SERVICE FEES

(valid from 2020-01-01)

Familial Hypercholesterolemia (FH) analysis

The MAF's FH genetic analysis involves two phases: The first entails the use of a mutation analysis panel using Agena MassARRAY system and iPLEX Gold chemistry (accredited according to ISO/IEC 17025 by SWEDAC). Samples for which no mutations are found are analyzed using Devyser's FH v2 amplicon sequencing methodology using Illumina MiSeq instrument. Findings from the latter analysis, which indicate the presence of a pathogenic variation, are confirmed using Sanger sequencing according to ISO/IEC 17025 accreditation.

The Karolinska FH_FM1 panel investigates 113 known mutations and deletions in the three most important FH genes, *LDLR, PCSK9* and *APOB* using the Agena MassARRAY system and iPLEX Gold chemistry.

Devyser's FH kit enables analysis of sequence variants associated with Autosomal Dominant Hypercholesterolemia.

FH analysis	Price (SEK)
Sample handling and genotyping with the Karolinska FH_FM1 panel	3200 SEK/sample
Above and Devyser directed sequencing (*LDLR, PCSK9* and exon 26-29 of *APOB*) in case of negative outcome on the panel	5700 SEK/sample
Carrier diagnostics: panel or Sanger sequencing	2700 SEK/sample

Concluding remarks