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ADVERSE IMPACT OF PREDISPOSITION TESTING ON MAJOR LIFE ACTIVITIES: LESSONS FROM *BRCA1/2* TESTING

KATHERINE A. SCHNEIDER, M.P.H.*

INTRODUCTION

Genetic testing for adult onset conditions is coming of age. There are currently several dominant hereditary syndromes for which testing can be performed prior to the onset of any signs of disease. The availability of predisposition genetic testing brings great hope regarding treatment of genetic diseases yet may also be creating a new genetic underclass of people, referred to by Paul Billings as "the asymptomatic ill."¹ Individuals who carry an inherited predisposition to a specific disease are not ill or disabled by standard definitions, yet they (and others around them) may perceive that their health status has been dramatically altered.

There have been concerns that knowing one carries an inherited predisposition to disease might adversely affect major life activities. Major life activities include day-to-day functioning such as caring for oneself, walking, talking, and breathing² but might also be expanded to include expected milestones such as getting married and having children, setting and achieving professional goals, and becoming financially secure. While the average person takes such milestones for granted, individuals with chronic illness or disabilities face significant obstacles along the way, including physical or psychological limitations and negative societal perceptions. An individual at increased risk for developing a future disease, based on a genetic test result, may face similar types of obstacles despite his/her current level of health. As Andrew Baum writes in his article on the stress of genetic testing, "[u]ncertainty about one's future can affect long-term planning, re-

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1. *Rising Cost of Private Health Insurance, 1991: Hearing Before the Subcomm. on Commerce, Consumer Protection, and Competitiveness of the House Comm. on Energy and Commerce*, 102d Cong. 12 (1991) (statement of Paul Billings, Vice Chairman, Dept. of Medicine, Pacific Presbyterian Medical Center).

2. See 29 C.F.R. § 1630.2(i) (1999).

productive decisions, health behaviors, and choices and expectations for career, family, and the future."³

In this Article, the author explores the potential adverse impact of predisposition testing on major life activities by using the example of *BRCA1/2* testing for breast cancer susceptibility. After providing a brief overview of *BRCA1/2* gene testing in Part I,⁴ the author discusses the potential short-term and long-term adverse effects of learning one's *BRCA1/2* gene status on emotional well-being in Part II.⁵ In Part III, the author discusses the adverse effects of testing on personal and familial relationships,⁶ while in Part IV, the author discusses the negative impact of gene testing on insurability and employment opportunities.⁷

I. BACKGROUND INFORMATION ABOUT *BRCA1/2* TESTING

This section provides a description of the *BRCA1/2* genes, associated cancer risks, and details about the testing process.

A. *Inheritance Pattern of the BRCA1/2 Genes and Associated Cancer Risks*

Approximately 5%-10% of breast cancer cases are due to an inherited factor, i.e. a germline mutation in a breast cancer susceptibility gene.⁸ The identification of two such genes, termed *BRCA1* and *BRCA2*, was completed approximately five years ago.⁹ Germline mutations in the *BRCA1* and *BRCA2* genes account for most, but not all, families with Hereditary Breast and Ovarian Cancer Syndrome (HBOCS).¹⁰ DNA errors have been found throughout the *BRCA1/2* genes, although three specific mutations seem to account for 90% of

3. Andrew Baum & Andrea L. Friedman, *Stress and Genetic Testing for Disease Risk*, 16 HEALTH PSYCHOL. 8, 9 (1997); see also THOMAS D. GELEHRTER ET AL., PRINCIPLES OF MEDICAL GENETICS 265 (2d ed. 1998).

4. See *infra* text accompanying notes 8-22.

5. See *infra* text accompanying notes 23-52.

6. See *infra* text accompanying notes 53-60.

7. See *infra* text accompanying notes 61-82.

8. See KENNETH OFFIT, CLINICAL CANCER GENETICS: RISK COUNSELING AND MANAGEMENT 72 (1998).

9. See Yoshio Miki et al., *A Strong Candidate for the Breast and Ovarian Cancer Susceptibility Gene BRCA1*, 266 SCIENCE 66, 66 (1994); Richard Wooster et al., *Identification of the Breast Cancer Susceptibility Gene BRCA2*, 378 NATURE 789, 789 (1995).

