

**UNDERSTANDING THE BELIEFS OF ASHKENAZI JEWISH INDIVIDUALS
REGARDING CANCER GENETIC COUNSELING SERVICES**

by

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Hereditary Breast and Ovarian Cancer Syndrome (HBOC) is caused by *BRCA1/2* gene mutations. *BRCA1/2* mutations are observed in 1 in 800 in the non-Ashkenazi Jewish population, while an individual of Ashkenazi Jewish (AJ) ancestry has an *a priori* risk of 1 in 40. This pilot study was designed to document common beliefs about cancer genetics services (CGS) and to identify the preferred methods of communication regarding cancer risks and inherited cancer predispositions to the AJ population. Participants were recruited on a voluntary basis from the Jewish community of Pittsburgh, PA to participate in an informal information session about cancer genetics at a local synagogue. Sixteen participants completed surveys with questions pertaining to basic genetics knowledge and beliefs regarding inherited cancer risks, genetic counseling and genetic testing for HBOC before and after the information session, thus allowing researchers to identify changes in genetic knowledge, as well as differences in perceptions about cancer genetics and CGS. Findings revealed that the main motivation to pursue CGS is if an individual perceives they are at a high-risk status to develop cancer based on personal or family history of cancer. The data shows that AJ individuals are aware of cancer genetics and risks associated with their ancestry, but do not pursue or participate in CGS due to a perception of lacking knowledge about general cancer genetics. Although 56.25% of respondents reported that their health care providers are not aware of their AJ ancestry, 75% reported that they preferred to learn of cancer genetics information and CGS through a healthcare provider or physician.

Between the pre- and post-information session surveys, analysis of the knowledge-based questions showed that the average correct response rate for every question increased (77% to 94%). The results of this study have important public health implications because they encourage the idea that a future preferred service delivery model for AJ population-specific genetic counseling may include informal community-based cancer genetics information sessions prior to traditional genetic counseling, thus allowing traditional counseling to focus on personalized risk assessment, benefits and limitations of testing, and potential psychosocial issues that are unique to each individual or family.

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1.0 INTRODUCTION

This study was undertaken to supplement current literature about cancer genetic counseling services (CGS) in the Ashkenazi Jewish (AJ) population and to determine if there are unique motivations for and perceived barriers to AJ individuals obtaining CGS.

For almost two decades, genetic counseling and testing for Hereditary Breast and Ovarian Cancer Syndrome (HBOC) has provided thousands of families with information about cancer risk and options for risk reduction and prevention. Mutations in *BRCA1* and *BRCA2* account for approximately 84% of hereditary breast cancers and over 90% of hereditary ovarian cancers (Petrucelli *et al.*, 2010). *BRCA1/2* gene mutations are observed in approximately 1 in 400 to 1 in 800 in the general population, while an individual of AJ ancestry has an *a priori* risk of 1 in 40, or 2.5% (Claus *et al.*, 1996; Ford *et al.*, 1994; Whittemore *et al.*, 1997; Petrucelli *et al.*, 2010; Foulkes, 2008; Metcalfe *et al.*, 2010). Increasingly, *BRCA* testing is helping individuals at increased risk make decisions regarding cancer surveillance and prevention, with the goal of reducing the incidence and mortality associated with these cancers.

According to the United Jewish Federation (UJF) 2002 Pittsburgh Jewish Community Study, there were approximately 54,000 individuals of AJ ancestry living in 20,900 Pittsburgh metropolitan area households (UJFGP, 2002). Given the statistics from the UJF study and statistically expecting 2.5% to carry a *BRCA1/2* gene mutation, the expected number of AJ individuals in Pittsburgh with a *BRCA1/2* mutation would equal 1,305.

Several published studies have focused on the beliefs of AJ individuals regarding genetic testing for HBOC; however, studies focusing specifically on the unique motivations for and perceived barriers to AJ individuals obtaining genetic counseling are limited. This study was designed to facilitate awareness of CGS, document common beliefs about CGS in the AJ population and identify preferred methods of communication regarding cancer risks and inherited cancer predispositions in the AJ population. The primary aim of this research is to aide in the identification of a preferred service delivery model for genetic counseling specific to the AJ population with respect to HBOC.

1.1 SPECIFIC AIMS

Specific Aim 1: To identify the current awareness, as well as facilitate awareness, of cancer genetic counseling services in the Ashkenazi Jewish population in the Pittsburgh area.

Hypothesis: Individuals of Ashkenazi Jewish ancestry are not aware of cancer genetic counseling services.

Plan: Surveys will inquire about participant's knowledge (or lack of knowledge) of cancer genetic counseling services, including the initial source of knowledge, previous experiences with genetic counseling and any prior awareness of available genetic testing for hereditary cancer predisposition syndromes.

Specific Aim 2: To identify the unique motivations for and perceived barriers to Ashkenazi Jewish individuals obtaining cancer genetic counseling services.

Hypothesis: Personal perception of increased risk to develop cancer based on family history or Ashkenazi Jewish ancestry is the main motivator for seeking cancer genetic counseling services. Lack of information about cancer genetics, cancer risk and recommendations for genetic counseling is the main barrier to obtaining cancer genetic counseling services.

Plan: Survey responses to questions specifically inquiring about motivations for and barriers to obtaining cancer genetic services will be analyzed to try and identify common trends.

Specific Aim 3: To identify the preferred methods of communicating information about inherited predispositions to cancer to the Ashkenazi Jewish population.

Hypothesis: Individuals of Ashkenazi Jewish ancestry often have close family relationships; therefore, communication between informed relatives may be the preferred strategy for disseminating information about cancer genetic services.

Plan: Survey responses to questions specifically inquiring about communication preferences and main sources of support will be analyzed to see if a preferred communication method emerges.

1.2 BACKGROUND OF HEREDITARY BREAST AND OVARIAN CANCER

In the United States, approximately half of all men and one third of all women will develop cancer during their lifetime (ACS¹, 2011). A carcinogenic event can occur as the result of a germline or somatic mutation in a major cancer predisposition gene that is responsible for cell growth or repair, including: proto-oncogenes, tumor suppressor genes and DNA repair genes (Schneider, 2002; Trepanier *et al.*, 2004).

Breast cancer is the second most common type of cancer in American women, behind only skin cancer. In the general population, breast cancer occurs in approximately 1 in 8 women (12%) with a median age of onset of 61 years (ACS², 2011). Generally, the 5-year survival rate for breast cancer in females ranges from 88% (stage I) to 15% (stage IV) (ACS², 2011). Male breast cancer is rare, accounting for approximately 1% of all breast cancers. The 5-year survival rate for breast cancer in males ranges from 96% (stage I) to 24% (stage IV) (ACS², 2011).

Ovarian cancer is the ninth most common cancer in American women, not including skin cancer (ACS³, 2011). In the general population, ovarian cancer occurs in approximately 1 in 70 women (1.5%) and typically develops after the age of 60 (ACS³, 2011). Generally, the 5-year survival rate for ovarian cancer ranges from 89% (stage I) to 18% (stage IV) (ACS³, 2011).

1.2.1 Categories of Cancer

As illustrated in Figure 1, there are three main etiologies of cancer development: a sporadic occurrence, familial predisposition and hereditary predisposition (Claus *et al.*, 1996).

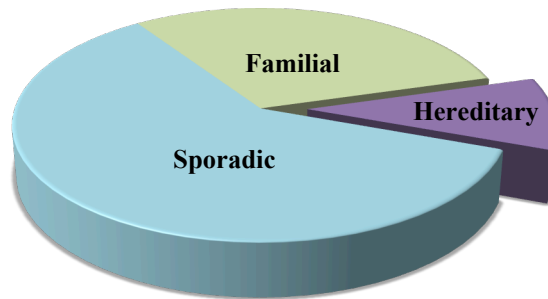


Figure 1. Categories of Cancer

The majority of cancer, approximately 60%, is sporadic, or occurs by chance. Sporadic cancers occur as the result of somatic mutations in major cancer predisposition genes (Trepanier *et al.*, 2004). Somatic mutations are acquired, resulting from lifestyle factors and environmental exposures during the normal aging process, or caused by unknown factors (Amos, 1994; Chen *et al.*, 1994; Trepanier *et al.*, 2004). There are many lifestyle factors and environmental exposures that influence cancer development. The most significant risk factors for the development of breast cancer are gender (being female) and aging (ACS², 2011). Additional risk factors (and risk modifiers) for the development of breast cancer in the general population include:

- Hormonal Factors
 - Early menarche (<12 years) and older age at menopause (> 55 years) (Hulka *et al.*, 2001; Kelsey *et al.*, 1993)
 - Recent, long-term hormone replacement therapy (Chlebowski *et al.*, 2010; Collaborative Group on Hormonal Factors in Breast Cancer, 1997; Beral *et al.*, 2011)

- Age at first live birth (>30 years) and number of pregnancies (nulliparity) (Kelsey *et al.*, 1993; Lambe *et al.*, 1994)
- Oral contraceptive use (risk returns to baseline 10 years after use) (Collaborative Group on Hormonal Factors in Breast Cancer, 1996)
- Breastfeeding has been shown to decrease breast cancer risk (4.3% for every 12 months of breastfeeding), with greater benefit associated with longer duration (Collaborative Group on Hormonal Factors in Breast Cancer, 2002)
- Clinical Factors
 - Benign breast conditions
 - Lobular carcinoma *in situ* (LCIS) can increase breast cancer risk 8 to 10 times that of women of average-risk (Oppong *et al.*, 2011)
 - Atypical ductal/lobular hyperplasia can increase breast cancer risk 4 to 5 times that of women of average-risk (Hartmann *et al.*, 2005; Tamimi *et al.*, 2005; Ashbeck *et al.*, 2007)
 - High breast tissue density can increase breast cancer risk 4 to 6 times that of women with less dense breasts (Cummings *et al.*, 2009; Ginsburg *et al.*, 2008; Boyd *et al.*, 2007; McCormack *et al.*, 2006)
 - High bone density post menopause (Chen *et al.*, 2008; Zmuda *et al.*, 2001; Zhang *et al.*, 1997; Cauley *et al.*, 1996; Kerlikowske *et al.*, 2005)

- Obesity can increase the risk of postmenopausal breast cancer; however, obesity is considered protective against premenopausal breast cancer (ACS², 2011)
- Personal history of breast cancer (especially early-onset, <40 years of age) can increase the risk for subsequent breast cancer 4.5 times that of women without a personal history (ACS², 2011)
- Exposures
 - High-dose radiation between the ages of 10-30 years, most often related to the treatment of lymphoma (ACS², 2011)
 - Exposure to diethylstilbestrol (DES) in utero increases breast cancer risk by 3.9% (Hoover *et al.*, 2011)
 - Alcohol use (more than 2 drinks per day) can increase breast cancer risk by 21% (Singletary *et al.*, 2001)
- Family history (ACS², 2011; Trepanier *et al.*, 2004; Chen *et al.*, 1994)
 - The risk of breast cancer is 1.7 times higher for women with one 1st-degree female relative with breast cancer, nearly 3 times higher for women with two relatives with breast cancer and nearly 4 times higher for women with three or more relatives with breast cancer (Collaborative Group on Hormonal Factors in Breast Cancer, 2001)
 - Family history of ovarian cancer (ACS², 2011)
 - Hereditary cancer syndromes predisposing to breast cancer

Figure 2 illustrates the comparison between the relative risks associated with specific hormonal, clinical and family history factors that can contribute to the development of breast cancer.

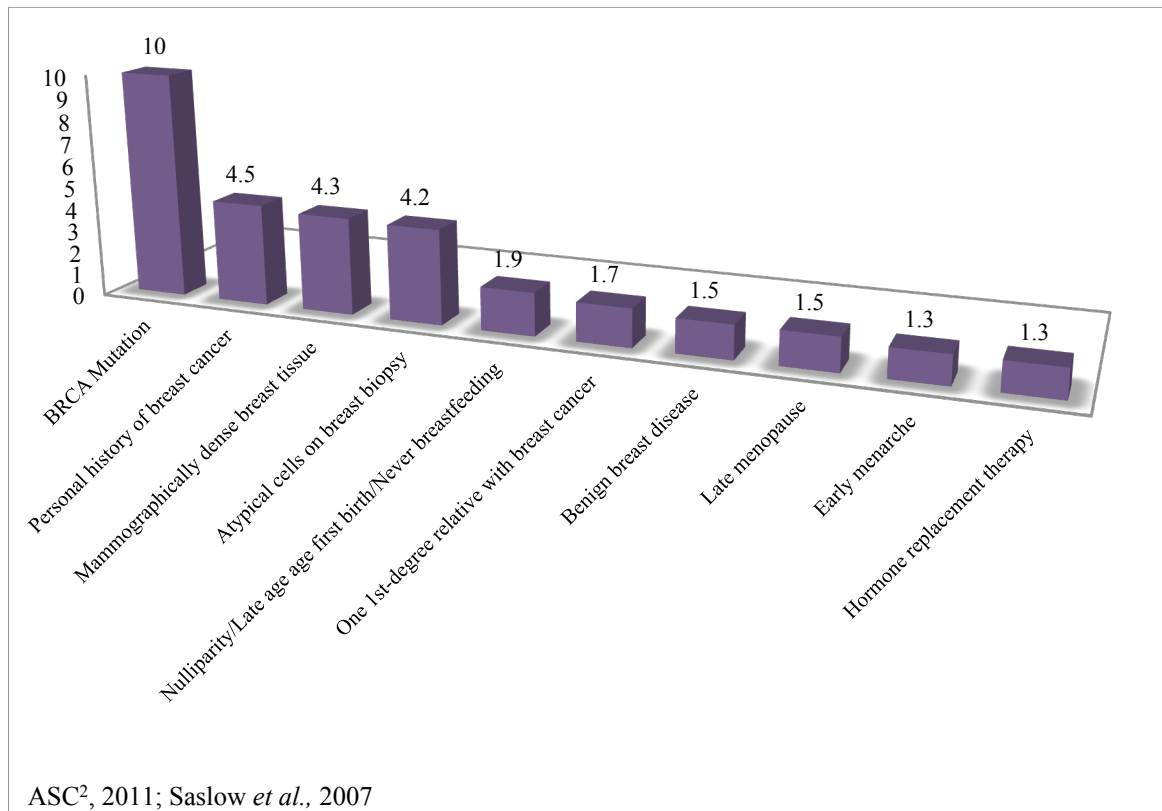


Figure 2. Comparison of Risk Factors for Breast Cancer (Relative Risk)

Environmental and lifestyle risk factors (and risk modifiers) for ovarian cancer in the general population include:

- Hormonal Factors
 - Age of first pregnancy (a pregnancy >35 years of age is twice as protective against ovarian cancer as a pregnancy <25 years of age) and total number of pregnancies (one pregnancy lowers risk by as much as 33% and the reduction in risk increases with each additional pregnancy)

(Whittemore *et al.*, 1992; Risch *et al.*, 1983; Cramer *et al.*, 1983; Joly *et al.*, 1974; Hildreth *et al.*, 1981; Whiteman *et al.*, 2003)

- Infertility is associated with a 2-fold increased risk of ovarian cancer; however, it is unclear whether this risk is due to infertility alone or conflicting data on the impact of fertility drugs (Tworoger *et al.*, 2007; Rossing *et al.*, 2004; Jensen *et al.*, 2009)
- Breastfeeding (<18 months) may decrease the risk for ovarian cancer by as much as 34% (Danforth *et al.*, 2007)
- Oral contraceptive use is associated with a 30 to 50% decreased risk of ovarian cancer if taken for three or more years (Whittemore *et al.*, 1992; Weiss *et al.*, 1996; Beral *et al.*, 2008)

- Clinical Factors

- Increasing age (> 50% of all cases occur in women over the age of 63 years) (ACS³, 2011)
- Obesity (Olsen *et al.*, 2007)
- Long-standing history of ovarian endometriosis (Munksgaard *et al.*, 2011)
- Long-standing history of Pelvic Inflammatory Disease (Lin *et al.*, 2011)
- Regular paracetamol use is associated with a decreased risk for ovarian cancer (Bonovas *et al.*, 2006)
- Hysterectomy and tubal ligation are associated with a 34% reduction in the risk of developing ovarian cancer (Whittemore *et al.*, 1992; Cibula *et al.*, 2011)

- Exposures
 - Talcum powder use (in the perineal area) increases risk by 33% due to contents of asbestos (Cramer *et al.*, 1982; Chang *et al.*, 1997; Harlow *et al.*, 1992; Huncharek *et al.*, 2003)
- Family History (ACS³, 2011)
 - The risk of ovarian cancer is 3.6 times higher for women with one 1st-degree relatives with ovarian cancer and 2.9 times higher for women with one 2nd-degree relative with ovarian cancer (Schildkraut *et al.*, 1989)
 - Hereditary cancer syndromes predisposing to ovarian cancer

Approximately 30% of cancers are “familial”, meaning there is a clustering of cancer within a family or more cases than would be expected by chance. There is no single explanation for these cancers, but they probably result from multiple factors in the environment (in the absence of an identifiable carcinogenic exposure) and multiple genetic factors interacting over time (Trepanier *et al.*, 2004). Familial cancers tend to have a variable age of onset, but may be slightly younger than the general population and are multifactorial in origin. Familial cancer could also represent a clustering of sporadic occurrences (Berliner *et al.*, 2007).

