Mutation Analysis Facility

Newsletter Summer 2022

The purpose and mission of the Mutation Analysis core Facility (MAF) is to help scientists and clinicians at academic institutions and hospitalsto perform genetic, epigenetic and genetic-epidemiological research and clinical testing in order to identify and assess genetic factors involved in different disease processes. Our services encompass the platforms Agena MassARRAY, Illumina iScan and MiSeq.

Familial hypercholesterolaemia (FH) genetic analysis

Peter Benedek successfully defended his thesis: ”Familial hypercholesterolemia in Sweden: Genetic and clinical studies” in December 2021. MAF has largely contributed to the development of the FH genotyping panel and the directed sequencing used in the thesis. Peter is now working as our FH specialist physician.

The described method is implemented at MAF and we perform an estimated 70% of all FH genetic analysis in Sweden. For more information: https://www.maf.ki.se/familial-hypercholesterolemia-fh-analysis/

Agena Bioscience news

In September 2021 Agena Bioscience joined Mesa Laboratories.
Agena increased their oncology portfolio with the ClearSEEK™ PIK3CA Panel that enables highly sensitive detection of PIK3CA variants in breast cancer specimens. This panel targets 20 variants in PIK3CA associated with response to targeted therapy, including the activating mutations recommended by the National Comprehensive Cancer Network Guidelines. More information on all applications: https://www.agenabio.com/applications/

Sequencing news

MAF has implemented an **HLA typing** method from CareDX, AlloSeq Tx17, with very high resolution of 17 loci. This innovative NGS HLA typing solution uses Hybrid Capture Technology, eliminating inefficiencies and limitations of traditional Long-Range PCR methods.

We perform **amplicon sequencing** projects using ready-made and validated kits from different vendors. Please contact us for more information.

HPV proficiency study

MAF participated in the 2021 Global HPV Proficiency Study using the Agena methodology. MAFs dataset was proficient for detection of 14 subtypes with no false positives. The assay is commercially available from Agena.

Illumina array news

Global Clinical Research Array (GCRA) and Global Screening Array (GSAv4) utilizes the new, smaller bead type called Excalibur. This advancement translates into either twice the number of samples per bead chip or twice the SNP density per array. GCRA contains 1.3M SNPs, 24 samples per chip; GSAv4 contains 850K SNPs, 48 samples per chip. Both arrays are expanded versions of the previous GSAv3 (650K SNPs), contain expanded GWAS backbones to improve genome coverage, and have the option to include a comprehensive PGx content. Both arrays will be available in Autumn 2022. In 2023 the Excalibur array for methylation screening will be released, designed for ulta-high troughput, low cost and large scale EWAS studies or biobank applications. Browse all available kits: https://emea.illumina.com/products/by-type/microarray-kits.html

Accreditation and quality assurance

MAF was subject to a SWEDAC audit in February and passed with flying colors. The accreditations encompass the organization and the Agena genotyping method

Personnel news

Päivi Kiviluoma has left MAF for a new position at Karolinska University Hospital. We wish her good luck!

Summer at MAF

MAF will be closed week 29-31. Only FH samples will be accepted during this time. Please e-mail maf@biosci.ki.se to reach available staff.

MAF homepage and contact

Please visit our for more information and current news: www.maf.ki.se. For more information concerning the applications in the newsletter, please contact Kristina Duvefelt +46 8 585 83693, Kristina.Duvefelt@ki.se.