10. See OFFIT, *supra* note 8, at 66-155.

inherited breast cancers among individuals of Ashkenazi (Eastern European) Jewish ancestry.¹¹

Hereditary Breast and Ovarian Cancer Syndrome follows an autosomal dominant pattern of inheritance.¹² Both men and women can carry *BRCA1/2* mutations and can pass them on to their offspring.¹³ Each child has a 50% chance of inheriting the *BRCA1/2* mutation.¹⁴

Lifetime cancer risks for people with *BRCA1/2* mutations are still being established. Women have high lifetime risks of breast cancer (approximately 50-85%) and increased risks of ovarian cancer (approximately 20-40% if *BRCA1*-positive and 10-20% if *BRCA2*-positive).¹⁵ Women may also have slightly increased risks of other types of cancer.¹⁶ Men may have small, increased risks of specific cancers as well, notably, breast and pancreatic cancer in *BRCA2* mutation carriers, but their daughters bear the most significant cancer risks.¹⁷ It is important to emphasize that most men and some women with *BRCA1/2* mutations never develop cancer.¹⁸

B. Testing Options for *BRCA1/2* Mutations

BRCA1/2 testing is available in both clinical and research testing programs. Individuals undergoing testing are recommended to receive genetic counseling both prior to testing and at results disclosure. The genetic testing process begins by analyzing the entire *BRCA1* and *BRCA2* genes in one family member, ideally someone likely to have a mutation.¹⁹ This means that the initial test is usually performed on a

11. See generally Dvorah Abeliovich et al., *The Founder Mutations 185delAG and 5382insC in BRCA1 and 6174delT in BRCA2 Appear in 60% of Ovarian Cancer and 30% of Early-Onset Breast Cancer Patients Among Ashkenazi Women*, 60 AM. J. HUM. GENETICS 505, 505 (1997).

12. See GELEHRTER ET AL., *supra* note 3, at 265.

13. See Deborah Ford et al., *Risks of Cancer in BRCA1-Mutation Carriers*, 343 LANCET 692, 694 (1994).

14. See Henry T. Lynch et al., *DNA Screening for Breast/Ovarian Cancer Susceptibility Based on Linked Markers*, 153 ARCHIVES INTERNAL MED. 1979, 1979 (1993).

15. See Douglas F. Easton et al., *Breast and Ovarian Cancer Incidence in BRCA1-Mutation Carriers*, 56 AM. J. HUM. GENETICS 265, 270 (1995); The Breast Cancer Linkage Consortium, *Cancer Risks in BRCA2 Mutation Carriers*, 91 J. NAT'L. CANCER INST. 1310, 1314 (1999); Jeffrey P. Struewing et al., *The Risk of Cancer Associated with Specific Mutations of BRCA1 and BRCA2 Among Ashkenazi Jews*, 336 NEW ENG. J. MED. 1401, 1401 (1997).

16. See Abeliovich et al., *supra* note 11, at 513.

17. See Noralane M. Lindor et al., *The Concise Handbook of Family Cancer Syndromes*, 90 J. NAT'L CANCER INST. 1039, 1048 (1998).

18. See *id.* at 1047-48.

19. See Katherine Schneider, *Genetic Counseling for BRCA1/BRCA2 Testing*, 1 GENETIC TESTING 92 (1997).

woman who has had ovarian and/or pre-menopausal breast cancer.²⁰ Finding a specific *BRCA1/2* mutation in this woman positively establishes a link between the breast/ovarian cancer in the family and the underlying genetic cause. It is then possible to offer site-specific genetic testing to other blood relatives, including those who have not had cancer.

To illustrate this point further, consider the example of two sisters who are eager to have *BRCA1/2* testing because their mother died from breast cancer at age thirty-five and their maternal aunt was just diagnosed with ovarian cancer. Genetic testing for this family would ideally begin with the aunt to establish a link between the cancer and a specific *BRCA1/2* gene mutation. If the aunt does have a specific mutation, then either or both sisters can decide to be tested for the familial mutation. The tests for the sisters would be considered definitive, i.e. either the mutation is present (a positive result) or absent (a true negative result). However, if the aunt refuses to have *BRCA1/2* testing, either or both sisters can still opt to be tested, but they would need to undergo a full analysis of both genes. Furthermore, they would be cautioned that a negative result in this scenario (termed indeterminate negative) would not rule out the possibility of an inherited susceptibility, although a positive result would continue to be meaningful.

C. Uptake of *BRCA1/2* Testing

Despite widespread availability of *BRCA1/2* testing and high anticipated uptake based on attitudinal surveys, only about half of the individuals eligible for testing elect to learn their gene status.²¹ People decide not to have *BRCA1/2* testing for a multitude of reasons. These include a lack of interest in obtaining genetic information, uncertainty that results would change their screening practices in any way, fear that results could be positive, concerns about possible insurance discrimination, and inappropriate timing because of seriously ill family members or other stresses.²²

20. KATHERINE A. SCHNEIDER, COUNSELING ABOUT CANCER: STRATEGIES FOR GENETIC COUNSELORS 122-23 (1994) [hereinafter COUNSELING ABOUT CANCER].

