When it comes to genetic diseases, family history is a significant risk factor that one may not have total control over. Although genes can determine similarities in appearance, they may also lead to a risk in the family for developing certain health conditions.

We speak to doctors and counsellors from Raffles Hospital to learn more about some of the common genetic diseases that may run in families, and to empower you with knowledge on how you can prevent illness by being aware of your family health history and by making healthy choices.

Like it or not, each and every one of us belongs to a family. You have a mother and a father, grandparents, and perhaps children of your own. You might have gotten your round eyes from your mother and your prominent nose from your grandfather. You may not think of them, but you also have hundreds of ancestors – people you have never met – whose genes you carry and may pass down to descendants in years to come. These are not the only things you may have inherited from your family, however. Many medical conditions including heart disease, cancer and diabetes have also been shown to be passed down through families.

Your genetic make-up — that is, the genes you inherited from your parents, holds the secret to your destiny. Not only the colour of your eyes and hair and your musical or athletic abilities but also thousands of other factors related to your appearance, your health, and how you interact with the world are all determined, in whole or part, but your genetics.

Most importantly, your genes determine whether you are at risk for specific diseases. Some diseases are caused when there is a change in the instructions in a gene and this is known as a mutation. Every person has many mutations. Sometimes these changes have no effect or are even slightly helpful, but sometimes they can cause disease.

Most common diseases are caused by a combination of mutations, lifestyle choices and your environment. Even people with similar genes may or may not develop an illness if they make choices or live in a different environment. On the other hand, some rare diseases are caused by a specific change in the DNA of a single gene. These conditions usually develop when an individual is born with a mutated gene.

Did You Know?

- A person has two copies of each gene - one from the mother and one from the father.
- Genes carry instructions that tell your cells how to work and grow.
- A complete set of genes is known as a genome. Humans have approximately 20,000 genes in their genome.
- Cells are the building blocks of the body and every part of the body is made up of billions of cells working together.
- Genes are arranged in thread-like structures called chromosomes. Humans have 23 pairs of chromosomes and copies of the chromosomes are found in each cell.
- Chromosomes are made up of DNA, which is the special code in which the instructions in your genes are written.
Cancer is a genetic disease and cancerous cells arise because the genes controlling cell growth has malfunctioned. The reasons why the genes become abnormal is due to three reasons closely interacting with each other namely the ageing process, environmental factors (smoking and viral infections for example) and inherited genetic syndromes predisposing to cancers.

All of us are at risk of cancer and the risk is about one in three in one’s lifetime. Inherited cancer or familial cancers account for less than five per cent of all cancer occurrences. Depending on the type of cancer and how many family members are affected, one may opt to have genetic testing done. If it is a common cancer like colorectal cancer in a first-degree family member, without other members being affected, the risk may not be more than 1.5 to two times the general population.

Many of the cancer-causing gene mutations are known. Some directly result in cancers in specific sites such as breast and ovarian cancers (BRCA 1 and 2). Carriers of these genetic mutations have up to 80 per cent chance of developing these cancers in their lifetime. Many solid and hematological cancers have a genetic basis which may be used to clinch the diagnosis by way of genetic testing of tumors excised during surgery. These genetic changes may also guide therapy by way of utilising targeted therapy.

Modification or avoidance of the environmental factors that result in the cancer-related genetic changes will lower risk. The main external factor that causes cancer is smoking. It is estimated that if everyone is to stop smoking immediately, the cancer incidence across all types will drop to less than 50 per cent of current incidence within one generation.

Regular cancer screening for breast, colorectal and cervix cancer do help prevent late stage disease from shortening life prematurely and human papillomavirus vaccines do help to prevent onset of cancer of the cervix in females. Prophylactic removal of organs such as mastectomy and ovariectomy in those who are detected to have the cancer genes are controversial; these measures do not completely eliminate cancers from forming.
MENTAL ILLNESS

The prevalence of mental illness among psychological dysfunctional families and those with genetic predisposition is expected to be higher than the general population. Typically, it is noted that families with history of mental illness and of lower socio-economic status are more at risk. For most, the chance of developing schizophrenia or bipolar disorder is only around one per cent. However, for families with a close relative such as a parent or sibling suffering from mental disorder, the average risk rises to about 10 per cent.

The most common view is that mental disorders tend to result from genetic dispositions, environmental stressors and individual’s lack of stress coping mechanism or adequate support. Family-linkage and twin studies have indicated that genetic factors often play an important role in the development of mental disorders.

Genes associated with mental disorders do not always show the same degree of penetrance, which is defined as the frequency with which a gene produces its effects in a specific group of people. For example, a gene for manic depression may have 20 per cent penetrance, which means that 20 per cent of the members of the family being studied are at risk of developing the disorder.

