- Heart—Diseases

These heart conditions are all in the family Genetic heart disease can live on for generations, here's how you and the clan can manage them

Manila Bulletin · 22 Mar 2022 · B-8

When it comes to maintaining a healthy heart, we all know the drill by now: Exercise regularly. Eat a healthy diet. Quit smoking. Drink moderately. Avoid stress.

Yet there are certain heart conditions that do not necessarily respond to tweaks in lifestyle habits. "Genetic heart disease is an umbrella term that covers a wide range of heart conditions passed on from parent to child," says Dr. Raul Lapitan, a cardiologist of the Section of Cardiology at Makati Medical Center (MakatiMed). "Genetic heart disease is caused by a mutation or a faulty gene. The parent with the faulty gene has a 50 percent chance of passing it onto their children."

Early diagnosis is important as it prevents the condition from becoming more serious, even life-threatening. "Unfortunately, because some of these heart conditions do not have any symptoms, or they can go undetected for years until it's too late," he explains.

Learning about your family's heart health history is a start. Supplement that with knowing the symptoms and treatments of these three genetic heart diseases.

Hypertrophic cardiomyopathy (HCM) "is the abnormal thickening of the heart muscle," says Dr. Lapitan. "This makes it harder for the heart to pump blood."

People with this condition usually exhibit shortness of breath, chest pain, lightheadedness, dizziness, or fainting spells. "Or they may have no symptoms at all," he points out. "This means early recognition, accurate diagnosis, and proper treatment are crucial. Without these, a patient becomes vulnerable to heart failure, arrhythmias, and even at risk of sudden death."

Genetic testing, while recommended, doesn't always yield a definitive diagnosis. This prompts doctors to rely on other means to detect HCM. "We typically use an echocardiogram to diagnose HCM," explains Dr. Lapitan "This diagnostic procedure uses sound waves to measure the thickness of your heart muscle and evaluates if your heart's chambers and valves are pumping blood efficiently.

"An electrocardiogram (ECG or EKG) detects abnormal heart rhythms and the possibility of heart enlargement through electrodes attached to your chest. We also use a cardiac MRI, which produces images of your heart that allows us to study the heart

muscle and how well your heart and its valves work."

Treatment, according to Dr. Lapitan, depends on your symptoms. "There are medicines to address your heart rhythms or prevent blood clots. Open-heart surgery (Septal myectomy) to remove part of the thickened heart muscle to reduce the obstruction is an invasive option when medicines do not improve your condition."

Familial Dilated Cardiomyopathy (DCM) is another genetic form of heart disease and happens when the left ventricle of the heart—the heart's main pumping chamber—is dilated, and thinned out. "This causes the other chambers to stretch as they hold more blood to pump through the body. Eventually, the heart muscle walls weaken and are not able to pump as efficiently," Dr. Lapitan explains. "This leads to fluid retention that pool in the legs, ankles, feet, lungs, and other organs—or what's known as congestive heart failure."

Apart from the signs of congestive heart failure, the genetic disorder manifests itself in the following symptoms: shortness of breath, fatigue, weight gain due to fluid retention, palpitations, dizziness, and sometimes fainting spells.

DCM is diagnosed by checking a patient's family history and through blood tests, chest X-ray, ECG, echocardiogram, CT scan, MRI, and the like. Medication to improve heart-pump function and remove excess fluid to reduce swelling in the feet and legs are prescribed.

Lifestyle changes can help ease symptoms too. "We recommend a low-salt diet with fluid restriction to alleviate shortness of breath, fatigue, and fluid retention," says Dr. Lapitan. "Depending on your DCM, we encourage engaging in non-competitive, low impact aerobic exercise."

Familial Hypercholesterolemia "is a metabolic disorder due to a genetic defect in your Chromosome 19 wherein you have an unusually high level of low-density lipoprotein cholesterol (LDL or bad cholesterol) in your blood, due to a defect in your chromosome 19," Dr. Lapitan says. "This makes you develop premature atherosclerosis (narrowing of the arteries due to plaque) early and puts you at risk for a heart attack even when you are young."

Besides the high level of LDL (at least 190 mg/dl) in your blood, the genetic disorder has visible symptoms. "Cholesterol deposits form bumps around the eyes, knuckles, elbows,

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knees, and swollen or painful Achilles tendon," he shares. "Indications of high cholesterol also appear in the eye itself, as a white or gray ring around the iris."

Pills like statins (medicines that block a cholesterol-producing substance in the liver) and Ezetimibe (a drug that limits the absorption of cholesterol from the food you eat) and injectable proprotein convertase subtilisin/Kexin type 9 serine protease (PCSK9) inhibitors are prescribed to patients with this condition. "For extreme cases, a procedure that filters excess cholesterol from a patient's blood or a liver transplant are recommended," says Dr. Lapitan.

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