21. See Caryn Lerman et al., *BRCA1 Testing in Families with Hereditary Breast-Ovarian Cancer: A Prospective Study of Patient Decision Making and Outcomes*, 275 JAMA 1885, 1890 (1996).

22. See Katherine Schneider et al., *Testing for Cancer Genes: Decisions, Decisions*, 1 NATURE MED. 302, 303 (1995).

II. ADVERSE EFFECTS ON EMOTIONAL WELL-BEING

At the extreme end of the spectrum, adverse emotional states, such as anxiety, distress, or depression, can lead to feelings of hopelessness and suicidal ideation.²³ However, even moderate levels of these emotions can, over time, hamper one's ability to function on a daily basis or impede personal or professional happiness.

In a thoughtful discourse on genetic testing, Andrew Baum writes, "[a]s has often been the case, society's technological capabilities have outpaced its understanding of the psychological consequences."²⁴ Although *BRCA1/2* testing programs have been reassuring about the lack of serious adverse psychological effects,²⁵ many individuals do experience some degree of psychological distress following the disclosure of their genetic test results.

Part II of this Article discusses the impact of genetic testing on the emotional well-being of patients. The author discusses the baseline emotional vulnerability of patients before testing and patients' anticipated reactions to test results. Also described are the actual reactions to test results and the short-term and long-term impact of learning results, including distress, depression, guilt, disease monitoring behaviors, lowered self-esteem, goals and expectations, as well as the effects of a negative test result.

A. Emotional Vulnerability at Baseline

Baseline levels of heightened anxiety and cancer-related distress have been observed in both high risk clinics and genetic testing programs. One program reported that 13% of their high risk patients had symptomatology consistent with an affective disorder,²⁶ and in our own *BRCA1* testing program, we found 21.7% who, at baseline, met criteria for needing further psychological assessment.²⁷ The increased anxiety and cancer-related distress can be due to a variety of causes, but the most significant variable appears to be the person's prior experiences with cancer in close relatives.²⁸

23. See Baum & Friedman, *supra* note 3, at 8-9.

24. *Id.* at 9.

25. See Lynch et al., *supra* note 14, at 1983 (noting that, to date, there is no evidence that disclosure leads to serious emotional disturbances).

26. See Penelope Hopwood et al., *Psychological Support Needs for Women at High Genetic Risk of Breast Cancer: Some Preliminary Indicators*, 7 *PSYCHO-ONCOLOGY* 402, 405 (1998).

27. See Stephanie A. Kieffer et al., *Assessment and Management of Psychologically Challenging Cases in Predisposition Testing for Cancer Susceptibility*, abstract presented at the National Society of Genetic Counselors 15th Annual Education Conference (Oct. 26, 1996).

28. See Ora Gilbar, *Women with High Risk for Breast Cancer: Psychological Symptoms*, 80 *PSYCHOL. REP.* 800, 801 (1997).

It is not uncommon for cancer risk counselors to refer patients to a mental health professional for more in-depth counseling. Reasons for making a psychological referral include the following: clinical levels of depression, guilt, or anxiety; unresolved grief; suicidal ideation; obsessive, intrusive thoughts about cancer or loss; overzealous health vigilance; severe sleep or eating disturbance; disruptions in sexual function and satisfaction; and relationship problems.²⁹

In a study of 256 women with at least one first-degree relative with breast or ovarian cancer, higher levels of general distress were observed in three groups of women: women who were not partnered, women who were less optimistic, and women who had exaggerated perceptions of their cancer risks combined with feelings that they had little control over whether breast cancer occurred.³⁰

These observations suggest that at least some individuals presenting for *BRCA1/2* testing will be psychologically vulnerable and that there are a variety of factors influencing reactions to the test results.

B. Anticipated and Actual Reactions from Test Result

Because *BRCA1/2* testing is still new, there has been great interest in identifying possible adverse reactions to learning test results. Among the factors that may influence the reactions to test results are biologic proximity and emotional ties to affected relatives, beliefs and fears about cancer, history of childhood loss, major life transitions, outcomes of cancer diagnoses, and family communication styles.³¹ Studies have examined both anticipated and actual reactions upon disclosure of *BRCA1/2* results.

1. Anticipated Reactions

Lerman and colleagues surveyed 105 first-degree female relatives of breast cancer patients for interest in *BRCA1* testing and anticipated

29. See generally Hopwood et al., *supra* note 26, at 404 (describing psychiatric problems of subjects who participated in study); June A. Peters & Jill E. Stopfer, *Role of the Genetic Counselor in Familial Cancer*, 10 *ONCOLOGY* 159, 160 (1996) (indicating several areas in which genetic counselors incorporate active counseling); Jeffrey R. Botkin et al., *A Model Protocol for Evaluating the Behavioral and Psychological Effects of BRCA1 Testing*, 88 *J. NAT'L CANCER INST.* 872 (1996) (describing a model testing program which includes a psychological assessment and plan for providing mental health referrals).

