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Genetic Screening Guidelines: Looking Back, Looking Forward

### GENETIC SCREENING GUIDELINES

Every New Year we look to the future and dream of what is possible. ReachMD Radio is proud to present our special series Focus on Future Medicine.

The National Academy of Science has published a report on genetic screening guidelines in 1975 that is considered a classic set of recommendations. Decades later, its genetics have been incorporated into all areas of medicine; are these guidelines still pertinent and what update should be made.

You're listening to ReachMD XM160, The Channel for Medical Professionals. Welcome to The Clinicians Roundtable, I am your host Dr. Lee Freedman and joining us to discuss genetic screening guidelines with is Dr. Artemis Simopoulos, founder and president of the Center for Genetics, Nutrition and Health, Nonprofit Educational Organization in Washington DC.

## DR. LEE FREEDMAN:

Thank you for being with us, Dr. Simopoulos.

## DR. ARTEMIS SIMOPOULOS:

It is a pleasure.

## DR. LEE FREEDMAN:

In 1975.

**DR. ARTEMIS SIMOPOULOS:**

A longtime isn't it.

**DR. LEE FREEDMAN:**

Yes 3 decades ago, I wouldn't have thought that genetics would have been a thought of its needing a set of recommendations, can you tell us how these came about and what some of the principles of that classic work are.

**DR. ARTEMIS SIMOPOULOS:**

Yes, the committee was established by the National Research Counsel of the National Academy of Sciences at the request of the American Society of Human Genetics. Back in the early 70s, there was a great deal of interest in genetics because the research clearly shows that is an area where more genetic tests are going to be developed.

**DR. LEE FREEDMAN:**

Hmm, hmm.

DR. ARTEMIS SIMOPOULOS:

And will be of use to the whole population and at that time, the tests were carried out in newborn babies.

**DR. LEE FREEDMAN:**

Hmm, hmm.

DR. ARTEMIS SIMOPOULOS:

And it was felt necessary that because this is an area where that's going to expand. It is important that a committee is established to evaluate very carefully, #1 the need and the standardization of test for genetic screening and secondly what are some of the issues both in terms of the legal, ethical and scientific aspects of genetic screening that need to be carefully looked at because until then, no one had looked at the field from all these standpoints.

## **DR. LEE FREEDMAN:**

Very interesting and from what I have read this was referred by phenylketonuria testing in infants.

## **DR. ARTEMIS SIMOPOULOS:**

Yes, so the very first genetic test that was carried out in newborns was the test for the diagnosis of phenylketonuria or PKU as people referred to it because it's due to a deficiency of an enzyme due to a genetic defect and it was also known at that time that a proper diet will definitely prevent the development of the mental retardation.

## **DR. LEE FREEDMAN:**

Hmm, hmm.

## **DR. ARTEMIS SIMOPOULOS:**

PKU was the most common cause of mental retardation in children. It was therefore, very important to review and evaluate how the program was developed and how in the future additional programs could be developed that would not have some of the drawbacks of the PKU program and some of the drawbacks where that they still had not really defined carefully what sort of diet needs to be developed, how soon after that needs to be started, who was going to follow the metabolic changes, and then in many states began to develop laws making it mandatory and the laws that were developed, they used as a model, infectious disease model which of course, the committee felt and agree with them that it's not an appropriate law because genetic diseases are not contagious where infectious disease is, and at that time, people were worried very much about Syphilis and gonorrhea, that these are very different and when it comes to genetics, nothing should be mandatory, but this would be left up to the individual and the family to agree for the test to be carried out and that the test also should be standardized, should be accurate, should be reproducible.

## **DR. LEE FREEDMAN:**

The infectious disease model was not appropriate, a new model, a new-way of looking at this type of test was necessary and it is remarkable as I read it how forward thinking you were in addressing some of these things like legal issues, ethical issues.

