

Dear Practitioner,

Advances in genetic research, along with greater effectiveness and lower cost of the associated technologies, are leading to the inclusion of genetic tests as part of standard medicine. This questionnaire focuses on genetic susceptibility tests, i.e., tests that assess the risk that healthy individuals will develop pathology.

Traditionally, genetic counseling and genetic testing for risk assessment are offered to individuals who are (or have been) ill themselves, or to healthy individuals with a suspect family history.

The purpose of this questionnaire is to examine the opinions of practitioners in various specialties on expanding the availability of genetic counseling and testing to **the (healthy) general Ashkenazi-Jewish population**, even in the **absence** of a suspect family history, in order to assess the individual's risk of morbidity.

We believe that the opinions of clinicians who deal with late-onset conditions are important in determining policy in this area, since the possibility of genetic testing of this type is already being studied in Israel and elsewhere.

The study has been approved by the Helsinki Committee of the Hadassah Medical Center in Jerusalem. Completing this anonymous questionnaire requires only a few minutes of your time.

The questionnaire focuses on two diseases: cancer and Parkinson's disease.

Thank you in advance for your cooperation,

Dr. Gili Ofer-Bialar, Specialist and Supervisor in Family Medicine, Maccabi Healthcare Services, and Sackler Faculty of Medicine, Family medicine department & medical education, Tel Aviv University

Prof. Vardiella Meiner, Director, Center for Clinical Genetics, Hadassah Medical Center

Dr. Ronit Calderon-Margalit, School of Public Health and Community Medicine, Hadassah-Hebrew University

Shiri Shkedi-Rafid, Hebrew University of Jerusalem

Enclosed is a self-addressed, stamped envelope in which to return the completed questionnaire.

What is your area of specialization?

- Family medicine Gynecology Oncology Neurology
 Clinical genetics Genetic counseling Other

Where did you study medicine/genetic counseling?

- Israel Elsewhere _____

How many years have you been working in the field of medicine/genetic counseling?

- 0-4 5-9 10-14 15-20 ≥ 21

In what setting do you work (you may choose more than one)?

- HMO Private clinic Hospital Other

The first section of the questionnaire deals with predictive genetic tests to assess the risk of cancer.

1. In an average month during the past year, how many individuals with cancer have requested genetic testing of the genes associated with increased risk of cancer?

- None 1-5 6-20 21-50 >50

2. In an average month during the past year, how many individuals who do not have cancer have requested genetic testing of the genes associated with increased risk of cancer?

- None 1-5 6-20 21-50 >50

2a. In your opinion, how many of them approached you as the result of a family history of cancer?

- The majority Half Only a minority

3. One in 40 individuals in the Ashkenazi Jewish population carries a mutation of the BRCA1/2 genes (one of three known mutations in this population). Those who are carriers have a 60%-80% risk of developing breast and/or ovarian cancer. Carriers are advised to be monitored for early detection of cancer, and are offered "preventive" surgery (such as prophylactic oophorectomy).

According to the accepted guidelines in Israel and worldwide, patients who themselves have cancer, or healthy individuals with a family history of cancer, should be referred for genetic counseling and testing.

Despite the genetic basis, roughly one half of carriers have no suspect family history, and the fact that they are carriers is discovered only after they have developed cancer.

To what extent do you agree or disagree with expanding genetic testing for the three known mutations to the general Ashkenazi population, i.e., to healthy individuals as well, who do not have a suspect family history?

- Strongly agree
- Tend to agree
- Tend to disagree
- Strongly disagree
- Not sure

4. To what extent do you agree or disagree with the following statements regarding the expansion of genetic testing for the three known prevalent BRCA1/2 mutations to the general Ashkenazi-Jewish (AJ) population, even in the absence of a suspect family history?

	Strongly agree	Tend to agree	Tend to disagree	Strongly disagree	Not sure
1. The benefits of population screening outweigh the costs					
2. Population screening will result in reduced morbidity and mortality					
3. Since most cancer patients are non-carriers, population screening is unjustified					
4. Carriers will feel greater confidence, since they will be able to protect themselves against future illness					
5. Testing will lead to stigmatization of the AJ population					
6. A negative test result will lead to false reassurance, and neglect of surveillance for early detection					
7. People may feel social pressure to be tested even if they do not wish to be					
8. Genetic information of this type will culminate in discrimination in insurance and/or employment					
9. Carriers will experience significant, long-term psychological distress					
10. Population screening will lead					

to an unjustified increase in termination of pregnancies of carrier embryos					
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5. On the assumption that testing would be offered to the general Ashkenazi-Jewish population, what do you feel would be the optimal age range at which to refer for testing?

