BioISI Research Seminar

Functional genomics of Familial hypercholesterolaemia

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Familial hypercholesterolaemia (FH) is an autosomal genetic disorder of lipid metabolism and the most common monogenic disorder that confers increased cardiovascular risk. Patients with FH present very high LDL levels (>190 mg/dL), a family history of severe hypercholesterolaemia and in several cases a family or personal history of coronary heart disease (CHD). The majority of FH patients present a variant in LDL receptor gene (LDLR) and less than 10% present a variant in APOB or PCSK9 genes. More. About 1900 variants have been described in LDLR but less than 20% have been proved to be cause of disease. In the scope of the Portuguese FH Study, since 1999, about 120 different variants have been found in LDLR of more that 700 index cases studied. From these, 44 are exclusive of the Portuguese population, the rest have been described worldwide. Our lab has been developing in vitro assays to characterize all Portuguese mutations. Assays to access small deletions and missense variants including splicing variants in LDLR variants have been developed as well as for APOB and PCSK9 variants. From the 120 variants, 29 are large rearrangements, nonsense or frameshift mutations that are considered deleterious, a total of 64 variants have been characterized by our group or others, and we are currently developing assays to characterize the remaining. The correct evaluation of each variant enables a correct diagnosis and better cardiovascular risk stratification with the implementation of a more personalized treatment.

Host: **Astrid Moura Vicente**
BioISI BTR

When: 7 April 2016, 12h00
Where: Building C8, Room 8.2.30

Faculdade de Ciências da Universidade de Lisboa -Campo Grande, Lisboa (Portugal)

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