Approximately 7 to 10% of cancers are hereditary, or are caused by inheriting a single gene mutation. Inherited germline mutations in tumor suppressor genes, as well as the acquisition of somatic mutations in the same cell, cause hereditary cancer to develop (Foulkes, 2008). This phenomenon, first described by Dr. Alfred Knudson in 1971, is known as the Two Hit Hypothesis. Knudson theorized that individuals with a germline mutation (first hit) would only need to acquire a somatic mutation in that cell (second hit) to lose control of cell division and lead to carcinogenesis (Knudsen, 1971). Knudson’s hypothesis proved true and led to the

realization that hereditary cancers caused by mutations in specific genes can be associated with some characteristic features, including early age of cancer onset, related cancers found in the same family (in the same bloodline) and unusual or rare tumors (NCCN, 2011).

1.2.2 Hereditary Breast and Ovarian Cancer Syndrome (HBOC)

Hereditary Breast and Ovarian Cancer Syndrome, caused by mutations in the tumor suppressor genes *BRCA1* and *BRCA2*, accounts for approximately 3-5% of all breast cancers and 10% of all ovarian cancers (Petrucelli *et al.*, 2010; ACOG, 2009). *BRCA1/2* mutations occur in all ethnic and racial populations (Claus *et al.*, 1996; Ford *et al.*, 1994; Whittemore *et al.*, 1997).

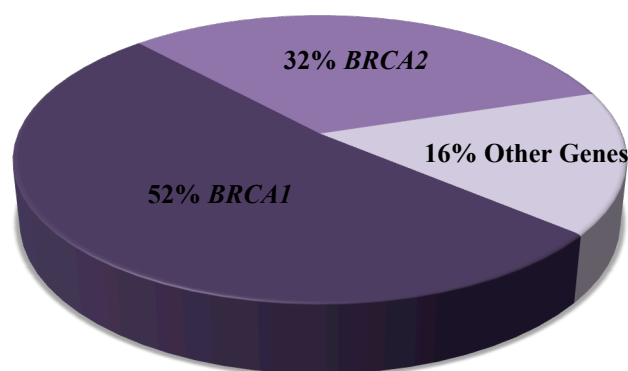


Figure 3. Gene Mutations Leading to Hereditary Breast and Ovarian Cancer

As illustrated above in Figure 3, mutations in *BRCA1/2* account for approximately 84% of hereditary cases of breast cancer, and approximately 16% of HBOC cases are attributed to genes that have yet-to-be discovered (Ford *et al.*, 1998; Robson *et al.*, 2001; Risch *et al.*, 2006; Rubin *et al.*, 1998; Claus *et al.*, 1996; ACOG, 2009). Features suggestive of HBOC include early-onset breast cancer (<50 years), bilateral breast cancer, ovarian cancer, breast and ovarian cancer in the same individual and male breast cancer.

1.2.2.1 The *BRCA* Genes

The *BRCA1* gene, located on chromosome 17q21, was isolated in 1990 and then found to be associated with HBOC in 1994 (Hall *et al.*, 1990; Miki *et al.*, 1994). *BRCA1* encodes the breast cancer type 1 susceptibility protein that interacts with other proteins involved in cell cycle progression, gene transcription regulation, DNA damage response and ubiquitination (Deng, 2006; Rosen *et al.*, 2006, Hall *et al.*, 1990). The *BRCA2* gene, located on chromosome 13q12.3, was identified in 1995 (Wooster *et al.*, 1995). *BRCA2* encodes the breast cancer type 2 susceptibility protein that is involved in the DNA repair process of double-strand breaks (Zhang *et al.*, 1998; Venkitaraman, 2001; Wooster *et al.*, 1994).

There are thousands of unique reported mutations in *BRCA1/2* that can lead to an HBOC-phenotype. Over 1,600 mutations have been identified in *BRCA1* and over 1,800 mutations have been identified in *BRCA2* (Petrucelli *et al.*, 2010). The most common types of deleterious mutations are those that result in missing, abnormal or nonfunctional proteins (nonsense), which account for approximately 88% of the mutations in *BRCA1/2*. Deletions and duplications account for the remaining 12% of deleterious mutations seen in *BRCA1/2* (Walsh *et al.*, 2006).

1.2.2.2 Phenotypes Associated with *BRCA* Mutations

An abundance of studies have been conducted to understand penetrance associated with *BRCA1* and *BRCA2* mutations. As seen in Table 1, multiple studies have shown that *BRCA1/2* mutations have the most significant impact on breast and ovarian cancer risk. The lifetime risk of breast and ovarian cancer for a woman with a *BRCA* mutation has a range of 50 to 87% and 27 to 44%, respectively. The range of risk is a result of incomplete penetrance in some families. It is known that the risk of both breast and ovarian cancer is higher in individuals with *BRCA1* mutations than with *BRCA2* mutations (Antoniou *et al.*, 2003).

Table 1. Lifetime Cancer Risks Associated with *BRCA1/2* Mutations (Breast and Ovary)

Cancer	Population Risk	<i>BRCA1/2</i> Mutation
Breast	8-12% ₁	50-87% ₄
Ovary	<2% ₂	27-44% ₅
2 nd Breast	<10% ₃	2-3% per year ₆
_{1.} ACS ⁴ , 2012 _{2.} ACS ³ , 2011 _{3.} ACS ⁴ , 2012 _{4.} Ford <i>et al.</i> , 1994 _{5.} Ford <i>et al.</i> , 1998 _{6.} Breast Cancer Linkage Consortium, 1999 _{7.} Thompson <i>et al.</i> , 2001 (adapted)		

Several additional studies have revealed that mutations in the *BRCA* genes can increase the lifetime risk for other types of cancer (Thull *et al.*, 2004). Carriers of *BRCA1* mutations can have an increased risk for cancers of the fallopian tube, uterus, cervix, prostate, pancreas, stomach and colon (Ford *et al.*, 1998; Thompson *et al.*, 2002). Carriers of *BRCA2* mutations can have an increased risk for early-onset prostate cancer (before age 55), male breast cancer, melanoma, ocular melanoma and cancer of the pancreas, stomach, gallbladder and bile duct (Breast Cancer Linkage Consortium, 1999; Easton *et al.*, 1997; Edwards *et al.*, 2003).

Many studies have been conducted to understand specific tumor pathology associated with *BRCA1* and *BRCA2* mutations. *BRCA1*-related breast tumors are most likely to be of high histologic grade (medullary histopathology) and are most likely to be classified as “triple negative” breast tumors, meaning that the tumor is estrogen and progesterone receptor-negative and does not demonstrate HER2/neu overexpression (Rakha *et al.*, 2008; Petrucelli *et al.*, 2010). *BRCA2*-related breast tumors do not seem to have a characteristic histopathology or classification, although the information regarding *BRCA2*-related breast tumors is limited (Petrucelli *et al.*, 2010). The only ovarian lesions associated with *BRCA1* and *BRCA2* mutations are invasive, non-mucinous epithelial ovarian tumors of high histologic grade and serous

adenocarcinomas (Petrucelli *et al.*, 2010). The most commonly associated ovarian tumors are serous papillary cystadenocarcinomas. Benign ovarian cysts (e.g. cystadenomas) and borderline tumors (or tumors of low malignant potential) are not part of the *BRCA* spectrum.

1.2.2.3 Ashkenazi Jewish (AJ) Founder Mutations

The majority of hereditary diseases occur across ethnic and racial populations, although there are some populations in which they occur at a much higher prevalence than expected (Petrucelli *et al.*, 2010; Claus *et al.*, 1996; Ford *et al.*, 1994; Whittemore *et al.*, 1997). This is known as the ‘founder effect’, or “the chance presence of certain mutant alleles among the ‘founders’ or ancestors who emigrated to [a particular location] and whose descendants constitute [a certain ethnic or racial group]” (Charrow, 2004).

Several hereditary diseases occur at a higher frequency in the AJ population due to common founder mutations. Individuals of AJ ancestry originated from Eastern Europe, particularly: Hungary, Poland, Russia, Lithuania, Italy, Portugal and Spain. It is hypothesized that the AJ founder effect began in 1500 CE, when Christians ruled medieval Europe. During that time, Jewish individuals were the minority and as a result were genetically and culturally isolated (Hamel *et al.*, 2011). Population genetics concepts such as population bottleneck effect, positive assortative mating, admixture and genetic drift all try to explain various founder mutations that occur in the AJ population (Im *et al.*, 2011; Hamel *et al.*, 2011; Risch *et al.*, 1995, 2003; Ostrer, 2001).

With an estimated prevalence of 1 in 40 or 2.5%, individuals of AJ ancestry are at a significantly increased risk over the general population to have *BRCA1/2* gene mutations due to founder effects (Petrucelli *et al.*, 2010; Foulkes, 2008; Metcalfe *et al.*, 2010). There are three specific HBOC-related AJ founder mutations including two in *BRCA1* (187delAG and 5385insC,

also known as 185delAG and 5382insC, respectively) and one in *BRCA2* (6174delT) (Roa *et al.*, 1996). In individuals of AJ ancestry, the 187delAG mutation and 5385insC mutation in *BRCA1* have been estimated to occur with a frequency of about 1.1% and 0.1-0.15%, respectively (John *et al.*, 2007; Oddoux *et al.*, 1996; Struewing *et al.*, 1995; Roa *et al.*, 1996). The 6174delT mutation in *BRCA2* has been estimated to occur with a frequency of about 1.5% in individuals with AJ ancestry (Struewing *et al.*, 1997; Oddoux *et al.*, 1996; Struewing *et al.*, 1995; Roa *et al.*, 1996). Table 2 outlines the estimated cancer risks associated with the three HBOC-related founder mutations in the AJ population.

Table 2. Lifetime Cancer Risks Associated with *BRCA1/2* Mutations (AJ Founder Mutations)

Cancer	187delAG	5383insC	6174delT
Breast	64%	67%	43%
Ovary	14%	33%	20%
adapted from Antoniou <i>et al.</i> , 2005			

In addition to the three founder mutations in *BRCA1/2*, there are almost 20 different genetic diseases that have associated AJ founder mutations. The most commonly known syndromes associated with AJ ethnicity are different from HBOC in regards to inheritance pattern, penetrance and onset. While HBOC is inherited in an autosomal dominant pattern, the common syndromes associated with AJ ethnicity are inherited in an autosomal recessive pattern and predominately occur during childhood. Some of the syndromes associated with AJ ethnicity and their associated founder mutations are outlined in Table 3, which was adapted from the Victor Centers for Jewish Genetic Diseases and Charrow, 2004.

Table 3. Autosomal Recessive Ashkenazi Jewish Genetic Diseases

Disease	Gene	AJ Carrier Frequency	Clinical Features
Gaucher Disease	<i>GBA</i>	1/14	Hepatosplenomegaly, bone disease, fatigue
Tay-Sachs	<i>HEXA</i>	1/25	Neurodegeneration
Cystic Fibrosis	<i>CFTR</i>	1/26	Pulmonary disease, malabsorption
Familial Dysautonomia	<i>IKBKAP</i>	1/30	Sensory and autonomic neuropathy
Canavan Disease	<i>ASPA</i>	1/40	Neurodegeneration
Spinal Muscular Atrophy	<i>SMN1</i>	1/41	Progressive muscle degeneration
Hyperinsulinemia	<i>ABCC8</i>	1/66	Hypoglycemia, FTT, neurologic damage
Glycogen Storage Disease 1A	<i>G6PC</i>	1/71	Hypoglycemia, hepatomegaly, seizures, short stature
Maple Syrup Urine Disease	<i>BCKDHB</i>	1/81	FTT, neurodegeneration
Fanconi Anemia C	<i>FANCC</i>	1/89	Bone marrow failure, high risk of malignancy, dysmorphic features
Niemann-Pick A	<i>SMPD1</i>	1/90	FTT, hepatosplenomegaly, neurodegeneration
Joubert Syndrome	<i>TMEM216</i>	1/92	Brain malformation, hypotonia, dysmorphic features
Dihydrolipoamide Dehydrogenase deficiency	<i>DLD</i>	1/96	FTT, neurodegeneration
Bloom Syndrome	<i>BLM</i>	1/100	High risk of malignancy
Usher Syndrome III	<i>CLRN1</i>	1/107	Progressive hearing and vision loss
Nemaline Myopathy	<i>NEB</i>	1/108	Muscle disease, absent deep tendon reflexes
Mucopolidosis IV	<i>MCOLN1</i>	1/125	Neurodegeneration, eye findings
Usher Syndrome IF	<i>PCDH15</i>	1/141	Congenital hearing loss, adolescent-onset vision loss
adapted from Victor Centers for Jewish Genetic Diseases, 2012; Charrow, 2004			

1.2.3 Genetic Counseling for HBOC

The goal of cancer genetic counseling is to help “empower the patient to make informed decisions regarding screening, prevention and genetic testing by providing him or her with the necessary genetic, medical and psychosocial information” (Berliner *et al.*, 2007; Lerman *et al.*, 1995, 1997; Bernhardt *et al.*, 2000; Lobb *et al.*, 2001; Meiser *et al.*, 2002; Pasacreta, 2003). Genetic counseling for HBOC typically consists of both pre- and post-test counseling and begins with family history interpretation, cancer risk assessment and psychosocial assessment.

If the individual is an appropriate candidate for *BRCA1/2* testing, informed consent should be obtained (ASCO, 2003). At a minimum, informed consent should include patient education of cancer genetics, discussion of medical management guidelines, discussion of the genetic testing process, benefits and limitations of genetic testing, discussion of possible psychosocial issues associated with genetic testing and identifying relevant resources and support in the community (Berliner *et al.*, 2007; Stopfer, 2000).

1.2.3.1 Family History Interpretation and Cancer Risk Assessment

Cancer genetic risk assessment is the process of identifying individuals at an increased risk to develop cancer due to hereditary components. A complete risk assessment for HBOC involves analysis of the family pedigree, discussion of an individuals' personal medical history and relevant exposures and appropriate use of risk models.

The family pedigree is the most important aspect to assessing the probability of a hereditary component to cancer. Berliner *et al.*, 2007, states that there are several indicators for HBOC in a family, including:

- Pre-menopausal breast cancer
- Ovarian cancer
- Bilateral breast cancer, or breast and ovarian cancer in the same individual
- Male breast cancer
- Two or more individuals in the family with breast and/or ovarian cancer
- AJ ancestry

There are some instances when the family pedigree is not helpful in assessing risk for HBOC. These complications include: limited family history information; small sized families; early deaths in the family (unrelated to cancer diagnosis); predominantly male relatives in the family; and adoption (Trepanier *et al.*, 2004; ACOG, 2009; Weitzel *et al.*, 2007).

There are several different risk calculation models available to assess likelihood of identifying a *BRCA1/2* mutation in the patient or family. Two such risk models used in clinical practice include the Myriad Prevalence Tables (myriadtests.com/provider/brca-mutation-prevalence.htm) and BRCAPRO (Parmigiani *et al.*, 1998). Myriad Genetics Laboratories, a molecular diagnostic company, publishes mutation prevalence tables using data gained from clinical testing services. Myriad tables estimate *BRCA1/2* mutation probability based on Myriad prevalence rates, the individual's cancer history, family history and AJ ethnicity. BRCAPRO uses a Bayesian analysis of conditional probabilities to estimate the likelihood of a *BRCA1/2* mutation based on the individual's cancer history, family cancer history and AJ ethnicity.

The combined information from a cancer risk assessment calculation tool provides an estimate for an individual's probability to carry a *BRCA1/2* mutation. An estimated probability of 10% or greater to have a *BRCA1/2* mutation is often used to determine appropriateness of HBOC genetic testing (ASCO, 2003). Cancer genetic risk assessment also helps determine appropriate candidates for genetic testing (Berliner *et al.*, 2007).

1.2.3.2 Differential Diagnosis

While HBOC accounts for the majority of hereditary breast cancers and ovarian cancers, there are additional, more rare, hereditary cancer predispositions to breast and ovarian cancer (Claus *et al.*, 1996; ACOG, 2009; Thull, 2004). Similar to HBOC, these other hereditary cancer

syndromes are associated with early-onset cancers, high penetrance and follow an autosomal dominant inheritance pattern (Trepanier *et al.*, 2004). See Appendix A for a brief overview of the other hereditary predispositions to breast and ovarian cancer.

1.2.3.3 Medical Management Options

While the cancer risks for individuals with a *BRCA1/2* mutation are significantly increased over those of the general population, there are medical management options available for risk reduction and prevention. The options include increased screening/surveillance, chemoprevention and risk-reducing surgeries. The National Comprehensive Cancer Network 2011 Guidelines are outlined in Table 4 and Table 5 (NCCN, 2011).