30. See Janet Audrain et al., *Psychological Distress in Women Seeking Genetic Counseling for Breast-Ovarian Cancer Risk: The Contributions of Personality and Appraisal*, 19 *ANNALS BEHAV. MED.* 370, 372-75 (1997).

31. See Peters & Stopfer, *supra* note 29, at 163.

reactions to test results.³² Not surprisingly, most women expected to have some degree of psychological sequelae³³ from a positive test result, including increased anxiety (83%), depression (80%), and impaired quality of life (46%).³⁴ Interestingly, 72% of the women thought that they would still worry about their cancer risks even if their results were negative.³⁵ Another study reported that women who anticipated heightened anxiety with a positive test result were significantly more anxious and depressed at baseline than the others surveyed.³⁶

2. Actual Reactions

Lynch and colleagues provided genetic counseling and testing to 388 individuals within 14 families with HBOCS.³⁷ Of the 78 individuals who learned they had positive results, the initial reactions included sadness (36%), anger (6%), and guilt (8%).³⁸ Nineteen percent had no apparent reaction, which could, at least in some, be indicative of a shock response.³⁹ This is borne out by findings in a separate study, which showed that individuals who exhibited lower anxiety and depression scores at results disclosure seemed less willing or able to discuss the implications of the test results at that moment.⁴⁰

C. Short and Long Term Impact on Emotional Well-Being

Genetic testing research programs have observed and reported several specific psychological responses to learning *BRCA1/2* results, including distress, depression, guilt, a negative impact on monitoring behaviors, lowered self-esteem, goals, and expectations, and the adverse effect of a negative test result.

32. See Caryn Lerman et al., *Interest in Genetic Testing Among First-Degree Relatives of Breast Cancer Patients*, 57 AM. J. MED. GENETICS 385, 388 (1995).

33. "Sequela" is defined as a morbid condition following or occurring as a consequence of another condition or event. See MILLER-KEANE ENCYCLOPEDIA & DICTIONARY OF MEDICINE, NURSING, AND ALLIED HEALTH 1354 (Michael J. Brown, ed., 5th ed. 1992).

34. See Lerman, *supra* note 32, at 388.

35. See *id.*

36. See C. Julian-Reynier et al., *Attitudes Towards Cancer Predictive Testing and Transmission of Information to the Family*, 33 J. MED. GENETICS 731, 733 (1996).

37. See Henry T. Lynch et al., *A Descriptive Study of BRCA1 Testing and Reactions to Disclosure of Test Results*, 79 CANCER 2219, 2220 (1997).

38. See *id.* at 2223.

39. See *id.*

40. See A. Christine Dudok de Wit et al., *Distress in Individuals Facing Predictive DNA Testing for Autosomal Dominant Late-Onset Disorders: Comparing Questionnaire Results with In-Depth Interviews*, 75 AM. J. MED. GENETICS 62, 71 (1998).

1. *Distress*

Croyle and colleagues found that the twenty-five *BRCA1* gene mutation carriers in his series demonstrated higher levels of test-related psychological distress when compared to the thirty-five noncarriers.⁴¹ The highest levels of overall distress were in women who have never had cancer and who had never undergone prophylactic surgery.⁴² Another study found that levels of "cancer-specific" distress (i.e. distress specifically related to fears about developing cancer) were more likely to be influenced by *BRCA1* testing than overall or "global" levels of distress.⁴³ It is reassuring to note that the levels of distress associated with receiving positive *BRCA1/2* results appear to significantly decline by six months as people become adjusted to the information.⁴⁴

2. *Depression*

One *BRCA1* testing program of 279 individuals did not find any increased depression and functional impairment among the 53 with positive results, although individual scores over time were not reported.⁴⁵ It is encouraging that *BRCA1/2* testing does not appear to cause major psychological sequelae, yet all testing programs have clinical experiences that remind us of the power of this information. Anecdotally, one woman tested through our program, who had not anticipated a positive result, later admitted she could not recall any of our lengthy discussion nor even who was present in the room during the disclosure. She said that it took weeks before she could focus on anything else. Another woman, whose husband and daughter both carry *BRCA1* mutations, related in a follow-up conversation that she and her daughter were tearful for months afterwards as they struggled to come to terms with the information.

3. *Guilt*

One of the most emotionally charged aspects about carrying an inherited predisposition to breast cancer is that one's children or siblings might have the disposition as well. Women who accept with

41. See Robert T. Croyle et al., *Psychological Responses to BRCA1 Mutation Testing: Preliminary Findings*, 16 HEALTH PSYCHOL. 63, 67 (1997).

