BioMed Research International

Special Issue on
Familial Hypercholesterolemia

CALL FOR PAPERS

Familial Hypercholesterolemia (FH) is one of the most common inherited conditions affecting humans and consists in an impaired LDL metabolism, leading to a life-long elevation in LDL-cholesterol (LDL-C) and development of premature atherosclerotic cardiovascular disease.

FH is caused by mutations in one of three genes involved in LDL metabolism: (1) *LDLR*, encoding the Low-Density Lipoprotein Receptor; (2) *APOB*, encoding apolipoprotein B, the predominant protein component of the LDL particle; and (3) PCSK9, the gene coding for Proprotein Convertase Subtilisin-Kexin 9, a protein involved in the degradation of the LDLR.

Estimates indicate that as many as 1 in 250-500 people of all ethnicities may have FH.

FH can be considered as a reference model for inherited disorders since it is a common disease, it has well-established genetic bases; its clinical diagnosis is relatively simple and can be treated with cholesterol lowering drugs like statins and newly approved anti-PCSK9 antibody. Patients can be provided with a laboratory and clinical assessment and a genetic diagnosis, an appropriate therapy can be established, and the clinical targets are monitored. Moreover, novel treatments start to be available for this condition that could be useful especially for complex patients.

Despite the high prevalence and potential health impact of FH, it is often not identified as the cause of high cholesterol levels or major coronary events by primary care providers or cardiologists. Less than 10% of individuals with FH have been properly diagnosed worldwide, although it is difficult to estimate the exact degree of underdiagnosis, due to gaps in screening, recognition, and classification of FH. Untreated FH patients have 20 times the risk of developing coronary artery disease compared with the general population so FH has to be considered as a major concern to public health.

This special issue is intended to present and discuss arguments related to FH, a common but underdiagnosed and often poorly managed genetic disorder associated with greatly increased mortality from coronary artery disease. Presentation of data and dissemination of knowledge about this condition could help in reducing the burden of young, undiagnosed FH patients needing treatment for premature myocardial infarction.

Potential topics include, but are not limited to:

- Identification and reporting of mutations causing FH
- Use of next generation sequencing (NGS) for the identification of FH mutations
- Application of genetics screening within high risk populations
- Evaluation of mutation pathogenicity (i.e., functional assays and bioinformatics predictions)
- Evaluation of the role of common variants to FH development (polygenic hypothesis)
- Identification of other genetic mechanisms leading to FH (i.e., epigenetics, miRNAs, splicing regulation, and regulation of gene expression)
- Correlations between phenotype and genetic background
- New or underappreciated phenotypic consequences of FH
- Programmes of lipid screening among adult and pediatric populations and model of care for FH people
- Construction of regional and national registries for FH patients and families
- Strategies for prevention of cardiovascular disease in FH patients and families
- Issues related to therapy in adults and children, especially for complex patients with attention to the genetic status (i.e., FH Homozygotes)
- Use of novel therapeutic agents (i.e., Ab anti-PCSK9) for the treatment of FH

Authors can submit their manuscripts via the Manuscript Tracking System at http://mts.hindawi.com/submit/journals/bmri/genetics/lemia/.

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