# GENETIC SCREENING – A PROMISING TOOL OF THE FUTURE

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**Abstract**

*Genetic screening or testing has evolved rapidly in the past three decades with scientists developing amazing new techniques that have revealed details about how genes work and how they are linked to diseases. This exciting spectrum of new diagnostic tests is made possible with the unraveling of the human genome. The techniques have provided convincing explanations of genetic factors responsible for certain diseases and have made possible, the genetic testing of asymptomatic individuals. The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a person's chance of developing or passing on a genetic disorder. Hundreds of genetic tests are currently available, and more are being developed. Effort has been made to discuss the various applications of genetic screening as a promising tool in medicine.*

***Keywords**: Genetic screening, biotechnology, disease.*

## INTRODUCTION

In medicine, modern biotechnology finds promising applications in areas such as drug production, pharmacogenomics, gene therapy and genetic screening (or genetic testing). Some diseases in humans have their roots in our genes. The efficiency of an individual's body system to process food, detoxify poisons, and respond to infections is determined by the individual's genes, through the proteins they encode.

The concept of genetic screening will be better understood with the background knowledge of its crucial factors. The foundation of genetic screening is based on the knowledge and function of Deoxyribonucleic acid (DNA). Thus, DNA is the only crucial requirement for genetic screening. In most organisms, including humans, DNA serves as the molecule storing genetic information (Klug & Cummings, 1997). It is usually a double–stranded molecule organized as a double helix. Contained within each DNA are hereditary units called genes, which are part of a larger element, the chromosome. Although there are some exceptions, members of most species have a specific number of chromosomes present in each somatic cell called the diploid number (2n). In man, the number is forty–six (46) (i.e. 2n) that form twenty three pairs (i.e. n) (Klug & Cummings, 1997).

Interactions of genes control the physical and structural traits or characteristics of organisms that are expressed differently. Scientists have already mapped the complete human chromosomes (i.e. the Genome Project) (Thieman & Palladino, 2008). Thus, the information from the sequencing of the human chromosome can be compared and contrasted with the individual that possesses a certain trait. This helps geneticists to determine which gene or set of genes control a trait, after which a genetic test could be developed to compare the sequences of an individual and determine if the individual is predetermined to develop a trait, disease or disorder.

Genetic screening has raised many questions such as who should be treated, when should an individual be treated, who should perform such tests, who should know about the results, who should counsel patients about their genetic status, should law be passed to protect people against genetic discrimination by private entities, how can genetic profile be kept confidential and how should people react to the knowledge of their particular genetic makeup? These and other questions play a role in how public opinion is formed and constitute some of the challenges of genetic screening. In the past three decades, scientists have developed amazing new techniques that have revealed details about how genes work and how they are linked to disease (Althoff, 1999). These techniques have provided convincing explanation of genetic factors responsible for certain diseases and have made possible, the genetic testing of asymptomatic individuals and thus, predict their risk of certain diseases. Predisposition for certain diseases as well as pathogen infections can be detected using very sensitive techniques.

The results of a genetic test can confirm or rule out a suspected genetic condition or help determine a

person's chance of developing or passing on a genetic disorder. Hundreds of genetic tests are currently available, and more are being developed.
In this paper, we are going to discuss the following areas:

- The definition of genetic screening
- Screening techniques
- Direct–to–Consumer (DTC) genetic testing
- Applications of genetic screening (testing)
- Medical procedure
- Challenges of genetic screening
- Our opinion

**Definition of Genetic Screening**
Genetic screening or testing is "the analysis of chromosomes (DNA), proteins, and certain metabolites in order to detect heritable disease–related genotypes, mutations, phenotypes, or karyotypes for clinical purposes" (Jorge & Barbara, 2008). It can provide information about a person's genes and chromosomes throughout life. A scientist scans a patient's DNA sample for mutated sequences. For example, to test a developing fetus for Down syndrome, Amniocentesis and chorionic villus, sampling can be used (Thieman & Palladino, 2008). Genetic testing identifies changes in chromosomes, genes, or proteins. Most of the time, testing is used to find changes that are associated with inherited disorders.

**Screening Techniques**
Molecular genetics methods are one of the most promising tools for diagnosis purposes in the near future. There are basically two major types of gene tests. In the first type, a researcher may design short pieces of DNA ("probes") whose sequences are complementing to the mutated sequences. These probes will seek their complement among the base pairs of an individual's genome. If the mutated sequence is present in the patient's genome, the probe will bind to it and flag the mutation. In the second type, a researcher may conduct the gene test by comparing the sequence of DNA bases in a patient's gene to disease in healthy individuals of their offspring.