Table 4. Medical Management Options for HBOC-associated Breast Cancer

Increased Surveillance	Chemoprevention	Prophylactic surgery
Monthly breast-self exams beginning between age 18 to 25 years	Medications (Tamoxifen/ Arimidex) can reduce breast cancer risk by 50% when taken for 5 years	Preventative mastectomy can reduce breast cancer risk by 90%
Annual or semiannual clinical breast exams beginning between age 25 to 35 years		Preventative removal of the ovaries before menopause can reduce breast cancer risk by 50%
Annual mammogram beginning between age 25-35 years		
Annual breast MRI		
adapted from National Comprehensive Cancer Network, 2011		

Table 5. Medical Management Options for HBOC-associated Ovarian Cancer

Increased Surveillance	Chemoprevention	Prophylactic surgery
Annual or semiannual transvaginal ultrasound	Oral Contraceptives can reduce ovarian cancer risk by 60% when taken for ≥ 5 years	Preventative removal of the ovaries can reduce ovarian cancer risk by as much as 96%
CA-125 blood test		
adapted from National Comprehensive Cancer Network, 2011		

1.2.4 Genetic Testing for HBOC

Molecular genetic testing for HBOC has been utilized for over a decade, first becoming clinically available in October 1996. Diagnosis of symptomatic individuals is the main clinical use of molecular genetic testing for HBOC, although predisposition testing for at-risk relatives is also utilized (Petrucelli *et al.*, 2010).

1.2.4.1 Recommendations and Guidelines for Genetic Testing

The American Society of Clinical Oncology (ASCO, 2003) outlines basic recommendations for genetic testing for cancer susceptibility. The recommendations state that genetic testing should be offered when there is:

- Personal or family history features suggesting a genetic cancer susceptibility
- The genetic test can be adequately interpreted
- The results will aid in the diagnosis or influence the medical management of the patient or family members at hereditary risk of cancer

Testing criteria specific to HBOC are published by the National Comprehensive Cancer Network (NCCN, 2011). The testing criteria include:

- Individual from a family with a known *BRCA1/2* mutation
- Personal history of breast cancer (IDC or DCIS) AND 1 or more of the following:
 - Diagnosed ≤ 45 years
 - Diagnosed ≤ 50 years with ≥ 1 close relative with breast cancer aged ≤ 50 AND/OR ≥ 1 close blood relative with ovarian cancer at any age
 - Two breast primaries when 1st diagnosis occurred ≤ 50 years
 - Diagnosed < 60 years with a triple negative breast cancer
 - Diagnosed < 50 years with a limited family history
 - Diagnosed at any age with ≥ 2 close blood relatives with breast cancer AND/OR ovarian cancer at any age
 - Male relative with breast cancer
 - Personal history of ovarian cancer
 - Ashkenazi Jewish or other high-risk background
- Personal history of ovarian cancer
- Personal history of male breast cancer
- Personal history of breast and/or ovarian cancer at any age with ≥ 2 close blood relatives with pancreatic cancer at any age
- Personal history of pancreatic cancer at any age with ≥ 2 close blood relatives with breast and/or ovarian and/or pancreatic cancer at any age

- Family history only
 - 1st- or 2nd- degree relative meeting any of the above criteria
 - 3rd- degree relative with ≥ 2 close blood relatives with breast AND/OR ovarian cancer (≥ 1 close blood relative with breast cancer aged ≤ 50 years)

1.2.4.2 Genetic Testing Methodologies

There are several clinical methods available for molecular genetic testing of the *BRCA* genes, including sequence analysis, targeted mutation analysis and deletion/duplication analysis. Testing is preformed using DNA obtained from a sample of peripheral blood or an oral sample obtained by a buccal rinse and results are usually available within two weeks. The majority of genetic testing for the *BRCA* genes is performed at Myriad Genetic Laboratories in Salt Lake City, Utah due to patent rights of the sequence analysis. However, there are currently seven laboratories in the United States that perform targeted mutation analysis: Myriad Genetic Laboratories, Boston University School of Medicine, Fox Chase Cancer Center in Philadelphia, PA, New Jersey Medical School, University of California Los Angeles, University of California San Francisco, University of Chicago and University of North Carolina Hospitals.

Targeted mutation analysis looks for the three HBOC-related founder mutations in individuals reporting AJ ancestry, detecting an estimated 90% of mutations in this population (Petrucelli *et al.*, 2010). Site-specific testing identifies the presence or absence of a known familial mutation without having to sequence the entire gene (NCCN, 2011; Petrucelli *et al.*, 2010). Targeted mutation analysis ranges in cost from \$325 to \$2,975 depending on the lab performing the test and is usually covered by most insurance plans (Deegan *et al.*, 2010; Myriad², 2010).

Comprehensive analysis, termed “BRACAnalysis” by Myriad Genetic Laboratories, has a detection rate of 85% and consists of full sequencing, as well as detection of five common large genomic rearrangements in *BRCA1* (Petrucelli *et al.*, 2010; Frank *et al.*, 1998; Walsh *et al.*, 2006; Unger *et al.*, 2000).). The total cost is \$3,340 and 90% of health insurance plans will cover the cost at 90% or better (Myriad², 2010). BRACAnalysis Large Rearrangement Test, termed “BART” by Myriad Genetic Laboratories, is complementary to comprehensive analysis and looks for additional large genomic rearrangements in *BRCA1* and *BRCA2* if no mutations are detected with standard comprehensive analysis. BART examines all coding exons and promoters of *BRCA1* and *BRCA2* for deletions and duplications, picking up an additional 3-4% of mutations (Petrucelli *et al.*, 2010; Myriad¹, 2010). For patients meeting specific criteria established by Myriad Genetic Laboratories based on both a personal diagnosis of cancer and family history, BART is free of charge. There is an additional charge of \$700 for individuals that do not meet these criteria and to date, many insurers do not cover this additional testing.

1.2.4.3 Genetic Testing Strategies and Result Interpretation

To ensure adequate interpretation of test results, it is standard of practice that genetic testing for *BRCA1* and *BRCA2* be initiated in a member of the family with a history of breast and/or ovarian cancer whenever possible because: a) It will clarify the risk of other HBOC-associated cancers for the affected family member; b) If no mutation is identified, testing unaffected family members is not necessary or useful (thus conserving healthcare resources); and c) If a mutation is identified, it makes test results in unaffected family members more informative and allows for site-specific testing of family members.

For individuals of AJ ancestry, it is recommended that initial testing consist of targeted mutation analysis for the three specific founder mutations. If no mutations are detected with the three-site panel and the individual meets NCCN Testing Criteria for HBOC despite their AJ ancestry, reflex to comprehensive analysis is performed due to the possibility that an AJ individual could have a *BRCA* mutation that is not one of the three founder mutations (NCCN, 2011; Petrucelli *et al.*, 2010). It is suggested that other individuals within a family of AJ descent who wish to undergo genetic testing include analysis for all three common AJ founder mutations rather than just being tested for the mutation previously identified in the family because of reports of coexistence of two founder mutations in some AJ families due to the frequency of these mutations (Lavie *et al.*, 2011; NCCN, 2011; Petrucelli *et al.*, 2010).

There are four possible test results from analysis of the *BRCA* genes. A true positive test result means that an individual is a carrier of a mutation in *BRCA1/2*, which increases the risk for HBOC-associated cancers. A true negative result indicates that an individual is not a carrier of a *BRCA1/2* mutation previously identified in the family. A “no mutation detected” result describes an individual who was not found to be a carrier of a *BRCA1/2* mutation and the carrier status of other family members may either be positive, negative or unknown. Lastly, a Variant of Uncertain Significance (VUS), result offers information about an alteration in *BRCA1/2* for which the risk for HBOC-associated cancers with the particular alteration is unknown.

1.2.4.4 Understanding the Benefits and Limitations of Genetic Testing

An integral component to pre-test counseling for HBOC is a discussion of the benefits and limitations of testing. The main benefit is a personalized risk assessment. Information about mutation status can help in making informed choices regarding medical management strategies.

Another benefit of genetic testing for HBOC includes an accurate risk assessment for other members of the family; identifying a *BRCA* mutation in one family member enables other relatives to determine whether or not they share the same cancer predisposition.

Genetic testing for HBOC is not perfect and it is necessary that the limitations be presented to the patient as well as the benefits. The detection rate for *BRCA* testing is 85 to 90%, depending on the population and test method; however, not all mutations can be detected using current testing technologies (Walsh *et al.*, 2006). For this reason, a negative test result is most informative when there is a known mutation in the family. It is possible that the genetic test will detect a VUS and the contribution of the variant to cancer risks is unknown. In the event that a VUS is detected, medical management decisions and further testing options are based on personal and family history. In addition, the *BRCA* genes are not the only genes that contribute to hereditary breast cancer and hereditary ovarian cancer (Berliner *et al.*, 2007).

The theoretical concern of genetic discrimination is often discussed during cancer genetic counseling sessions. A long-time barrier of genetic testing has been fear of insurance discrimination based on genetic test results. In 1996, the government enacted the Health Insurance Portability and Accountability Act (HIPAA), which protects patient's privacy and provides some protection against genetic discrimination with regard to health insurance for individuals with group policies (Fleisher *et al.*, 2001; Trepanier *et al.*, 2004). In 2008, the government enacted the Genetic Information Nondiscrimination Act (GINA), which protects patients from potential discrimination from health insurances and employers based on genetic information (Petrucelli *et al.*, 2010).

1.2.5 Psychosocial Issues Related to HBOC

There is a wide array of psychosocial issues that can arise before, during and after genetic counseling and/or testing for HBOC. When individuals receive the results of their genetic test and are informed of their mutation status, they will no longer be at an arbitrary “increased risk” status; they will be faced with the knowledge of actual cancer risks (whether increased or decreased, based on testing results). An individual’s personal cancer history, as well as stage of life, can have a large impact on the reaction to their test results as well as their thoughts concerning medical management strategies.

For individuals who have had an HBOC-associated cancer, a positive test result can bring mixed emotions. For some, a positive test result can bring a sense of relief because it provides an “explanation” for the cancer diagnosis. For others, a positive result may bring a sense of anxiousness, sadness, or fear because the individual is now faced with additional cancer risks. Some individuals can experience both relief and anxiousness after receiving a positive result (Douglas *et al.*, 2009).

For an individual that has never had cancer, learning that they are positive for a *BRCA* mutation places that otherwise healthy individual at substantially increased risk for potentially life-threatening illnesses. “Previvor” is a term coined in 2000 by individuals on the Facing Our Risk Of Cancer Empowered (FORCE) community blog describing “unaffected carriers” of *BRCA* mutations as survivors of a predisposition to cancer. Cancer previvors face unique challenges and stress over the difficult decisions that come with the many medical management options for those with a *BRCA* mutation (FORCE, 2012).

Just like a positive result, learning that no mutation in *BRCA* was detected can bring mixed emotions. For most, a negative test result is reassuring and brings relief because the

individual is not known to have substantially elevated risks for the development of HBOC-related cancers (Lerman *et al.*, 1998). For some, a negative result can be associated with “survivor guilt”, especially if a mutation has previously been identified in the family and was inherited by siblings but not themselves (Wagner *et al.*, 2000; Tibben *et al.*, 1992; Huggins *et al.*, 1992).

An individual’s stage of life can also have a large impact on their thoughts concerning medical management strategies and influence their decision making process with regards to surveillance versus prophylactic surgeries (Wagner *et al.*, 2000).

Regardless of personal cancer history or stage of life, many individuals who test positive for a *BRCA* mutation contend with possible guilt and worry over passing a mutation to children, as well as having concern for creating guilt for a parent or grandparent from whom the mutation was inherited. Individuals may feel a strong psychosocial burden over having to inform the family of a mutation and being the “bearer of bad news” (Lubinsky, 1994). Individuals who learn they carry a *BRCA* gene mutation may experience depression. Although most *BRCA* mutation carriers can cope with this information over time, some individuals experience prolonged periods of depression or are unable to adjust to this genetic diagnosis and require referral for more involved psychosocial support.

1.2.5.1 Patient Support and Resources

There are many different sources of support for individuals and families with HBOC. Numerous websites, outreach programs and support groups are dedicated to helping individuals wanting support and information about HBOC.

FORCE is a nonprofit organization dedicated to providing information, resources and support to individuals and families facing hereditary breast and/or ovarian cancer

(www.facingourrisk.org). Bright Pink is a nonprofit organization dedicated to helping young women who are at high risk for breast and ovarian cancer (brightpink.org). Bright Pink aims to provide education to empower women to take control of their health and medical management strategies. Sharsheret, meaning “chain” in Hebrew, is a nonprofit organization dedicated to helping Jewish young women and their families who face breast cancer (www.sharsheret.org). Sharsheret helps young women with breast cancer or a *BRCA* mutation to make “culturally-relevant individualized connections” to peers, support groups, health professionals and other resources. FORCE, Bright Pink and Sharsheret are just a few of the numerous organizations that provide excellent resources for the HBOC-community.

1.3 PREVIOUS RESEARCH OF POPULATION-SPECIFIC GENETIC TESTING AND COUNSELING

It is important for genetic counselors to be aware of a patient’s ethnic, racial and religious background because it can provide important information about how a patient will interpret and utilize genetic information, as well as allow for multi-culturally sensitive and specific genetic counseling (Berliner *et al.*, 2007; Mitchell, 1998; Trepanier *et al.*, 2004). When counseling individuals of AJ ancestry, it may be important to be aware of the ethical understandings of genetic testing from the Jewish code of ethics, as well as to be aware of previous research pertaining to the beliefs of AJ individuals in regards to genetic testing and counseling for HBOC.

1.3.1 Ethical Implications of Genetic Testing in the AJ Population

The approach to health care and decision-making in western culture is driven by autonomy, meaning that the individual has the right to make their own decisions concerning health care (Callahan, 2003). Under Jewish law, health care and decision-making is driven by obligation and responsibility to protect one's health, not an individual's "right" to make their own decisions (Steinberg, 2003). Under Jewish law (Deuteronomy 4:9; 4:15), having a diagnosis of cancer and not seeking treatment or undergoing routine screenings recommended by a physician would be defiant because it would be going against the law of "guard[ing] [one's] health" (Mor *et al.*, 2008; Fisch, 1984). Genetic testing for HBOC does not specifically fall under this tenant of Jewish law because having a mutation in a *BRCA* gene implies a predisposition to cancer, not a diagnosis of cancer. However, rabbinical consensus is consistent with western medical professionals' recommendations: in most cases where there appears to be a hereditary predisposition for breast and/or ovarian cancer, there is an obligation to test (Mor *et al.*, 2008; Steinberg 2003).

While the decision, or "obligation", to test is supported by Jewish law in the Orthodox community, the decision to undergo risk-reducing surgeries is not as straight forward and the idea of sharing genetic information, even with family members, is sometimes considered a breach of Jewish law (Mor *et al.*, 2008). Sharing information with Orthodox family members about a *BRCA* mutation could impact marriage prospects for others in the family, bring fear of social discrimination and label the family as "defective" (Rosner, 1998; Mor *et al.*, 2008). Unless a genetic counselor has prior knowledge of possible social implications for Orthodox Jewish

women, genetic counseling and testing for HBOC in this population may not be effective. Reports in the literature are limited in regards to the ethical implications of *BRCA* testing for individuals with Reform Judaism or Conservative Judaism beliefs.

1.3.2 Previous Research of CGS for HBOC in the AJ Population

Genetic counselors having a prior awareness of possible motivations for and unique barriers to cancer genetic counseling and testing in the AJ population may be a helpful aide for implementing more effective counseling strategies. There have been several studies conducted that focus on the beliefs of AJ individuals regarding genetic testing for HBOC; however, studies that focus specifically on genetic counseling for HBOC in the AJ population are limited.

Phillips *et al.*, 2000, conducted a multicenter study of 134 Canadian women with AJ ancestry using questionnaires that examined the factors that influenced their decision to undergo genetic testing for *BRCA1/2*. This study found that the main motivating factors for testing included [their] desire to contribute to research, implications for family members and “the need to know”. Study participants felt that the main discouraging factors to testing included fear of insurance discrimination, potential impact on marriage prospects and concern about the negative focus on the Jewish community.

Lehmann *et al.*, 2002, conducted a population-based study of 200 AJ women through telephone surveys that examined their attitudes towards genetic discrimination and *BRCA1/2* testing, as well as perceived advantages and disadvantages of *BRCA1/2* genetic testing. Their study revealed that there is significant variation among AJ women’s interest in *BRCA1/2* testing, however, the majority of participants were not concerned about group discrimination based on *BRCA1/2* test results. In fact, 95% of the study population felt that research focused on Jews was

either neutral or good. The main perceived advantages of *BRCA1/2* genetic testing included obtaining information about [their] children's risk of disease and valuing information for its own sake. The main perceived disadvantages of *BRCA1/2* genetic testing included fear of insurance discrimination and increased anxiety from knowing mutation carrier status.

Kelly *et al.*, 2004, conducted a study using repeated-measures surveys that examined cancer genetics knowledge and beliefs before and after traditional genetic counseling and their relationship to receipt of results for *BRCA1/2* mutations in 120 highly educated AJ individuals. The study population included individuals who had a personal or family history suggestive of a *BRCA1/2* mutation. Their study revealed that genetic counseling is helpful in improving overall knowledge of cancer genetics even for highly educated AJ individuals, although continued communication regarding the implications of genetic risk may require additional educational materials and may need to be conducted over time.

Bowen *et al.*, 2003, conducted a study of 221 AJ women from Seattle, WA that explored the connections between Jewish identity (cultural identification and religious practice) and interest in screening behaviors (mammography, breast self-exam, genetic testing). This study was part of a larger study that examined the efficacy of two counseling methods for AJ women (Bowen *et al.*, 2006). Study participants completed surveys and multiple regressions were examined. Findings revealed that cultural identity positively predicted interest in testing, whereas religious identity was inversely related. Religious identity was a significant predictor of intention to adhere to mammography recommendations. Findings show that culture and religion, although correlated, may have different associations with health attitudes.

