

Master's thesis topic:

**Accreditation of genetic testing for Familial Hypercholesterolemia according to ISO 15189**

Familial hypercholesterolaemia (FH) is a common genetic disorder of lipid metabolism characterized by substantially increased plasma concentrations of low-density lipoprotein cholesterol (LDL-C) since birth, leading to premature atherosclerosis. FH is associated with an increased risk of premature coronary heart disease, however the early diagnosis of FH and the lowering of LDL-C values over the lifespan can reduce that risk and offer health and socioeconomic gains. Incorporating the FH genetic testing as the standard care for patients with clinical diagnosis of FH, as well for their at-risk relatives, will improve the clinical outcomes of these patients. This way, healthcare providers play a crucial role in identifying FH patients and providing appropriate management, but the clinical laboratories play the fundamental role in identifying the pathogenic or probably pathogenic variants affecting these individuals, and for the trust in these results there is a standard that must be used: the ISO 15189. This international standard is critical for ensuring the accuracy and reliability of laboratory test results, which are used for patient diagnosis and treatment, and the laboratories that meet these standards are recognized for their ability to produce consistent, accurate, and trustworthy results.

Instituto Nacional de Saúde Dr. Ricardo Jorge, through Departamento de Promoção da Saúde e Prevenção de Doenças Não Transmissíveis (DPS), provides genetic testing services for patients with clinical diagnosis of FH and is currently adding this test to its ISO 15189 accredited list. For this, we offer a master's thesis topic, in which the master's student, at the DPS, in the Cardiovascular Research Group, will identify and manage the research and work already developed, develop necessary documentation, and, in contact with the Quality Manager, implement this genetic test in the Department's Quality Management System. At the end of the project, it is expected that the master's student will develop skills in the area of quality management in clinical laboratories, namely in the cardiovascular area, skills that are highly sought after in the labor market.

**Bibliography:**

Bourbon e Rato 2006

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Mariano, C.; Alves, A.C.; Medeiros, A.M.; Chora, J.R.; Antunes, M.; Futema, M.; Humphries, S.E.; Bourbon, M. "The familial hypercholesterolaemia phenotype: Monogenic familial hypercholesterolaemia, polygenic hypercholesterolaemia and other causes". *Clinical Genetics* 97 3 (2020): 457-466. 10.1111/cge.13697.

ISO 15189:2012 Medical laboratories - Requirements for quality and competence. International Organization for Standardization.

**Thesis supervisor:** Paulo Dario (PhD), Departamento de Promoção da Saúde e Prevenção de Doenças Não Transmissíveis, Unidade de Investigação e Desenvolvimento, INSA.

paulo.dario@insa.min-saude.pt

**Thesis co-supervisor:** Mafalda Bourbon (PhD), Departamento de Química e Bioquímica,  
Faculdade der Ciências da Universidade de Lisboa.