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*Original Paper*

# At What Age Should Information on Genetic Testing be Accessible in Order to Raise Awareness and Prevent Genetic Disease among the General Population?

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***Abstract***

*Hereditary genetic diseases are illness that may be early identified, and there is agreement of the World Health Organization on the tests' importance and its effectiveness. However, the screening test' performance, as it is currently carried out, is affected by lack of information, which its damages range between lack of general knowledge on screening test' performance to a situation in which the screening test are performed in the absence of informed consent. Therefore, there is a need to promote knowledge about the actual exams performance and their significance among general population. The question is what is the right age and manner to make this knowledge available in order to obtain a reasonable level of knowledge sufficient for informed consent. As of today there is not much literature dealing with this question and there is no consensus concerning the way of making the information available. Therefore, there is a need in additional research and extensive public debate concerning the promotion of knowledge about screening test, even only to provide informed consent in situations where the exam performance is mandatory and unavoidable.*

***Keywords***

*information, awareness, lack knowledge, genetic screening test, Hereditary diseases*

## 1. Introduction

Genetic disease is an illness resulting from a genome disorder. While these are often low-frequency diseases that may not be predicted, there is a group of genetic diseases whose source is known and can be early diagnosed. These are genetic diseases that result from a family hereditary defect, when parents who are carriers or patients transmit a defective gene to their offspring (WHO, 2018). There are methods early diagnosis of these hereditary diseases, the best known is genetic mapping, which is

directed to known hereditary diseases in the risk population (Okazaki et al., 2016). There is agreement in professional literature about the criteria to performing these tests, which have been updated over the years, but still include a requirement to defined target audience, economically feasible, the tests should be accepted by the target population (Andermann, Blancquaert, Beauchamp, & Déry, 2008). And while there is an agreement in literature about the need to advance knowledge about these tests, the question arises what is the right stage and age to begin promoting the implementation of screening tests in target population.

## 2. Performing Screening Tests

Due to global awareness to hereditary diseases, many countries facilitate the selected tests' performing according to the prevalence and cost-benefit criteria to the general population. There are countries as Cyprus or the Palestinian Authority, where genetic testing is mandatory before marriage, according to a known law (Cousens et al., 2010). On the other hand, there are countries like Israel, where performing screening tests are payed by the state, but it is not mandatory, and public's access to knowledge is usually done in first pregnancy follow-up (Ministry of Health, 2014). While the official attitude is that hereditary screening tests are performed only in feasibility state, i.e., a situation in which the test reduces the disease incidence is economically feasible (Andermann et al., 2008), there are those who claim that the knowledge and hereditary screening tests performance do not necessarily guarantee a decline in hereditary disease, often due to the population's attitude toward the test's significance. Reviewing seven programs of promoting awareness to hereditary disease in inbred marriages showed that, despite of promoting knowledge, none of the clinics in Arab countries have succeeded in canceling over 65% of marriages at risk, a goal determined by previous studies (Saffi & Howard, 2015). It should be noted that this is still an improvement, since datum in Saudi Arabia in earlier research was 90% marriage despite diagnosis and risk awareness (Cousens et al., 2010).

However, in addition to performance feasibility, making the screening test acceptable by the target population, is another criterion for making the tests performance possible (Andermann et al., 2008). Some countries enforce this "acceptance" through laws, while the refusal results are ranged from non-abrogation by church in highly religious state to reasonable cause for divorce (Cousens et al., 2010). On the other hand, there are countries in which tests are performed by choice (Ministry of Health, 2014). In any case, this choice must meet the requirement of informed consent. In case of genetic testing, this requirement does not necessarily meets reality.