Bowen *et al.*, 2006, conducted a study of 221 AJ women with average or moderately increased risk of breast cancer to test the efficacy of two counseling methods: individual genetic

counseling and psychosocial group counseling. Individual genetic counseling emphasized information and explaining hereditary cancer risk. Psychosocial group counseling emphasized discussion of emotions, distress, and coping with cancer risks. Researchers reported that many AJ women initially overestimated their risk but that genetic counseling lowered risk perceptions. The study revealed that providing AJ women who are of average or moderately increased breast cancer risk with either traditional genetic counseling or psychosocial group counseling reduced worry about cancer, lowered inflated perceptions of breast cancer risk and decreased interest in having genetic testing.

1.3.3 Previous Research of Population-specific Genetic Counseling for HBOC

Several research studies have shown lower rates of cancer genetic counseling and *BRCA1/2* testing in individuals belonging to minority populations, specifically the African American (AA) population, than those in the general population (Forman *et al.*, 2009). With the knowledge that diligent screening and early detection of cancer increases the chances for a better health outcome, this health disparity encouraged researchers to understand the knowledge, attitudes and emotional barriers to cancer genetic counseling and testing in the AA population specifically (Lerman *et al.*, 1999; Williams, 1999; Kendall *et al.*, 2007).

Researchers discovered perceived barriers for obtaining CGS in the AA population, including: concern for ethical implications and discrimination, differing levels of genetic knowledge, resistance to risk-reducing strategies and the belief that [their] health was “in God’s hands” (Thompson *et al.*, 2003; Singer *et al.*, 2004; Kendall *et al.*, 2007). While concerns of

blame and guilt are not unique to the AA population, these barriers have frequently been reported in regards to genetic testing for hereditary cancer predispositions (Thompson *et al.*, 2003; Kendall *et al.*, 2007).

Lerman *et al.*, 1999, conducted a randomized trial of two hundred twenty-eight Caucasian women and 70 African American (AA) women with a family history of breast or ovarian cancer to investigate racial differences in response to two alternate pretest education strategies for *BRCA* genetic testing: a standard education model and an education plus counseling model. The standard education model only provided information about genetic testing to participants, and the education plus counseling model incorporated the information with additional discussion relating to psychosocial issues in genetic testing. This study found that the effects of the interventions on testing intentions in AA women differed significantly from Caucasian women. The education plus counseling model in the AA women led to greater increases in intentions to be tested than the education only model. In Caucasian women, there was no notable difference on outcome despite different interventions. These findings are indicative that genetic counselors need to be aware of the unique aspects to cancer counseling in the AA population, as well as tailoring genetic counseling for individuals who report AA ancestry.

While the information that was gained from these studies supports the idea of population-specific genetic counseling, there has been little research conducted specifically on AJ individual's beliefs regarding CGS for HBOC (Lehmann *et al.*, 2002). In order to best serve the AJ community, genetic counselors need to learn from the research done in the AA population and embrace the idea of AJ population-specific genetic counseling.

2.0 EXPERIMENTAL DESIGN AND METHODS

This pilot study, which culminated in an information session, was conducted on December 11, 2011 from 10:00-11:30am at Rodef Shalom Congregation in Pittsburgh, Pennsylvania. The study was a collaboration of efforts between the University of Pittsburgh and the Cancer Genetics Program of the West Penn Allegheny Health System, which provides genetic counseling and testing services, as well as outreach genetic counseling services in Western Pennsylvania.

The study facilitated analysis of the current awareness of, perceived beliefs regarding and preferred methods of communicating cancer genetics information in the AJ population. Surveys completed by the study participants before and after the information session helped to assess knowledge gained and a difference in opinions regarding CGS. Approval was obtained from the Institutional Review Boards at the University of Pittsburgh and Allegheny General Hospital of the West Penn Allegheny Health System (Appendix B).

2.1 PROTOCOL

2.1.1 Advertisement and Recruitment

The information session was advertised as a program about Jewish genetic diseases, specifically HBOC. Advertisement for the information session began two months prior to the event and

continued until the day before the event. The various forms of advertisement can be found in Appendix C and consisted of: an advertisement in the Rodef Shalom Bulletin; an article in the Jewish Chronicle; flyers that were distributed at Rodef Shalom, Allegheny General Hospital and a Jewish Genetic Disease Screening event at Hillel Jewish University Center of Pittsburgh in November; and by word of mouth.

Two genetic counselors greeted all attendees before the information session and briefly explained the study and invited them to participate. If the individual expressed interest in the study, they were given a packet of information containing the pre- and post-information session surveys (Appendices F and H), a copy of the PowerPoint presentation about HBOC given during the session (Appendix G), several educational brochures about HBOC (described in section 1.2.5.1), a handout discussing the Jewish perspective of taking care of the body (Appendix D) and the program agenda (Appendix E). The individuals who declined to participate in the study were still provided with the educational brochures, program agenda and a copy of the PowerPoint presentation. In accordance with Institutional Review Board (IRB) regulations for pilot studies, a maximum of 25 individuals could participate in the research study. However, no attendees were turned away from participating in the event.

2.1.2 Surveys

Participants were given a pre-information session and post-information session survey to complete. The pre-session survey was to be completed before the start of the information session and included questions pertaining to demographics and basic genetic knowledge including inherited cancer risks, genetic counseling and genetic testing for HBOC. The post-session survey was to be completed following the information session and asked the same questions as the pre-

session survey, allowing researchers to identify changes in genetic knowledge, as well as differences in perceptions about cancer genetics and genetic counseling services from before the information session. Participants were given the option to stay after the session and complete the post-session survey at Rodef Shalom, or return the post-session survey using a provided pre-paid and pre-addressed envelope.

2.1.3 Participants

The study participants were AJ individuals with an interest in HBOC. The participants were recruited on a voluntary basis from the Jewish community of Pittsburgh, Pennsylvania, with the majority of participants learning of the event through the advertisement in the Jewish Chronicle. A large proportion were also members of Rodef Shalom Congregation. Participants understood that they would not be compensated for the study. The only incentive for the participants was gaining knowledge of hereditary cancer information and the services available to assess cancer risks and medical management options.

Inclusion criteria included AJ ancestry by birth (as reported by the participant) and participants had to be over age 18 because genetic testing for inherited susceptibilities to HBOC in children is not indicated. Gender and race were not included as inclusion or exclusion criteria. Exclusion criteria included mentally incompetent individuals or members of any other legally restricted group.

2.1.4 Information Session

The session, led by genetic counseling intern Rachel Pearlman, consisted of guest speakers telling their personal stories, a PowerPoint presentation on HBOC and concluded with a panel Question and Answer session.

The session opened with an introduction from Rabbi Amy Hertz, followed by guest speakers Mr. Jay Rogal and Mrs. Barbara Rogal and Mrs. Kathy Pattak. Rabbi Hertz briefly spoke about the Jewish perspective “Shmirat HaGuf”, which is Hebrew for “taking care of the body”. Mr. and Mrs. Rogal spoke about their personal experience with Gaucher’s Disease, as well as their involvement in Jewish Genetic Disease Screening within the Jewish community of Pittsburgh. Mrs. Kathy Pattak, an AJ individual who carries a *BRCA2* mutation, spoke about her personal experience with genetic testing, counseling and medical management decisions.

The information that was included in the PowerPoint presentation on HBOC consisted of an overview of genetics, inherited cancer syndromes, risk assessment, available medical management options, available genetic testing, benefits and limitations of testing and access to these services (Appendix G).

The program concluded with a panel Question and Answer session consisting of two genetic counselors, a genetic counseling intern and Rabbi Hertz.

2.2 DATA ANALYSIS

After the information session was complete and the surveys were returned, the collected data from the pre- and post-session surveys were analyzed using a qualitative descriptive method. Descriptive statistics were produced for selected characteristics of the sample; means, ranges and frequencies, were reported where appropriate. Likert scales were used to analyze information and themes were identified.

3.0 RESULTS

3.1 RESPONSE RATE

Forty-five individuals attended the event. In accordance with the IRB, 25 individuals were consented to participate in the study. Of the 25 individuals who agreed to participate, 16 individuals completed and returned the pre-session survey (64%). Of the 16 individuals who returned the pre-session survey, 8 individuals also completed and returned the post-session survey (50%). Therefore, 32% (8/25) of individuals who originally agreed to participate in the research study completed both the pre- and post-information session surveys.

3.2 DEMOGRAPHICS

Data from 16 individuals that attended the information session and completed the questionnaires were used in this study. Table 6 illustrates the characteristics of the participants by several categories including: gender, age, marital status, family status, education level and religious affiliation.

Table 6. Characteristics of Participants (Demographics)

Variable (n=16)	Category	Number of Responses	(%)
Gender	Female	11	68.75
	Male	5	31.25
Age	18-25	1	6.25
	26-30	0	0
	31-40	2	12.5
	41-50	0	0
	51-60	6	37.5
	61-70	5	31.25
	70+	2	12.5
Marital Status	Single	1	6.25
	Married	11	68.75
	Divorced	1	6.25
	Widowed	1	6.25
	In a relationship	2	12.5
Family Status	Children	13	81.25
	No children	3	18.75
Education level	Some high school	0	0
	High school graduate	1	6.25
	Some college	1	6.25
	College graduate	6	37.5
	Graduate/professional school	8	50
Religious status	Reform	10	62.5
	Conservative	3	18.75
	Orthodox	2	12.5
	Jewish	1	6.25
	Other	0	0
	None	0	0

All individuals were of AJ ancestry. Twice as many females (68.75%) participated in the study than males (31.75%). The ages of study participants were grouped and the groups ranged from 18 years to 70+ years. The majority of the participants, 37.5%, were between 51 and 60 years of age. The majority of participants were married (68.75%), had children (81.25%) and had a post-High School education. 50% had a graduate/professional degree and 37.5% had a college degree. The majority of participants identified with Reform Judaism (62.5%).

Table 7 illustrates cancer-specific characteristics of the study participants including: personal diagnosis of cancer, family history of cancer diagnosis (first-degree or second-degree), type of cancer (breast and/or ovary cancer or other type of cancer) and age range of cancer diagnosis (>50 or <50).

Table 7. Characteristics of Participants (Cancer-specific)

Variable (n=16)	Category	Number of Responses	(%)
Personal cancer diagnosis	Br/Ov cancer > 50	2	12.5
	Br/Ov cancer < 50	0	0
	Other types of cancer	1	6.25
	No cancer	13	81.25
First-degree relative with cancer diagnosis	Br/Ov cancer > 50	2	12.5
	Br/Ov cancer < 50	5	31.25
	Other types of cancer	3	18.75
Second-degree relative with cancer diagnosis	Br/Ov cancer > 50	2	12.5
	Br/Ov cancer < 50	3	18.75
	Other types of cancer	1	6.25

The majority of individuals that participated (81.25%) had never been diagnosed with cancer. There were no participants that were diagnosed with breast or ovarian cancer before age 50. 31.25% of participants reported having a first-degree relative diagnosed with breast and/or ovarian cancer under the age of 50 years and 18.75% of participants reported having a second-degree relative diagnosed with breast and/or ovarian cancer under the age of 50 years.

3.3 AWARENESS OF CGS

Study participants were asked a variety of questions in the pre-session survey concerning awareness of CGS, specifically; genetic counseling, general genetic testing and genetic testing for hereditary cancer predispositions. Table 8 shows participant responses of awareness prior to attending the information session.

Table 8. Awareness of CGS

Variable (n=16)	Category	Number of Responses	(%)
Previous knowledge of GC	Yes	14	87.5
	No	0	0
	No response	2	12.5
If yes: Reason for previous knowledge of GC	Personal experience	5	35.7
	Healthcare provider	2	14.3
	Relative	6	42.85
	Friend	4	28.6
	Newspaper/magazine	2	14.3
	Internet	2	14.3
	Synagogue	4	28.6
	Other (work)	1	7.1
Previously had genetic testing	Yes	11	68.75
	No	5	31.25
	Unsure	0	0
Previously had genetic counseling	Yes	9	56.25
	No	7	43.75
	Unsure	0	0
Previous knowledge of testing for hereditary cancer predisposition	Yes	12	75
	No	1	6.25
	No response	3	18.75

The majority of individuals (87.5%) reported that were previously aware of genetic counseling and 75% were aware of testing for hereditary cancer predisposition before attending the information session. Two individuals who completed the pre-session survey did not respond to this question, so they were not included in the analysis.

Of the 14 individuals that reported having prior knowledge of genetic counseling, the main source was communication with relatives (42.85%). 35.7% of respondents reported having a previous personal experience with genetic counseling. Communication with friends and information from the synagogue tied with 28.6% for the third most common reason for prior knowledge of genetic counseling. 68.75% of respondents previously underwent genetic testing and 56.25% of respondents participated in genetic counseling. It should be noted that some individuals selected more than one answer for how they learned of genetic counseling.

3.4 BELIEFS REGARDING CGS

Study participants were asked to rate a variety of possible motivations for and perceived barriers to obtaining CGS for HBOC using a five-level Likert scale. Participants were asked to do the same in the post-session survey so that changes due to information gained during the information session could be analyzed.

To analyze motivations for seeking CGS, Table 9 illustrates the participant's personal perceived risk for cancer development based on their personal and family cancer history. Analysis of the pre-session surveys revealed that 6 of the 16 participants responded to the question inquiring about personal perceived cancer risk, so the individuals who did not respond were not included in the analysis of personal perceived cancer risk.

Table 9. Perceived Cancer Risk Based on Personal and Family History

Variable (n=16)	Level of Perceived Risk									(%)
Perceived cancer risk	Low (0-3)			Neutral (4-6)			High (7-10)			
	Pre	Post	Diff	Pre	Post	Diff	Pre	Post	Diff	
Personal cancer diagnosis (br/ov)	0	0	0	0	0	0	1	1	0	16.6
No family history	1	1	0	0	0	0	0	0	0	16.6
First-degree relative with br/ov cancer diagnosis	1	1	0	0	0	0	1	1	0	33
Second-degree relative with br/ov cancer diagnosis	1	1	0	0	0	0	0	0	0	16.6
First-degree relative with cancer diagnosis (not br/ov)	0	1	0	1	0	0	0	0	0	16.6
Second-degree relative with cancer diagnosis (not br/ov)	0	0	0	0	0	0	0	0	0	0

As would be anticipated, the participants with a personal history of cancer perceived their cancer risk as high. The participant with no family history of cancer perceived their risk as low. One participant with a 1st-degree relative with breast and/or ovarian cancer perceived their cancer risk as high, while another participant with a 1st-degree relative with breast and/or ovarian cancer perceived their cancer risk as low. Analysis of the post-session surveys revealed that one participant (16.6%) went from feeling that their cancer risk was neutral to feeling that their cancer risk was low.

Participant's motivations for participating in cancer genetic counseling and testing are illustrated in Table 10. It should be noted that 4 of the 16 individuals who completed the pre-session survey did not respond to the questions inquiring about perceived benefits to CGS, so they were not included in the results. Because not all participants completed the post-session survey, Table 10 is split into analysis of participants who completed the pre-survey only (n=12) (top portion of table) and analysis of participants who completed both the pre- and post-survey (n=8) (bottom portion of table).

Table 10. Benefits for Seeking CGS for HBOC

Benefit (n=12)	Disagree (1-2)			Neutral (3)			Agree (4-5)		
	Pre	Post	Diff	Pre	Post	Diff	Pre	Post	Diff
Knowledge of increased risk is useful	1			0			3		
Knowledge of mutation status is useful	1			0			3		
Knowledge of risk would influence medical management decisions	1			0			3		
Knowledge of increased risk is useful	0	0	0	0	0	0	8	8	0
Knowledge of mutation status is useful	0	0	0	1	0	-1	7	8	+1
Knowledge of risk would influence medical management decisions	0	0	0	2	0	-2	6	8	+2

Analysis of the pre-session surveys revealed that the majority of participants agreed that the greatest benefit to undergoing genetic counseling for HBOC was the usefulness of knowing increased cancer risk status (91.6%). The majority of participants also felt that knowledge of mutation status would be beneficial (83.3%) and knowledge of cancer risk status would influence their medical management decisions (75%).

Analysis of the post-session surveys revealed that one person (12.5%) went from feeling neutral to agreeing that knowledge of mutation status would be useful. Two (25%) people went from feeling neutral to agreeing that knowledge of cancer risk status would influence their medical management decisions. The post-session survey revealed that 100% of the participants felt that knowing increased cancer risk status, mutation status and their influences on medical management decisions were equally beneficial.

Questions dedicated to ascertaining perceived barriers to participating in cancer genetic counseling and testing were also analyzed and are displayed in Table 11. It should be noted that 8 of the 16 individuals who completed the pre- survey did not return the post-survey. Because

not all participants returned the post-survey, Table 11 is split into analysis of participants who completed the pre-survey only (n=16) (top portion of table) and analysis of participants who completed both the pre- and post-survey (n=8) (bottom portion of table).