## **DR. ARTEMIS SIMOPOULOS:**

Yes, I am glad you mentioned that because, at that time I was working at a division of Medical Sciences at the National Academy of Sciences. So, when we received the letter from the American Society of Human Genetics, we saw it was a very important issue of national importance and eventually international and that the National Academy of Sciences was very appropriate place to put that experts from this country and Canada to evaluate not only the scientific and the managerial aspect, but the legal, the ethical, the social, and the economic aspect because when we are testing people for genetic diseases, we have to bear in mind that all of us have some kind of a genetic defect, that in a special environment will manifest itself, so that it becomes ethical to give the opportunity to people to agree or disagree if their wish to be tested. Because we don't want to have anything that will interfere with their privacy or that the presence of defective gene will increase their health insurance premiums and at that time, we were very much aware of that and we emphasize the need for the development by the Federal Government or protection of individuals. So, that nothing will work against them.

## **DR. LEE FREEDMAN:**

If you're just tuning in, you're listening to The Clinicians Roundtable on ReachMD XM160, The Channel for Medical Professionals. I am your host Dr. Lee Freedman and joining me to discuss genetic screening guidelines is Dr. Artemis Simopoulos founder and president of the Center for Genetic Nutrition and Health in Washington DC.

So, Dr. Simopoulos you did 30 years ago, tried to take into account this in a very holistic way, looking at whether someone should be forced to do this, the accuracy of the test, or reproducibility, what to do with the results, several social and legal and educational ramification. How do you think these guidelines have been accepted and followed and where do we need to put more emphasis as we go forward.

## **DR. ARTEMIS SIMOPOULOS:**

Yes, first of all, I should tell you that we even carried out our own research to define how physicians felt about genetic screening, because if you're going to develop a genetic screening program, you want to have acceptance by the public, by the physician, and another health personnel and we found that the more physicians knew about genetics, the more interested were in carrying out genetic screening, therefore, the education of physicians about genetics. The education of the public about genetics were 2 of the major recommendations that we made because it is important to have knowledge. Because knowledge influences attitude towards genetics screening. So, the report was received very well and got excellent reviews in fact I will just look into find comments about the report and this particular one, actually was published in the official journal of the National Federation of Catholic Physicians' Guilds and it says that the final section of the report is synopsis of all it's major points and is available as a separate pamphlet useful in its own right. So, we

made every effort to make the report user friendly and we did develop a pamphlet and so they're going to say genetics screening is achievement of great value to anyone interested in the subject from what ever perspective, medical practice, public health policy, legal or ethical or sociological implications of health care system and research. We can simultaneously serve the practicing physician who desires deeper inside into the genetic aspects of health problem. So, it was very well accepted, but it was not really acted upon and by that I mean, you know the Federal Government did not move into setting-up some kind of an agency that would then be working with the various regions of the country rather than individual states, because of frequency of genes and the population varies in the Unites States.

## **DR. LEE FREEDMAN:**

Hmm, hmm.

### **DR. ARTEMIS SIMOPOULOS:**

So, we have recommended the establishment of regional center and a federal agency that will make sure that the tests are properly standardized that there is quality control, that are reproducible. We also recommended that in High Schools, students are taught a little bit more about cell biology and genetics. We also recommended our programs in Medical School and Schools of Public Health, in order for all of us to be ready for it.

## **DR. LEE FREEDMAN:**

Hmm, hmm.

### **DR. ARTEMIS SIMOPOULOS:**

Because, research base was very active in the 70s and we knew that a lot of data got to be coming out that need to be interpreted. What happened is, the various administrations and secretaries of health, setup additional committees and committees, none of which will evaluate in depth as did the study we carried out at the National Academy of Sciences.

### **DR. LEE FREEDMAN:**

So, we really haven't seen the government.

### **DR. ARTEMIS SIMOPOULOS:**

Were not practiced.

**DR. LEE FREEDMAN:**

And now, as I am aware many practicing doctors are, capitalism is a kind of leading the way instead of science it seems. There are of lot companies that are offering genetic screening for more common complex illnesses, not the simple single gene mutation illnesses like PKU. How do feel about that?

**DR. ARTEMIS SIMOPOULOS:**

Yes, well let me tell you. The PKU is a very good example, of course it is a single gene effect and we looked at the whole issue of common diseases like heart disease, and cancer, and diabetes, which are due to multiple genes, okay and the principles that we studied and recommended are the same whether you are dealing with a single gene defect or multiple genes in terms of a chronic diseases.

