# Special Issue

# Familial Hypercholesterolemia: Genetics and Emerging Therapies

## Message from the Guest Editor

Familial hypercholesterolemia (FH) is an inherited autosomal dominant metabolic disorder characterized by lifelong exposure to highly elevated cholesterol levels, with an estimated prevalence of 1 in 200–400 people. Those with FH carry a significantly higher risk of premature coronary artery disease (CAD). However, early diagnosis and initiation of optimal therapeutic strategies may normalize life expectancy. The most common variants involve mutations of the low-density lipoprotein receptor (*LDLR)* gene, followed by mutations of the apolipoprotein B-100 (*APOB*) and proprotein convertase subtilisin/kexin type 9 (*PCSK9*) genes. Genetic testing leads to improved FH diagnosis, improved adherence to treatment, improved LDL and total cholesterol levels, accessibility to genetic counseling services, etc. Our aim is to give an overview of the current status of FH genetic testing and its potential future applications, as well as challenges and pitfalls. We call for reviews on the current technologies, such as targeted next-generation sequencing, current state and the potential clinical utility of genetic testing for FH; as well as original research articles.

## Guest Editor

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## Deadline for manuscript submissions

closed (15 May 2022)

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Genes are central to our understanding of biology, and modern advances such as genomics and genome editing have maintained genetics as a vibrant, diverse and fastmoving field. There is a need for good quality, open access journals in this area, and the *Genes* team aims to provide expert manuscript handling, serious peer review, and rapid publication across the whole discipline of genetics. Starting in 2010, the journal is now well established and recognised.
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## Editor-in-Chief

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