MARCOS DÍAZ GAY

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SUMMARY

I am a postdoctoral researcher in the group of Prof. Ludmil B. Alexandrov at the University of California, San Diego. Originally trained in engineering, I have focused my career on computational biology, with a strong interest in cancer genomics data analysis. My research focuses on developing new computational methodologies that allow the discovery of the underlying mechanisms of cancer predisposition and progression. Making this knowledge accessible to the research community and understanding the dynamics and evolution of human cancer are my main goals at the Alexandrov lab.

CURRENT POSITION

University of California, San Diego
Feb 2020 – present

POSTDOCTORAL SCHOLAR (Alexandrov lab)
Main project: "Mutational signatures dynamics in human cancers".

EXPERIENCE

August Pi i Sunyer Biomedical Research Institute
Aug 2016 – Nov 2019

PhD STUDENT (Genetic Predisposition to Gastrointestinal Cancer group)
PhD thesis: "Identification of new candidate genes for germline predisposition to familial colorectal cancer using somatic mutational profiling".

University of California, San Diego
Feb 2019 – May 2019

VISITING GRADUATE STUDENT (Alexandrov lab)
VGS project: "Advanced detection of mutational signatures in colorectal cancer".

August Pi i Sunyer Biomedical Research Institute
Nov 2015 – Jul 2016

MSc STUDENT (Genetic Predisposition to Gastrointestinal Cancer group)
MSc thesis: "Identification of novel genes involved in familial colorectal cancer: integrated analysis of germline and tumor DNA".

University of A Coruña
Dec 2013 – Jul 2015

RESEARCH ASSISTANT (Water and Environmental Engineering group)

EDUCATION

University of Barcelona
2016 – 2019

PhD | MEDICINE AND TRANSLATIONAL RESEARCH

University of Barcelona / Open University of Catalonia
2017 – present

MSc | BIOINFORMATICS AND BIOSTATISTICS
In progress (67 % completed).

University of Barcelona
2015 – 2016

MSc | BIOMEDICINE

University of A Coruña
2009 – 2015

BSc & MSc | CIVIL ENGINEERING

PUBLICATIONS

Total number of publications: 17 (1 preprint) | Citations: 237 (Google scholar), 154 (WOS) | h-index: 9 (Google scholar), 8 (WOS)

Original articles as first author

Uncovering novel mutational signatures by *de novo* extraction with SigProfilerExtractor
Islam S.M.A.*, **Díaz-Gay M.*** *et al. BioRxiv.* 2022:12.13.422570 (preprint).
*shared first authors

Using linkage studies combined with whole-exome sequencing to identify novel candidate genes for familial colorectal cancer
Toma C.*, **Díaz-Gay M.*** *et al. International Journal of Cancer*. 2020;146(6):1568–1577.

Identification of a novel candidate gene for serrated polyposis syndrome by performing linkage analysis combined with whole-exome sequencing
Toma C.*, **Díaz-Gay M.*** *et al. Clinical and Translational Gastroenterology*. 2019;10(10):e00100.

Integrated analysis of germline and tumor DNA identifies new candidate genes involved in familial colorectal cancer
Díaz-Gay M. *et al. Cancers*. 2019;11(3):362.

Mutational signatures in cancer (MuSiCa): a web application to implement mutational signatures analysis in cancer samples
Díaz-Gay M. *et al. BMC Bioinformatics*. 2018;19(1):224.

Reviews and book chapters as first author

Unraveling the genomic landscape of colorectal cancer through mutational signatures ***[book chapter]***
Díaz-Gay M. & Alexandrov L.B. *Advances in Cancer Research: "Novel Approaches to Colorectal Cancer"*. 2021;151:385-424.

Somatic mutational signatures in polyposis and colorectal cancer ***[review]***
Grolleman J.*, **Díaz-Gay M.*** *et al. Molecular Aspects of Medicine*. 2019;69:62–72.

Original articles in collaboration

Comprehensive genomic characterization of early-onset Lynch-like syndrome colorectal cancers
Golubicki M. *et al. Cancers*. 2021.;13(6):1259.

Germline and somatic WES identifies new candidate genes involved in familial predisposition to serrated polyposis syndrome
Soares de Lima Y. *et al. Cancers*. 2021;13(4):929.

Identification of new genes involved in germline predisposition to early-onset gastric cancer
Herrera-Pariente C. *et al. International Journal of Molecular Sciences*. 2021;22(3), 1310.

Germline biallelic mutations in *MCM8* are associated with early-onset lynch-like syndrome
Golubicki M. *et al. JCI Insight*. 2020;5(18):e140698.

Germline mutations in *FAF1* are associated with hereditary colorectal cancer
Bonjoch L. *et al. Gastroenterology*. 2020;159(1):227-240.

Colorectal cancer genetic variants are also associated with serrated polyposis syndrome susceptibility
Arnau-Collell C. *et al. Journal of Medical Genetics*. 2020;57(10):677-682.

CNApp, a tool for the quantification of copy number alterations and integrative analysis revealing clinical implications
Franch-Expósito S. *et al. eLife*. 2020;9:e50267.

Quantitative analysis of somatically-acquired and constitutive uniparental disomy in gastrointestinal cancers
Torabi K. *et al. International Journal of Cancer*. 2019;144(3):513–524.

Rare germline copy number variants in colorectal cancer predisposition characterized by exome sequencing analysis
Franch-Expósito S. *et al. Journal of Genetics and Genomics*. 2018;45(1):41–45.

***POLE* and *POLD1* screening in 155 patients with multiple polyps and early-onset colorectal cancer**
Esteban-Jurado C. *et al. Oncotarget*. 2017;8(16):26732–26743.

FUNDING, SCHOLARSHIPS & AWARDS

University of Barcelona Nov 2019	**CUM LAUDE HONORS (PhD)** Faculty of Medicine.
University of Barcelona Dec 2018	**SHORT-TERM INTERNATIONAL RESEARCH STAY FELLOWSHIP** Faculty of Medicine.
University of Barcelona Apr 2018	**EXTRAORDINARY MASTER'S DEGREE AWARD** Faculty of Medicine.
Regional Government of Catalunya Feb 2017	**PhD FELLOWSHIP FI-2017** Agency for Management of University and Research Grants (AGAUR).
University of A Coruña Jan 2016	**EXTRAORDINARY END-OF-DEGREE AWARD** School of Civil Engineering.

SOFTWARE

Co-author and maintainer of **MuSiCa R-Shiny web application** and **SigProfilerExtractor Python package** for mutational signature analysis. Co-author and curator of the **COSMIC Mutational Signatures** website.

REFERENCES

Dr. Ludmil B. Alexandrov, PhD Cellular and Molecular Medicine and Bioengineering departments, University of California, San Diego. Moores Cancer Center, 3855 Health Sciences Dr, 92093 La Jolla, CA (United States of America). l2alexandrov@ucsd.edu

Dr. Sergi Castellví-Bel, PhD Genetic Predisposition to Gastrointestinal Cancer group, August Pi i Sunyer Biomedical Research Institute (IDIBAPS), CIBERehd, Hospital Clínic. Rosselló 153, 08036 Barcelona (Spain). sbel@clinic.cat

Dr. Francesc Balaguer, MD, PhD Gastroenterology department, Hospital Clínic, CIBERehd, IDIBAPS. Villarroel 153, 08036 Barcelona (Spain). fprunes@clinic.cat