**DR. LEE FREEDMAN:**

Hmm, hmm.

**DR. ARTEMIS SIMOPOULOS:**

Still, people have to be knowledgeable. The physicians are to have more expertise. There should be quality control, the federal government should have an active role in quality control and you have to make sure that the rights of individuals are not inappropriately harmed and emphasized that genetic variation occurs in all of us and under certain circumstances there is interaction of the genes with the environment that cause disease.

**DR. LEE FREEDMAN:**

So, the same principles are

**DR. ARTEMIS SIMOPOULOS:**

The principles are the same.

**DR. LEE FREEDMAN:**

They are the same.

**DR. ARTEMIS SIMOPOULOS:**

Exactly, the same.

**DR. LEE FREEDMAN:**

What you feel in terms of the legislation that George Bush signed last year that genetic information and nondiscrimination act.

**DR. ARTEMIS SIMOPOULOS:**

Right, the GINA that was signed last May. I think, it was way overdue and I think, it's a very good beginning and it will be interesting to see how they're going to begin to implement that, because that will take quite a bit of time, you know interpreting the various aspects of the act, but this you know, could have been done 20 years ago.

**DR. LEE FREEDMAN:**

That's why; here a little bit of frustration in your voice that we still have some of the same challenges.

**DR. ARTEMIS SIMOPOULOS:**

Yes, no question about that. Because what is very important is you don't really want to harm anyone.

**DR. LEE FREEDMAN:**

Hmm, hmm.

**DR. ARTEMIS SIMOPOULOS:**

And they have gone to carry out genetic screening and let's say you are going to discover a group of genes that are associated or increased the risk of let's say heart disease. You want to be sure that the information is precisely interpreted and given to the individual and that most importantly it is the privacy of that individual is safeguarded and that you have genetic counselors, who are going to further explain the meaning of the genetic findings. Because genetic counseling is going to be very important in sustaining this individuals.

**DR. LEE FREEDMAN:**

Hmm, hmm.

**DR. ARTEMIS SIMOPOULOS:**

And then the physicians usually works with the genetic counselor in both explaining the risk and then begin to consider what sort of treatment they are going to recommend.

**DR. LEE FREEDMAN:**

And I imagine there with treatment there is the great risk economically of launching into screening or treatments that really have no scientific basis, but in this medicolegal climate, you find out to have genetic predisposition as well lets do the test, lets do the treatment, that's dangerous.

**DR. ARTEMIS SIMOPOULOS:**

Well, it's very important to have that centers or clinics in Medical Schools or other places where the tests are accurate. The information is properly explained, genetic counseling takes place, and an appropriate management is beginning to be carried out by the physician. This idea of direct to consumer genetic testing is not appropriate.

**DR. LEE FREEDMAN:**

As you looked to the future, what do you see?

**DR. ARTEMIS SIMOPOULOS:**

Oh, I'm very optimistic and I'll tell you why I'm very optimistic because I think, what happened is certain industry groups move very fast, you know 2 or 3, 4 years ago, and the fact that they moved in a direction of which is not I would say the most ethical approach to it, began to raise questions and I think that's why GINA was signed into law by the President last May because all these other things were coming up. Mostly, I think, it's very important to consider the fact, the tests should be ordered by the physician or a healthcare person, if not a physician and the test should be properly carried out. The test should be properly interpreted and whatever form of treatment or management should be carried out. Then, I think, it's important to maintain privacy. The test results should be given to the individual that was tested. This is not for other people to have knowledge. Maintaining privacy and the ethical approach to genetic screening are very important and this cannot be carried out by marketers.

**DR. LEE FREEDMAN:**

I would like to thank my guest, Dr. Artemis Simopoulos, who has been discussing with us genetic screening guidelines. It's fascinating to me that the principles that were outlined in her community's report of 1975 are still very pertinent and very important. Thank you for listening to The Clinicians Roundtable. This is ReachMD XM160, The Channel for Medical Professionals.

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