centre nacional d'anàlisi genòmica
centro nacional de análisis genómico

cnag

2013 CNAG CALL: 300 EXOMES TO ELUCIDATE RARE DISEASES

Description

The Centro Nacional de Análisis Genómico (CNAG) is a genomic research platform with the mission to carry out large scale projects in DNA sequencing and analysis in collaboration with researchers from Catalonia, Spain and from the international research community in order to ensure the competiveness of our country in the strategic area of genomics.

The CNAG is offering a unique opportunity to carry out Whole Exome Sequencing and Bioinformatic Analysis for disease gene identification in rare disorders using Whole Exome Capture and Illumina HiSeq 2000/2500 sequencing (WES). The program will provide free-of-charge Whole Exome Sequencing and Bioinformatic analysis to 20-30 selected projects (indicative 5-30 samples per project).

Objective

The goal of this program is to promote the utilization of cutting-edge next-generation sequencing technology for the identification of genes involved in rare disorders with a clear genetic basis and to provide support for the Spanish research community. The results will be published as a collaborative effort between the applicant's group and the supporting investigators.

Eligibility

1. Researchers working at Spanish universities, public research institutes and hospitals.
2. Applicants must hold a PhD or MD degree and a work contract with the host institution at the time of application.
3. Applicants must have their own research lines and experience in leading research groups.
4. Applicants MUST NOT hold awards directly related to Whole Exome Sequencing in the proposed group of disorders.

Application Process:

Download and complete the **Submission form** and save it as pdf with the following title: Name and Surname of applicant_CNAG2013_Form. Then send it to info_cnag@pcb.ub.cat with "2013 CNAG CALL: 300 EXOMES TO ELUCIDATE RARE DISEASES" in subject line.

Important: If working with extended families, please enclose a separate pdf file (document title: Name and Surname of applicant_ CNAG2013_Pedigree) with the pedigree structure indicating available DNA samples and with appropriate legends.

One applicant can submit only one project proposal to this call. Applications should be written in English to allow international review. The **submission deadline** is 15/01/2014 (23:59, GMT+1 hour).

Internet Explorer is the recommended browser to download the submission form, however if you have any trouble at this step, please send an email to info_cnag@pcb.ub.cat and you will be sent the editable pdf submission form as an email attachment.

Review Process

A selection committee comprised of CNAG and CIBERER members, as well as external referees, will review the proposals and select the award recipients within 4 weeks of submission deadline. The selection results will be communicated by mail to the principal investigator. The title of the selected projects and name of their principal investigator will be published on the web site of the CNAG (http://www.cnag.cat).

Projects will be chosen based on scientific quality and on the chances of success of the exome sequencing approach based on sample availability, pedigree structure, clinical phenotype, availability of additional information, etc.

DNA samples

All samples must have been obtained with the corresponding approval of the Bioethics Committee and signed "informed consent" from each donor, both for collection and for their use, including conservation and manipulation/sequencing by entities such as CNAG, and controlled access distribution in the databases mentioned below. Samples should be anonymized and preferably linked to diagnostic categories and Human Phenotype Ontology (HPO) terms.

A minimum of 6 ug of high quality DNA at around 200ng/ul is required for Whole Exome Sequencing. DNA samples that fulfill the quality requirements **MUST BE AVAILABLE** at time of project submission. All samples from one study will be processed as a single batch.

Databases

Exome Sequencing results will be deposited at the European Genome-to-phenome Archive (EGA, https://www.ebi.ac.uk/ega/), as well as in the CNAG internal database. Inclusion to additional databases such as RD-Connect (rd-connect.eu) will also be encouraged.

Acknowledgement Policy

The CNAG support and collaboration must at least be recognized in the acknowledgments section of any resulting publication.