Table 11. Perceived Barriers to Obtaining CGS for HBOC

Barrier (n=16)	Disagree (1-2)			Neutral (3)			Agree (4-5)		
	Pre	Post	Diff	Pre	Post	Diff	Pre	Post	Diff
Painful/difficult	5			0			1		
Increase anxiety/worry	3			1			2		
Feel guilty if passed to children	3			1			1		
Unable to find a GC in the area	4			0			1		
Fear of insurance discrimination	2			0			2		
Feel ashamed/singled-out in community	3			0			1		
Fear of knowing cancer risk	2			0			4		
Lack of genetic information	2			0			5		
Unsure of family history	2			2			3		
Test result would be beneficial	1			0			4		
Cost	4			1			1		
Painful/difficult	6	6	0	1	0	-1	1	2	+1
Increase anxiety/worry	2	2	0	1	1	0	4	5	+1
Feel guilty if passed to children	3	4	+1	2	1	-1	3	3	0
Unable to find a GC in the area	8	7	-1	0	1	+1	0	0	0
Fear of insurance discrimination	6	5	-1	1	2	+1	1	1	0
Feel ashamed/singled-out in community	8	7	-1	0	0	0	0	0	0
Fear of knowing cancer risk	1	1	0	1	2	+1	6	5	-1
Lack of genetic information	1	3	+2	1	0	-1	6	5	-1
Unsure of family history	1	3	+2	2	0	-2	5	5	0
Test result would be beneficial	0	0	0	0	0	0	8	8	0
Cost	6	4	-2	1	1	0	1	2	+1

Analysis of the pre-session survey revealed that the majority of participants agreed that the greatest barrier to seeking CGS was lack of genetic information (68.75%). Fear of knowing cancer risk (62.5%) and not knowing family history (50%) were also among the greatest perceived barriers. The majority of participants disagreed that fear of pain or difficulties obtaining testing were barriers (68.75%). The majority of participants disagreed that cost (62.5%), inability to find a genetic counselor (75%), fear of insurance discrimination (50%) and fear of feeling ashamed or singled-out in the Jewish community (68.75%) were barriers.

Analysis of the post-session survey revealed that the majority of participants agreed that the greatest barriers to obtaining CGS for HBOC included lack of genetic information (62.5%), an increase in anxiety or worry (62.5%), fear of knowing cancer risk (62.5%) and uncertainty of family history (62.5%). In addition, analysis of the post-session survey revealed that the majority of participants disagreed that the inability to find a genetic counselor (100%), fear of insurance discrimination (87.5%) and fear of feeling ashamed or singled-out in the Jewish community (100%) were barriers to obtaining CGS for HBOC.

3.5 PREFERRED COMMUNICATION STRATEGIES

As illustrated in Table 12, study participants were asked a variety of questions in the pre-session survey concerning preferred methods of communication in regards to cancer risk and family history, as well as main sources of support (other than family) and physician awareness of AJ ancestry.

Table 12. Communication Strategies

Variable (n=16)	Category	Number of Responses	(%)
Best method for communicating risk	Healthcare provider	12	75
	Relative	6	37.5
	Friend	4	25
	Newspaper	2	12.5
	Internet	4	25
	Synagogue	5	31.25
	Other	0	0
Main sources of support (other than family)	Healthcare provider	2	12.5
	Religious organization	6	37.5
	Support group	4	25
	Colleagues	1	6.25
	Friendships	8	50
Physician awareness of AJ ancestry	Yes	3	18.75
	No	9	56.25
	Unsure	1	6.25
	No response	3	18.75

The majority of participants (75%) felt that their healthcare provider was the best method for communicating information about cancer risk and family history. Other preferred methods of communication included relatives (37.5%) and the synagogue (31.25%). Other than family, participants reported that their religious organization (37.5%) was their main source of support in terms of communicating cancer risk and family history. 56.25% of individuals reported that their health care providers were unaware of their AJ ancestry.

3.6 CHANGE IN KNOWLEDGE

The participants were asked a series of knowledge-based questions, found in the pre- and post-session surveys. The majority of questions were multiple-choice with a few true/false questions. The answers to all questions were provided during the information session. Table 13 illustrates the comparison of the participant's answers from the pre- and post-surveys (n=8), as well as an analysis of gain in knowledge.

Table 13. Assessment of Knowledge

Question	Correct (n=8)		
	Pre	Post	Difference
1. The majority of all cancer is hereditary (T/F)	7	8	+1
2. How common is breast cancer in the general population?	5	6	+1
3. How common is hereditary breast cancer in the general population?	3	6	+3
4. How common is hereditary breast cancer in the AJ population?	5	8	+3
5. Cancer is only passed through your mother (T/F)	7	8	+1
6. Having a cancer gene means you will definitely develop cancer (T/F)	8	8	0
7. Inheriting a mutation in a cancer gene increases the risk that you will develop specific types of cancer (T/F)	7	8	+1
8. How many genes must someone inherit to have HBOC?	5	7	+2
9. How can you tell if someone carries a gene that increases the risk for HBOC?	6	8	+2
10. If a person has genetic testing and no mutation is found in a cancer gene, that person will never develop cancer (T/F)	8	8	0
11. A mutation in a breast cancer gene also contributes to a higher risk of ovarian cancer (T/F)	6	8	+2
12. Our genetic code (DNA) consists of A, T, G, C (T/F)	5	8	+3
13. According to HIPAA, genetic testing results can be used for insurance discrimination (T/F)	8	8	0

Analysis of the pre-survey revealed that the majority of the questions posed had at least a 62.5% (5/8) correct response rate, with an average correct response rate of 77% (6.15/8). Question 3, inquiring about the incidence of hereditary breast cancer in the general population, had the lowest correct response rate of 37.5% (3/8). All participants (100%) correctly answered questions 6, 10 and 13 on the pre-survey.

Analysis of the post-surveys revealed that a gain in knowledge occurred with every question (excluding the three questions (6, 10 and 13) that all participants answered correctly on the pre-survey), with an average correct response rate of 94% (7.5/8). The questions with the greatest gain in knowledge (3, 4 and 12) had an increase in correct responses by 37.5% (+3/8).

4.0 DISCUSSION

This study was designed to document common beliefs about CGS and identify preferred methods of communication regarding CGS in the AJ population; in addition, a second goal of the information session was to facilitate awareness of CGS. The implications of the study findings are vast and with further studies, will aid in the future identification of a preferred service delivery model of AJ population-specific genetic counseling with respect to HBOC.

4.1 AWARENESS OF CGS

The first aim of the study was to identify the current awareness of CGS in the AJ population. It was hypothesized that individuals of AJ ancestry were not aware of cancer genetic counseling or testing. Analysis of the surveys completed by study participants revealed that the majority of participants *were* aware of the availability of cancer genetic counseling, with the majority stemming from family communication. This understanding allows focus to be placed on the preferred method of delivery rather than facilitating awareness of genetic counseling for cancer genetics.

It should be noted that 35.7% of participants reported their prior awareness of CGS due to a previous personal experience with genetic counseling, so perhaps individuals attending this information session were coming for additional information and the targeted population of those

who were unaware did not attend. In addition, it is possible that the individuals who participated in the study did so because they had prior awareness of CGS. Perhaps a greater portion of AJ individuals than are represented in this study are actually not aware of cancer genetic counseling and testing for HBOC and that is why they did not attend the information session.

Interestingly, 28.6% of participants reported their prior awareness of CGS was from communication with friends, as well as the synagogue. One bias of this study could be that the information session was advertised by and held at Rodef Shalom Congregation. Perhaps individuals who are affiliated with an institution (like Rodef) may feel a community connection to it and would be more inclined to participate in information sessions that were in the community setting they were already comfortable with. The results of the information session may (or may not) have been different had the information session been hosted at a location that has a more sporadic attendance (like a library or an informal meeting hall) instead of regular gatherings and a sense of community.

4.2 BELIEFS REGARDING CGS

The second aim of the study was to identify any preconceived beliefs (motivations and barriers) about CGS. It was hypothesized that the main motivation for AJ individuals to seek CGS was perceived high-risk status to develop cancer based on family history or AJ ancestry and survey responses were in support of this hypothesis. It was also hypothesized that lack of one's personal knowledge about cancer genetics was the main barrier preventing AJ individuals from obtaining

CGS and survey analysis found that the participant's own perception of lacking knowledge about general cancer genetics is a self-identified barrier that is contributing the underutilization of CGS in the AJ community.

Analysis of the pre- and post-session surveys revealed that the information session did change the perception of one participant in terms of perceived cancer risk (the individual went from feeling neutral risk to feeling low risk). This finding, which supports the idea of AJ population-specific genetic counseling, is reminiscent of the Bowen *et al.*, 2006 study, which reported that genetic counseling for AJ women of average or moderately increased breast cancer risk reduced worry about cancer, lowered inflated perceptions of breast cancer risk and decreased interest in having genetic testing.

When reporting on individual's perception of cancer risk, it is especially important to recognize that an individual's perception of "high-risk" or "low-risk" based on family history can be extremely variable, as seen with the participants of this study. One individual with a 1st-degree relative with breast and/or ovarian cancer perceived their personal risk as high, while another individual with the same family history perceived their risk as low. Another participant with a 2nd-degree relative with breast cancer and a 2nd-degree relative with ovarian cancer perceived their risk as low, saying "Since I am 76 and my sons are 51 and 49 and our grandchildren range from 8 to 12 (all our sons), I haven't given much thought to genetic testing". Therefore, genetic counseling can provide formal risk assessment and discuss the differences between an individual's perception of risk versus calculated risk based on family history.

Other than personal perceived high-risk status, analysis of the pre- and post-session surveys found that the information session did change some opinions of the participants for certain perceived benefits. One participant went from feeling neutral to agreeing that knowledge

of mutation status would be useful and influence their medical management decisions. Two participants went from feeling neutral to agreeing that knowledge of cancer risk status would influence their medical management decisions. This finding, an interest in medical management decisions, is also important to keep in mind when designing a preferred service delivery model for AJ-specific genetic counseling. Because AJ individuals are at an increased risk over the general population to carry a *BRCA1/2* mutation, emphasizing the different screening, chemoprevention and surgical options to help manage cancer risk can empower AJ individuals once they know their mutation status (whether positive or negative).

The information session did change some opinions of the participants for certain perceived barriers. One participant went from feeling neutral to disagreeing that the possibility of feeling guilty if a gene mutation was passed to their children was a perceived barrier, suggesting an increased understanding of the transmission of *BRCA* mutations (feelings of guilt were reduced with the understanding that parents cannot control the genes they pass to their children as discussed during the informational portion of the event). Two participants felt like cost was a perceived barrier after the information session, suggesting that these individuals were not aware of the cost of genetic testing prior to the information session. Two participants went from agreeing or feeling neutral to disagreeing that feeling unsure of how to find a genetic counselor and lack of genetic information were perceived barriers, suggesting that the information session provided adequate information to the participants and helped them feel prepared for seeking traditional genetic counseling.

Analysis of the data from pre- and post-session surveys shows that the majority of AJ individuals are aware of cancer genetics and risks associated with their ancestry, but 68.75% of participants do not pursue or participate in genetic counseling due to their own perception of

lacking knowledge about general cancer genetics. This self-identified barrier is difficult to interpret as the average correct response rate of the pre-session survey was 77% and all participants (100%) correctly answered questions 6, 10 and 13 on the pre-session survey. The high correct response rate on the pre-session survey implies that the participants do not actually lack knowledge about general cancer genetics. However, the perception of a lack of knowledge is a reported barrier of the participants and is a factor in not seeking CGS.

The participant's perception of lacking knowledge about general cancer genetics may reflect some form of self-doubt or uncertainty about cancer genetics, or the participants may be avoiding seeking CGS for a reason unrelated to "lack of genetic knowledge". Perhaps the emotional impact of testing, in contrast to knowledge about testing, could also be contributing to the participant's self-doubt or uncertainty about cancer genetics. Reasons such as fear of knowing actual cancer risk, which was reported as the second main barrier to seeking CGS in this study, or fear of knowing test results, which was not reported in this study, could be contributing to the participant's avoidance of CGS. The potential impact of testing or the testing process on the entire family, as well as fear of the unknown or issues relating to uncertainty rather than genetic knowledge may be a barrier to seeking CGS.

Finding that the majority of participants do not pursue or participate in genetic counseling due to their own perception of lacking knowledge about general cancer genetics may suggest that providing education and emotional support in a "nontraditional" setting, reminiscent of the Bowen *et al.*, 2006 study, could provide the information necessary for individuals to feel prepared to have genetic counseling. Future questionnaires or discussions with individuals in the AJ community might elucidate a greater understanding about what information the community desires so that this barrier might be removed or better understood. Community-based information

sessions, similar to that conducted for this study, may be a good way to provide genetic information and emotionally prepare individuals before genetic counseling and therefore removing the barrier of the perception of lacking knowledge or fear of knowing test results.

4.3 PREFERRED COMMUNICATION STRATEGIES

An additional goal of this study was to identify the preferred method of communicating information regarding cancer risks and genetic counseling in the AJ population; assuming that discussion with informed relatives would be the preferred method of communication. Contrary to what was hypothesized, survey responses revealed that the majority of respondents (75%) felt that discussion with a healthcare provider was the best method for communicating information, while a minority of respondents (37.5%) obtained this information from their families.

Knowing that AJ individuals prefer to discuss this information with their health care providers suggests that emphasis should be placed on physician education. Physicians should also be encouraged to provide their patients with an explanation as to why a referral for CGS is appropriate, as well as to provide anticipatory guidance as to what they can expect at their appointment. If AJ individuals understand the importance and meaning of CGS when they are referred due to “high-risk” status, they may be more inclined to participate.

The majority (56.25%) of participants reported that their physicians are unaware of their AJ ancestry. Ricker *et al.*, 2006, conducted a study that examined the development of a free cancer genetics clinic for an underserved, primarily Hispanic population. As part of the development of the clinic, 371 participating providers received CME credits for attending five educational seminars about genetics and referral guidelines. Surveys revealed that, prior to the

seminars; providers answered only 22% of questions correctly. Post seminars, there was a documented 94% improvement in knowledge. In addition to the impressive increase in knowledge, physicians also began to incorporate this information into practice. Whether religious and ethnic background are not routinely obtained in clinic, or AJ individuals did not understand the importance of reporting ancestry, continued education within the medical community about the importance of discussing family history and AJ ancestry are necessary. The themes identified in this study support information from the Ricker study and encourage the idea that physician education may be a successful route to take in terms of facilitating communication of cancer risk and genetic information between physicians and AJ patients as well as ensuring that physicians are making appropriate referrals for genetic counseling.

During the Question and Answer session, the participants asked many excellent questions and provided many relevant personal experiences, the majority of which concerned frustration with physicians not discussing individuals increased risk for HBOC due to AJ ancestry. Participants seemed confused as to why their physicians never offered them “carrier screening” for *BRCA1/2* because of their AJ ancestry.

Due to the AJ founder effect, genetic testing in the AJ population is not a new idea. Carrier Screening Programs for recessive diseases like Tay-Sachs disease and Gaucher disease (as seen in Table 3) have been common in this community for many years (Charrow, 2004). The Jewish community as a whole has embraced the idea of carrier screening, aiming to prevent the births of children with genetic disease. It is important to recognize that carrier screening is different from disease screening or predisposition screening, which aims to identify individuals with a disorder at an early or even pre-symptomatic stage so that the condition can be managed more effectively (Charrow, 2004; Levine, 1999).

Currently, an evaluation of family history and gene testing are the only options available to genetic counselors and physicians to help identify AJ individuals at increased risk to develop HBOC-related cancers. To date, *BRCA1/2* testing is recommended only for those with a convincing family history of HBOC-associated cancer and ideally begins with testing an affected family member (NCCN, 2011). Although AJ population-based *BRCA* testing is currently contraindicated outside of a research setting, there have been discussions as to whether this type of screening would be appropriate and worthwhile to implement because relying solely on family history to predict mutation status can limit interpretation (Rubenstein, 2004; Hartge *et al.*, 1999).

It should be noted that other preferred methods of communication include exchanges between relatives, as well as communication with the synagogue. While information sessions at a synagogue would be ideal, it may be beneficial to educate religious officials to mention CGS to their congregants and stress the importance of discussing family history and AJ ancestry with their healthcare providers. One interesting aspect of this study was that the majority of participants identified with Reform Judaism (62.5%). As mentioned before, Orthodox Jews view genetic testing for *BRCA1/2* mutations as an “obligation” to one’s health; however, having a known mutation in the family can have substantial consequences for other family member’s future marriage prospects. While the reports in the literature regarding Reform and Conservative Judaism beliefs are limited, it would be interesting to investigate the beliefs of the other branches of Judaism to see if the “obligation” to one’s health holds for all branches of Judaism or just the Orthodox branch.

Recalling the Bowen *et al.*, 2003 study that explored the connections between Jewish identity and interest in screening behaviors, the finding that cultural identity positively predicted

interest in testing whereas religious identity was inversely related is very interesting and something that religious officials may want to consider when they explain CGS to their congregants.

4.4 CHANGE IN KNOWLEDGE

In order to determine the effectiveness of the general genetics discussion, a series of knowledge-based questions were provided to participants both before the information session and after the information session. Gain in knowledge was analyzed by comparing the participant's answers from the pre- and post-session surveys. With an average correct response rate of 77% on pre-session surveys to 94% on post-session surveys, analysis of the compared surveys proved that the information session was an effective tool for educating the AJ population about cancer genetics.

Understanding that communicating genetic information in a group setting outside of a hospital facility is an effective method for learning in the AJ population has important implications for the future development of a preferred service delivery model of AJ population-specific genetic counseling. Recalling the results of the Lerman *et al.*, 1999 study, AA women had greater intentions to undergo genetic testing for HBOC after receiving informal education in addition to traditional genetic counseling, and that same theme was observed in this study.

