

The Technology and Innovation Unit of the National Institute of Health: A sequencing and bioinformatics core facility specializing in public health genomics

Authors: Catarina Silva^{1,2}, Daniel Sampaio¹, Joana Mendonça¹, Dina Carpinteiro^{1,2}, Sílvia Duarte¹, Paula Barreiro¹, Joana Isidro^{1,3}, Miguel Machado¹, Luís Vieira^{1,2}

Affiliation: ¹Technology and Innovation Unit, Department of Human Genetics, National Institute of Health, Lisbon, Portugal; ²Centre for Toxicogenomics and Human Health (ToxOmics), Genetics, Oncology and Human Toxicology, NOVA Medical School, FCM, UNL, Lisbon, Portugal; ³Bioinformatics Unit, Department of Infectious Diseases, National Institute of Health, Lisbon, Portugal.

The National Institute of Health (INSA) is the state laboratory in the health sector. INSA has a long tradition in investigating the molecular etiology of genetic and complex diseases and in the identification of pathogenic organisms responsible for disease outbreaks and environmental imbalances. These activities benefit greatly from the existing centralized sequencing services provided by the Technology and Innovation Unit (UTI). Its mission includes performing sequencing and genotyping assays in the framework of research, diagnosis and epidemiological surveillance, as well as implementing data analysis pipelines for the study of variation in human genes. The equipment portfolio includes two next-generation sequencers and two capillary electrophoresis instruments for Sanger sequencing/fragment analysis, that altogether process an average of 36.000 samples/year. The team performed over 300 next-generation sequencing workflows for small genomes, amplicons, gene panels, clinical exome, *16S rRNA* gene and RNA/microRNAs. Standard of operation procedures are conducted by trained technicians under a quality control system that includes external quality assessment and ISO 15189 accreditation. UTI plays a key role in public health genomics, providing state-of-the-art equipment, centralized resources, technical expertise and short response times for public health problems (This work was supported by Centre for Toxicogenomics and Human Health - UID/BIM/00009/2019 - and GenomePT project – POCI-01-0145-FEDER-022184).