A good understanding of one’s family-of-origin’s mental health history is crucial as it can highlight any biological predisposition due to possible shared genes. One should review his or her lifestyle to ensure balance and build up his or her mental resilience through psycho-education like mental health awareness talks and counselling. It is important for one to develop positive coping skills to handle stressful life events and have good social and emotional support from loved ones and friends.

DIABETES & OBESITY

Diabetes

Diabetes is a chronic disease and those with genetic predisposition may not necessarily develop the condition. Environmental factors such as obesity, lack of physical activity, and unhealthy diet are important in facilitating the development of diabetes. Having a first-degree family history of type 2 diabetes (non-insulin dependent) may increase the chance of developing diabetes by about two to three folds compared to the general population.

Those who are obese, lack exercise, have unhealthy diet and have family members with diabetes are at risk of contracting the disease. While genetic risks cannot be lowered, environmental factors such as obesity, sedentary lifestyle and poor diet can be improved. If a person is obese, he should reduce weight. Regular exercise and adhering to a low sugar and low fat diet can be useful in preventing type 2 diabetes. Therefore, it is even more important that the person should adopt healthy lifestyle at an early age.

There are many genes found to be linked to type 2 diabetes. However, diabetes is a multi-factorial and polygenic disease. In other words, many genes and their interactions with the environment are responsible in the development of diabetes. Therefore, it is still premature to contemplate the use of genetic testing at present.

Obesity

Obesity is a complex condition and the interaction of genetic and environmental factors is currently believed to be the basis for most cases of obesity. Environmental factors include an unhealthy diet and a sedentary lifestyle. The genetic role of obesity is less well understood. It is very difficult to conclude whether it is genetics or the environment, which are conferring the risks within a family. This is because family members share not just genes, but often also share the same environment.

The risk is obviously greater, but the exact risk is greatly determined by the environmental factors. While we cannot change our genes, we can change our environment. Start adopting an active lifestyle by exercising frequently. Eat well by having a low fat, low sugar and high fibre diet.

It is better to know the genetic predisposition so that we can start implementing a healthier lifestyle early. Similar to other chronic diseases, there are many genes linked to obesity. However, these genes and their interactions with the environment are very complex.

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HEART DISEASE

Virtually all heart disease result from susceptibility genes, defective single genes, or genes interacting with various environmental factors. One may have been born with a genetic defect, also referred to as a congenital heart defect, that led him or her to develop genetic heart disease or one may have been predisposed to developing this condition due to family history. For instance, if one’s parents developed coronary artery disease at a young age, one has a much higher risk of developing genetic heart disease at a younger age, such as younger than 65 for females and younger than 55 for males.

Two of the most common genetic disorders of the heart are coronary artery disease (CAD) and high blood pressure (hypertension). In CAD, the arteries that supply blood to the heart muscle can get hard and narrow due to a build-up or plaque cholesterol on the inner walls. As the heart gets less blood, less oxygen is delivered to the heart muscles and one can develop a heart attack. On the other hand, high blood pressure is a measure of how hard one’s heart is working to push blood through the arteries. Over time, high blood pressure can cause kidney failure, heart attacks, strokes and other health problems.

CAD is caused by a combination of genetic background, lifestyle choices and your environment. Since one’s genetic background cannot be changed, some people need additional medical assistance such as medication to lower their risk of having a heart attack. Likewise, a family history of high blood pressure increases one’s risk for developing it at a younger age. Greater risk comes with increasing age, being overweight or having a family history of hypertension.

To lower risk, one should ensure that he or she has a healthy diet. Get active and exercise regularly as obesity increases one’s risk. Exercise such as walking or jogging can strengthen the heart and blood circulation throughout the body. Stop smoking and avoid stress because when a person is stressed, the body will release the hormone cortisol, which can cause the blood vessels to become stiff.

Know Your Family Health History

Although genes are beyond your control, other things – such as what you eat, whether you smoke or exercise and what you do for a living – can be influenced by the choices you make. If you discover that you are at increased risk for a disease, there are actions you can take to decrease the likelihood of getting the disease or to limit its impact on you if it should ever manifest.

Looking at your family history may offer a somewhat more accurate predisposition for particular diseases, because members of your immediate family do share some of your genes, and they often share similar habits and lifestyle. It will help you to identify risks due to shared genes, understand better what lifestyle and environmental factors you share with your family, understand how healthy lifestyle choices can reduce your risk of developing a disease, talk to your family about your health and share your family health history with your healthcare provider.

Like in the case of cancer, Dr Poon explains: “One cannot choose one’s parents. So knowing family health history can guide one to go for earlier cancer screening and possibly gene mutation testing but these will have to be carefully discussed with an oncologist. We cannot change the genetic disorder and mutations but knowing what cancers the individual is at risk for helps to direct screening and preventive measures with careful counselling and discussion.”

Accepting the fact that genetics determine much of who you are is the first step toward outsmarting your genes and conquering disease. The more information you gather about your family health history, the more informed you will be about your medical heritage. Remember, what you learn could literally save your life!

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