## 3. The Knowledge Aspect in Performing Screening Tests

Regarding the existence of sufficient knowledge for informed consent in genetic tests, two extreme cases must be examined. The first case is not performing hereditary tests due to lack of knowledge about its existence. In fact, one of the major factors for not performing hereditary screening tests is the lack of knowledge of the parents to be for the hereditary screening test's availability, purpose and

existence. A research performed among women of Pakistani origin in England found that there was a lack of knowledge about thalassemia screening tests among the population at risk, and that there is wide demand to knowledge on this subject (Ahmed, Green, & Hewison, 2005). In addition, promoting knowledge among public is not necessarily effective. A research performed in Western Australia among 633 mothers showed that women who had private health insurance generally had higher attitudes toward screening and more knowledge. On the other hand, women with higher education and a wide knowledge about the tests had more negative attitudes towards the tests. In addition, the average score of women in screening tests questionnaire was 62%. This means that even among educated women who are accessible to knowledge about the tests, the knowledge quality was not in a level that promoted positive attitudes, and the provided knowledge level is not sufficient to obtain full informed consent (Rostant, Steed, & O'Leary, 2003).

Many factors influence parental knowledge on genetic testing. A research performed in Australia showed that higher education and exposure to genetic issues are positively correlated to the reported awareness level to genetics basic concepts and correlation between genetics and morbidity (Karger & Basel, 2008). Knowledge promotion in these situations may be significant even by providing information bulletin during pregnancy surveillance. A survey performed on this issue in Israel demonstrated a positive correlation between providing pamphlets about screening tests and women response to performing the tests (Romano-Zelekha & Shohat, 2012). On one hand, it is intervention performed during pregnancy, but on the other hand, this is an effective demonstration of intervention that narrows information gaps to promote compliance to screening tests.

However, in some countries, there may be a situation in which the screening tests are performed in general public and even by law, and there may be a situation in which the tests are performed without the informed consent or even without parents' consent. There are only few researches on this issue, but a research performed in 200 women in Australia showed that only 26.5% of the participants knew their baby had postnatal screening and which hereditary diseases were examined (Suriadi, Jovanovska, & Quinlivan, 2004). In some states in the United States, it is assumed that the parent agrees, or that he may agree without defining an informed consent (written or not, refusal or not, for whatever reason). Contrary to certain medical procedures, the common view is that no informed consent is required for hereditary screening procedures "because it takes time" (Ross, Saal, David, & Anderson, 2013).

What these two extreme cases have in common is the lack of knowledge on which the informed consent should be based on, and according to which the screening test will be performed. Table 1 summarizes researches dealing with existing information gaps and their impact on attitudes and performing hereditary screening tests. It can be seen that the knowledge about the tests is usually obtained by independent means (for example, on the basis of higher education), not always available to the general population, and is not provided in a manner that allows informed consent as required.

**Table 1. Summary of researches on knowledge related to performing screening tests**

| The research editors | Year of research | Number of participants | Findings |
|---|---|---|---|
| Romano-Zelekha et al. | 2012 | 768 | There is a significant correlation between getting a pamphlet about screening tests and performing the tests. |
| Ahmed et al. | 2005 | 110 | 77.4% of the participants were not aware to Thalassemia screening tests, and 85.8% of the participants were interested to get information about screening tests. |
| Karger et al. | 2008 | 1009 | Higher knowledge on genetics and health among educated, high-income population, women aged 18-44, and people who were exposed to genetics in previous conversation. |
| Rostant et al. | 2003 | 633 | The accessible knowledge does not sufficiently promote positive attitudes toward screening tests, nor is it sufficient for reasonable level of informed consent. |
| Suriadi et al. | 2004 | 200 | 26.5% of the participants knew which screening tests were performed to their baby after birth and for which diseases. |

In this context we may see the need in accessing information about hereditary screening tests to the public. The question is what is the right age and manner to make this knowledge available.

