Know your family medical history: your health could depend on it, and so could the well-being of your children, doctors say

- Most people have a parent or grandparent who suffered from a chronic disease – cancer, heart disease or diabetes; this could make it more likely you will too
- Your doctor can check for signs of them. If there is a rare genetic disease in your family, a test can show if you carry it and could pass it to your children

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When you visit a doctor, you usually answer questions about your extended family's health. What conditions do your parents suffer from? Did your grandparents have chronic illnesses?

The answers are helpful in safeguarding your own health – and that of your family members.

Most people have a family health history of at least one chronic disease, such as cancer, heart disease, or diabetes. My maternal grandmother suffered from macular degeneration, which causes retinal damage and vision loss, so I know I need to be mindful of this. My paternal grandmother suffered from high blood pressure – so I monitor my own.

I cannot change my genetic make-up, but knowing these two facts about two women whose DNA I share is important.

Often, good lifestyle choices can reduce your vulnerability to the diseases other family members have – and often not just because you share a genetic map; you may share lifestyle choices and habits, and even environment.

Regular screening for the same illnesses they have can be a powerful adjunct to this – for example, blood sugar testing if there's a susceptibility to diabetes, or mammograms to screen for breast cancer.

A family history can provide information about the risk of rarer conditions caused by variants (mutations) in a single gene, such as cystic fibrosis and sickle cell disease.

"It provides important information about the pattern of transmission of a heritable disease and the risk of having that condition," says Dr Ivan Chow, a Hong Kong-based specialist in family medicine experienced in chronic-disease management.

"Family history can be used as a diagnostic tool and help guide decisions about genetic testing for the patient and at-risk family members. In addition, a family history can even help to exclude genetic diseases."

There are more than 6,000 known genetic disorders, and around 1 in 50 people are affected by a known single-gene disorder, and 1 in 263 by a chromosomal disorder.

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Chow lists the most common:

- congenital hypothyroidism;
- glucose-6-phosphate dehydrogenase, or G6PD, deficiency – a genetic disorder that mostly affects males and which compromises the way red blood cells work;
- inherited metabolic disorders such as familial hypercholesterolaemia which results in a significantly elevated level of low-density lipoprotein (LDL) cholesterol or "bad cholesterol", and predisposes sufferers to an increased risk of early onset of coronary artery disease;
- Wilson's disease, in which the body doesn't eliminate copper so it accumulates to life-threatening levels, manageable with medication;
- inherited blood disorder thalassaemia that causes a person to have less haemoglobin than normal; and
- cystic fibrosis causes many bodily secretions – like mucus and digestive juices – to become thick and sticky and can severely damage the lungs and other systems.

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Arthur Bozikas, who was born with thalassaemia in the 1960s when screening and treatment were far more rudimentary than they are today, published Iron Boy, a book about living with the condition. It describes a life of "over 8,600 needle sticks, 700 blood transfusions, and 2,200 blood packs".

He was diagnosed at one, had his spleen removed by the time he was four and at 20 never imagined living into old age. At 21 though, a new treatment was developed. Bozikas is 60 today.

The diagnosis – and treatment or management of heritable disorders – can begin much earlier than it did in Bozikas' case now, says Chow, as a result of newborn screening. All babies born in public hospitals in Hong Kong are tested for congenital hypothyroidism and G6PD deficiency, using their umbilical cord blood.

Genetic testing – or premarital screening – is also widely available. This helps couples learn if they carry a gene for genetic diseases and is typically offered to those with a family history of a genetic disorder – which is why it's important to know your own history – and to people in ethnic groups with an increased risk of specific genetic conditions.

Thalassaemia – Bozikas' diagnosis – for example, affects people of Mediterranean, Asian or African descent most often. Bozikas is of Greek descent. If both parents are tested, the test can provide information about a couple's risk of having a child with a genetic condition. Today, to assess the risk of having a baby with thalassaemia, says Chow, parents' haemoglobin patterns can be assessed before conception.

Genetic testing isn't routinely done, explains Chow – but a doctor may advise a person to have it if they have a family history of a genetic disorder.

People in Hong Kong, says Chow, generally aren't aware of heritable diseases simply because many are not that common – for example, congenital hypothyroidism occurs in 1 in 2,000 to 1 in 4,000 newborns.

G6PD deficiency is much more common and is believed to affect more than 400 million people worldwide – it affects about 10 to 14 out of every 100 African-American men, for example – though most people remain asymptomatic.

Genetic disorders vary widely in the ratio of people affected: familial hypercholesterolaemia is thought to present in about 1 in 300 people, cystic fibrosis in about 1 in 3,000; Wilson's disease affects 1 in 30,000. Thalassaemia presents in 5 per cent of the global population.

None of these rarer genetic disorders is present in my family. So for now, I'll keep up those eye tests and keep off the salt.

https://www.scmp.com/lifestyle/health-wellness/article/3169245/know-your-family-medical-history-your-health-could-depend