**Mini Review**
Volume 16:12, 2022

Journal of Molecular and Genetic Medicine

ISSN: 1747-0862 Open Access

# Genetic Screening for Inherited Familial Hypercholesterolemia in Cardiovascular Organ Transplants

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## Abstract

Familial hypercholesterolemia (FH) is an inherited condition that causes high levels of low-density lipoprotein cholesterol (LDL-C) in the blood. The condition is caused by mutations in genes responsible for regulating the metabolism of cholesterol in the liver. FH affects approximately 1 in 200 people worldwide, and is associated with a higher risk of premature cardiovascular disease (CVD), such as heart attacks and strokes. Heart transplantation is a life-saving procedure for patients with severe heart disease, but it is not without its risks. In particular, heart transplant recipients are at an increased risk for CVD, including accelerated atherosclerosis, which can lead to transplant failure and death. FH is a significant risk factor for accelerated atherosclerosis in heart transplant recipients, and managing cholesterol levels in these patients is critical to their long-term outcomes.

**Keywords:** Hypercholesterolemia • Genetic Screening • Heart transplantation

## Introduction

Familial hypercholesterolemia (FH) is an inherited condition that causes high levels of low-density lipoprotein cholesterol (LDL-C) in the blood. The condition is caused by mutations in genes responsible for regulating the metabolism of cholesterol in the liver. FH affects approximately 1 in 200 people worldwide, and is associated with a higher risk of premature cardiovascular disease (CVD), such as heart attacks and strokes. Heart transplantation is a life-saving procedure for patients with severe heart disease, but it is not without its risks. In particular, heart transplant recipients are at an increased risk for CVD, including accelerated atherosclerosis, which can lead to transplant failure and death. FH is a significant risk factor for accelerated atherosclerosis in heart transplant recipients, and managing cholesterol levels in these patients is critical to their long-term outcomes.

FH is inherited in an autosomal dominant pattern, which means that a person only needs to inherit one copy of the mutated gene from either parent to develop the condition. This results in high levels of LDL cholesterol from birth, which can lead to premature CVD if left untreated. In heart transplant recipients with FH, the risk of accelerated atherosclerosis is even higher than in the general population. This is because the transplant process itself can trigger an immune response that increases inflammation and promotes the development of atherosclerotic plaques. In addition, the use of immunosuppressive medications to prevent rejection of the transplanted heart can also increase cholesterol levels and contribute to the development of atherosclerosis.

## Literature Review

FH is caused by a mutation in one of three genes: the LDL receptor (LDLR), apolipoprotein B (APOB), or proprotein convertase subtilisin/kexin type 9 (PCSK9). These genes are involved in the metabolism of LDL cholesterol, which is the "bad" cholesterol that contributes to the buildup of plaque in the arteries. Heart transplant patients are a high-risk group for CVD, as they have undergone a major surgical procedure and often have underlying cardiovascular disease. Opportunistic genetic screening for FH in heart transplant patients could help identify individuals with the condition who may be at increased risk of CVD and who could benefit from early intervention [1-3].

This essay will explore the benefits and challenges of opportunistic genetic screening for FH in heart transplant patients, and discuss the ethical considerations that must be taken into account when implementing such a screening program.

### Opportunistic genetic screening for FH in heart transplant patients

Heart transplant patients are a high-risk population for developing cardiovascular disease due to their underlying cardiac disease, the surgical intervention, and the immunosuppressive therapy they receive after transplant. Opportunistic genetic screening for FH in heart transplant patients may help identify individuals who are at increased risk of developing cardiovascular disease and enable early intervention to prevent adverse outcomes.

### Benefits of opportunistic genetic screening

One of the main benefits of opportunistic genetic screening for FH in heart transplant patients is the early identification of individuals with the condition. Early identification can allow for early intervention, such as lifestyle modifications and medication, which can help prevent or delay the onset of CVD. In addition, identifying individuals with FH can also allow for screening of family members, who may also be at risk of the condition. Early identification and intervention in family members can also help prevent or delay the onset of CVD [4,5]. Opportunistic genetic screening for FH in heart transplant patients can also provide valuable information for clinicians in terms of treatment planning. Individuals with FH may require more aggressive lipid-lowering therapy, and this information can help clinicians tailor treatment plans to individual patients.

## Discussion

### FH and cardiovascular disease

FH is a common genetic disorder that affects approximately 1 in 200-500 individuals worldwide. Individuals with FH have a 20-fold increased risk of

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**Received:** 01 December, 2022, Manuscript No: jmgm-23-92277; **Editor Assigned:** 03 December, 2022, Pre-QC No. P-92277; **Reviewed:** 15 December, 2022, QC No. Q-92277; **Revised:** 20 December, 2022, Manuscript No: R-92277; **Published:** 27 December, 2022, DOI: 10.37421/1747-0862.2022.16.587

developing cardiovascular disease compared to the general population. The risk of developing cardiovascular disease is particularly high in individuals who have already undergone cardiac surgery, such as heart transplant patients, as they have additional risk factors, such as immunosuppressive therapy, that increase their risk of developing cardiovascular disease. FH is diagnosed using a combination of clinical criteria and genetic testing [6,7].

Clinical criteria include family history of premature cardiovascular disease, such as myocardial infarction or stroke, and high levels of LDL cholesterol in the blood. Genetic testing can confirm the diagnosis by identifying mutations in the LDL receptor gene. Treatment of FH involves aggressive lipid-lowering therapy, such as statins, to reduce LDL cholesterol levels and prevent the development of cardiovascular disease. Lifestyle modifications, such as dietary changes and increased physical activity, are also recommended.

### Challenges of opportunistic genetic screening

One of the main challenges of opportunistic genetic screening for FH in heart transplant patients is the potential for psychological harm. Receiving a diagnosis of FH can be distressing, and individuals may experience anxiety or depression as a result. In addition, there may be concerns about stigmatization, particularly if the individual's genetic information is shared with others, such as employers or insurers. Another challenge is the potential for overdiagnosis and overtreatment. Not all individuals with FH will develop CVD, and there is a risk of unnecessary medication and lifestyle modifications for those who are identified as having the condition but may not actually develop CVD.

### Ethical considerations

Opportunistic genetic screening for FH in heart transplant patients raises a number of ethical considerations that must be taken into account when implementing such a screening program. One consideration is the need for informed consent. Individuals must be fully informed about the potential benefits and risks of genetic screening, as well as the implications of receiving a positive diagnosis. This includes the potential for psychological harm, the possibility of overdiagnosis and overtreatment, and the risk of stigmatization.

Another consideration is the potential for discrimination. Genetic information is sensitive and can be used to discriminate against individuals, such as in employment or insurance decisions. It is important to ensure that individuals' genetic information is protected and that appropriate measures are in place to prevent discrimination. A third consideration is the importance of confidentiality. Genetic information is personal and sensitive, and individuals have a right to privacy with respect to their genetic information. It is important to ensure that appropriate measures are in place to protect individuals' genetic information and that only authorized individuals have access to this information.

## Conclusion

Opportunistic genetic screening for FH in heart transplant patients has the potential to identify individuals with the condition who may be at increased risk of CVD and who could benefit from early intervention. However, there are also potential challenges, including the risk of psychological harm, over diagnosis, and overtreatment. Ethical considerations must also be taken into account, including the need for informed consent, the potential.

## Acknowledgement

None.

## Conflict of Interest

Authors declare no conflict of interest.

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**How to cite this article:** Ritchie, Krista. “Genetic Screening for Inherited Familial Hypercholesterolemia in Cardiovascular Organ Transplants.” J Mol Genet Med 16 (2022): 587.