42. See *id.* at 69.

43. See Caryn Lerman et al., *The Influence of Psychological Distress on Use of Genetic Testing for Cancer Risk*, 65 J. CONSULTING AND CLINICAL PSYCHOL. 414, 417 (1997).

44. See A.Christine Dudok deWit et al., *Course of Distress Experienced by Persons at Risk for an Autosomal Dominant Inheritable Disorder Participating in a Predictive Testing Program: An Exploratory Study*, 60 PSYCHOSOMATIC MED. 543, 545 (1998).

45. See Lerman et al., *supra* note 21, at 1885.

equanimity the risks to themselves may find the same risks untenable in their daughters. The initial person in the family who tests positive may also feel guilty about "opening Pandora's box" within the family and having to inform other members of the family that they are also at risk. Conversely, family members who test negative for a familial mutation may also experience guilt, termed survivor guilt,⁴⁶ because they were spared from the inherited cancer predisposition while others in the family were not.

4. *Negative Impact on Monitoring Behaviors*

Nancy Wexler writes, "[w]ith a positive test, the ambiguity of waiting for the disease may be unbearable. Gene carriers prematurely become 'patients,' possibly forfeiting years of otherwise good health."⁴⁷ Ms. Wexler is referring to individuals at risk for Huntington's Disease (HD), but this statement also applies to women with a positive *BRCA1/2* test result. A positive result may exacerbate a woman's feelings of vulnerability about developing cancer. In extreme cases, this could either lead to a preoccupation with disease-related signs or to a complete avoidance of physicians. Anecdotally, women with *BRCA1/2* mutations identified through our program have indicated that their routine cancer screening appointments are now more anxiety-provoking. This is concerning, because in a study of 180 women, higher levels of breast cancer worry were associated with poorer adherence to mammography screening.⁴⁸

5. *Lowered Self-Esteem*

Upon being told of her positive *BRCA1/2* result, one woman in our testing program remarked that the result made her feel like "damaged goods." Self-esteem is influenced by multiple factors, including past experiences, personality type, coping mechanisms, support network,⁴⁹ and perhaps, in the future, by one's genetic make-up. Words can invoke powerful images and considering the manner in which susceptibility genes are described—examples include damaged, defective, bad, mutated—it is certainly conceivable that a positive result

46. "Survivor guilt" is a guilty feeling individuals may experience when contrasting their own good health with relatives who have developed cancer. See COUNSELING ABOUT CANCER, *supra* note 20, at 108.

47. Nancy S. Wexler, *Disease Gene Identification: Ethical Considerations*, HOSP. PRAC., Oct. 15, 1991, at 147.

48. See Caryn Lerman, *Adherence and Psychological Adjustment Among Women at High Risk for Breast Cancer*, 28 BREAST CANCER RES. & TREATMENT 145, 148 (1993).

49. See DEBORAH Y. ANDERSON & CHRISTOPHER L. HAYES, GENDER, IDENTITY, AND SELF-ESTEEM xiii-xiv, 58-59, 142-43 (1996).

could cause or reinforce lowered self-esteem. This is an area that deserves greater attention as more individuals with inherited susceptibilities to disease are identified.

6. *Lowered Goals and Expectations*

BRCA1/2 testing is available to at-risk individuals who are 18 years old or older. Thus, individuals being tested may still be in college or in the midst of making career choices. Researchers do not yet know how genetic test results will affect these decisions, although Nancy Wexler and others have raised concerns about the impact of a positive genetic test result, “[w]ill a young person, age 21, now aware that there is a 95% probability of carrying the gene, expend the time, money, and energy to achieve a chosen career?”⁵⁰

7. *Adverse Effects of a Negative Test Result*

A subset of patients who receive negative results will also experience adverse psychological sequelae. Contrary to expectations, negative results may not always relieve the individual’s level of distress or significantly improve his or her quality of life. In addition to experiencing intense survivor guilt,⁵¹ individuals receiving so-called “good news” may have difficulty assimilating the information after considering themselves at high risk for so long or may regret major life decisions (such as choice of partner or career) that were influenced by their fears of developing cancer.⁵²

III. ADVERSE EFFECTS ON PERSONAL AND FAMILIAL RELATIONSHIPS

June Peters and Jill Stopfer, both cancer genetic counselors, write, “[t]he emotional impact of cancer goes beyond the person diagnosed with the malignancy; family and friends also are deeply affected.”⁵³ This statement is equally apropos for individuals found to have an inherited predisposition to cancer. In this section, the author discusses the impact of testing on family relationships as well as decisions about marriage and child-bearing.

