



**Research Project Proposal**  
Academic year 2020-2021

***Máster en Investigación Biomédica***

<b>Project Nº 51</b>	
<b>Title: Clinical Utility of a Comprehensive Next-Generation Sequencing Panel for Hereditary Cancer Syndromes</b>	
<b>Department/ Laboratory: CIMA LAB Diagnostics. Laboratorio Tumores Sólidos y Enfermedades Hereditarias</b>	
<b>Director:</b> Gorka Alkorta-Aranburu <b>Contact:</b> galkorta@unav.es	
<p><b>Summary:</b> Pathogenic sequence changes in more than 90 hereditary cancer genes explain a small fraction of all diagnosed cancer. However, identifying high risk individuals whose cancer is caused by an inherited pathogenic sequence change is essential. This identification allows enhanced surveillance improving early detection and selection of the most adequate risk reduction measure on the patient as well as family members. This is because once the first individual in a family carrying a pathogenic sequence change is identified, the high or average risk status of each family member can be genetically assessed and appropriate care established. Until very recently, Sanger sequencing was used to screen one single gene at a time; but due to time and monetary constrains, most of the genes were not tested even in patients with strong family history of cancer. In addition, latest studies agree that lack of family history should not exclude patients with early onset or bilateral cancer from comprehensive genetic testing. Fortunately, over the last 4 years, with the introduction of next-generation sequencing (NGS), we have been able to screen 91 hereditary cancer genes over 600 patients with or without strong family history. Therefore, the goal of this research proposal is to evaluate the clinical utility of our NGS-based testing strategy in patients with or without family history of cancer while understanding the technical limitations realated with studying particular genes and identifying certain types of sequence changes with NGS.</p>	
yes	<input type="checkbox"/>
no	<input checked="" type="checkbox"/>
<b>Does the project include the possibility of supervised animal manipulation to complete the training for animal manipulator?</b>	