Genetic testing techniques can also be grouped into two main categories: screening and scanning. Screening methods involve probing specific genes for previously identified mutations (Eng & Jan, 1997). These methods are however, unrealistic for such diseases which are characterized by a large number of disease–causing mutations that can be virtually anywhere in the genes coding region or regulatory region. For example, in breast cancer, mutations can be found anywhere within the large BRCAI and BRCAZ genes, two of the many genes that have been implicated in breast cancer. Development of a separate DNA probe for each mutation has proven to be too expensive and time consuming to make screening methods feasible. Scanning methods involve testing the gene or genes having no assumptions about any mutations (Eng & Jan, 1997). Another amazing scanning technique is the use of DNA chips. This was initially developed to enhance genomic sequencing projects, especially the Human Genome Project (Althoff, 1999). DNA chips are finding applications throughout the field of molecular biology. Gene scanning techniques that are based on oligonucleofide arrays called DNA chips, provide a rapid method to analyze thousands of genes simultaneously. DNA chips are thus potentially very powerful tools for gaining insights into the complexity of gene expression, detecting genetic variations, making new gene discoveries, fingerprinting and developing new diagnostic tools (Althoff, 1999).

About a decade ago, specific chips were available for as little as $100, but cost thousands of dollars, once custom–made chips were available (Singh & Sangeet, 1999). Presently, chips have been designed using the computer, instead of doing it manually, by hand. These have greatly sped up the process allowing companies to make custom chips in one day, as opposed to months, which has lowered the cost of production. Consequently, DNA chips are cheap, providing access to scientists regardless of their funding situation. For instance, IMMUNIQ, a company in Poland has test kits that can be acquired for research and routine. Among their new products is a complex diagnostic panel (chip based) with sufficient MutaCHIP$^{(R)}$ detection system. It is a combination of PCR with hybridization – ELISA and subsequent software in a small self–sufficient reader system (simple, cost–effective, comfortable) and reported with therapy recommendation for the doctor.

**Direct –to–consumer (DTC) Genetic testing**
This is a type of genetic test that is accessible directly to the consumer without having to go through a health care professional. Usually, to obtain a genetic test, health care professionals such as doctors require the permission of the patient and order the desired test. DTC genetic tests, however, allow consumers to bypass this process and order one themselves. Some pharmaceutical companies deliver the test kits unrestricted only in patent free countries, with reservation to proof of individual case. However, in some countries that are not patent free, to purchase a PCR and certain diagnostic reagents, the user has to pay attention to the respective national patent situation concerning PCR instrument and belonging analysis methods. There are a variety of DTC tests, ranging from testing for breast cancer alleles to mutations linked to cystic fibrosis. Benefits of DTC testing are the accessibility of tests to consumers, promotion of proactive healthcare and the privacy of genetic information.

**Applications of Genetic Screening (Testing)**

Genetic screening is currently available for the following:

i. Neonatal Genetic screening: This is also referred to as new born screening. This type of screening is done just after birth to identify genetic disorders that can be treated early in life. For instance, inborn errors of metabolism are test run on all infants in some developed countries like the USA and UK to determine rare disabilities or chemical disorders like phenylketonuria (a genetic disorder that causes mental illness if left untreated), congenital hypothyroidism (a disorder of the thyroid gland), sickle cell disease, HIV and cystic fibrosis.
ii. Carrier screening: The identification of unaffected individuals who carry one copy of a gene for a disease that requires two copies for the disease to manifest.
iii. Forensic/identity testing: This type of screening uses DNA sequences to identify an individual for legal purposes. Examples include detection of crime or catastrophe victims, rule out or implicate a crime suspect, or establish biological relationship between individuals such as paternity.
iv. Prenatal diagnostic screening: Prenatal genetic screening is used to detect changes in a fetus' genes or chromosomes before birth. This type of screening is usually offered to couples with an increased risk of having a baby with a genetic or chromosomal disorder. However, not all possible inherited disorders or birth defects can be detected. In some cases, such information helps some parents to decide whether to abort the pregnancy.

Blatt (1996) reported the following examples of genetic screening:

**Maternal serum alpha–fetoprotein (MSAFP):** This is a common blood–screening test that is performed early in pregnancy to determine the chance of fetus carrying neural tube conditions.

**Enhanced MSAP:** This is a genetic screening test that measures biologic markers to determine the possibility of fetal Down syndrome.