- 20-30
 31-40
 41-50
 51-60
 61 and over

The second section of the questionnaire deals with predictive genetic tests to assess the risk of Parkinson's disease.

1. In an average month during the past year, how many individuals with Parkinson's disease have requested genetic testing of the genes associated with increased risk of Parkinson's?

- None
 1-5
 6-20
 21-50
 >50

2. In an average month during the past year, how many healthy family members, who do not have Parkinson's disease, have requested genetic testing of the genes associated with increased risk of Parkinson's?

- None
 1-5
 6-20
 21-50
 >50

3. Mutations of the LRRK2 gene are associated with increased risk (30%-70%) of developing Parkinson's disease. It has been demonstrated in the US and Israel that 2.2% of the Ashkenazi Jewish population carry a prevalent mutation of this gene. The average age of onset of symptoms among carriers and non-carriers of the mutation is similar (the late fifties).

To what extent do you agree or disagree that genetic testing for the known mutation of the LRRK2 gene should be offered to the general Ashkenazi population, that is, to healthy individuals as well, without a family history, in order to assess the risk of morbidity?

- Strongly agree
 Tend to agree
 Tend to disagree
 Strongly disagree
 Not sure

4. To what extent do you agree or disagree with the following statements regarding the expansion of genetic testing for the known LRRK2 mutation to the general Ashkenazi-Jewish population, even in the absence of a suspect family history?

	Strongly agree	Tend to agree	Tend to disagree	Strongly disagree	Not sure
1. The benefits of population screening outweigh the costs					
2. Population screening will result in reduced morbidity and mortality					
3. Since most Parkinson's disease patients are non-carriers, population screening is unjustified					
4. Carriers will feel greater confidence, since they will be able to protect themselves against future illness					
5. Testing will lead to stigmatization of the AJ population					
6. People may feel social pressure to be tested even if they do not wish to be					
7. Genetic information of this type will culminate in discrimination in insurance and/or employment					
8. Carriers will experience significant, long-term psychological distress					
9. Population screening will lead to an unjustified increase in termination of pregnancies of carrier embryos					

5. Assuming that testing would be offered to the general Ashkenazi-Jewish population, what do you feel would be the optimal age range to refer for genetic counseling regarding genes that increase the risk of developing Parkinson's disease?

- 20-30
 31-40
 41-50
 51-60
 61 and over

6. Assuming that predictive genetic tests would be offered to the general Ashkenazi-Jewish population, in your opinion is there a need for genetic counseling prior to testing?

- Testing should be performed only after counseling
(Who should provide the counseling? _____)
- It should be offered, but not mandatory
- Counseling should be given only to those identified as carriers (following testing)
- There is no need at all for genetic counseling

7. Assuming that predictive genetic tests would be offered to the general Ashkenazi-Jewish population, who in your opinion should deliver the test results and their interpretation to the patient (you may choose more than one)?

- Family practitioner
- Specialist (in accordance with the disease)
- Genetic counselor/medical geneticist
- Genetic counselor/medical geneticist together with a specialist (in accordance with the disease)
- Other _____

8. Are there other implications, not cited above, that you feel are important to note in the context of predictive genetic tests offered to the healthy general Ashkenazi-Jewish population?

In conclusion, please provide the following information about yourself to enable us to analyze the findings.

Sex: Male Female

Age: 25-45 46-65 66 and over

Religion/Ethnicity: Jewish Ashkenazi (partial or full) Jewish non-Ashkenazi
 Muslim Christian Druze Other _____

How would you define your level of religiosity?

Secular Traditional Religious Ultra-Orthodox Other _____

Were you born in Israel?

Yes No (If so, in what country? _____ Year of aliyah _____)

Are you:

Single Married or living together Divorced Widowed

Do you have children?

Yes No

Do you have first-degree relatives with breast and/or ovarian cancer?

Yes No

Do you have first-degree relatives with Parkinson's disease?

Yes No

Would you be interested in being tested for BRCA1/2 mutations?

Yes No Have already been tested Not sure

Would you be interested in being tested for mutations that increase the risk of Parkinson's disease?

Yes No Have already been tested Not sure

Do you refer the relevant age group for fecal occult blood testing?

Always Almost always Occasionally Rarely No

Thank you for your time and participation!