**EXP. CRG06-16**

**PLIEGO DE PRESCRIPCIONES TÉCNICAS**
**CONTRATACIÓN ARMONIZADA**

# SUMINISTRO DE CONSUMIBLES DE LABORATORIO CUSTOMIZADOS PARA LA CAPTURA DE REGIONES GENÓMICAS PARA LA UNIDAD CENTRO NACIONAL DE ANÁLISIS GENÓMICO DEL CENTRO DE REGULACIÓN GENÓMICA (CNAG-CRG)

Introduction

CNAG-CRG is a partner in the B-CAST project (Breast Cancer Stratification), an H2020 EU project, coordinated by the Netherlands Cancer Institute Antoni Van Leeuwenhoek and that runs through September 2020. The project targets to assess risk and prognosis modelling of breast cancer. One of the aims of the project is generating new genomic information based on a highly targeted sequencing approach by analysing genetic variation in specific genomic regions on a subset of ~11,000 tumour and paired germline DNA samples. CNAG-CRG is interested in the delivery of Highly Multiplexed Custom Capture for Targeted next-generation DNA sequencing.

Technical specifications

The bid is open for a cost-effective, high-throughput, targeted genomic solution of library preparation, custom designed for a pre-selected panel of genomic intervals to efficiently explore (somatic) genetic variation within the framework of the BCAST project. Specifically, we are interested in sequencing 107 genes, exons and regions suspected to be associated with the breast cancer. The selected targets cover ~550kb of human genome cumulative sequence. All the tumour samples (11.000) will be derived from FFPE (Formalin Fixed Paraffin Embedded) material and the paired germline DNA samples (11.000) will originate from whole blood, fresh frozen source.

The product must fulfil the following requirements:

- The product is a custom design of a panel with detailed description of the theoretical coverage guaranteed. The proposal must exemplary demonstrate robustness in specificity and uniformity in particular for the FFPE samples. The product must allow to be used with low input amounts (down to 10 ng). A comprehensive solution must be in particular given for processing the FFPE extracted material with fragment sizes below 300bp and anticipation of FFPE DNA related biases, such as resolution of cytosine deamination.

The protocol must be simple and with a limited number of steps in order to facilitate the processing of a very large number of samples. The protocol should allow processing at least 300 reactions in 2 working days by one FTE.

- The method must have an Indexing scheme supporting up to 96 samples per sequencing lane and must include unique molecular identifiers (UMIs) allowing to uniquely identify copies derived from each molecule.
- The product must be compatible with sequencing on Illumina HiSeq4000 and with data analysis applications available on Illumina's BaseSpace cloud computing platform.

- The proposal must include an established protocol for automated sample preparation on Perkin Elmer Sciclone and Zephyr robots. Methods that require purchase or lease of additional equipment (6 PCR thermocyclers available) or incompatibility with automation already installed (Perkin Elmer Sciclone G3 and Zephyr NGS) will not be considered.
- The product must be capable to detect germline and somatic (down to 5%) single nucleotide variations and insertion/deletions, and must have the potential to call copy number variants.
- The product must cover more than 95% of the uniquely mappable target area with a minimum coverage equal to 20% of the mean coverage over the entire uniquely mappable target area. The uniquely mappable target area is defined as the part of the target area that is mappable with a MAPQ >= 20 using the requested capture and sequencing technology.
- Suggested method should not have any previously described GC bias or risk of missing genetic variation due to lack of coverage and location in the vicinity of the restriction-enzyme digestion site, causing also bias towards wild-type base calls.
- The proposal must include a fully integrated DNA-to-data solution with automated data analysis that allows to easily review project data, including variant and mutation detected and threshold determination, across multiple runs.
- The proposal must include dedicated assistance of an expert for the kit design, with direct contact and full assay optimization support is also requested for the successful solution.

Barcelona, July 29, 2016.

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