specific mutation with a single site analysis.

Please contact Myriad Professional Support at 1-800-469-7423 to discuss any questions regarding this result.

Michael Rhode

Benjamin B. Roe, Ph.D.
Diplomate ABMG
Laboratory Director

Richard J. Wenstrup, MD
Diplomate ABMG
Chief Medical Officer

These test results should only be used in conjunction with the patient's clinical history and any previous analysis of appropriate family members. It is strongly recommended that these results be communicated to the patient in a setting that includes appropriate counseling. The accompanying Technical Specifications summary describes the analysis, method, performance characteristics, nomenclature, and interpretive criteria of this test. This test may be considered investigational by some states. This test was developed and its performance characteristics determined by Myriad Genetic Laboratories. It has not been reviewed by the U.S. Food and Drug Administration. The FDA has determined that such clearance or approval is not necessary.