




DtCOGt

Direct-to-consumer Genetic Tests



A variety of services and tests related to genetics and genomics are recently made available in the market. A lot of these are direct-to-consumer genetic tests (DTCGTs), which have raised public concern worldwide.

This user guide aims at informing the public about the potential risks associated with DTCGTs, and the precautions that should be taken.



## WHAT ARE GENETIC DISEASES ?

Genetic diseases refer to disorders caused by mutations in an individual's genetic material. Genetic testing is generally used for screening or diagnostic purposes, with the aim to rule out, confirm or predict the development of particular genetic disorders.

## WHAT ARE DIRECT-TO-CONSUMER GENETIC TESTS ?

DTCGTs are genetic testing products that consumers can purchase directly over the counter or online. The entire process from publicity, marketing to delivery is done directly by the service provider to the consumers without involving healthcare professionals.

From a medical point of view, the public should first consult healthcare professionals and obtain their assessment and opinion before deciding whether they should undertake any diagnostic or therapeutic investigation (including genetic testing).

## Are you interested in DTCGTs?

There are two main types of DTCGTs, namely **health-related** genetic tests and **non-health related** genetic tests.

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### Health-related genetic tests

- Diagnostic test for heritable diseases (e.g. cystic fibrosis, spinal muscular atrophy (SMA), etc.)
- Genetic susceptibility/predisposition testing for common and complex diseases (e.g. cancer, cardiovascular diseases, and diabetes, etc.)
- Carrier testing for genetic diseases inherited in X-linked recessive (e.g. haemophilia) and autosomal recessive manner (e.g. Thalassaemia)
- Pharmacogenomic testing for guiding choice of medications and dosage

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### Non-health related genetic tests

- Ancestry
- Talents
- Athletic performance
- Fun traits (e.g. eye colour)
- Nutritional needs

## Are you considering a DTCGT product?

### WHAT SHOULD I BE MINDFUL OF ?

Before purchasing a DTCGT product or service, consumers should have a clear purpose, and understand the scope covered by the test, which can include:

- Diagnostic or predictive testing for a specific genetic disorder
- Determination of genetic susceptibility or predisposition to common yet complex diseases (cancer, cardiovascular diseases, and diabetes, etc.)
- Testing for non-health related traits (e.g. athletic performance, talents, etc.)

### WHAT ARE THE USUAL PROCEDURES ?

Different service providers may have different procedures in their service delivery. Consumers should ask the service provider to obtain a clear understanding of the testing arrangement details, including:

- Whether the test product can be obtained over the counter or online
- Detailed instructions on the self-collection of sample (e.g. hair and saliva samples)
- How the sample is sent to the laboratory that performs the test (e.g. by courier)
- How the test results are returned to the consumer (e.g. through mobile app, email, or service provider's website)






## Do you know the possible impact of DTCGT result on you?

### ARE DTCGTs ACCURATE?

DTCGTs have varying degrees of credibility; there is sufficient scientific and clinical evidence available for some genetic tests but not for others. Therefore, different types of genetic tests have variable levels of accuracy, uncertainty and clinical utility.

**Seeking healthcare professionals' opinions can assist in determining the clinical utility of the DTCGT results.**



### WOULD I GET THE SAME RESULT IF I PURCHASE DTCGTs FROM DIFFERENT SERVICE PROVIDERS?

For the same disease, tests provided by different service providers may differ in content, hence produce different test results.

### CAN I USE THE DTCGT RESULTS TO UNDERSTAND MY DISEASE RISKS AND THEREBY TAKE APPROPRIATE PRECAUTIONS?



For most genetic diseases, finding a genetic diagnosis does not necessarily mean there is a cure for the disease. Nevertheless, a genetic diagnosis may help doctors in drawing up a more suitable management plan for patients.

For health-related genetic tests that are predictive in nature, asymptomatic individuals may or may not be able to find preventive measures based on such test results.

## Now you have received the health-related genetic test result. What if.....

### WHAT ACTION SHOULD I TAKE IF THE RESULT IS POSITIVE ?

If the genetic test is done to diagnose a particular genetic disease, a positive result usually, though not always, means confirmation of a particular genetic disease, or prediction of its development in the future. However, a lot of times the test result may not be able to predict disease severity and prognosis. When a test result is positive, the test subject should seek opinion from a medical specialist relevant to the particular disease at his/her earliest convenience.

The interpretation of genetic susceptibility testing for common and complex diseases (e.g. cancer, cardiovascular diseases, and diabetes, etc.) has greater uncertainty. The accuracy in predicting the development of such diseases, either positive or negative, is usually low. Therefore, the clinical utility of such genetic susceptibility testing is also low. The test subject needs not overly worry. He/she may adopt suitable lifestyle modifications to mitigate the risk of the relevant disease. When appropriate, he/she can inform his/her family doctor or other medical specialists, so that appropriate surveillance for the particular disease can be introduced.

### AM I EXEMPTED FROM DEVELOPING A PARTICULAR GENETIC DISEASE IF MY RESULT IS NEGATIVE ?

A negative test result indicates that the test subject is unlikely to suffer from the genetic disease in question, but it does not entirely exclude such possibility.

Uncertainty exists in all results, be it positive or negative, and its magnitude depends on various factors, such as the complexity of the genetic disease, the testing technology adopted, the stringency of the testing procedures and so on.

**If genetic testing is accompanied by counselling and explanation on the results , you can gain a better understanding of the result and its impact.**

## HOW SHOULD I INTERPRET THE RESULT OF A NON-HEALTH RELATED GENETIC TEST ?

Non-health related genetic tests target various traits including athletic performance, talents, etc. Anyone receiving such genetic tests needs to be mindful that these traits are the result of complex intrinsic and environmental factors. Therefore the accuracy of these tests are generally low. The test subject should be aware of the limited utility of such tests.

## AS A CONSUMER, HOW SHOULD I TREAT THE DTCGT RESULTS ?

The results of genetic testing are usually more complex and more difficult to understand than those of other medical testing. To comprehend the results of genetic testing, other information such as personal medical history, family history and reasons for taking the test need to be taken into consideration as well.

Misunderstanding of the genetic test results could create unnecessary anxiety and psychological burden to the individual and his/her relatives, and in extreme cases, lead to ill-informed medical decisions based on the test results.

**It is important to seek advice from healthcare professionals both before and after undertaking a genetic test.**





## Have you realised DTCGT might possibly bring about.....

### PRIVACY ISSUE ?

Genetic data is as unique as fingerprints, and can potentially be traceable to the individual being tested.

Compared to fingerprints, genetic data also reveal information related to personal traits and health conditions.

**Consumers should be mindful of how the service provider protects the privacy of their customers. Details regarding how the company stores, uses, safeguards and shares the data should also be clarified.**

### IMPACT ON YOUR DAILY LIVES ?

Consumers should beware of the possibility of genetic discrimination, which refers to the differential or unfair treatment by one's employer or insurance company because he/she is found to carry a gene mutation that causes or predisposes to an inherited disorder.



# CONCLUSION

From a medical point of view, you are recommended to consult healthcare professionals before undertaking any medical test (including genetic test).

Healthcare professionals may arrange suitable tests based on the patient's health condition and clinical assessment, and provide follow-up or treatment as needed. Appropriate genetic counselling can help patients or their relatives to understand the genetic disease in question, e.g. the inheritance pattern and recurrence risk, and provides advice on issues regarding genetic tests.

