Technological Platform / Bergonié Institute

Name	**Molecular Genetic Lab & OncoGenetic Unit**	Bergonié UNICANCER NOUVELLE-AQUITAINE
Platform coordinator(s)	**Pr. Nicolas SEVENET, PU-PH & Dr. Michel LONGY, MD** Coordinator of the Molecular Genetic Lab of Bergonie & Coordinator of OncoGenetic Unit Vice-Dean, Pharmalogical Sciences, College of Health Sciences INSERM U1218 - Biopathology Department –Bergonié Institute N.Sevenet@bordeaux.unicancer.fr/ +33(0)5 56 33 04 41 website: https://www.bergonie.fr/diagnostic-et-traitements/laboratoire/	
Type of services	**Routine diagnostics & Research projects**	
Activities	As a medical discipline, Oncogenetics is focused on managing the familial risk of cancers. In fact, almost 5% of diagnosed cancers are linked to the presence of constitutional alterations of the genome. The Bergonié Institute has a well-recognized expertise in the genetic diagnosis of hereditary form of breast cancers and the ovarian cancers (*BRCA1*, *BRCA2*, *PALB2* & other HRD genes), of Lynch syndrome predisposing to colorectal cancers and of the endometrium (*MLH1*, *MSH2*, *MSH6* & *PMS2* genes), of Cowden disease (*PTEN* gene) and of Gorlin syndrome (*PTCH1* gene). **Evaluation of the individual predisposition to cancer** • **Clinical Evaluation** Oncogenetic consultations are carried out by MDs and genetic counselors, in patient displaying a greater familial aggregation of cancers as compared to the risk evaluated in the general population. At the end of this investigation, the risk individual can be assessed. • **Biological Confirmation** Genomic material is extracted from blood in most cases and for *BRCA* genes in a somatic context too, for theranostics. The genetic characterization of familial tumor aggregation is performed using capture techniques followed by sequencing by synthesis on Illumina pltaform. PCR amplification followed by sequencing techniques provides valuable insight in the patient genome and DNA fragment of interest. Those fragments are analyzed based on the presence of two types of alterations: point mutations (substitution, duplication, deletion) and large rearrangements (deletion or duplication of large gene fragments).	
Key technologies	• **Genomic material extraction from blood** • **NGS Sequencing techniques (capture protocol & SBS on Illumina platform) on constitutional and somatic genomic DNA** • **CGH array** • **Sanger sequencing technology** • **PCR/RT-PCR, splicing studies** • **Cell culture** • **Digital droplet PCR for mosaic syndrom**	
Please state no more than 5 facts illustrating the appeal or reputation of the platform	• Participation on national and international research projects in collaboration with academic structure and/or private companies • Participation in oncogenetic multidisciplinary board and oncogenetic tumor board sessions • Active participation in University Teaching: University diplomas in oncogenetic and genetic pathologies, pharmacology and medical sciences. • Center of reference for Medical student, PhDs Student training in oncogenetic. • Participation and intervention at national and international meetings	
Please state the platform's major publications	Leman et al, BMC Genomics 2020 Lichou F et al, Human Genomics 2019 Renaux-Petel et al, . J Med Genet 2018 Bubien V *et al* . Gene Chr Cancer 2017. Crivelli L, *et al* . Eur J Hum genet 2017. (within the last 5 years)	
People in the platform with their degree	PharmD : Nicolas SEVENET (PhD) / MD : Virginie BUBIEN (PhD), Emmanuelle BAROUK SIMONET, Claire SENECHAL DAVIN, Michel LONGY (PhD) / Genetic counselors: Eglantine JOLLY, Angela BADIN, Anais DUPRE / Biological Ingenior (PhD) : Natalie JONES, Francoise BONNET / Bioinformaticien: Mme Jennifer CHIRON / Technicians: Bernadette GASTALDELLO, Gaelle GENESTE, Delphine LAFON, Laurène DUFIN, Mélanie MULLER, Valerie DAPREMONT / Manager: Karine PEUTAT / Quality manager: Anulka KESTEMONT.	
Innovation technologic or research in the future 5 years	Long read NGS sequencing technology & Haplotype phasing HiC capture and sequencing for TAD exploration CGH by NGS (Oneseq) & exome NGS sequencing for dedicated patient Ultra-deep sequencing & digital droplet PCR for tissue mosaicism annotation in syndromic context	