**4. When Is It Proper to Start**

The target age for guiding screening tests' performance was not widely discussed in the literature. In Israel, the first exposure to issues of diseases' genetics and heredity begins not as a discussion of hereditary diseases and screening tests, but rather as a discussion of heredity and morbidity, including the connection to inbred marriage, within biology matriculation studies (Ministry of education, 2015). This exposure does not deal with ethical dilemmas, but is accessible only to those who chose to study biology in $10^{th}$ to $12^{th}$ grades and only as something that exists and can be prevented by avoiding inbred marriage. There is no knowledge promotion of this issue except for pamphlet offered during pregnancy follow up and usually after the pregnancy has already begun—as parents' screening, not as a clarification of the newborn's condition (Ministry of Health, 2014.) There are also communities in Israel where awareness is only prior to marriage, when calling hereditary information line and having a "recommended or not recommended" answer after firs meeting in traditional match, i.e., the "generation of the righteous" project. In this situation, the future couple does not know which the tested diseases are, but only that the match is "not recommended". On one hand, this has led to a significant reduction in hereditary morbidity in the extra orthodox community in Israel, on the other, it is a promotion of a lack of knowledge about hereditary tests, since the information is given in a single

telephone call from a "hot line" rather than proper information (Leiman, 2006).

In other countries, like Cyprus, performing genetic tests is a condition to get state marriage license, and therefore is part of the compulsory information to young couples before marriage. Promoting knowledge about tests performance is part of the citizen's awareness to the state's laws and not a separate focus on the disease or its consequences (Cousens et al., 2010). In another case, in Iran, education on hereditary morbidity and the importance of screening for thalassemia, a common hereditary disease in this population, is performed twice for men in the country. The first time in high school, and the second time in young men enlisted in the army. These screening tests are mandatory by law, and while in 90% of cases it does not prevent marriage of couples at risk and does not promote termination of sick fetuses' pregnancy. Moreover, a 10% reduction in the marriage of couples in risk is also a datum that must not be ignored (Cousens et al., 2010).

In Europe screening tests' knowledge promotion is performed by brochures in marriage registration offices, public media and public's training. The result, for example, is that over 80% of Italians in Italy have heard of Thalassemia and the importance of screening tests, compared to 19% of the Italian-origin population in the United States (Armeli, Robbins, & Eunpu in Cousens et al., 2010). There is no definition of target audience's age, but every couple at marriage age can be exposed to this knowledge on their way to couples' registration.

Therefore, it may be seen that there is an agreement about the importance of advancing screening tests knowledge for couples before marriage, although there is a lack of consensus regarding the age at which the knowledge should be accessed. And although there are populations that for various reasons try to minimize the detailed knowledge in the tests, the young couples are informed about the tests' existence even before marriage.

## 5. Summary

While the World Health Organization publishes guidelines and recommendations for performing screening tests, the criteria for testing include acceptance by the target population, including informed consent. However, the existing information gaps in screening tests performance policy create a situation where even in countries where screening is mandatory, and where there is awareness to the need of informed consent, the informed consent is not achieved, or it is treated as a time-consuming nuisance. The result is two extreme cases: a group that does not even know that there are screening tests and how important they are, and a group that knows that they have to do a screening test, but does not know why, and therefore performed without informed consent. The common denominator of these two groups is the lack of knowledge about the importance of performing tests. This is despite the fact that the existence of written knowledge is known to improve the population's responsiveness to performing screening, even when the test is not mandatory. Moreover, the issue of informed consent to perform hereditary screening tests was not widely researched, and was not discussed as an ethical issue, even though the consent itself involves criteria which determines the screening tests' performance.

Due to the need for available knowledge to the general public, the question arises is how to make this knowledge accessible. There are countries in which knowledge is part of compulsory curriculum, in some it is only a content of choice with information on morbidity, but not the tests. There are countries in which knowledge is legally accessible, without explanations, and there are countries in which there is extensive publicity and public training. In any case, the prevailing consensus is that until a young couple decides to marry the information should already be available and clear.

Therefore, there is a need to examine the existing policy of performing screening tests. On one hand the question of informed consent, and on the other hand, the question of the right age the content should be made available. There is also a need in a research that will examine the question of awareness to screening tests by age, and the issue of informed consent to hereditary screening tests, emphasizing the knowledge element.

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