This knowledge can encourage genetic counselors to continue to conduct group information sessions at synagogues or Jewish community centers, which can facilitate the education of many AJ individuals in a condensed period of time, as well as overcome some of the barriers to participating in “traditional” genetic counseling. Following an education session,

participants may feel better prepared to participate in traditional genetic counseling by already having a fundamental cancer genetics background and therefore being able to concentrate on a personalized risk assessment, benefits and limitations of testing and potential psychosocial issues that are unique to that individual or family.

4.5 PARTICIPANT AND COMMUNITY FEEDBACK

The information session was well received by Rodef Shalom Congregation. In addition to the large attendance, participants, organizers, synagogue Clergy and the greater community provided very positive feedback about the event.

The individuals who attended the event were attentive throughout the entire session and many individuals took notes on the copy of the PowerPoint handout that was provided in the attendee's folders. Participants shared personal stories about their own experiences, asked for clarification regarding misconceptions and asked follow-up questions about information in the presentation.

Two individuals provided responses to the open-ended portion of the post-session surveys. The surveys requested that participants "share [with us] additional comments regarding genetic counseling and testing". One individual wrote, "Your presentation was great" and the other individual wrote, "Great presentation... Well informed, knowledgeable speakers; each of whom presented an interesting aspect that led to an overall discussion about Jewish Genetic Diseases."

4.6 CONCLUSIONS

This pilot study was undertaken to aid in the future identification of a preferred service delivery model of population-specific genetic counseling for HBOC in the AJ population. The study was designed to facilitate awareness of CGS, document common beliefs about CGS and identify preferred methods of communication regarding CGS in the AJ population.

The first aim of the study was to identify the current awareness, as well as facilitate awareness, of cancer genetic counseling services in the AJ population in the Pittsburgh area, and this aim was achieved. Aim 1 hypothesized that individuals of AJ ancestry were not aware of cancer genetic counseling services. The study showed that AJ individuals are aware of cancer genetic counseling and testing. Therefore, lack of awareness of CGS in this study population is not the main reason for the underutilization of services.

The second aim of the study was to identify the unique motivations for and perceived barriers to AJ individuals obtaining cancer genetic counseling services, and this aim was also achieved. Aim 2 hypothesized that personal perception of increased risk to develop cancer based on family history or AJ ancestry was the main motivator for seeking cancer genetic counseling services, and lack of information about cancer genetics, cancer risk and requirements for genetic counseling was the main barrier to obtaining cancer genetic counseling services. The study found that the perception of high-risk status to develop cancer based on personal or family history of cancer is a significant motivating factor for seeking CGS. The participant's perception of lacking knowledge about general cancer genetics is a self-identified barrier that is contributing to the underutilization of CGS in the AJ community.

The third aim of the study was to identify the preferred methods of communicating information about inherited predispositions to cancer to the AJ population, and this aim was

achieved as well. Aim 3 hypothesized that individuals of AJ ancestry often have close family relationships; therefore, communication between informed relatives may be the preferred strategy for promoting cancer genetic services. The study found that AJ individuals prefer that a healthcare provider or physician be their main source for information and communication about cancer risk, genetic information and referrals for genetics services.

This study was a collaboration between health care providers and a smaller Jewish community organization and received a great amount of community support. Analyzing the results of this study and recalling the results of the Lerman *et al.*, 1999 study that revealed that AA women had greater intentions to undergo genetic testing for HBOC after receiving informal education followed by genetic counseling, encourages the idea that in order to best serve the AJ community, an “education plus counseling model,” similar to that proposed for the AA population would also be successful for the AJ population.

The conclusions gained from this study suggest that informal community-based information sessions prior to formal or traditional genetic counseling could be a successful method to overcoming preconceived perceptions about genetic counseling for this specific population. In response to the positive results of this study, additional information sessions personalized for the AJ community should be conducted to continue to educate this population about AJ-specific cancer genetics.

4.7 STUDY LIMITATIONS

Certain limitations of the study should be noted. The primary limitation of this study was the small sample size, which negatively affects the ability to make statistically significant conclusions that are applicable to the AJ population as a whole. Because this was a pilot study, only 25 participants could be included in the study in accordance with IRB standards for Pilot studies.

The survey response was another limitation impacting the significance of the study. While all 25 possible participants agreed to participate in the study, only 16 of them completed the pre-session survey even though they were given instructions when they enrolled in the event and were encouraged to do so by the genetic counseling intern walking about the room and interacting with participants prior to the start of the event. Of those individuals, only 8 completed the post-session survey. The option of taking the post-survey home and mailing it back upon completion inhibited the return rate. If the participants were not given that option, the response rate would likely increase, though many participants were ready to leave at the conclusion of the session and some had other obligations to attend. The response rate was further impacted due to some participants not answering all of the questions on the surveys.

It is possible that the location of the information session biased the results of this study. The results of the information session may (or may not) have been different had the information session been hosted at a location with more sporadic attendance instead of regular gatherings and a sense of community like Rodef Shalom Congregation.

4.8 FUTURE RESEARCH OPPORTUNITIES

Given the results of this study, many opportunities exist for future research studies. While this study provided insight into the perspectives of AJ individuals with respect to CGS for HBOC, it would be useful to investigate additional community-based educational opportunities, as well as the perceptions of physicians, other healthcare professionals, and religious officials.

The study found that the majority of participants do not pursue genetic counseling due to their own perception of lacking knowledge about general cancer genetics; however, the high correct response rate on the pre-session surveys implies that the participants do not actually lack knowledge about general cancer genetics. A future qualitative study investigating the thought process and emotional impact of testing, in contrast to knowledge about testing, might help elucidate a greater understanding of AJ individual's perception of lacking knowledge about general cancer genetics, and whether or not that perception may reflect some form of self-doubt or uncertainty about cancer genetics, or if the participants are avoiding seeking CGS for a reason unrelated to "lack of genetic knowledge". Future qualitative investigations of the AJ population regarding the perception of lacking knowledge about general cancer genetics may reveal what information the community actually desires so that this barrier might be removed or better understood.

This study found that physicians are the preferred method of communicating cancer risk and genetic information in the AJ population. Understanding physician's current practice of obtaining (or not obtaining) their patient's ethnic background and religious identity in the clinic, as well as understanding physician awareness (or lack of awareness) of the importance of eliciting a basic family history and recognition of indicators of a hereditary predisposition would provide additional insight for conducting future physician-based education programs. Genetic

counselors participate and conduct many community, medical and non-medical education programs, but perhaps more emphasis should be placed on eliciting ancestry in primary care offices and educating about the relationship between the AJ population and HBOC, as well as who to refer, how to refer and when to make appropriate referrals for CGS.

This study found that another method of preferred communication involved synagogues. While this study proved that information sessions at synagogues are well received, there may be future educational opportunities at Jewish Community Centers, perhaps mirroring the information sessions at synagogues. Conducting information sessions on a larger scale, like that of a community center, would help to determine success and interest in large scale community-based cancer genetics educational programs.

Future religious official-based education programs may be helpful for clergy members regarding how to discuss genetic topics with their congregants, how to explain the importance of discussing cancer history with family members and how to explain the importance of discussing AJ ancestry with physicians. Religious official-based education programs may also provide the opportunity for genetic counselors and clergy members to discuss the Jewish perspective (of all branches of Judaism) on genetic testing for hereditary cancer predispositions and the implications for the individual and family, providing invaluable insight and feedback for genetic counselors designing a preferred service delivery model for AJ individuals.

The conclusions gained from this study provide suggestions that could lead to a successful method to overcoming preconceived perceptions about genetic counseling for the AJ population.

APPENDIX A: DIFFERENTIAL DIAGNOSIS

Other than HBOC, the most significant hereditary cancer syndromes leading to an increased risk for breast cancer include Li-Fraumeni syndrome (LFS), Cowden syndrome and Peutz-Jeghers syndrome (PJS). LFS, caused by mutations in *p53*, increases the risk for multiple types of cancer, including: breast cancer, soft tissue sarcomas, osteosarcomas, leukemia, adrenocortical carcinomas and brain tumors (Malkin, 2011). Cowden syndrome, caused by mutations in *PTEN*, is associated with an increased risk for breast cancer, uterine cancer and thyroid cancer. Cowden syndrome is also associated with characteristic benign hamartomatous lesions of the skin, oral mucosa and intestinal mucosa, as well as benign breast and thyroid disease (Eng, 2000; Trepanier *et al.*, 2004). PJS, caused by mutations in *STK11/LKB1*, is associated with an increased risk for breast cancer, pancreatic cancer, prostate cancer and benign ovarian tumors. PJS is also associated with characteristic benign hamartomatous polyps of the GI tract, ureter, bladder, renal pelvis, bronchus and nasal passage, as well as melanin spots on the lips, oral mucosa and fingers (Trepanier *et al.*, 2004).

Lynch syndrome accounts for approximately 7% of hereditary predispositions to ovarian cancer. Lynch syndrome, also known as Hereditary Non-polyposis Colorectal Cancer (HNPCC), is caused by mutations in one of several genes, including: *MLH1*, *MSH2*, *MSH6*, *PMS2* and *EPCAM*. Lynch syndrome increases the risk for colon cancer, uterine cancer, ovarian cancer, small bowel cancer, gastric cancer, pancreas cancer, ureter cancer and renal pelvis cancer (Lynch, 2000).

APPENDIX B: LETTERS OF IRB APPROVAL



University of Pittsburgh
Institutional Review Board

3500 Fifth Avenue
Pittsburgh, PA 15213
(412) 383-1480
(412) 383-1508 (fax)
<http://www.irb.pitt.edu>

Memorandum

To: Rachel Pearlman, BS
From: Sue Beers, PhD, Vice Chair
Date: 5/19/2011
IRB#: [PRO11040034](#)
Subject: Health Beliefs and Barriers Inhibiting Individuals of Ashkenazi Jewish Ancestry from Seeking Cancer Genetic Counseling Services

The above-referenced project has been reviewed by the Institutional Review Board. Based on the information provided, this project meets all the necessary criteria for an exemption, and is hereby designated as "exempt" under section 45 CFR 46.101(b)(2).

The IRB has approved the advertisement that was submitted for review as written. As a reminder, any changes to the wording of the approved advertisement would require IRB approval prior to distribution.

Please note the following information:

- If any modifications are made to this project, use the "**Send Comments to IRB Staff**" process from the project workspace to request a review to ensure it continues to meet the exempt category.
- Upon completion of your project, be sure to finalize the project by submitting a "**Study Completed**" report from the project workspace.

Please be advised that your research study may be audited periodically by the University of Pittsburgh Research Conduct and Compliance Office.



**ALLEGHENY-SINGER
RESEARCH INSTITUTE**

WEST PENN ALLEGHENY HEALTH SYSTEM

320 EAST NORTH AVENUE, PITTSBURGH, PA 15212-4772

412-359-3156

INSTITUTIONAL REVIEW BOARD 01
FWA00015120

Certification of Exemption

April 26, 2011

Megan Marshall, M.S.
Department of Human Oncology

RE: **RC-5186** "Health Beliefs and Barriers Inhibiting Individuals of Ashkenazi Jewish
Ancestry from Seeking Cancer Genetic Counseling Services"

Dear Ms. Marshall:

The Institutional Review Board (IRB) is in receipt of the updated information for the
above referenced protocol.

The IRB has reviewed this information and finds it continues to qualify for exempt status
according to the following category in the Code of Regulations: 45 CFR 46.101 (b)
Category (2).

Please retain this letter as evidence of IRB review and determination of exempt status for
this research.

Annual review of this research is not required provided the research is conducted as
proposed. If there are modifications or changes to this study, the Investigator must have
the IRB review the study prior to initiating the changes.

If you have any questions, please contact the IRB office.

Sincerely,

Athanasios Colonias, MD,
Vice-Chairman
Institutional Review Board

AC/sbg

APPENDIX C: ADVERTISEMENTS



Rodef Shalom
CONGREGATION

Jewish Family Concern Series

Presents:

Through the Lens of *Shmirat HaGuf* (Taking Care of the Body): What you NEED to Know About Jewish Genetics

When: Sunday, December 11, 2011

Where: Rodef Shalom Congregation, Room ALC 1

Time: 10:00 am - 11:30 am

In this session participants will receive important facts about Jewish genetic diseases and learn what every person needs to know about risks, treatment, and prevention.

A panel of experts will present valuable new information and answer questions.

Speakers include Rodef Shalom members Jay and Barbara Rogal who will share their personal stories relating to Jewish genetic diseases.

Our rabbis will speak about taking care of our bodies from a Jewish perspective

Co-sponsored with Rodef Shalom Brotherhood and Sisterhood and community partners:



The series continues.....

January 22, 2012 - A Jewish response to Bullying with Psychotherapist and Teen Expert Barbara Wollman, LCSW, BCD of Jewish Family and Children's Service of Pittsburgh

April 15, 2012 - How to Talk about Life's BIG Questions with our Rabbis and Educators



24 CHESHVAN 5772 • NOVEMBER 21, 2011



RODEF SHALOM
CONGREGATION

REconnecting...

81-WEEKLY SUPPLEMENT TO HAKESHER BULLETIN OF RODEF SHALOM CONGREGATION



Health & Healing...Jewishly

Blood Drive

Sunday, December 4

9 a.m.-1:30 p.m.

Freehof Hall

Support Brotherhood's blood drive! The Talmud says, "And whoever saves a life, it is considered as if he saved an entire world." Do what you can to save a life by donating blood now.

To make an appointment, call Richard Meritzer at (412) 494-7609. Walk-ins are welcome. Open to all. Be sure to invite your friends and family.

► See inside

Closure: End of Life Decision Making Series continues on Monday, November 21.

Health Insurance Essentials

L'Chaim: To Your Good Health Project

Thursday, December 8 • 1-2:30 p.m. • ALC 1

Learn ways to stay healthy at this informal conversation with physicians and other healthcare professionals. *Continues 1/12, 2/9.*

Jewish Genetics: What You Need to Know

Jewish Family Concerns:

Through The Lens of Shmirat HaGuf (Taking Care of the Body)

Sunday, December 11 • 10-11:30 a.m. • ALC 1

Come receive important facts about Jewish genetic diseases and learn what every person needs to know about risks, treatment, and prevention. A panel of experts will present valuable, new information and answer questions. Speakers include Barbara and Jay Rogal, who will share their personal stories, and Rodef Shalom rabbis, who will discuss taking care of our bodies from a Jewish perspective. Co-sponsored with Brotherhood, Sisterhood, and community partners. Series continues: 1/22, 4/15.

THE WORLD OF SHOLOM ALEICHEM

Sunday, December 11 • 7:30 p.m. • Levy Hall

Your ticket to fun!



Don't miss this delightful adaptation of three classic Yiddish stories produced for television in 1959. In English, the film features celebrated Jewish actors of the last century including Zero Mostel, Sam Levene, Gertrude Berg, and Morris Camovisky. No charge. The series will conclude on February 12.

PRESENTED BY SISTERHOOD MOVIE NIGHT SERIES

Local woman made extreme decisions following genetic testing results

by Toby Tabachnick

Staff Writer

12.08.11 - 03:34 pm

Kathy Pattak did not have ovarian or breast cancer. Nevertheless, in 1999, she opted to have a hysterectomy, and in 2005, she underwent a prophylactic double mastectomy, as well.

Pattak chose to have the procedures as preventative measures because genetic testing indicated she had an 87 percent chance of developing breast cancer, and a 25 percent chance of developing ovarian cancer, if she did not have the surgeries.

"I have no regrets," said the former physical education teacher from the Mt. Lebanon Area School District. "The key thing is you need to be proactive. You need to get the [genetic] testing. Then there are things you can do."

Pattak will be one of the featured speakers this Sunday, Dec. 11, at Rodef Shalom Congregation's Jewish Family Concerns Series. The program, entitled "Through the Lens of Shmirat HaGuf (Taking Care of the Body): What You Need to Know about Jewish Genetic Diseases," begins at 10 a.m., and is open to the public.

The program aims to educate its audience on the importance of Jewish genetic testing, focusing on screening for BRCA 1 and BRCA 2 gene mutations, which are predictors for breast and ovarian cancer, and which are prevalent among Ashkenazi Jews.

"About one person in every 500 to 800 who are not Jewish have the mutation, compared to one out of every 40 for Ashkenazi Jews," according to Megan Marshall, a genetic counselor who will be speaking Sunday at the program.

While women in the general population have about an 8 percent chance of developing breast cancer by age 70, the risk of developing breast cancer for a woman with a BRCA gene mutation is about 10 times as high.

Those odds were enough to convince Pattak to have the surgeries, thus reducing her risk of developing breast and ovarian cancer by about 95 percent.

Ashkenazi women, with family histories of breast or ovarian cancer, should consider getting tested for the BRCA gene mutation, Pattak said, but making such a decision is not always easy.

One has to be prepared to accept the results.

Pattak had a strong family history of both cancers. Her mother, aunt and grandmother all had breast cancer, and her mother and aunt also had ovarian cancer. Pattak's sister, after getting tested, found she had the BRCA mutation, and tried to convince Pattak to get tested as well.

"I didn't want to at first, because I didn't know what I would do with the results," Pattak said. "My sister kept telling me to stop being an ostrich, putting my head in the sand."