50. Wexler, *supra* note 47, at 147.

51. See *supra* note 46.

52. See COUNSELING ABOUT CANCER, *supra* note 20, at 129-30 (discussing risks associated with negative results regarding the carrying of altered genes).

53. Peters & Stopfer, *supra* note 29, at 163.

A. *Impact on Family Relationships*

Relationships within a family can change dramatically as family members share their test results with one another. Individuals may have complicated reactions to learning a relative's result because it has serious implications both for their relatives and for themselves. Blood relatives, some of whom may have only vaguely been aware of the cancer history, may suddenly be faced with being at increased risk of cancer and having to make unwelcome decisions about testing.⁵⁴

At the Dana-Farber Cancer Institute genetic testing program, we have witnessed serious family rifts develop as a consequence of genetic testing. This has occurred, for example, when the only living affected family member either declines to be tested to the great disappointment of at-risk relatives or feels as though he or she is being coerced into testing by relatives. Testing may also cause family members to share and discuss information that had never been talked about. In one of the families with whom I worked, the mother had concealed her extensive personal and familial history of cancer from her five adult children until she received a positive genetic test result; disclosure of this information caused a severe family rift. Family conflicts may also arise as family members make different decisions about being tested or about their medical management post-disclosure.⁵⁵

Learning one's test results can also cause a certain awkwardness when talking to other family members or friends. As examples, one testing participant in her twenties stated that she had no one to talk to about her positive *BRCA1/2* result; her husband and family members became upset whenever she mentioned it, and her friends, who never worried about cancer themselves, had difficulty relating to her situation. Another woman, with a negative result, spoke of feeling "left out" when her sister and female cousin spoke of issues relating to their positive results.

B. *Impact on Marriage and Child-Bearing Decisions*

The effect of genetic testing on marital and childbearing practices is not yet known. In a survey of 105 women, about one fourth thought that learning their *BRCA1* results would be important in making marital decisions and about one half thought it would be important in making childbearing decisions.⁵⁶

54. See COUNSELING ABOUT CANCER, *supra* note 20, at 125-26.

55. See *id.* at 121-39.

56. See Lerman et al., *supra* note 32, at 388.

1. *Decisions about Marriage*

As stated earlier, women with higher levels of general distress were more likely to be unmarried.⁵⁷ It is not yet known whether women who test *BRCA1/2* positive will be less likely to seek marriage after learning their results or whether they could face greater difficulty finding partners. The latter may be especially problematic for women in cultures where marriages are frequently arranged and may cause these women to decline testing for fear of stigmatization.⁵⁸

2. *Child-Bearing Decisions*

Women with positive *BRCA1/2* results are offered the option of prophylactic oophorectomy (surgical removal of ovaries) to reduce their risks of ovarian cancer.⁵⁹ Thus, women may decide to have their children at younger ages rather than delaying childbearing until their late thirties or early forties. Some women may elect to have fewer children or not to have children at all, either because of fears that they might pass on the gene mutation to their offspring or fears that they might not live long enough to raise a child to adulthood.⁶⁰

IV. ADVERSE EFFECTS ON INSURANCE AND EMPLOYMENT

A. *Genetic Discrimination*

In an article discussing the potential ramifications of genetic testing, Marvin Natowicz warns, “[t]he practice of genetic discrimination has the potential of creating a new group of disadvantaged people who will need the same protections now accorded those suffering from race and sex discrimination.”⁶¹ Genetic discrimination is defined as:

discrimination against an individual or against members of that individual’s family solely because of real or perceived differences from the “normal” genome in the genetic constitution of that individual

. . . .

People at risk for genetic discrimination are (1) those individuals who are asymptomatic but carry a gene(s) that

57. See Audrain et al., *supra* note 30, at 372.

58. See Renee Royak-Schaler & Beryl Lieff Benderly, *The Role of Risk in Daily Life*, in CHALLENGING THE BREAST CANCER LEGACY 132 (1992).

59. See Lerman et al., *supra* note 21, at 1891.

60. See COUNSELING ABOUT CANCER, *supra* note 20, at 110.

61. Marvin R. Natowicz et al., *Genetic Discrimination and the Law*, 50 AM. J. HUM. GENETICS 465, 465 (1992).

increases the probability that they will develop some disease, (2) individuals who are heterozygotes (carriers) for some recessive or X-linked genetic condition but who are and will remain asymptomatic, (3) individuals who have one or more genetic polymorphisms that are not known to cause any medical condition, and (4) immediate relatives of individuals with known or presumed genetic conditions.⁶²

