A new protocol to quantify the alterations (copy number changes) in cancer samples. Chromosomal instability (CIN), a type of genomic instability, favours changes in chromosome number and structure and is associated with the progression and initiation of multiple diseases, including cancer. Therefore, CIN identification and analysis represent a helpful tool for cancer diagnosis and treatment. In 2021, we reported an optimised molecular cytogenetic protocol to detect CIN in formalin-fixed, paraffin-embedded mouse and human tissues, using fluorescent in situ hybridization to visualise and quantify chromosomal alterations such as amplifications, deletions, and translocations.

Technological and translational activities. Our Unit offers rapid, precise, and affordable technologies to analyse cancer cells at the chromosome level and to functionally interrogate the cancer genome. We provide state-of-the-art molecular cytogenetic and genome editing services. The Unit focuses on making available a complete repertoire of gene editing tools for cellular and genetic manipulation and an array of delivery vehicles, offering a flexible, modular platform for precision genome manipulation. The Unit provides molecular cytogenetic technologies for human and mouse chromosomes analysis, including conventional karyotyping, FISH, SKY and CGH array. In 2021, we carried out over 2,500 assays for experimental and clinically oriented projects.

We also participate in collaborative projects with clinical and basic science investigators across the CNIO and other institutions.

**HUMAN CANCER GENETICS PROGRAMME | MOLECULAR CYTOGENETICS UNIT**

**OVERVIEW**

Recurrent chromosomal rearrangements — changes in the structure of native chromosomes — are very common and well-known hallmarks of cancer. A better understanding of these cancer-causing mechanisms will lead to novel therapeutic regimens to fight cancer. The research activity of the Molecular Cytogenomics and Genome Editing Unit focuses on increasing the knowledge about the role of chromosomal rearrangements in cancer development and progression and discovering new therapeutic targets. With the combined use of CRISPR genome editing and cytogenetic technologies, we are creating models that recapitulate chromosomal and genetic cancer alterations. The goal of the Unit is to provide CNIO and external researchers that recapitulate chromosomal and genetic cancer alterations.

**MOLECULAR CYTOGENETICS UNIT**

Sandra Rodríguez-Perales Sh Head

Staff Scientist

Raf Torres

Graduate Student

Maria-Cruz Casado (since May), Pilar Puig

Technicians

M. Carmen Martín, Marta M. Carmen Martín, Marta

Students in Practice

Alessandro Alcains (since September)
(Maastricht University, The Netherlands), Alejandro Nieto (since November) (Universidad Autónoma de Madrid, Spain), Paula M. Spina (February-June) (Universidad de Alcalá de Henares, Spain), Beatriz Gómez (March-August) (Universidad Politécnica de Valencia, Spain)

**RESEARCH HIGHLIGHTS**

A new protocol to quantify the alterations (copy number changes) in cancer samples. Chromosomal instability (CIN), a type of genomic instability, favours changes in chromosome number and structure and is associated with the progression and initiation of multiple diseases, including cancer. Therefore, CIN identification and analysis represent a helpful tool for cancer diagnosis and treatment. In 2021, we reported an optimised molecular cytogenetic protocol to detect CIN in formalin-fixed, paraffin-embedded mouse and human tissues, using fluorescent in situ hybridization to visualise and quantify chromosomal alterations such as amplifications, deletions, and translocations.

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**PUBLICATIONS**

- Mano R; Rodríguez-Perales S, Torres-Ruíz R, Santacana C, Rodríguez-Philha SM (2021). PD-L1 expression in peripheral T-cell lymphomas is not related to either PD-L1 gene amplification or rearrangements, copy number variations, and CIN. Nat Commun 12, 6910.

**AWARDS AND RECOGNITIONS**

1st prize “Paper of the Year 2020-2021” contest in the gene therapy category for the article: in vivo CRISPR/Cas9-mediated fusion of oncogenes for selective elimination of cancer cells (Nature Communications, 2021, bestowed by The Spanish Society for Gene and Cell Therapy (SFCGTC)).

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