It wasn't until 1998, when Pattak heard of a study at the University of Pittsburgh that focused on the mental and emotional process of being tested and dealing with the results, that she decided to do what she had been avoiding.

"Once I signed up for the study, I decided to have the testing done," she said. "I didn't make the decision to be tested until I was ready to accept the results, assuming they would be positive."

Pattak did, in fact, test positive for the BRCA gene mutation, and in 1999, had a hysterectomy.

"It was easier to decide to have the hysterectomy than the mastectomy because it was internal," she said.

But in the early 2000s, her gynecologist suggested she speak to a breast surgeon to discuss the possibility of a mastectomy as well. Pattak also spoke to a plastic surgeon about reconstruction.

"Once I knew what was going to happen, and what was involved, I decided to go ahead and have the surgery," she said.

"This is something more and more Jewish families need to know about," Pattak said. "They need to know about the risks, and that with this you can be proactive. There is something you can do to reduce your risks of breast or ovarian cancer."

Sheila Solomon, one of the genetic counselors scheduled to speak at Sunday's program, called genetic testing "a very important issue in the Jewish community."

Nevertheless, she noticed while working with Marshall at Allegheny General Hospital several years ago that few Jewish patients were opting for it.

"We wondered why those in the Jewish community were not coming in for testing for hereditary cancers," she said. "We wanted to become informed on what are the barriers, and what are the best methods to get the information for the community."

"We don't see the correct percentage of Jews coming in for genetic counseling," Marshall said.

Ashkenazi ancestry by itself does not necessarily indicate a need for genetic testing, according to Marshall. Age and family history are also factors.

"Having this information is really important," said Rabbi Amy Hertz of Rodef Shalom Congregation, who will also be speaking at Sunday's program. "How we

take care of our bodies is a Jewish value."

Other speakers at the Sunday program include Rachel Pearlman, a graduate student in genetic counseling, and Barbara and Jay Rogal, whose daughter lives with Gaucher, another genetic disease prevalent among Ashkenazi Jews.

The genetics event is co-sponsored by Allegheny General Hospital, University of Pittsburgh, Rodef Shalom Brotherhood, and Rodef Shalom Sisterhood.

(Toby Tabachnick can be reached at tobytt@thejewishchronicle.net.)

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APPENDIX D: TEN MINUTES OF TORAH



Jewish Genetic Disease: A Congregational Discussion

by Rabbi Lawrence R. Sernovitz

We recently heard the news that a young family in our congregation gave birth to a child with a Jewish genetic disease that we had never heard of. When it comes to Jewish genetic diseases, we weren't aware that there were others beside Tay Sachs. The family is devastated with the diagnosis and is trying to figure out what to do and how to cope. The child is failing to thrive and it is becoming increasingly difficult for her parents to provide adequate nutrition.

This raises many questions for us as a congregational community. How do we react to the news? We have no idea what to expect and want to be there for the family. How can we support them? We are truly at a loss. Any help you can give us would be most appreciated.

There is much that congregations can do and your question is an important one. Expectations are very difficult as this depends on each individual family. The most important course of action that you can do is to listen, to provide support, and be compassionate. Reach out to the family and be there for them during this difficult time.

I give this advice based on personal experience. For the first four months of my son's life, he was unable to suck and swallow, inhibiting his ability to get nutrition and gain weight. He was diagnosed with "failure to thrive" and suffered immensely. After visiting many specialists, at four months old, he was diagnosed with a Jewish genetic disease called Familial Dysautonomia. Like many of you, we had never heard of this disease and had no idea what to expect. It was devastating and we knew our son would not be like the children born to our friends. Would our son have a normal childhood? Would he be able to participate in activities along with his peers? How would his disease affect his education? These were just a few of the questions we had.

What we needed the most was a support system to help us through the difficult times that were to come. Besides our close friends and family, who else could we count on? Our congregational community was truly there for us. Soon after his diagnosis, I send out a letter to our congregation, informing them of Sam's diagnosis. One part of the letter read as follows,

It is very difficult for me to write these words to you today. However, as members of this caring community, Becky and I feel it is important to share with you the events that have recently taken place, and which have changed our lives forever.

Martin Buber once wrote, "Children have a future, a destiny that is all their own. But they will not have to face the infinite universe alone, nor worry that they will be unprepared, for they are surrounded by loving family and friends who care deeply for them. Each of us brings them the gift of our love and our dedication, helping them to feel understood, supported and special as they go through life."

We have no doubt that our son will have the love and support of our community and for that we offer your our heartfelt thanks. We will certainly need your support and understanding as the months and years go by and will keep you advised of any new developments

Since our son was born, almost three years ago, we have been overwhelmed by the kindness and generosity of our congregational community. I have always understood the synagogue to be a *kehilah kedoshah*, sacred communities where people look out for one another. When one individual in our community suffers, we all suffer. As Jewish tradition teaches, "*Kol Yisrael arevim zeh b'zeh*," all Israel looks out for one other.

The needs of each family will be different and the best thing to do is reach out and ask how you can help. In many cases, families will tell you they don't need anything, when in reality they don't even know where to start. Offer a meal, help with errands, or just stop by to lend your support or to play with their child. Many times, just being there can be all the support that they need.

Beyond this personal support, here are three easy ways that your congregation can do can make a difference:

1. **EDUCATION AND AWARENESS:** Educate yourself, the board, and your congregation about Jewish Genetic diseases. The Victor Center for Jewish Genetic Diseases at Albert Einstein Medical Center in Philadelphia is a great place to start. Their website is filled with useful information. You can also visit the website for the Jewish Genetic Disease Consortium (JGDC). Both organizations have pamphlets that you can have on display and personnel that give educational presentations.
2. **PREVENTION:** Emphasize to your clergy the importance of pre-marital screening. Speak with them and make sure they have up to date information. Make sure that they encourage, if not require all engaged couples to be screened before marriage. You can visit the **National Society of Genetic Counselors** website for a listing of genetic counselors and screening centers nationwide. All counselors are accredited and are excellent resources for you and your couples.
3. **SCREENING:** Host a genetic screening in your community with other synagogues, or by yourself if you are the only one in the area. You can make it a family fun day for the community, with games and activities, to create additional awareness.

Did you know that 1 out of 5 Ashkenazi Jews are carriers for at least one of 19 severe and many life threatening Jewish Genetic diseases. We are the only ethnic group with this reality. The only way to know if you are a carrier is to be screened, i.e. have a simple blood test or give birth to a child with the disease.

How can you help ensure the birth of healthy babies? Be screened, inform everyone you know to be screened prior to each pregnancy, organize a community screening. A little effort can go a long way.

Rabbi Larry Sernovitz serves Old York Road Temple-Beth Am in Abington, PA

- Join us for the Biennial December 14-18, 2011 in Washington D.C. Biennial is where Reform Jews gather to learn, pray, share ideas, dance and sing, hear from inspiring guest speakers, reunite with old friends, make new connections, and make decisions about the policies of the Reform Movement.

10/28/11 Friday - Reform Judaism Q & A



Ten Minutes
of Torah



REFORM JUDAISM Q & A

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October 28, 2011 | 30th Tishri 5772

The Link between Wedding Planning and Genetic Testing: A Jewish Perspective

by Rabbi Lawrence R. Sernovitz

E-MAIL TO THE RABBI

Re: Getting Tested for Jewish Genetic Diseases

Dear Rabbi,

Thank you for meeting with us the other day. We really appreciate your time and the meeting was very meaningful. We are so excited to stand under the *chuppah* with you! However, we both are not convinced of the need to get tested for Jewish Genetic Diseases before we get married. We have too much to do before the wedding anyway and we just don't have time to get it done. Whatever our carrier status happens to be, it isn't going to prevent us from getting married so why should we get tested anyway? See you at the next meeting!

Jonathan and Rachel

Dear Jonathan and Rachel,

Planning for a wedding can be a very time consuming process, especially for a young couple. Juggling a busy work schedule while at the same time scheduling meetings with the venue, the caterer, the florist, the photographer, the videographer, and the rabbi can be challenging. Then there is planning the engagement party, the bridal shower, the rehearsal dinner, and of course, the invite list and the seating chart. Oh, and you can't forget the all important registry. And, what about the wedding and bridesmaid dresses. OY! So much to do and so little time! All of this can be extremely overwhelming. For those going through this at the moment, I am sure that thinking about all this just brought on more anxiety of all that needs to be done, according to that all important wedding notebook. Yes, there is much to do but know that it will all get done. It usually does.

However, genetic testing should not be tossed off the list just because there are so many other things to be done. In fact, it should be a priority on that ever growing list. Most young couples do not think about family planning because they are so caught up in the wedding planning itself. In fact, many couples forget about nurturing their own relationship during the planning process, let alone thinking about kids down the line. But, the harsh reality is that if they you don't get tested prior to marriage and you are both carriers, the caterer, the wedding dress, and the fond memories of the day will be meaningless when your child is born with one of 19 Jewish genetic diseases. Instead of filling your

world with memorabilia from the wedding, it will be filled with doctors appointments, medical supplies, and many tears. I know as I speak from experience. (See the August 26, 2011 Ten Minutes of Torah for more of my story.)

Getting a simple blood test can make all the difference. If one partner is a carrier of any of the 19 Jewish genetic diseases, the other partner should be screened as well. If both partners are carriers, there is a 25% chance the couple will give birth to an affected child. Once you have this knowledge, you can make intelligent decisions regarding the future. This can include in-vitro fertilization, virtually eliminating the chances of having an affected child, or having a natural pregnancy with the knowledge that a tough decision might have to be made down the road. If this is the chosen route, having a CVS test (Chorionic villus sampling, a prenatal test that detects chromosomal abnormalities such as Down syndrome as well as genetic diseases) completed around 11-12 weeks can identify genetic diseases in the fetus and give you the knowledge to make an informed decision.

Unfortunately, once pregnant with an affected child, a couple has two choices; to continue with the pregnancy and give birth, or to stop the pregnancy.

Don't take the risk! Here is what you can do:

Get in touch with the Victor Center for Jewish Genetic Diseases. They have centers in Philadelphia, Boston, and Miami. Their website is <http://www.victorcenters.org/> and it is filled with useful information. They will give you all the information you need to make informed decisions and to get tested.

Additionally, for information about genetic counselors nationwide, you can visit the National Society of Genetic Counselors website at NSGC.org for a listing of genetic counselors and screening centers nationwide. All counselors are accredited and are excellent resources for you.

One day, please God, you will be holding a little healthy baby in your arms, continuing the legacy that your parents created for you. And, at that moment, you will be thankful that you did everything you could to ensure that your precious little one will have every opportunity to pursue their hopes and their dreams. Mazal Tov on your upcoming wedding! I look forward to standing with you and your family under the chuppah. See you at our next meeting!

L'Shalom,

Rabbi Larry Sernovitz

Rabbi Larry Sernovitz serves Old York Road Temple-Beth Am in Abington, PA.

Join us for the Biennial December 14-18, 2011 in Washington D.C. Biennial is where Reform Jews gather to learn, pray, share ideas, dance and sing, hear from inspiring guest speakers, reunite with old friends, make new connections, and make decisions about the policies of the Reform Movement. **Register now!**

Nov. 13-14: Intensive 2-Day Adult Learning Workshop - Only \$36! Highly subsidized by the Covenant Foundation, this URJ workshop for congregational leaders will explore adult learning theories, modalities and curricula to help leaders find the best approach for their adult learning programs. Limited to 30 participants, space still available. **Register today!**

APPENDIX E: INFORMATION SESSION AGENDA

Jewish Genetic Disease Program: What you need to know!
AGENDA

Rodef Shalom Synagogue

December 11, 2011

Room: ALC1

10:00-11:30am

Guest speakers

Barbara and Jay Rogal

Kathy Pattak

Presenter

Rachel Pearlman

Panel

Rachel Pearlman

Megan Marshall

Rabbi Hertz

Barbara and Jay Rogal

10:00am	Introduction/Welcome	Rabbi Amy Hertz
10:05am	Guest Speakers -The Rogals will share their personal experience with Jewish Genetic Diseases and speak about the screenings they help organize in PGH	Barbara and Jay Rogal
10:20am	Guest Speaker -Kathy will share her personal experiences with genetic counseling and testing for hereditary breast and ovarian cancer	Kathy Pattak
10:35am	Hereditary Breast and Ovarian Cancer presentation -Rachel will discuss the difference between hereditary and sporadic cancer, hereditary breast and ovarian cancer in the Ashkenazi Jewish population, medical management options and genetic testing	Rachel Pearlman
11:10am	Q/A, summary panel	

APPENDIX F: PRE-SESSION SURVEY

CANCER GENETICS PROGRAM
WEST PENN ALLEGHENY HEALTH SYSTEM

Pre-Cancer Genetics Information Session Survey

Date: / /

CG ID#

Please answer the following cancer genetics questions to the best of your knowledge.

Demographics questions:

The following questions tell us more about you. Please circle or fill in the answer that best describes you.

1. Sex: M F

2. I am ____ years old: 18-25 25-30 31-40 40-50 50-60 60-70 70+

3. I am ____: Single Married Divorced Widowed In a relationship

4. I have ____ children: 0 1 2 3 4 5+

5. The highest level of school I have finished is ____: Some high school
High School graduate
Some college
College graduate
Graduate/professional school

6. Religious Affiliation: Reform__ Conservative__ Orthodox__ Jewish__ Other__ None__

7. Do you belong to a synagogue? Yes ____ No ____

8. Have you ever participated in genetic counseling? Yes ____ No ____ I don't know__

9. Have you ever participated in genetic testing? Yes ____ No ____ I don't know__

Family history questions:

1. Do you consider yourself to be an individual of Ashkenazi Jewish ancestry? Yes ____ No ____

2. What country are your ancestors from?

3. Have any of your close relatives been diagnosed with cancer? Yes ____ No ____

Type:

- a. Breast
- b. Ovarian
- c. Colon
- d. Pancreatic
- e. Melanoma
- f. Other: ____

How are you related?

4. Have any of your close relatives been diagnosed with cancer **before age 50**? Yes ____ No ____

How are you related?

5. Have you ever been diagnosed with cancer? Yes ____ No ____

Type:

- a. Breast
- b. Ovarian
- c. Colon
- d. Pancreatic
- e. Melanoma
- f. Other: ____

6. Were you diagnosed before age 50? Yes ____ No ____

The following are questions about hereditary breast-ovarian cancer. Please read each question carefully and circle the BEST answer for each question.

1. True or False The majority of all cancer is hereditary

2. How common is breast cancer in the general population?

1 in 8 1 in 40 1 in 100 1 in 800

3. How common is hereditary breast cancer in the general population?
 1 in 8 1 in 40 1 in 100 1 in 800

4. How common is hereditary breast cancer in individuals of Ashkenazi Jewish ancestry?
 1 in 8 1 in 40 1 in 100 1 in 800

5. True or False Cancer is only passed through your mother (mother's side of the family)

6. True or False Having a cancer gene means you will definitely develop cancer

7. True or False Inheriting a mutation (change) in a cancer gene increases the risk that you will develop specific types of cancer

8. How many genes must someone inherit to have Hereditary Breast-Ovarian Cancer?
 a. Zero, it is not caused by genes b. One from mom c. One from mom or dad
 d. Two, one from mom and one from dad e. None of the above

9. How can you tell if someone carries a gene that puts them at an increased risk for Hereditary Breast-Ovarian Cancer?
 a. They look sick b. They have been diagnosed with breast and/or ovarian cancer
 c. With a simple blood test d. There is no way to know e. None of the above

10. True or False If a person has genetic testing and no mutation is found in their cancer genes, that person will never develop cancer

11. True or False A mutation in a breast cancer gene also contributes to a higher risk of ovarian cancer

12. True or False Our genetic code (DNA) consists of A, C, T, G

13. True or False According to HIPAA (Health Information Portability and Accountability Act), genetic testing results can be used for insurance discrimination

14. What do you believe is your estimated risk of cancer (1 to 10, with 10 being the highest)? _____

Communication questions:

1. What method is the best way to communicate information about cancer risk and family history?

☐ Health care provider
☐ Relative
☐ Friend
☐ Newspaper/magazine
☐ Internet
☐ Synagogue/religious organization
☐ Other: _____

2. Do you think healthcare professionals are aware of your Ashkenazi Jewish ancestry when discussing health issues, especially relating to cancer?

Health belief questions:

For the following questions, please tell us whether you:

5. STRONGLY AGREE
4. AGREE
3. NEUTRAL
2. DISAGREE
1. STRONGLY DISAGREE

Severity

1. Hereditary Breast-Ovarian Cancer is a serious disease _____
2. I think passing a gene mutation to my children would be scary _____
3. My life would change if I had a gene mutation that predisposed me to Hereditary Breast-Ovarian Cancer _____

Susceptibility

1. I feel I am at risk for developing Hereditary Breast-Ovarian Cancer _____
2. There is a genetic cause to the cancers in my family _____
3. I carry a genetic susceptibility to Hereditary Breast-Ovarian Cancer _____

Benefit

1. It is useful to know whether I have an increased risk to develop breast or ovarian cancer _____
2. It is useful to know whether I have a gene mutation that causes an increased risk to develop breast or ovarian cancer _____
3. Knowing the risk of developing breast or ovarian cancer would change my medical management _____

Barriers

1. Testing for a gene mutation known to cause breast and/or ovarian cancer is **painful and difficult**_____
2. Testing for a gene mutation known to cause breast and/or ovarian cancer would **increase my anxiety and make me worry** about other family members_____
3. Testing for a gene mutation known to cause breast and/or ovarian cancer would **make me feel guilty if my children were found to inherit the gene mutation**_____
4. I would not want to pay for genetic testing if it is not paid for by my insurance_____
5. I do not know how to find a genetic counselor in my area_____
6. I am worried about health insurance, life insurance and job discrimination_____
7. I would feel ashamed, singled-out and/or viewed negatively in my community if I participated in genetic testing_____
8. I think **fear of knowing cancer risk** is a block to genetic counseling for breast cancer_____
9. I think **lack of genetic information** is a block to genetic counseling for breast cancer_____
10. I think **not knowing family history** is a block to genetic counseling for breast cancer_____
11. This information would provide a benefit to my family and me_____
12. I know how to find more information_____

Please share with us additional comments regarding genetic counseling and testing that were not addressed in the questions above.