1. *Extent of Genetic Discrimination*

To date, there have been few reported cases of genetic discrimination among individuals with *BRCA1/2* mutations, although until recently, most testing occurred within protected research programs. With the advent of commercial testing has come increased physician referrals for genetic testing (which may be duly noted in medical charts) and increased requests for reimbursement of testing-related expenses.⁶³ Thus, it will be important to continue monitoring this issue over time. There have been several well-documented cases of genetic discrimination experienced by individuals with a variety of genetic disorders, including one case of a woman with a family history of Huntington's Disease, who was told that she would qualify for a life insurance policy only if she underwent genetic testing and was found to be negative.⁶⁴

2. *Patients' Perception of Genetic Discrimination*

Individuals considering *BRCA1/2* testing consistently identify health insurance discrimination among their major concerns about testing. In one testing program, 34% of individuals reported that the possibility of losing health insurance was a major risk of having *BRCA1* testing.⁶⁵ This fear may discourage individuals from being tested and may also keep individuals from sharing positive test results—potentially important medical information—with their physicians.

B. *Insurance Discrimination*

Genetic discrimination by insurers may result in canceled or denied health, life, or disability insurance policies, limitations in cover-

62. *Id.* at 466.

63. See generally The Ad Hoc Committee on Genetic Testing/Insurance Issues, *Background Statement: Genetic Testing and Insurance*, 56 AM. J. HUM. GENETICS 327, 328-29 (1995) [hereinafter The Ad Hoc Committee on Genetic Testing/Insurance Issues] (discussing implications of genetic testing with respect to insurance policies).

64. See Paul R. Billings et al., *Discrimination as a Consequence of Genetic Testing*, 50 AM. J. HUM. GENETICS 476, 480 (1992).

65. See Lerman et al., *supra* note 21, at 1890.

age, or higher premiums for coverage. At present, insurers do not themselves conduct genetic testing for applicants, although they may be interested in learning the test result.⁶⁶ Insurers can gain access to genetic test results through physician records or by asking the individual for the result during the application process.⁶⁷ Individuals are required to answer such questions honestly or they are at risk for policy cancellation or legal prosecution for insurance fraud.⁶⁸

1. *The Practice of Underwriting*

Individual rates of insurance are determined by considering the person's overall risk factors and chances of morbidity or untimely death.⁶⁹ Individuals with few risk factors pay average rates, while individuals with unique or multiple risk factors pay higher rates or are denied policies altogether.⁷⁰ Thus, higher rates are charged to smokers, diabetics, and motorcyclists. However, this practice becomes more problematic when considering individuals who are asymptomatic yet carry inherited susceptibilities to disease. For individuals with *BRCA1/2* mutations, two central questions are raised: does a germline *BRCA1/2* mutation constitute a pre-existing condition?; and do insurers have the right to base policy decisions on possible future disease?

a. *Does a germline BRCA1/2 mutation constitute a pre-existing condition?*

It is true that *BRCA1/2* mutations, which are associated with HBOCS, are present from birth. However, individuals with *BRCA1/2* mutations are not born with any signs or symptoms of HBOCS and, in fact, may not develop cancer for several decades (if at all).⁷¹ Thus, it does not seem appropriate to label a person as having a pre-existing condition years before the onset of symptoms. The condition should be considered present with the first signs of disease, not before. It is also inappropriate to label a person as having a "pre-existing condition" because of his/her genetic make-up (as opposed to behavior which is theoretically under the person's control), especially since all

66. See Gail Geller et al., *Genetic Testing for Susceptibility to Adult-Onset Cancer: The Process and Content of Informed Consent*, 277 JAMA 1467, 1471 (1997).

67. See *id.*

68. See *id.*

69. See The Ad Hoc Committee on Genetic Testing/Insurance Issues, *supra* note 63, at 327.

70. See generally *id.* (describing how standard premiums are calculated based upon the provider's expected outcome for different groups of individuals with similar risks).

71. See generally Lynch et al., *supra* note 37, at 2223 (noting that there is a limited ability to predict the age of cancer onset among *BRCA1*-positive individuals).

human beings are thought to carry deleterious genes that predispose to disease.

b. Do insurers have the right to base policy decisions on possible future disease?