**Amniocentesis:** This is a surgical tap into the uterus to obtain amniotic fluid. This fluid can be analyzed to determine genetic status of fetus.

**Pre–implantation genetic diagnosis:** This is used to detect whether or not gametes contain genes for a specific disease prior to determine the status of gametes and the gamete that will be most suitable to be used for fertilization.

- Pharmacogenomics: This type of genetic testing determines the influence of genetic variation on drug response.
- Detection of 'hard to find' genes found in RNA species using the DNA chips.
- Predictive and presymptomatic screening: These types of screening are used to detect gene mutations associated with disorders that appear after birth, often later in life.

Preductive screening can identify mutations that increase an individual's chances of developing of disorder with genetic basis e.g. an individual with a mutation in BRCAI has a 65 % cumulative risk of breast cancer. Presymptomatic testing can determing whether an individual will develop a genetic disorder, e.g. adult onset disorders such as Huntington's disease, hemochromatosis (an iron overload disorder), prior to appearance of any symptom or sign.

- Genealogical DNA test (for genetic genealogy purposes): Genetic screening can be used to determined a child maternity (genetic father) of an individual ancestry.
- Detection of more complex conditions such as breast, ovarian, and colon cancers.

**Medical Procedure**

The usual way of genetic tests involves the following:

**Consultation:** Once an individual decides to proceed with genetic testing, a medical geneticist, genetic counselor, primary care doctor or specialist can order the test after obtaining informed consent.

**Test samples:** The tests are usually performed on a blood sample, skin, hair, amniotic fluid (the fluid that surrounds a fetus during pregnancy), or other tissues.

**Actual screening/testing:** After sample collection, the sample is sent to the laboratory where technicians look for specific changes in chromosomes, DNA or protein, depending on the suspected disorder.

**Test results:** The laboratory reports the test results in writing to an individual's doctor or genetic counselor.

**Interpretation of results:** This is usually done by the individual's doctor, genetic counselor or other relevant healthcare professionals, who consider the individual's medical history and family history in the course of interpreting the test results. The results may be positive or negative. In some cases, depending on the purpose of the test, there could be need for further testing.

**Challenges of Genetic Screening**

In spite of the recent advances in genetic screening or testing, some of the following challenges exist:

- Genetic screening may not detect every mutation associated with a particular condition because many are as yet undiscovered, and the ones detected may present different risks to different people and populations (Robin, 1991).
- Ethical considerations – Ethical issue, like the decision to have a genetic test deserves careful preparation and thought.
- Clinical issues – These center on the capabilities and limitations of doctors and other health service providers, people identified with genetic condition, and the general public in dealing with

genetic information. For instance, the results of genetic tests are not always straight forward, which makes them difficult to interpret and explain.

- Public policy – Genetic discrimination is perhaps one of the most critical issues involved in genetic screening. The absence of privacy and anti–discrimination legal protections in most countries can lead to discrimination in employment or insurance. However, in the US, the legislation called the Genetic Information Nondiscrimination Act prohibits group health plans and health insurance from denying coverage to a healthy individual or charging that individual higher premiums based solely on a genetic predisposition to developing a disease in future. The legislation also bars employers from using individuals' genetic information when making hiring, firing, job placement or promotion decisions. The legislation was passed into law by President George W. Bush on May 21, 2008, but went into effect on November 21, 2009 (Keim, 2008).
- Effects on social institutions – Genetic tests reveal information about individuals and their families. Therefore, test results can affect the dynamics within social institutions, particularly the family.
- Some of the risks of DTC testing are the lack of governmental regulation and the potential misinterpretation of genetic information. DTC genetic testing has been controversial due to outspoken opposition within the scientific community. Critics of DTC testing argue against the risks involved, the unregulated advertising and marketing claims, and the overall lack of governmental oversight.

**Our Opinion**

In our opinion, genetic screening is a noble, technical advancement in sciences, especially with respect to biotechnology. However, before genetic tests become publicly available, specialists and society at large need to come to terms with major technical, ethical and economic concerns, through carefully conducted research programs and genetic counseling. Since genetic testing may open up ethical or psychological problems, genetic testing should be accompanied by genetic counseling. This would greatly reduce the mental and physical strains that would be put on an individual and members of his or her family. Therefore, with proper regulation and genetic counseling, genetic screening offers one of the most promising tools for meeting future demand for medical and pharmaceutical products to cope with genetic related diseases in the increasing global population.

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