Thank you for taking the time to complete this survey. If you have questions regarding the survey, the study, or for additional information, please call (412) 359-8064.

APPENDIX G: INFORMATION SESSION POWERPOINT

Jewish Genetic Disease: What you Need to Know!

Inherited Predispositions to Cancer

Who Are We?

Rachel Pearlman, BS
Genetic Counseling Intern
Master's Degree Candidate, University of Pittsburgh
Genetic Counseling Training Program

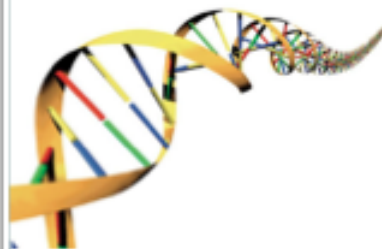
Megan Marshall, MS, CGC
Certified Genetic Counselor
Cancer Genetics Program
West Penn Allegheny Health System

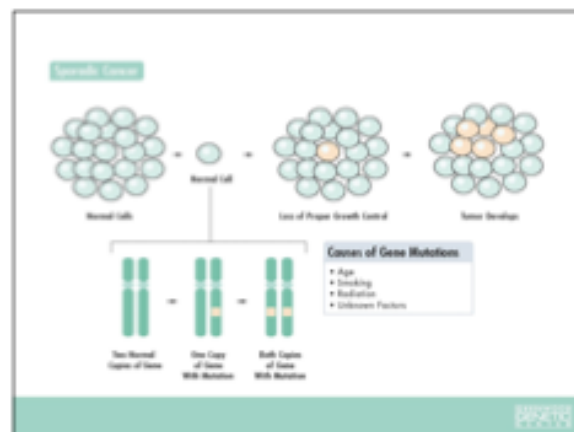
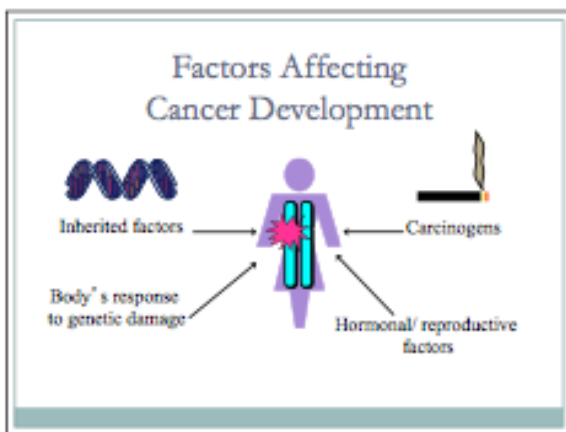
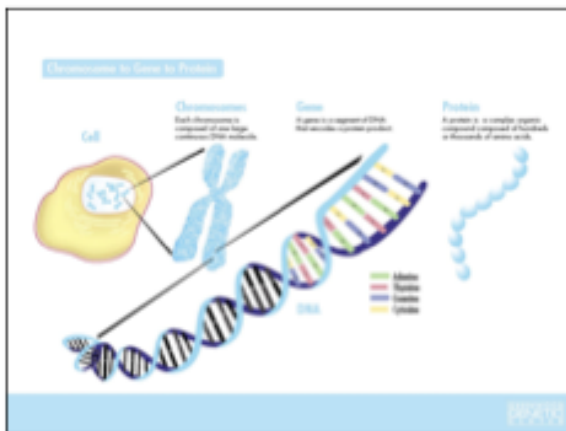
Have you ever wondered...

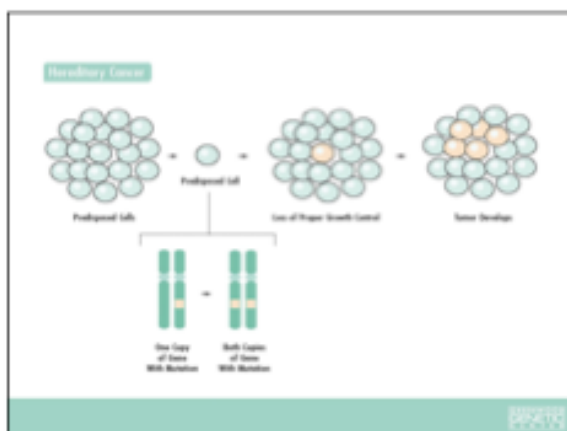
- What caused the cancer to develop in my family?
- What are my cancer risks?
- Are my children at risk?
- What can I do to help my family?



Brief Genetics Overview







Who Is at High Risk of Hereditary Cancer?



Hereditary cancer accounts for only a small proportion of all cancer

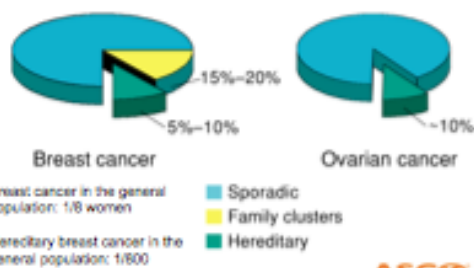
WHO. *Global Cancer Statistics*. World Cancer Research Fund. New York: Wiley-Liss, 1996.

Schneider B. *Counseling About Cancer: Strategies for Genetic Counseling*. 2nd ed. New York: John Wiley & Sons, 2001.

Fogelman B, Sklar J. *The Genetic Basis of Human Cancer*. New York: McGraw-Hill, 1998.

ASCO

How Much Breast and Ovarian Cancer Is Hereditary?



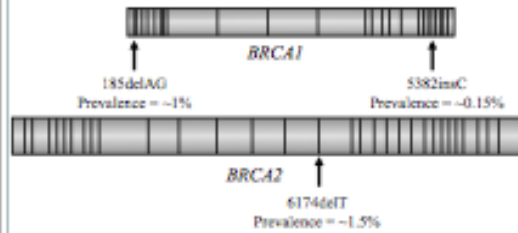
Cancer Risks for *BRCA1/2* Mutation Carriers

Cancer	General Population	<i>BRCA1</i>	<i>BRCA2</i>
Breast	12%	50-87%	50-87%
2 nd Breast	2-11%	48-64%	48-64%
Ovary	<2%	28-44%	27%
Male Breast	<1%	<7%	7%
Prostate	16%	20-32%	20-32%
Pancreas	<2%	2%	2-5%
Melanoma	<1%	not increased	5%

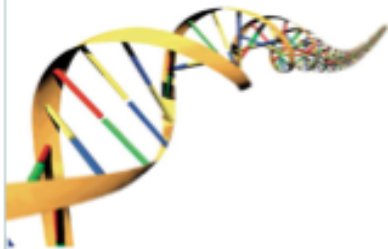
Features Suggestive of Hereditary Cancer Predisposition

- 2 or more closely related individuals with cancer before age 50
- Individuals with multiple primary tumors
- Several affected generations in the same bloodline
- Unusual tumors
- Ethnicity

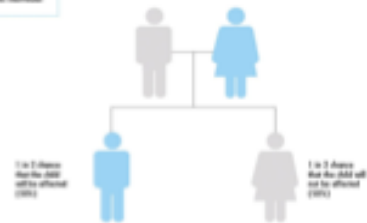
BRCA1 and *BRCA2* Mutations in the Ashkenazi Jewish Population



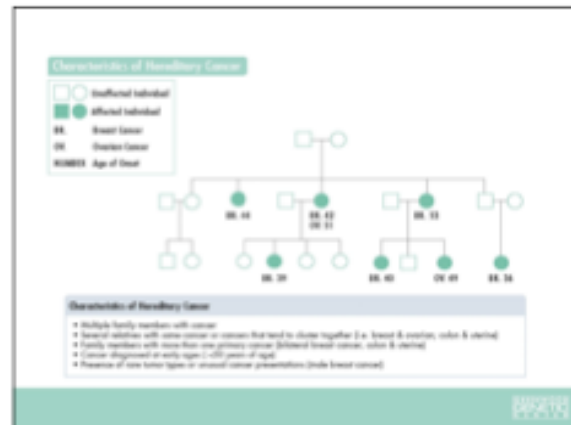
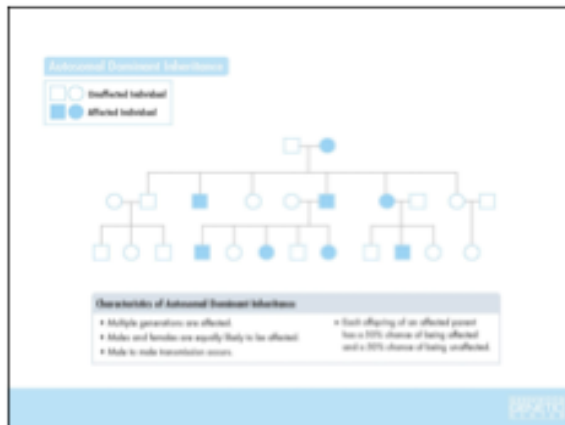
Inheritance Patterns



Autosomal Dominant Segregation, One Parent Affected



BRCA1



- Genetic Testing**
- Myriad Genetic Laboratory
 - Salt Lake City, Utah
 - Offered to individuals with > 10% chance of carrying a mutation
 - BRCAnalysis: \$3,340
 - Multisite 3 BRCAnalysis: \$575 (AJ panel)
 - Single Site BRCAnalysis: \$475

Genetic Counseling and Testing

• Benefits

- Personalized risk assessment allows for better informed medical management decisions
- Allows for accurate risk stratification
- Important information for family members
- Alleviates uncertainty and anxiety

• Limitations

- A negative result is most informative when there is a known mutation in the family
- Some genetic variants are of unknown clinical significance
- Genetic testing for the BRCA genes does not identify all causes of hereditary breast and ovarian cancer

Cancer Detection and Risk Reduction Options

Increased Surveillance

- Breast Cancer**
 - Monthly self breast exam beginning at age 18
 - Semi-annual clinical breast exam beginning at age 25
 - Annual mammogram and breast MRI beginning at age 25

Chemoprevention

- Breast Cancer**
 - Medications like Tamoxifen may reduce risk for breast cancer by as much as 50% when taken for 5 years
 - The efficacy of Tamoxifen for BRCA2 carriers is uncertain

Surgical Prevention

- Breast Cancer**
 - Option of bilateral mastectomies to reduce breast cancer risk by at least 90%

- Ovarian Cancer**
 - Semi-annual transvaginal ultrasound and CA-125 measurement beginning at age 35

- Ovarian Cancer**
 - Oral contraceptives may reduce risk for ovarian cancer by as much as 60% when taken for at least 5 years

- Ovarian Cancer**
 - Removal of the ovaries is recommended between the ages of 35 and 40 and reduces the risk for ovarian cancer by 95%
 - Removing the ovaries before menopause may also reduce the risk for breast cancer by as much as 50%

So now you are thinking,

Is Genetic Counseling & Testing for ME?



Let's find out the details...

Goals of Genetic Counseling

- Help patients, couples, and families understand the meaning of specific information about their genes and other genetic information and issues that may impact their life
 - Family history, inheritance, risk assessment
- Assist patients in deciding whether to have a genetic test performed and then what to do with information provided by such a test
 - Testing options, informed consent
- Help patients comprehend their diagnosis, prognosis, recurrence risks, reproductive choices, and to make the best possible life adjustment to the condition
 - Medical management options, psychosocial implications

Genetic Testing Has Implications for the Entire Family

- Consider the impact of testing on *all* family members
- Ultimately, testing is the individual's choice



...The Bottom Line



- Important to ask family history
- Options for cancer prevention and early detection are available for those at high hereditary risk
- Genetic counseling provides families with:
 - Opportunity to learn about cancer risk
 - Discussion of emotions about living with this risk
 - Options for cancer risk reduction and prevention

Who to Contact with Questions

Cancer Genetics Program

- Allegheny General Hospital
- Sharon Regional Health System
- Armstrong County Memorial Hospital
- Canonsburg General Hospital
- UPMC's Magee Women's Hospital
-
- Referrals: 359-8064 or mmarsha2@wpahs.org

THANK YOU!!!

Questions????

APPENDIX H: POST-SESSION SURVEY

CANCER GENETICS PROGRAM
WEST PENN ALLEGHENY HEALTH SYSTEM

Post-Cancer Genetics Information Session Survey

Date: / /

CG ID#

Please answer the following cancer genetics questions to the best of your knowledge.

Questionnaire for assessment of knowledge of Cancer Genetics:

The following are questions about hereditary breast-ovarian cancer. Please read each question carefully and circle the BEST answer for each question.

1. True or False The majority of all cancer is hereditary

2. How common is breast cancer in the general population?
1 in 8 1 in 40 1 in 100 1 in 800

3. How common is hereditary breast cancer in the general population?
1 in 8 1 in 40 1 in 100 1 in 800

4. How common is hereditary breast cancer in individuals of Ashkenazi Jewish ancestry?
1 in 8 1 in 40 1 in 100 1 in 800

5. True or False Cancer is only passed through your mother (mother's side of the family)

6. True or False Having a cancer gene means you will definitely develop cancer

7. True or False Inheriting a mutation (change) in a cancer gene increases the risk that you will develop specific types of cancer

8. How many genes must someone inherit to have Hereditary Breast-Ovarian Cancer?
a. Zero, it is not caused by genes b. One from mom c. One from mom or dad
d. Two, one from mom and one from dad e. None of the above

9. How can you tell if someone carries a gene that puts them at an increased risk for Hereditary Breast-Ovarian Cancer?

- a. They look sick b. They have been diagnosed with breast and/or ovarian cancer
- c. With a simple blood test d. There is no way to know e. None of the above

10. True or False If a person has genetic testing and no mutation is found in their cancer genes, that person will never develop cancer
11. True or False A mutation in a breast cancer gene also contributes to a higher risk of ovarian cancer
12. True or False Our genetic code (DNA) consists of A, C, T, G
13. True or False According to HIPAA (Health Information Portability and Accountability Act), genetic testing results can be used for insurance discrimination
14. What do you believe is your estimated risk of cancer (1 to 10, with 10 being the highest)? _____

Communication questions:

1. What method is the best way to communicate information about cancer risk and family history?
- ___ Health care provider
 - ___ Relative
 - ___ Friend
 - ___ Newspaper/magazine
 - ___ Internet
 - ___ Synagogue/religious organization
 - ___ Other: _____
2. Do you think healthcare professionals are aware of your Ashkenazi Jewish ancestry when discussing health issues, especially relating to cancer?

Health belief questions:

For the following questions, please tell us whether you:

- 5. STRONGLY AGREE
- 4. AGREE
- 3. NEUTRAL
- 2. DISAGREE
- 1. STRONGLY DISAGREE

Severity

1. Hereditary Breast-Ovarian Cancer is a serious disease_____
2. I think passing a gene mutation to my children would be scary_____
3. My life would change if I had a gene mutation that predisposed me to Hereditary Breast-Ovarian Cancer_____

Susceptibility

1. I feel I am at risk for developing Hereditary Breast-Ovarian Cancer_____
2. There is a genetic cause to the cancers in my family_____
3. I carry a genetic susceptibility to Hereditary Breast-Ovarian Cancer_____

Benefit

1. It is useful to know whether I have an increased risk to develop breast or ovarian cancer_____
2. It is useful to know whether I have a gene mutation that causes an increased risk to develop breast or ovarian cancer _____
3. Knowing the risk of developing breast or ovarian cancer would change my medical management_____

Barriers

1. Testing for a gene mutation known to cause breast and/or ovarian cancer is **painful and difficult** _____
2. Testing for a gene mutation known to cause breast and/or ovarian cancer would **increase my anxiety and make me worry** about other family members _____
3. Testing for a gene mutation known to cause breast and/or ovarian cancer would **make me feel guilty if my children were found to inherit the gene mutation** _____
4. I would not want to pay for genetic testing if it is not paid for by my insurance_____
5. I do not know how to find a genetic counselor in my area _____
6. I am worried about health insurance, life insurance and job discrimination _____
7. I would feel ashamed, singled-out and/or viewed negatively in my community if I participated in genetic testing _____
8. I think **fear of knowing cancer risk** is a block to genetic counseling for breast cancer _____
9. I think **lack of genetic information** is a block to genetic counseling for breast cancer _____

10. I think **not knowing family history** is a block to genetic counseling for breast cancer _____

11. This information would provide a benefit to my family and me _____

13. I know how to find more information _____

Please share with us additional comments regarding genetic counseling and testing that were not addressed in the questions above.

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