The entire premise of underwriting is based upon making predictions about future disease given certain characteristics or behaviors.⁷² The concern about basing decisions on genetic test results, such as a positive *BRCA1/2* result, is the inability to assign risks of disease or eventual outcome (i.e. death) with any precision. This may change in the future as our knowledge base grows, yet even with HD, a condition with 100% penetrance, it is not possible to predict the age at which symptoms will occur or how rapid the disease course will be.⁷³

2. The Issue of Adverse Selection

An alternative problem, from the perspective of the insurer, is adverse selection. Adverse selection occurs when individuals have more information about their risks of specific illnesses than do their insurance companies.⁷⁴ Not only does this allow these "high risk" individuals to purchase insurance at the same rates as those designated "average risk," but it might lead individuals to obtain more insurance than they otherwise would have. Adverse selection has the potential for jeopardizing the economic well-being of the insurance company or may lead companies to raise all premiums as protection.⁷⁵

C. Employment Discrimination

Genetic discrimination by employers refers to unfavorable treatment in hiring, promotion, assignment of duties, and privileges of employment.⁷⁶ There are concerns that individuals with positive *BRCA1/2* test results will be viewed as less desirable employees because of their potential for serious illness. Since a number of cancer survivors as well as individuals with genetic disorders have experienced workplace discrimination,⁷⁷ these concerns would appear to be valid. The Amer-

72. See The Ad Hoc Committee on Genetic Testing/Insurance Issues, *supra* note 63, at 327.

73. See GELEHRTER ET AL., *supra* note 3, at 28, 217.

74. See The Ad Hoc Committee on Genetic Testing/Insurance Issues, *supra* note 63, at 329.

75. See *id.*

76. See Council on Ethical and Judicial Affairs, American Medical Association, *Use of Genetic Testing by Employers*, 266 JAMA 1827, 1827 (1991).

77. See Billings et al., *supra* note 64, at 478; see also Mark A. Rothstein et al., *Are Cancer Patients Subject to Employment Discrimination?*, 9 ONCOLOGY 1303, 1306 (1995).

ican Medical Association Council on Ethical and Judicial Affairs has carefully considered potential uses of genetic testing by employers.⁷⁸ Their report discusses three major rationales that might be used to justify genetic discrimination by employers: 1) public safety issues; 2) employee safety issues; and 3) higher costs and lower productivity.⁷⁹

1. *Public Safety Issues*

There may be rare instances when the risk of illness or disability in the employee can negatively impact public safety. This is unlikely to be the case for employees with *BRCA1/2* mutations; however, it might be an issue for individuals at risk for other serious genetic conditions. For example, should a commercial airline continue to employ a pilot who is at 50% risk for developing Huntington Disease's (HD), a degenerative neurological condition? If this individual is showing signs of HD, then it seems clear that concerns about public safety should prevail, and the pilot should no longer be allowed to fly. However, consider an individual at risk for HD, who may not develop any symptoms of HD for ten, twenty, or even thirty years. Arguing that this individual is a public safety hazard based on "possible future illness" seems much less reasonable, especially since all commercial pilots are required to demonstrate physical and neurologic fitness on an annual basis. It would be difficult to prove that employees who might in the future develop a specific illness or disability pose a risk to public safety.

2. *Employee Safety Issues*

It is the employer's duty to protect employees from unnecessary harm. Thus, there may be instances when an employer is justified in basing employability or job assignments on an individual's genetic make-up or risk of future health problems. However, several conditions must be met before this is allowable: a) the individual's genetic make-up must be known to make him/her unusually susceptible to injury from chemicals or other substances in the workplace; b) health problems in other employees working with these chemicals or substances occur rarely or never; and c) the cost of reducing the exposure-related risks for the employee is prohibitive.⁸⁰ Given these stringent conditions, very few predictive genetic tests (if any) would

78. See Council on Ethical and Judicial Affairs, *supra* note 76, at 1827-30.

79. See *id.*

80. See *id.*

qualify, and therefore, it is unlikely that employee safety issues will prove to be valid justification for genetic discrimination.

3. *Higher Costs and Lower Productivity*

Employers, particularly in small businesses, may feel that it is prohibitively expensive to cover health, life, or disability insurance policies for employees with inherited susceptibilities to disease. Employees may also be less than enthusiastic about hiring individuals who may not be able to work as long or as productively because of the impending threat of illness or disability. However, since the Americans with Disabilities Act⁸¹ states that disabled individuals have the right to employment despite possible additional costs to the employer,⁸² this argument is certainly not valid for individuals who are currently healthy but predisposed to future disease.

CONCLUSION

In the words of Michael Yesley, “[g]enetic information will play a central role in health care, but it may destroy individuals’ self-esteem and be used to discriminate against them.”⁸³ This one sentence captures both the promise and the peril of predisposition genetic testing. The major challenges to health care providers and policy-makers are to ensure that individuals have adequate pre- and post-test counseling, access to appropriate medical resources, and protection from all forms of genetic discrimination.

81. See 42 U.S.C. §§ 12101-12213 (1994).

82. See Council on Ethical and Judicial Affairs, *supra* note 76, at 1828.

83. Michael S. Yesley, *Editor's Introduction* to Eric T. Juengst, *The Ethics of Prediction: Genetic Risk and the Physician-Patient Relationship*, 1 GENOME SCI. & TECH. 21 (1995).