# Understanding

# *Gene Therapy in Cardiovascular Disease*

## A DISCUSSION GUIDE FOR PATIENTS AND CAREGIVERS

### WHAT IS GENE THERAPY?

**Gene therapy is a type of biological medicine that works at the level of your genes – the instructions inside your cells that tell your body how to grow and function. It can also act at a cellular and protein level.**

Sometimes, a gene may not work as it should. This can lead to health problems, including cardiovascular disease.

Gene therapy aims to:

**CORRECT A FAULTY (DEFECTIVE) GENE OR PROTEIN SO IT WORKS PROPERLY**

**REPLACE A MISSING OR FAULTY GENE WITH A HEALTHY ONE**

**ADD A HEALTHY GENE THAT CAN HELP THE BODY FIGHT OR PREVENT DISEASE**

UNLIKE SOME TREATMENTS THAT RELIEVE SYMPTOMS,

**THE GOAL OF GENE THERAPY IS TO** *correct the root cause* **OF A CONDITION.**

**IN EUROPE, GENE THERAPY IS** *being studied* **FOR SEVERAL DISEASE AREAS, INCLUDING** *cardiovascular conditions.*

### WHAT IS A GENE?

- Your body is made of building blocks called **cells.**
- Inside most cells is a substance called **DNA**, which acts like an instruction manual.
- A **gene** is a small section of DNA that gives instructions to make something your body needs, usually a **protein**.
- Proteins help the body grow, repair itself, and stay healthy.

You inherit your genes from your parents — half from each. Together, all your genes form the **genome**, which is like a complete set of instructions for how your body develops and functions.

### HOW DOES A GENE CAUSE DISEASE?

Sometimes a gene can stop working properly. This is because the DNA inside it changes (mutates) and can no longer give the body the right information.

This can happen in two ways:

- **Hereditary changes** – passed down from one or both parents. These are called **inherited genetic diseases.**
- **Acquired changes** – develop later in life, as a result of aging or environmental factors like radiation or certain chemicals. These are called **acquired genetic diseases.**

**Some genetic diseases are caused by a single faulty gene, while others involve several. They can also be caused by faulty chromosomes.**

Both inherited and acquired faulty genes can lead to disease.

## GENE THERAPY FOR CARDIOVASCULAR CONDITIONS

Gene therapy can help correct the root cause of inherited cardiovascular diseases. However, researchers are also studying therapies that could help in non-genetic heart problems.

## GENETIC CONDITIONS

- Certain types of **cardiomyopathy** (hypertrophic or dilated), that weaken the heart muscle
- **Inherited arrhythmia disorders** like long QT syndrome, which affect the heart's rhythm
- **Familial hypercholesterolemia (FH),** causing very high cholesterol levels from birth
- **Hereditary transthyretin amyloidosis** (hATTR), which can damage the heart and other organs
- **Inherited aortic disease** like Marfan syndrome, affecting the body's main artery

These conditions may lead to other serious complications such as:

- **Heart failure**
- **Dangerous heart rhythms (arrhythmias)**
- **Heart attack**
- Or even **sudden cardiac death**

## NON-GENETIC CONDITIONS

- **Heart failure**
- **Ischemic and non-ischemic heart disease**

These experimental therapies aim to:

- ♡ Promote new blood vessel growth in the heart muscle.
- ♡ Repair or replace damaged heart muscle cells.
- ♡ Improve heart function.

While these approaches are still in research stages, some are already being tested in clinical trials, and more are expected in the future. Availability will depend on ongoing research results and regulatory approval for each country.

## HOW GENETIC DISEASES ARE DIAGNOSED

**CLINICAL GENETIC TESTING:** This test looks for specific changes in your genes. It is often recommended if your doctor suspects an inherited condition. If a genetic change is found, family members may also choose to be tested to see if they carry the same faulty genes.

**GENOME MAPPING:** This is a more detailed look at your genetic code. It helps specialists find where exactly the faulty gene is and understand how it causes disease. This information can confirm a diagnosis and guide future treatments, including gene therapy.

*Genetic testing*

**CAN PROVIDE ANSWERS NOT ONLY FOR YOU, BUT ALSO FOR YOUR FAMILY.**

## TYPES OF GENE THERAPY

**Once the faulty gene has been identified, there are different approaches to treatment.**

**GENE REPLACEMENT:** A healthy new gene is created in a lab and used to replace the faulty one. The new gene is usually delivered into the body using "carriers" called vectors (which are typically modified viruses that can no longer cause disease). In some cases, your own cells can be collected, genetically modified in a lab, and then returned to your body.

**GENE EDITING:** This approach uses advanced tools that can precisely modify and repair the DNA of the faulty gene directly. Think of this as a "cut-and-paste" for your DNA.

**GENE SILENCING (SWITCH OFF):** This technique targets the DNA instructions that cause a faulty gene to make harmful proteins, reducing or stopping their production.

**Different approaches: replacing, repairing, or switching off faulty genes.**

## RISKS AND SIDE EFFECTS

**As with any treatment, gene therapy has possible risks or side effects. These may include:**

**Immune responses** (such as chills, fever, headache, low blood pressure, nausea, or vomiting) can occur if the body reacts to the viral vector. Sometimes, the immune system sees this virus as an invader, even though it is not harmful, and attempts to attack it, which causes these side effects.

**Unintended effects** if the new gene accidentally interacts with other genes or damages healthy cells or organs.

**A risk of infection**, especially for patients with weak immune systems.

**Unknown long-term effects** – more data on other effects is being studied in ongoing clinical trials.

## BENEFITS

**Possible benefits of gene therapy for cardiovascular disease may include:**

- ♡ One-time treatment
- ♡ Correct the root cause of disease
- ♡ Improve heart function
- ♡ Improve quality of life

**LIKE ANY TREATMENT, GENE THERAPY HAS** *benefits* **AND** *risks*

## LOOKING AHEAD

Research into gene therapy in cardiology is ongoing. Because this field is still developing, talk to your healthcare team to learn more about:

- **Clinical trials that might be relevant to your condition**
- **Existing or newly approved treatments in your country**
- **Reliable resources to help you stay informed**
- **Patient networks that can help you navigate your journey with cardiovascular disease**

**Gene therapy research is ongoing in cardiovascular disease, and your healthcare team is the best source for updates relevant to your condition.**

## QUESTIONS TO ASK YOUR DOCTOR

- Am I eligible for any ongoing gene therapy trials for my condition?
- What are the potential benefits and risks of these treatments?
- What is the mode of delivery of this therapy?
- Will I need immunosuppression?
- How would gene therapy or a clinical trial affect my daily life?
- Are there long-term follow-ups or monitoring required after treatment?
- How can I stay updated on new therapies or approvals in my country?
- Should my family members consider genetic testing as well?
- Are there patient support resources for individuals undergoing gene therapy or taking part in clinical trials?

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